HELIXLINE

PERSONALIZED GENOMIC ANALYSIS

ANCESTRY REPORT



ORIGINS

NAME **Demo User** · SEX **Male** · ISSUED **October 2026**

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HELIXLINE

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⚠ FOR INFORMATIONAL USE ONLY · NOT A CLINICAL DIAGNOSTIC

Results are estimates. Personal findings appear with a confidence label that reflects how strongly the evidence supports them. Clearly separated research previews may also be shown, but are labelled and are not primary personal conclusions. As panels grow, results may shift - treat released findings as the best current estimate, not a final identity. · **Ancestry ≠ identity.** Genetic similarity does not determine state, community, caste, religion, language, or personal identity. · **Not a diagnostic tool.** Do not use for medical, legal, or insurance decisions. Consult a licensed clinician for health interpretation. · **Privacy:** your DNA is processed with explicit consent and never sold.

ANCESTRY AT A GLANCE

HELIXLINE

HOW TO READ THIS REPORT
Some findings come directly from your DNA, while others are estimates based on reference populations. **Research Preview** results are still developing. **Not Measured** means the available data could not support a dependable answer.

ACT I · HUMAN ORIGINS CONTEXT

Shared Out-of-Africa science - not a reconstruction of your personal migration route.

CONTEXT

ACT II · MEASURED ARCHAIC DNA

1.72%
Neanderthal estimate
SE ±0.33 points
Reference range 0.94-2.08%

Denisovan · below 2 SE
f4-ratio · 759,092 SNPs

PIGMENTATION MARKERS
38/41
markers read

ANCIENT REFERENCE SIMILARITY

How the tested groups rank

RANKED COMPARISON

A similarity test (f3) ranking how close your DNA sits to these ancient reference groups, using an unrelated African population (Mbuti) as a neutral yardstick - not ancestry percentages

1. Indus Periphery	0.07669
2. Steppe Middle/Late Bronze Age	0.07578
3. Iran Neolithic	0.07503
4. AASI-related proxy (Onge)	0.07410

Dot = f3, bar = ±1 SE; spacing is relative to these four only. **#1 and #2 overlap within 1 SE - their order is not resolved.**

ANCIENT MIXTURE MODEL

MODEL NOT REJECTED · RESEARCH PREVIEW

Sarazm-EN-related	49.6% ±7.1
AASI-related proxy	43.6% ±2.6
Steppe (MLBA)	6.8% ±6.3

qpAdm source-model coefficients; not literal ancestral fractions. Exact-vendor validation is still in progress.

POPULATION TIMING · PUBLISHED
~1,900-4,200 years ago
Population context - not your measured date.

ACT III · YOUR PRIMARY ANCESTRY RESULT

South Asian

HIGH CONFIDENCE

Called by our ancestry-detection model; this confidence covers only the broad-region call shown above. Tested against 1253 people with already-known ancestry, it got the right answer 99.76% of the time.

HOW CLEAN THE MATCH IS
91.8 / 100
How strongly your DNA fits the "South Asian" pattern in our 6-group model - not a percentage of ancestors.



91.8% South Asian model axis	3.7% West Asian trace / uncertain	2.0% European-labelled K=6 axis trace / uncertain	1.7% African trace / uncertain	0.5% Native-American-like axis trace / uncertain	0.2% East Asian trace / uncertain

ACT IV · BROAD REGIONAL SIGNAL

South India

MODERATE CONFIDENCE

South	62.4
West	36.7

Leads the broad-reference comparison by 25.7 points. Model scores, not genome percentages-not an exact state, community or caste determination. Validation 77.4% · n=31.

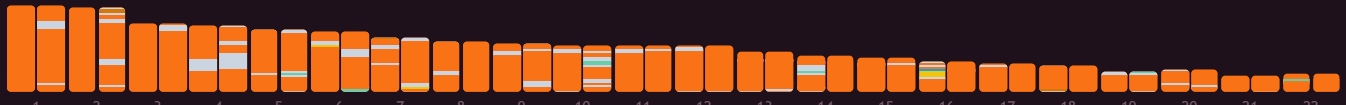
COMMUNITY · NOT INFERRED FROM DNA



States are shaded by which broad region they belong to. This is a broad-region result - no state-level prediction was made.

ACT V · THE MAKING OF YOU · DIRECT GENOME MEASURES

99.45%	2.82%	78th	22 / 22
CALL RATE	SHARED-ANCESTRY DNA (ROH)	F-ROH PERCENTILE	CHROMOSOME PAINTING · PREVIEW
616,214 positions assessed	36 runs ≥1 Mb - see Endogamy & ROH	against the South Asian reference cohort	89.7% posterior-passing · research only



Two statistical haplotypes per autosome; parent of origin is unknown and no tract boundary is exact. Bar height is proportional to chromosome length.

ACT VI · VII · YOUR TWO DIRECT LINEAGES

One direct maternal line and, where applicable, one direct paternal line-not your whole ancestry.

MATERNAL · mtDNA
M
Safe-clade placement · deeper branch provisional · 984 positions · 1/2 terminal markers

PATERNAL · Y-DNA
H1a1d2c1
94/100 defining positions derived · terminal marker observed

ACT VIII · YOUR EXTENDED FAMILY

Relative matching

COHORT SCREENED

No close relative crossed the reporting threshold among 127 consented samples. The report still shows the nearest pseudonymous comparison with its KING kinship value.

THE STORY OF YOU

"Every person alive today is part of a human story that began in Africa ~70,000 years ago. Across coasts and mountains, population movements into South Asia, followed by farmers, herders and later communities, shaped the genetic landscape you carry today."

YOUR HUMAN JOURNEY

300,000 YEARS AGO
Homo Sapiens Emerge
Anatomically modern humans appear in Africa - the origin of every lineage alive.

~70,000 YEARS AGO
Out of Africa
Population expansions from Africa contribute to the ancestry of people living outside Africa today.

~65,000 YEARS AGO
First Peoples Reach South Asia
Hunter-gatherers settle the subcontinent - an early layer in South Asian population history.

~50,000 YEARS AGO
Meeting Neanderthals & Denisovans
Population-level mixing with archaic humans left alleles that can still be measured today.

~9,000 YEARS AGO
Iranian-Related Farming Ancestry
Iranian-related ancestry becomes part of later South Asian populations - a reference label, not an ethnicity you belong to.

~4,000 YEARS AGO
Steppe-Related Ancestry & South Asian Structure
Steppe-related ancestry joins older South Asian layers; later populations sit along a broad ancestry spectrum, not one fixed group.

~2,000 YEARS AGO
South Asian Communities Take Shape
Long-standing population structure creates detectable group patterns, without making DNA equivalent to identity.

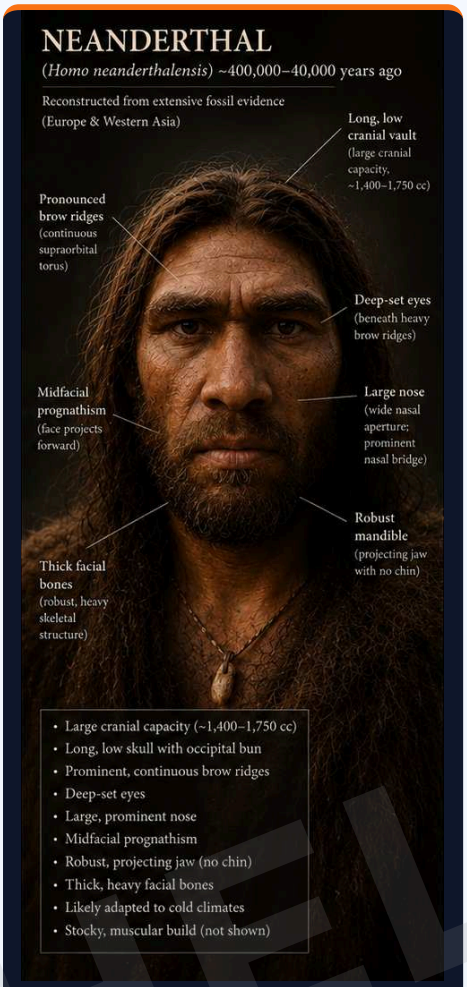
TODAY
YOU
Demo User
The newest branch of a 300,000-year human story.

HELIXLINE

THE HUMANS WHO SHAPED YOUR DNA

> *"Your ancestors didn't just live alongside other human species - they had children with them. 50,000 years later, those ancient encounters are still written in every cell of your body."*

ARCHAIC-HUMAN SIGNALS EVALUATED IN YOUR GENOME



NEANDERTHAL

Our closest known relative - lived until 40,000 years ago

1.7% f4-RATIO

MODERATE CONFIDENCE · ESTIMATE WITH JACKKNIFE SE

Measured directly against **real, sequenced Neanderthal genomes** (Altai, Vindija) across **759,092 DNA positions**, giving 1.72% ± 0.33 - run on your own DNA, not a population average or estimate, using the same peer-reviewed method that published Neanderthal-ancestry research relies on.



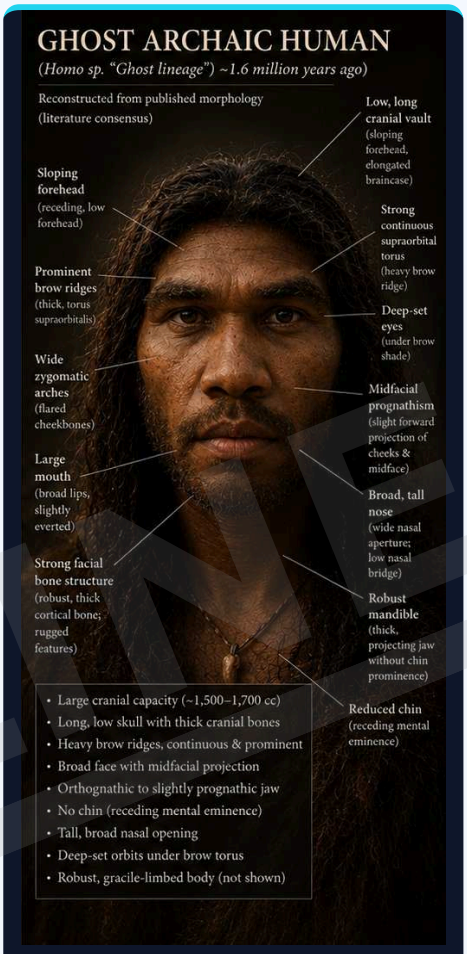
DENISOVAN

Discovered in 2010 from a single finger bone in a Siberian cave

BELOW REPORTING THRESHOLD

BELOW 2 SE · NOT A ZERO-ANCESTRY CLAIM

Your Denisovan result was too small to tell apart from zero (uncertainty ±0.41 percentage points) - most likely you carry very little or none, but the measurement isn't precise enough to fully rule out a small trace.



THE MYSTERY HUMAN

A population-level research hypothesis; no corresponding archaic genome has been sequenced

Research context not a personal result NOT QUANTIFIABLE

Population-level research has proposed deeply diverged ancestry in South Asian history, but no corresponding archaic genome has been sequenced. This is background context only and is not used for individual inference here.

ARE YOU ABOVE OR BELOW AVERAGE? - YOUR ANCIENT DNA VS THE WORLD

Neanderthal Heritage - Evolutionary Context

You	1.7%
South Asian average	1.5%
European average	1.7%
East Asian average	1.9%
African average	0.0%

Denisovan Heritage - Evolutionary Context

You	Below threshold
South Asian average	0.1%
East Asian average	0.2%
Papua New Guinea	4.8%
European average	0.05%

YOUR NEANDERTHAL REFERENCE AFFINITY

MODERATE CONFIDENCE · RANKING AMONG THREE TESTED REFERENCES ONLY

Altai Neanderthal (Denisova 5)
Denisova Cave, Altai Mountains · 91-130 kya
TESTED REFERENCE

Vindija 33.19 Neanderthal
Vindija Cave, Croatia · ~50 kya
STRONGEST RESOLVED AFFINITY

Chagyrskaya 8 Neanderthal
Chagyrskaya Cave, Altai Mountains · ~80 kya
TESTED REFERENCE

Of the high-coverage ancient Neanderthal genomes tested, your DNA looks most similar to Vindija 33.19 Neanderthal - meaning your Neanderthal ancestors were more closely related to that population than to the others tested. This is a similarity comparison, not descent from that individual or a DNA percentage. This comparison is limited to the three high-coverage genomes tested; it is not a direct-family relationship or an ancestry percentage.

THE TRAITS YOU INHERITED

HOW THESE VARIANTS SHAPE YOU TODAY

0 introgression-associated alleles observed · **2** associated loci genotyped ·

A single DNA marker isn't enough on its own to prove a full block of Neanderthal DNA at this spot - that takes several markers agreeing together.

COVID-19 severity haplotype (chr3p21, Neanderthal-introgressed)

NEAN. anc.

LZTFL1 rs35044562

ARCHAIC CC *(imputed)* Ancestral (non-risk) genotype - you do not carry this archaic variant.

Antiviral OAS1 p46 isoform (Neanderthal-introgressed in Eurasians)

NEAN. anc.

OAS1 rs10774671

ARCHAIC AA *(imputed)* Ancestral (non-risk) genotype - you do not carry this archaic variant.

Data from published South Asian selection scans (1000G SAS, HGDP). These marker associations are evolutionary observations, not clinical risk estimates. Zeberg & Paabo 2020 Nature 587:610

YOUR BROADER INHERITED MARKERS

ADULT LACTOSE DIGESTION

Common lactase non-persistence marker pattern

GG · DIRECT

This tests one major European-associated marker. Other South Asian lactase-persistence variants are not ruled out.

ALCOHOL FLUSH RESPONSE

Common alcohol-metabolism pattern at this marker

GG · DIRECT

The major East-Asian alcohol-flush-associated variant was not detected at this tested position.

MUSCLE-FIBRE PROTEIN

Alpha-actinin-3 deficiency-associated pattern

TT · DIRECT

This marker affects a fast-twitch muscle protein, but does not predict athletic ability, strength or endurance.

EARWAX AND UNDERARM ODOUR

Dry-earwax / reduced-odour-associated pattern

TT · DIRECT

A well-replicated single marker influences earwax type and average underarm odour production.

WHY EVOLUTION SHAPED YOUR DNA HERE

Three migration waves

Successive groups from Africa, the Middle East, and Central Asia each brought different variants - and selection picked the best of each for this environment.

Disease pressure

South Asia's warm climate drove strong selection for immunity against malaria, cholera, and parasites. Survivors passed resistance genes to you.

Diet shift

When agriculture arrived ~10,000 years ago, variants for plant-fat metabolism and lactose digestion gave a huge survival advantage.

Endogamy

Tight-knit communities amplified rare beneficial mutations quickly - leaving some of the strongest selection sweeps in the human genome.

HOW TO READ THESE RESULTS

Two evidence groups

The left column shows introgression-associated loci first, followed by broader modern adaptive markers that do not claim archaic origin.

Marker provenance

Each card says whether we read this DNA position directly off your genotyping chip, or predicted it statistically from nearby DNA (imputation) - both are reliable, but direct reads are the gold standard. A single marker is narrower than a complete phenotype model.

Published association

These loci have replicated biological associations, but an observed allele is not by itself a diagnosis or guaranteed visible trait.

Scope

Pigmentation duplicates and the diabetes-risk marker are kept on their dedicated appearance and health pages, not repeated here.

⚠ WHAT THIS DOES NOT MEAN

Carrying an archaic or adaptive allele tells you about your **ancestral history** - not your present-day health or fitness. Most selected variants gave a small survival advantage thousands of years ago; their effect in modern environments can be neutral or even maladaptive. These are a window into the past, not a verdict on the future.

ARCHAIC REFERENCE AFFINITY
ARCHAIC INTROGRESSION-ASSOCIATED LOCI
HIGH CONFIDENCE

EXPLORATORY
MODERATE CONFIDENCE
Neanderthal 1.72% ± 0.33 · Denisovan below detection / statistically consistent with zero

Vindija 33.19 Neanderthal
11-marker panel · all called

direct+imputed
direct
Direct + imputed · 759,092 markers

YOUR PIGMENTATION MARKER SIGNALS

SAMPLE-SPECIFIC MARKER ASSOCIATIONS · NOT A PHENOTYPE PROBABILITY

These cards look at specific DNA spots that published research has linked to eye, hair and skin colour, and report what **your own DNA** shows at each one. Each headline is a genuine finding from your genotype - but it's a scientific clue, not a guaranteed, complete picture of exactly how you look.

YOUR THREE MARKER-INFORMED RESULTS

EYE COLOUR

Mixed eye-colour pattern

Your strongest available eye-colour marker contains one allele from each association pattern.

HAIR COLOUR

No strong red-hair pattern detected

None of the three screened high-impact markers showed a strong red-hair-associated signal.

SKIN COLOUR

Mixed skin-pigmentation pattern

One of two copies follows the lighter-associated pattern; many other markers and non-genetic factors also matter.

How to interpret the cards: each headline is based on a DNA marker we could either read directly or confidently estimate from this sample. It's a useful genetic clue, not a guaranteed visible trait or a full prediction of your exact eye, hair or skin colour.

THE BIOLOGY BEHIND THESE MARKERS

HOW PIGMENT IS MADE

Human colouring comes from two pigments: dark **eumelanin** (brown-black) and reddish **pheomelanin**. HERC2/OCA2 control eye colour; MC1R shifts the balance toward hair colour; SLC24A5, SLC45A2 and TYR affect how much pigment your skin makes. The cards above show what we found in your DNA at these genes.

WHY MARKER PATTERNS HAVE LIMITS

Some DNA letters here were read directly; others were carefully estimated from the surrounding DNA, a standard method called imputation. A full appearance prediction would need a much bigger model trained on real photos across many ancestries - beyond what this test does - so we report only what these specific markers show. The displayed markers are not assigned invented weights.

WHAT WE ANALYSED

We checked 41 DNA positions that published research has linked to eye, hair and skin colour. 38 gave a usable result for this sample: 28 were read directly and 10 were estimated using quality-filtered imputation. The cards above summarise what those results show.

KEY GENOTYPES BEHIND THE CARDS

HERC2 · rs12913832 AG DIRECT	OCA2 · rs1800407 CC DIRECT	MC1R · rs1805007 CC DIRECT	MC1R · rs1805008 CC DIRECT
MC1R · rs1805009 GG DIRECT	SLC24A5 · rs1426654 AG DIRECT	SLC45A2 · rs16891982 CC DIRECT	TYR · rs1126809 GG DIRECT

Only the named markers above drive each headline above. The rest of the tested positions are counted in the 38/41 total shown earlier, but don't change the result. Key references: HERC2 rs12913832 for brown vs. blue eyes; MC1R rs1805007/8/9 for red hair; SLC24A5 rs1426654 for skin tone in South Asians.

FUNCTIONAL AND ADAPTATION-ASSOCIATED MARKERS
MODERATE CONFIDENCE 13-marker panel · 1 not called
MARKER EVIDENCE · MODERATE CONFIDENCE 38-marker panel · all called · 38 usable genotype(s)
direct
28 direct · 10 imputed

THE ANCIENT POPULATIONS IN YOUR DNA

HELIXLINE

ANCIENT REFERENCE SIMILARITY · REPORTABLE RESULT

A DNA similarity test called **f3** - shown as f3(Mbuti; Target, Reference) - run on markers read directly from your chip. A higher score means more shared ancient genetic history with that group **specifically**, not a percentage and not proof of one particular ancestor.

IMPORTANT

These raw f3 values are **not ancestry percentages**, are not 0-100 scores, and do not identify a recent ancestor. **This comparison does not identify caste, community, social status, or personal identity.**

CLOSEST TESTED MIXTURE PATTERN

LOW CONFIDENCE · EXPLORATORY VIEW



Sarazm-EN-related — 49.6% ±7.1

Ancestry related to early farming-period individuals from Sarazm used by the Cell-2025 Indian-cline qpAdm mod...

AASI-related proxy — 43.6% ±2.6

Affinity to the AASI-related proxy used in the selected ancient-source model; not an unadmixed sampled AASI g...

Steppe (MLBA) — 6.8% ±6.3

NOT SEPARATELY RESOLVED FROM ZERO

Allele-sharing with the Central Steppe Middle-to-Late Bronze Age proxy used in the selected source model; not...

How to read this: this model is shown as a preview only. It did not meet our bar for a primary, released result, so treat its percentages as tentative - they are simply the closest-fitting pattern among the models we tried, shown with how well it fit rather than as your confirmed ancestry percentages.

SHARED-DRIFT LADDER

The order and spacing below compare only the reference groups shown here, not the whole world. The values and error bars are computed shared-drift comparisons among these references; they are not ancestry percentages or proof of a unique history.

1 Indus Periphery	2 Steppe Middle/Late Bronze Age	3 Iran Neolithic	4 AASI-related proxy (Onge)
0.07669	0.07578	0.07503	0.07410
± 0.00069 SE	± 0.00070 SE	± 0.00073 SE	± 0.00077 SE
111456 SNPs · direct/imputed agree	111456 SNPs · direct/imputed agree	111355 SNPs · direct/imputed agree	111456 SNPs · direct/imputed agree

LOWER SHARED DRIFT — HIGHER SHARED DRIFT

MIXTURE-MODEL DECISION

THRESHOLD NOT MET

The ancient-similarity ranking above is still shown because it answers a different question. We only release a personal mixture percentage once a candidate model clearly passes our accuracy checks; none has for this sample yet.

52389 SNPs
6 candidate models
qpAdm input: direct + validated imputed allowlist

Your result uses the model fixed in advance for your ancestry, not whichever scored best. Ranges in brackets show where each figure could plausibly fall - a wide range means that model measures that part loosely.

MODEL DECISION LEDGER · WHY NONE WAS RELEASED

India cline (Sarazm) — p=0.01113
AASI proxy 43.6% (38-49%) · Sarazm EN 49.6% (36-64%) · Steppe MLBA 6.8% (0-19%) *too small to measure*

India cline (Sarazm, no Steppe) — p=0.01346
Leaves out Steppe MLBA - shares redistributed
AASI proxy 43.4% (38-49%) · Sarazm EN 56.6% (51-62%)

North-west (Indus west) — p=0.02842
Different source list - not directly comparable
AASI proxy 36.9% (29-44%) · Indus Periphery West 49.4% (33-66%) · Steppe MLBA 13.8% (1-26%)

North-west (Parkhai) — p=0.01882
Different source list - not directly comparable
AASI proxy 45.7% (41-50%) · Parkhai Anau EN 37.3% (28-46%) · Steppe MLBA 16.9% (8-25%)

India cline (East Asian) — p=0.007237
Not reported (p = 0.0072): the fitted source proportions fell outside the possible 0-100% range.
Adds East Asian - less precise
AASI proxy 56.0% (16-96%) · Sarazm EN 46.0% (28-64%) · Steppe MLBA 5.9% (0-19%) *too small to measure* · East Asian -7.9% (0-17%) *too small to measure · not reported*

India cline (East Asian, no Steppe) — p=0.01397
Not reported (p = 0.0140): the fitted source proportions fell outside the possible 0-100% range.
Different source list - not directly comparable
AASI proxy 64.5% (24-100%) · Sarazm EN 48.8% (33-65%) · East Asian -13.3% (0-12%) *too small to measure · not reported*

Source-panel sensitivity: India cline (Sarazm) is the displayed model under the declared primary/rotation rule; it is not the model chosen for having the largest p-value. The runner's routed candidate...

Modern stand-ins are explicitly labelled as proxies; all other displayed references are ancient AADR groups.

THE EVIDENCE BOUNDARY

SUPPORTED BY THIS SAMPLE

The order these reference groups rank in against your DNA; the exact similarity score and margin of error for each one; and whether directly-read and estimated genotypes agree on that ranking.

NOT ESTABLISHED BY THIS MODEL

Which named ancient culture you belong to, your single closest ancient individual, a migration date, a specific recent ancestor, or your exact state or community. Those all need separate, dedicated tests.

ALTERNATIVE ANCIENT-SOURCE MODEL SENSITIVITY	EXPLORATORY	6 alternative model(s) tested
ANCIENT REFERENCE AFFINITY	MODERATE CONFIDENCE	1. Indus Periphery · 2. Steppe Middle/Late Bronze Age · 3. Iran Neolithic
EXPLORATORY	Sarazm Eneolithic 49.6%±7.1 · AASI (proxy) 43.6%±2.6 · Steppe MLBA 6.8%±6.3 · fit p=0.01113 vs preferred p≥0.05 (below threshold)	model-derived model-derived Direct + imputed · 52,389 markers

YOUR ANCESTRAL TIMELINE & GENETIC SIGNATURE

ANCIENT SOURCE CONTEXT, ERA BY ERA

This page keeps two kinds of evidence separate: how closely your DNA matches ancient reference groups, and the percentage breakdown from a fitted ancestry-source model. No ancient-source model passed our checks for this sample, so we keep your measured broad ancestry profile and separately-released similarity ranking instead. A good statistical fit doesn't prove it's the only possible history. Your exact state, community and a personal mixing date are not worked out here.

★ HOW TO READ THE CARDS

Every ancestry-source percentage that passed our checks is shown with its margin of error and how well the model fit. The % number is the modelled share for a tested reference source, not a headcount of ancestors or a specific stretch of your chromosomes. A source that's absent simply wasn't part of the model shown - it isn't a measured zero.

ANCESTRAL TIMELINE

YOUR FITTED ANCIENT PROXY BREAKDOWN

⚠ LOW CONFIDENCE · BELOW RELEASE THRESHOLD · SHOWN FOR REFERENCE

The bars below are the closest coefficient-plausible candidate tested; it has not yet cleared the model's declared fit threshold. They show what that candidate model estimated; they are not confirmed ancestry proportions and must not be used as a personal ancestry conclusion.

Sarazm-EN-related

4th millennium BCE

50% ±7.1 pp

Ancestry related to early farming-period individuals from Sarazm used by the Cell-2025 Indian-cline qpAdm model; not identical to Iran-Neolithic ancestry Exploratory sensitivity result; the complete fit or sample-precision acceptance threshold was not met.

WHAT IT REPRESENTS

Affinity to the named Iranian-plateau, Central-Asian, or Indus-periphery reference used by the displayed model. A fitted coefficient cannot establish descent from a particular archaeological site or civilisation.

WHY THIS PROXY MATTERS

Iranian-farmer-related and Central-Asian references capture a widely shared allele-sharing stream in South-Asian models. The historical note supplies population context; the genetic fit does not assign one culture or innovation to a person.

AASI-related proxy

Model proxy

44% ±2.6 pp

Affinity to the AASI-related proxy used in the selected ancient-source model; not an unadmixed sampled AASI genome Exploratory sensitivity result; the complete fit or sample-precision acceptance threshold was not met.

WHAT IT REPRESENTS

Deep South Asian hunter-gatherer-related ancestry represented indirectly in the model. No unmixed AASI ancient genome has yet been sampled, so this is a reconstructed proxy rather than a named ancestral population.

WHY THIS PROXY MATTERS

AASI-related ancestry is an important component in models of South-Asian population history. The coefficient describes affinity to the tested proxy; it is not a pure ancestral population or a literal fraction of identifiable ancestors.

Steppe (MLBA)

2000-1500 BCE

7% ±6.3 pp

Allele-sharing with the Central Steppe Middle-to-Late Bronze Age proxy used in the selected source model; not a literal Yamnaya percentage Exploratory sensitivity result; the complete fit or sample-precision acceptance threshold was not met.

WHAT IT REPRESENTS

Affinity to the Bronze Age Steppe-related reference named in the displayed model. Source labels are statistical proxies; the result does not establish literal Yamnaya descent, a migration route, language, or culture.

WHY THIS PROXY MATTERS

Steppe-related ancestry is one tested stream in models of later South-Asian population formation. Linguistic and Y-lineage associations are population-level historical hypotheses, not personal-language or direct-lineage assignments.

Interpretation: This is a transparent research preview from the already-imputed genotyping-array analysis. The released ancient result is the shared-drift ranking; these source-model coefficients remain conditional on the tested proxies and are not suitable for genealogy or comparison with commercial ancestry percentages.

YOUR GENETIC CLUSTER

GENOME-WIDE POPULATION CLUSTER

South India - broad best fit

Broad regional best fit: South India. Cleanroom holdout top-1 label agreement was 77.4%; this remains an estimate. It is not a community, caste, language, religion, state, nationality, or identity finding.

ENDOGAMY SIGNATURE

★ WHAT IS ENDOGAMY?

Endogamy is repeated partnership within a population over many generations and can increase shared ancestry between parents. **Runs of homozygosity** are long stretches where both chromosome copies carry the same alleles, and **F-ROH** is the measured genome fraction in qualifying runs. A higher value means more qualifying homozygosity under this method; population history, long-standing endogamy, recent parental relatedness, marker density, and calling thresholds can all influence it. It does not identify a community or relationship by itself.

78th percentile in our cohort

F-ROH **0.0282**

Higher than 78% of South Asians.

VS COHORT N = 471

STRONGEST TESTED ANCIENT REFERENCE

REFERENCE SIMILARITY · NOT A CLOSEST INDIVIDUAL

Indus Periphery

Raw outgroup-f3: **0.07669** · SE 0.00069. Highest shared drift among the references tested; not proof of direct descent, an individual match, or an ancestry percentage.

HIGH CONFIDENCE

F-ROH 2.82% · 36 segments · 81.3 Mb total · longest 14.55 Mb

Directly genotyped · 616,214 markers

YOUR ENDOGAMY & ROH PROFILE

WHAT THIS MEASURES - IN PLAIN TERMS

Everyone inherits one copy of each chromosome from their mother and one from their father. A **run of homozygosity (ROH)** is a stretch where those two copies are identical - usually because, many generations back, an ancestor's DNA reached you twice, once through each side of your family. **F-ROH** is simply the share of your genome made up of these matching stretches. It does **not** determine caste, community, religion, or health, and it does **not** diagnose a specific relationship between your parents.

YOUR MEASURED F-ROH

2.82%

of the assessed autosomal genome

ROH SEGMENTS	TOTAL LENGTH	LONGEST RUN
36	81.3 Mb	14.55 Mb

MATCHED REFERENCE POSITION

HIGHER THAN TYPICAL

78th percentile among South Asians

More shared ancestry across the branches of your family tree than most South Asians - common in communities with a long tradition of marrying within the group.

Reference: 471 South Asians (1000 Genomes GIH/PJL/ITU/BEB/STU), same PLINK --homozyg parameters

WHAT A HIGH NUMBER DOES - AND DOESN'T - MEAN

What it usually means: a long history of people marrying within the same community, region, or group, across many generations - common worldwide, and especially common in parts of South Asia.

What it does not mean: this report cannot and does not tell you whether your own parents were related. That would need a separate, dedicated analysis your family would have to specifically request - it is never inferred from this number alone.

YOUR ROH LANDSCAPE

How qualifying runs are distributed by physical length.

1-2 Mb	2-4 Mb	4-8 Mb	8+ Mb
26 runs · 35.0 Mb	7 runs · 20.7 Mb	2 runs · 11.1 Mb	1 runs · 14.6 Mb

Shorter runs (left bars) are common and usually trace to ancestry many generations back. A larger share of longer runs (right bars) is what drives a higher overall F-ROH.

Largest measured chromosome burdens: **chr1** 20.8 Mb · **chr4** 17.9 Mb · **chr10** 6.0 Mb · **chr16** 5.5 Mb. This is descriptive segment context, not a pedigree diagnosis.

METHODOLOGY, FOR THE TECHNICALLY CURIOUS

Measured from direct autosomal genotype calls using plink1.9 homozyg: minimum run 1000 kb / 100 SNPs, 50-SNP windows, ≤1 heterozygous call per window. Imputed variants are not used.

HELIXLINE

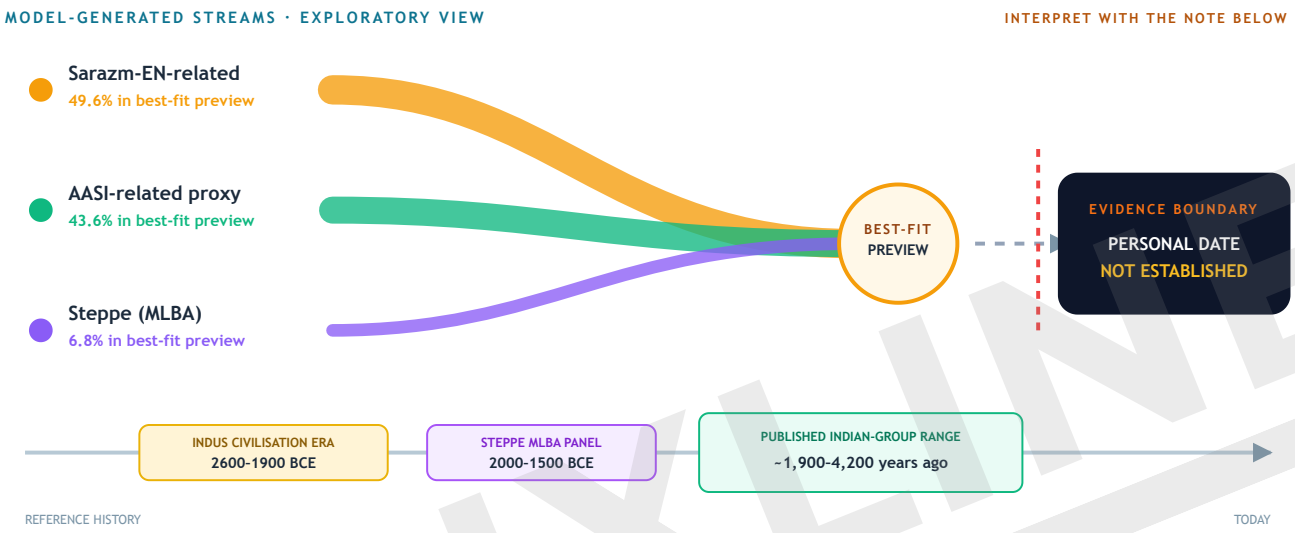
WHEN YOUR ANCESTRAL STREAMS MIXED

THE TIMING OF YOUR ANCESTRY

Our model search found the best-fitting combination of ancient sources for this sample, shown below as an exploratory view. Turning that into a real date needs a separate, validated group-timing result, so the historical bands are background context rather than a date assigned to this family line.

EXPLORATORY CONFLUENCE - YOUR BEST-FITTING MODEL VIEW

The stream widths show the closest-fitting combination our model search found. The full picture is shown below, with a confidence and interpretation note right underneath it.



How to read this: coloured width reflects the best-fitting preview our model found. The bottom bands mark reference material and published population history; they do not date when any source entered this family.

HOW TO INTERPRET THIS MODEL · LOW CONFIDENCE

This source combination was generated successfully and was the closest-fitting candidate among the models tested. Its statistical fit didn't reach the bar required for a primary personal ancestry result, so it's shown as an exploratory, sensitivity-only result rather than a confirmed percentage breakdown. A better-fitting, validated model may become available as our reference data and methods improve. The dated bands are historical background; no group-timing result has passed our separate checks yet.

PUBLISHED POPULATION CONTEXT

1,900-4,200

years ago

LITERATURE RANGE · NOT A PERSONAL DATE

What the published band means

Moorjani et al. analysed ancestry-LD in **571 people from 73 Indian groups** and reported group-level ANI-ASI-related mixture estimates spanning approximately 1,900-4,200 years ago. The green band is that published cross-group comparison-not a midpoint selected for you and not a measurement from this one genome.

Source: Moorjani et al., American Journal of Human Genetics 93, 422-438 (2013), doi:10.1016/j.ajhg.2013.07.006.

Separate result fields: no cohort-measured genetic date passed, so only the published population range is shown. qpAdm proportions and similarity rankings are not converted into a clock.

WHAT THIS ANALYSIS ADDS

WHAT IT ANSWERS	WHY IT MATTERS	LIMITS & UNCERTAINTY
A future released result will answer when a matched group's ancestries typically mixed together.	qpAdm tells us which ancestry sources are present and in what proportion. A separate DNA-timing method adds when that mixing happened, helping place the result in historical sequence.	It is not a date for one ancestor, migration or identity. The stated margin of error travels with the estimate; repeated waves of mixing over time can produce a date somewhere in between.

PUBLISHED POPULATION CONTEXT · PERSONAL DATE NOT ESTIMATED

NOT MEASURABLE FROM THIS FILE

Personal admixture timing was not measured from this file. Dating a mixture event needs the length distribution of ancestry segments, which this sample does not support at a usable precision. The population-history ranges on this page are published context, not your personal dates.

Published studies report broad mixing intervals for the relevant reference populations. Those are literature values for populations, not a date measured from your DNA.

YOUR GLOBAL GENETIC ADDRESS

WHERE DOES YOUR DNA SIT ON EARTH'S FAMILY TREE?

Human populations differ from each other in broad geographic patterns built up over tens of thousands of years. We compare your raw DNA to **six major world regions** to see which ones your genetic makeup most resembles. The percentages below always add up to 100% by design - they are not six literal ancestors, and a small percentage next to a region does not, by itself, mean you have a recent ancestor from there.

SOUTH ASIAN
Indian, Sri Lankan, Pakistani, Bangladeshi and Nepali populations. A finer breakdown into ancient ancestral groups is shown separately, only when a dedicated, more rigorous model can support it.

EUROPEAN
From Iberia to the Urals. Itself a mosaic of farmer + steppe + hunter-gatherer ancestries.

EAST ASIAN
Han Chinese, Japanese, Korean, Vietnamese. Diverged from West Eurasians ~40,000 years ago.

AFRICAN
The deepest-rooted human lineage. All non-Africans descend from a single population that left ~70,000 years ago.

NATIVE AMERICAN
A comparison point based on Indigenous American populations. A weak match here often just reflects overlap between reference groups, not a claim about your personal ancestry.

92%	4%	2%	2%	0%	0%
SOUTH ASIAN	WEST ASIAN (MIDDLE EASTERN)	WEST EURASIAN SIMILARITY AXIS	AFRICAN	NATIVE-AMERICAN-LIKE AFFINITY	EAST ASIAN

YOUR CONTINENTAL COMPOSITION

South Asian strongest signal — **91.8%**
Includes Indian, Sri Lankan, Pakistani, Bangladeshi and Nepali populations.

West Asian (Middle Eastern) TRACE / UNCERTAIN — **3.7%**
Middle Eastern and North African (MENA) populations.

West Eurasian similarity axis TRACE / UNCERTAIN — **2.0%**
A K=6 model axis trained with European-labelled references. It is not, by itself, a recent-European ancestor claim and is not converted into a Steppe, Iran_N or ANI percentage unless a separate ancient-source model passes.

African TRACE / UNCERTAIN — **1.7%**
Sub-Saharan African lineages. All humans trace their deepest roots to Africa.

Native-American-like affinity TRACE / UNCERTAIN — **0.5%**
A weak fit to the Indigenous-American-facing reference axis. At this level it is trace/uncertain model overlap, not evidence of a recent Native American ancestor.

★ QUICK GLOSSARY

kya = thousand years ago · **AASI-related** = deep South Asian hunter-gatherer-related ancestry · **Sarazm-EN-related** = the early farming-period Central Asian proxy used in this model · **Steppe MLBA-related** = Middle/Late Bronze Age pastoralist-related ancestry.

HOW TO READ THIS PAGE

1 · **Start with the dominant result.** This is our most reliable broad-region call for you.

2 · **Read the six percentages as one whole.** They always add up to 100% because of how the model works - not because you have exactly six ancestors.

3 · **Treat values below 7 points as too small to trust.** Do not read them as proof of one specific recent ancestor.

VALIDATION & MODEL IDENTITY

We tested this model on 1253 people whose ancestry was already known, and it named the right broad region **99.76%** of the time (95% confidence range 99.3-99.92%).
This page compares your DNA to **six fixed reference regions**, chosen in advance for every customer rather than customized to you.

WHEN YOUR ANCESTRY ASSEMBLED

These time bands place your ancestral source groups within known archaeological periods. Our model estimates the proportion from each group - not the specific date any mixing happened in your own family.

- **AASI-related** — deep prehistory
  South Asian hunter-gatherer-related source
- **Western farming-related** — Neolithic/Bronze Age
  Model-specific western farming source proxy
- **Steppe MLBA-related** — 2nd millennium BCE
  Middle/Late Bronze Age pastoralist-related source

SAMPLE AND INPUT OVERVIEW	HIGH CONFIDENCE	Helixline genotyping array · 616,214 usable markers	direct
GLOBAL GENETIC ADDRESS / REFERENCE AXES	EXPLORATORY	5 reference affinity axis/axes	model-derived
SUB-CONTINENTAL COMPOSITION	NOT APPLICABLE	Your report already provides the strongest supported broad and regional ancestry views. A separ...	model-derived
SUBMITTED ANCESTRY-BACKGROUND COMPARISON	HIGH CONFIDENCE	No linked family-background comparison was available; DNA results were unchanged	context
MODERATE CONFIDENCE	South Asian 91.8% · West Asian 3.7% · European 2% · African 1.7%		Model-derived · 616,214 markers

YOUR REGIONAL DNA PROFILE

THE REGIONAL PATTERN IN YOUR DNA

We compare the overall pattern across your DNA to broad South Asian regional groups. This gives you a general sense of geography while respecting how much genetic overlap exists between neighbouring populations.

South India
LEADING BROAD REGIONAL FIT

ALL REGIONAL SCORES · SHOWN TOGETHER



01 · GENOME-WIDE PROFILE

Many points across your whole DNA are combined together, which makes this more stable and reliable than looking at any single genetic marker alone.

02 · RELATIVE FIT

These scores show how you rank among the regional groups we tested, not a standalone match to just one of them. Closely related regions can share much of the same genetic history.

03 · USE WITH CONTEXT

Read this alongside family origins, chromosome painting and ancient-source results-not as an exact state, language or community assignment.

THE USEFUL TAKEAWAY

A clear margin supports a broad regional orientation. It still leaves room for recent migration, mixed family histories and communities spread across several states-the richness that documentary family history can add to the DNA result.

YOUR REGIONAL SIGNAL IN ONE SENTENCE

South is the leading broad fit, 25.7 points ahead of West.

This gap describes how clearly one region stands out from the next in this comparison; it is not a probability of your birthplace or identity.

BEST NEXT CONTEXT

Compare this broad signal with grandparents' birthplaces, languages and migration histories. Documentary detail can distinguish family stories that genetics alone intentionally leaves open.

HELIXLINE

YOUR VALIDATED ANCESTRY COMPOSITION

PRIMARY BROAD RESULT · HIGH CONFIDENCE

Predominantly South Asian

Your DNA's strongest broad match is with the **South Asian reference group**. The detailed percentages below break down the comparison behind that broad result.

91.8%

South Asian

model weight · not a single ancestor

HOW THE SIX REFERENCE AXES ADD UP



91.8% South Asian MODEL AXIS	**3.7%** West Asian TRACE / UNCERTAIN	**2.0%** European-labelled K=6 axis TRACE / UNCERTAIN	**1.7%** African TRACE / UNCERTAIN	**0.5%** Native-American-like axis TRACE / UNCERTAIN	**0.2%** East Asian TRACE / UNCERTAIN

WHAT 91.8% MEANS

It is your score on the South Asian reference group in this fixed six-region model. It supports your overall South Asian result.

WHY 2.0% IS NOT "RECENT EUROPEAN"

This is your score on the reference group built from European populations. It does not, by itself, mean you have a recent European ancestor or point to a specific ancient source.

A SEPARATE QUESTION · WITHIN SOUTH ASIA

LOW CONFIDENCE · DIRECTIONAL ONLY

South India

Broad-region best fit · not exact identity

The broad-region model's best fit is **South India**, ahead of West India by 25.7 model-score points. This is broad regional context, not an exact state, community, caste, language or identity label.

CONFIDENCE AT A GLANCE

HIGH · BROAD PLACEMENT

South Asian

We tested this model on 1253 people with already-known ancestry, and it got the broad region right 99.76% of the time.

VALIDATED MODEL OUTPUT

Six-region composition

This measurement is not quoted in this report because the test behind it is not available to cite.

LOW · REGIONAL CONTEXT

Broad region: South India

This is a broad DNA-region best fit only. It does not establish a state, community, caste, language or recent ancestor.

One rule for the whole page: these percentages describe how similar your DNA is to six fixed reference regions. They are not six literal ancestors, and small values below 7 points are uncertain.

REGIONAL, STATE & COMMUNITY CONTEXT

GENETIC GEOGRAPHY | EVIDENCE SCOPE

This page separates the DNA model's broad genetic-region best fit from exact state or social identity. The nearest community reference candidates are shown as an exploratory shortlist with qualitative confidence labels only; they are not a caste, religion, language, or identity determination.

BROAD GENETIC REGION · LOW CONFIDENCE

South India

South India 62.4 · West India 36.7 · model scores, not ancestry %

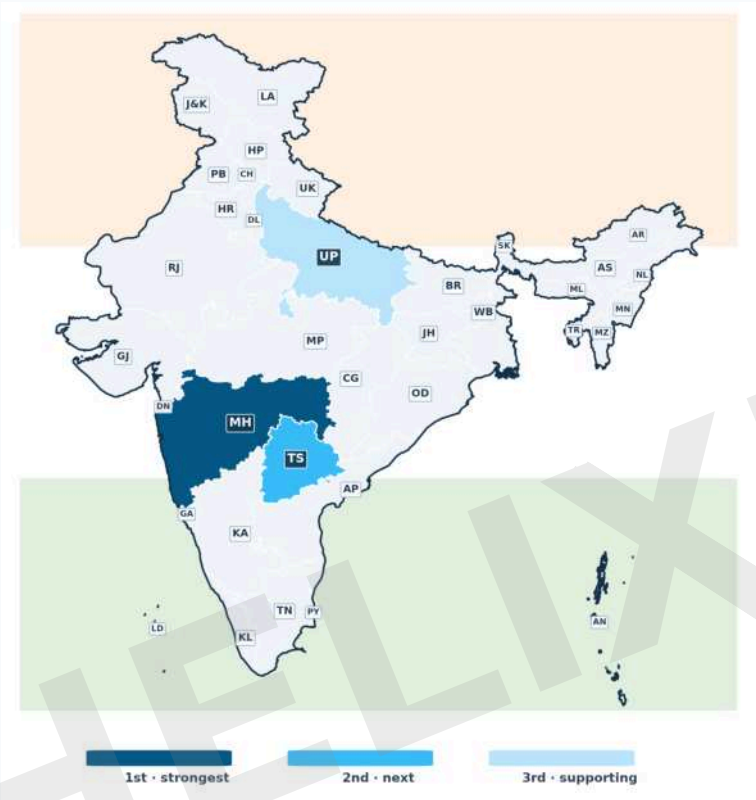
RELEASE CONFIDENCE

LOW · broad directional best fit

Fine-scale identity is not inferred

RANKED STATE SHORTLIST · LOW CONFIDENCE

TOP 3 STATE MATCHES · LOW CONFIDENCE



Low-confidence top three: dark blue marks first, medium blue second and light blue third. Read the shortlist together; it is directional, not a confirmed state origin.

COMMUNITY RESEARCH PREVIEW

RESEARCH PREVIEW · NOT A PERSONAL FINDING

These are nearest training-panel neighbourhoods, retained so you can compare them with genealogy you already know. Confidence is shown as qualitative labels, not percentages.

Maratha
Outside broad fit — LOW CONFIDENCE

Why it is not a result: the current cleanroom holdout reached 56.8% top-1 and 81.1% top-3 among only 37 evaluable samples (wide 95% intervals); family-grouped CV reached 39.6% top-1 (n=849). There is still no independently verified self-report truth for this customer cohort. It is not used in the main ancestry conclusion and does not establish community, caste, religion, language or social identity.
Improves with: balanced, verified four-grandparent truth and a sealed external test set.

SOUTH-WEST DECCAN REFERENCE CORRIDOR

Exact state unresolved

The broad-region result and the state-reference model point across neighbouring South and West Deccan panels. The original state model order is preserved below; regional tags explain the disagreement without turning it into an exact-state claim.

LEADING CANDIDATES (NOT INDEPENDENTLY VALIDATED)

Maharashtra	Outside broad fit	Low confidence
Telangana	Aligned with broad fit	Low confidence
Uttar Pradesh	Outside broad fit	Low confidence

Bands, not percentages: state-from-DNA saturates well below 100% top-1 accuracy even for a true match, so an exact number would overstate precision this model doesn't have.

HOW THE REGION MODEL WAS TESTED

Method: 5-fold StratifiedGroupKFold; KING kinship >= 0.0442 kept within fold. DNA projection features only; address, surname, caste, religion and language excluded.
Production classifier record: 77.4% broad-region label agreement (n=31; canonical cleanroom holdout). The strict audit below is a separate subset, not a pooled figure.
Strict geography-audit subset: 73.5% (n=913). Class-level support and recall were not recorded in this sealed artifact.
Limit: The sealed external set is underpowered; use this as a broad reference-fit estimate, not a geographic or identity fact. Sealed external check: 77.4% (n=31; underpowered; target n=150).
Improves with: balanced, independently verified four-grandparent geography across Punjab, UP, Bihar, Haryana, Kashmir, Himachal, the Northeast and under-represented states.

ANCESTRY BACKGROUND COMPARISON

No completed ancestry-background survey was available for comparison in this report.

Strongest region-aligned DNA directions
States: Telangana

When customer-provided background is available, it is displayed and compared separately. It does not alter, rerank, calibrate or rescue any DNA-derived result; current address and surname are excluded.

IMPORTANT INTERPRETATION NOTE

DNA may support broad regional similarity, but it **cannot reliably determine your caste, community, language, religion, home state or personal identity**. Any group names produced by the DNA models are research references—not conclusions about who you are or which community your family belongs to. Administrator-supplied context, when present, is separately labelled with its own evidence status.

CLOSEST STATE REFERENCE MATCHES	EXPLORATORY	1. Maharashtra · 2. Telangana · 3. Uttar Pradesh	model-derived
CLOSEST COMMUNITY REFERENCE MATCHES	EXPLORATORY	1. maratha	model-derived
MODERATE CONFIDENCE	South 62.4 · West 36.7 (model scores, not percentages)		Model-derived · 616,214 markers

CHROMOSOME PAINTING · RESEARCH PREVIEW

HELIXLINE

SAMPLE-SPECIFIC FLARE TRACT MAP

RESEARCH PREVIEW · NOT CONFIRMED AS A PERSONAL RESULT

This map shows our best current guess, chromosome by chromosome, for which broad ancestry group each stretch of your DNA most resembles. The colours and boundaries are the model's estimate - not an exact family tree, a list of recent ancestors, a community, a state, or which parent a segment came from. See the main ancestry-percentages page earlier in this report for your primary result. This chromosome map is shown for research interest only and is not confirmed as a personal result.

SAS South Asian | EUR European | EAS East Asian | AFR African | WAS West Asian / Middle Eastern | OCE Oceanian | UNCERTAIN Low-confidence / unassigned

22 / 22 AUTOSOMES - each card is an experimental broad-label map



PAINTING STATS

LABELLED TRACTS	193
METHOD	FLARE_v0.6
ANCESTRY CLASSES	7 / 5
HIGH-CONFIDENCE COVERAGE	89.7%

PILOT MODEL · EXPLORATORY VIEW

Think of this as an early, exploratory sketch of your ancestry across the genome, not your official result. A coloured section is the model's best guess for that stretch of DNA, not a confirmed, exact boundary.

RESEARCH-PREVIEW MAP SUMMARY

HAPLOTYPE 1

South Asian	89.9%
Low-confidence / unassigned	7.9%
East Asian	1.0%
European	0.6%

HAPLOTYPE 2

South Asian	87.0%
Low-confidence / unassigned	11.3%
European	0.9%

WHAT WOULD ADD FINER DETAIL

Breaking your South Asian result into smaller regional or community groups would need a much larger, evenly balanced set of comparison samples from each community, confirmed family trees going back generations, and thorough independent testing. Simply having more estimated (imputed) DNA data points is not enough on its own.

HOW TO READ THIS

Colours show the model's broad ancestry call for each of your two DNA copies at that spot. Grey regions mean the model wasn't confident enough to make a call there. **Why this can look different from your main ancestry result:** this chromosome map and your main ancestry percentages are two separate tests that ask different questions. Treat this page as a supporting, exploratory view; your main ancestry-percentages page remains the primary result.

• centromere · **H1/H2** statistical haplotypes; parent of origin is unknown · bar lengths proportional to chromosome size.

EXPLORATORY

22/22 autosomes · 193 segments · EUR, AFR, WAS, OCE, SAS, EAS, UNCERTAIN

Statistically imputed · 616,214 markers

INDEPENDENT EVIDENCE · ONE COHERENT STORY

HOW YOUR ANCESTRY RESULTS FIT TOGETHER

Your report uses several methods because no single model can answer every ancestry question. This page shows what each method found and how to read the results together.

BROAD ANCESTRY

South Asian 91.8%

Your strongest match in the broad continental reference model.

CHROMOSOME ANCESTRY

Broad methods agree

This chromosome map and your main ancestry result agree on your leading broad ancestry group. Their exact percentages still won't match, because the two methods work differently and handle uncertainty differently.

ANCIENT SIMILARITY

Indus Periphery

The ancient population your DNA most closely resembles, based on inherited genetic patterns you share with it; this is not a literal ancestor or a percentage of your DNA.

DIRECT-CALL CROSS-CHECK

Same leading ancient reference

Direct: Indus_Periphery · **Expanded:** Indus_Periphery
Ranking agreement: 100.0% · 111,649 direct and 126,226 expanded markers. This check confirms that adding the extra, statistically estimated markers doesn't change the ranking already visible from your directly-read DNA alone.

WHY THE NUMBERS CAN DIFFER

Each method below answers a different ancestry question using its own comparison samples. "Agreement" means they broadly point the same direction - their exact percentages are not meant to match, and should never be added, subtracted, or forced to line up.

Broad ancestry compares your DNA to present-day population groups around the world. Chromosome painting looks at individual stretches of your chromosomes one at a time. Ancient similarity measures how closely your DNA resembles specific ancient burial-site genomes, based on inherited patterns you share with them. qpAdm (a statistical modelling method) tests a specific, named mix of ancient ancestor groups against your DNA, the way a lab tests a hypothesis. None of these methods identifies caste, community, social status, or one named ancestor.

WHAT YOU CAN TAKE FROM THIS

- Your leading broad reference match
- Whether independent methods support the same broad pattern
- Which conclusions are measured, exploratory, or unavailable

WHAT REQUIRES CAUTION

- Similarity to a reference group is not your personal identity or nationality
- Ancient models describe stand-in source populations, not your actual named ancestors
- Different methods compare you to different populations and different points in the past

Best reading order: start with Broad Genetic Address, then compare the Chromosome Painting, Ancient Population Origins, and Ancient Ancestry Models pages. Each page states its own confidence and limitations.

HELIXLINE

YOUR MOTHER'S ANCIENT STORY

Mitochondrial DNA · Maternal lineage · One direct maternal line

This haplogroup follows one direct family line only. It represents one narrow branch among the thousands of people in your ancestry, not your complete maternal heritage.

HELIXLINE

M

MT-DNA · MOST RELIABLE DEPTH

The lineage above is your **most reliable maternal placement** for this array - the deepest point we're confident is correct. The deeper branch below is our best guess, but needs a few more DNA markers to confirm it.

BEST-FIT DEEPEST SUB-BRANCH **M3** PROVISIONAL SUB-BRANCH

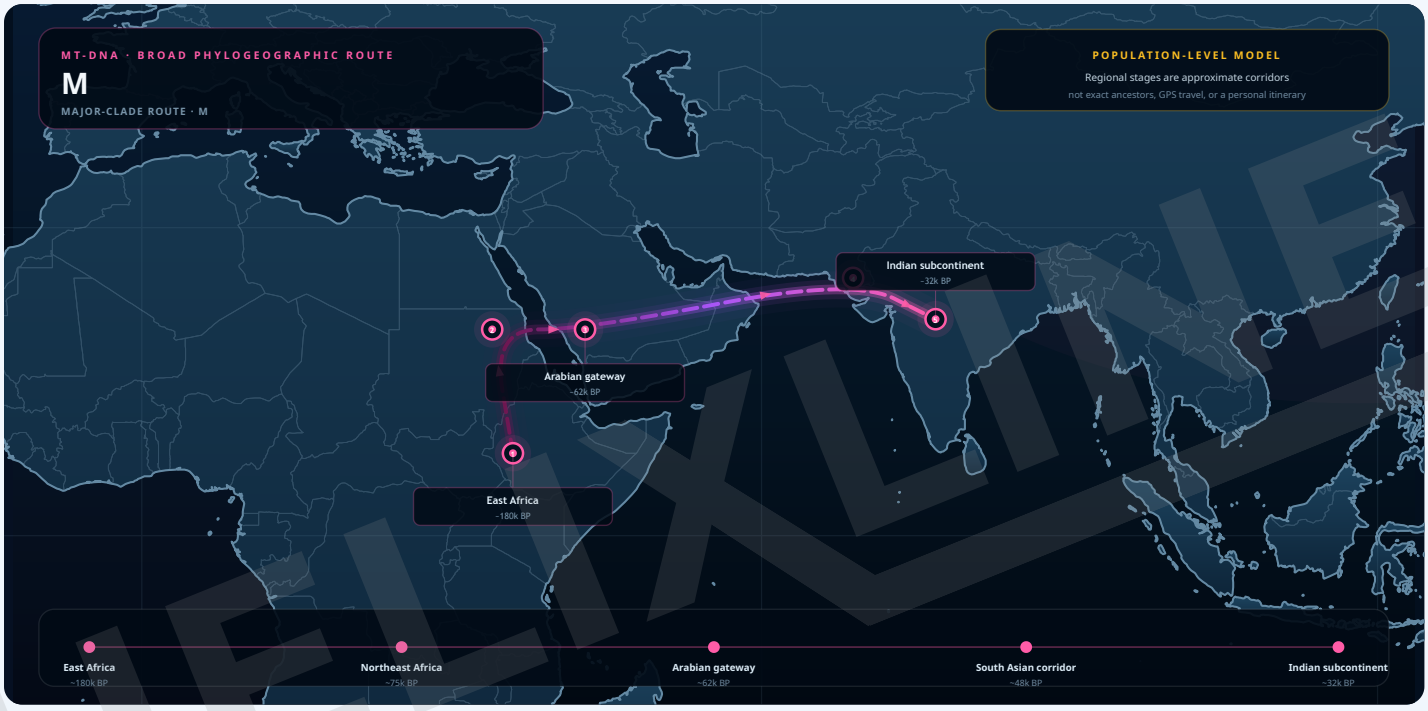
Placed using 984 mtDNA positions read directly from your chip; 1 of 2 of the final, most-specific markers were found. This is not full mitochondrial sequencing. The confidence above reflects how often this method got it right in testing on other people - not the odds that your own result is correct.

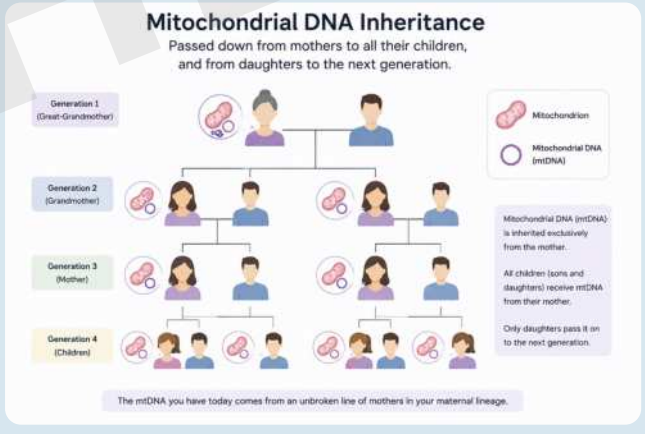
HOW WE DETERMINED YOUR MATERNAL HAPLOGROUP

Your mitochondrial DNA haplogroup M was placed by comparing the mtDNA positions read by this genotyping chip against the standard reference tree of human mtDNA (PhyloTree 17, via HaploGrep3). We only report as deep a branch as those chip positions can reliably support. Full mtDNA sequencing may confirm it or refine it further.

BROAD PHYLOGEOGRAPHIC ROUTE · HAPLOGROUP M

TRACED TO · M





MATERNAL LINEAGE · MT-DNA

EVOLUTIONARY PATH → YOUR HAPLOGROUP

Mitochondrial Eve → mtMRCA → L1'2'3'4'5'6 → L2'3'4'5'6 → L2'3'4'6 → L3'4'6 → L3'4 → L3 → M → M3

WHAT HAPLOGROUP M MEANS

Haplogroup M is a phylogenetic placement for one direct maternal line. The array supports the displayed branch at the marker resolution shown above. It does not establish a personal origin, migration route, archaeological culture, language, community, or the ancestry of the rest of the genome.

NOTABLE FACTS

- This haplogroup describes one direct uniparental line, not the rest of the genome.
- It does not determine community, caste, religion, language, culture, or nationality.

LINEAGE EVIDENCE AND PROVISIONAL DEPTH

LOW CONFIDENCE — maternal M (provisional M3), 1 defining marker(s) absent · paternal H1a1d2c1

LOW CONFIDENCE — M (provisional deeper: M3)

direct

Directly genotyped · 987 markers

YOUR MATERNAL-LINE PLACEMENT & EVIDENCE

ONE DIRECT MATERNAL LINE - NOT ALL OF YOUR MATERNAL ANCESTRY

Mitochondrial DNA is usually inherited from the mother and generally does not recombine, but mutations accumulate over generations. Haplogroup **M** shows where that one direct mother-to-child line sits on the human family tree of DNA. It does not describe all ancestors, establish ethnicity, or prove a personal migration route.

M	984	1/2	593/593	6
YOUR HAPLOGROUP · BROAD CLADE (PROVISIONAL)	DIRECT MT POSITIONS	CONFIRMING MARKERS FOUND	CORRECT IN TESTING	NEARBY BRANCHES

How we tested this: when we ran this same method on 593 other people with already-known DNA, it placed 593 of them at the correct depth (a reliability range of 99.36-100.0%). That's a track record for the method, **not** a 99% chance that your own result specifically is right. Observed derived defining marker(s): 16126C. Not present on this array: 482C.

YOUR LINEAGE PATH

This traces where the DNA markers read by your chip place you on the reference tree of mtDNA branches. Markers we couldn't read limit how deep a branch we can confidently show.

★ READING THE NOTATION

Each marker is written as *position + ancestral base > variant base*. For example, `m.10398A>G` reads as: at position 10,398 the **G** variant replaces the ancestral **A**. Each marker like this is a *defining mutation* that places your mtDNA in this haplogroup.

- Mitochondrial Eve
- mtMRCA
- L1'2'3'4'5'6
- L2'3'4'5'6
- L2'3'4'6
- L3'4'6
- L3'4
- L3
- M
- **M3**

WHAT IS MITOCHONDRIAL DNA?

Inside every cell are tiny power-plants called mitochondria. Each has its own DNA - a small circular molecule of **16,569 base pairs** - inherited only from your mother. The mitochondrial genome is generally inherited as one unit, so variants can support placement on a direct maternal phylogeny.

Inheritance	Maternal-only
Genome size	16,569 bp
Mutation rate	Not inferred here
Common ancestor	Not a sample estimate
Number of genes	37 (13 protein-coding)
Copies per cell	100 - 10,000

NOTABLE FACTS ABOUT M

- This haplogroup describes one direct uniparental line, not the rest of the genome.
- It does not determine community, caste, religion, language, culture, or nationality.

PHYLOGENETIC SCOPE OF M

Not inferred

The displayed label is a phylogenetic branch name. This array result does not infer the branch's geographic origin, age, ethnolinguistic identity, or worldwide frequency.

SISTER & NEARBY MATERNAL BRANCHES

Close relatives of **M3** on the DNA family tree. A direct sister shares the immediate parent branch; a nearby cousin branches off one level further back.

M1'20'51	DIRECT SISTER BRANCH	M → M1'20'51
M10	DIRECT SISTER BRANCH	M → M10
M11	DIRECT SISTER BRANCH	M → M11
M12'G	DIRECT SISTER BRANCH	M → M12'G
M13'46'61	DIRECT SISTER BRANCH	M → M13'46'61

Tree source: PhyloTree Build 17 RSRS (HaploGrep3 resource). These are your place on the DNA family tree - not people who live near you or share your surname.

YOUR FATHER'S ANCIENT STORY

Y-Chromosome DNA · Paternal lineage · One direct paternal line

This haplogroup follows one direct family line only. It represents one narrow branch among the thousands of people in your ancestry, not your complete paternal heritage.

HELIXLINE

H1a1d2c1

Y-DNA

HIGH MARKER EVIDENCE

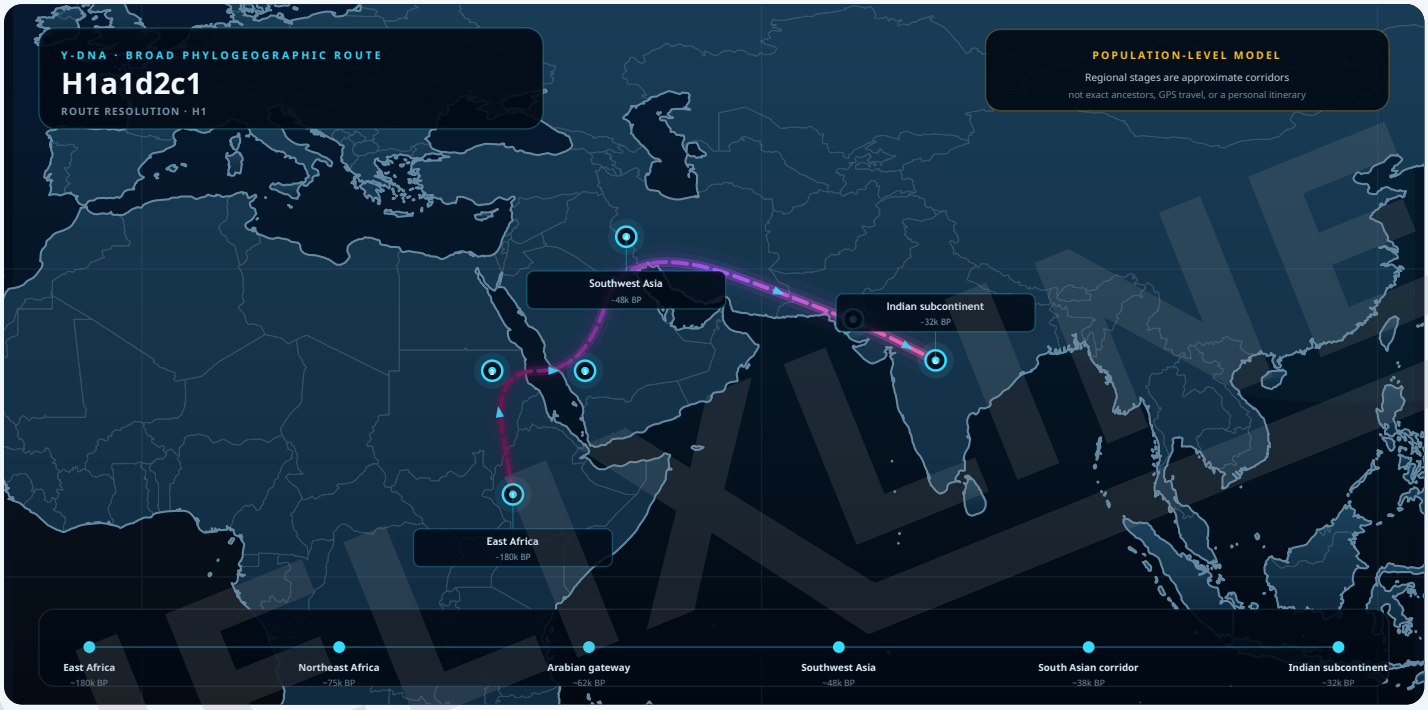
94 of 100 defining markers checked confirm this branch · the most specific marker was confirmed in your data

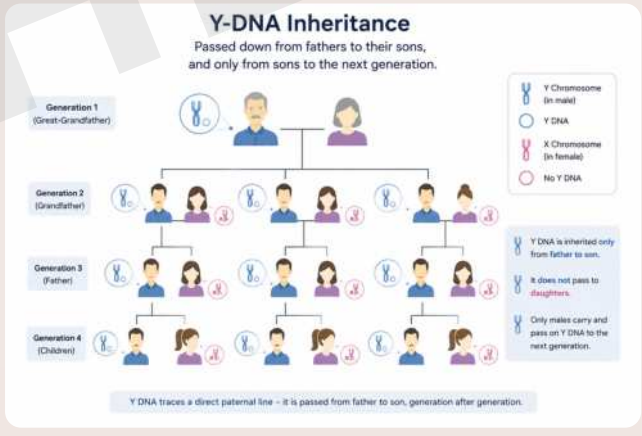
HOW WE DETERMINED YOUR PATERNAL HAPLOGROUP

Your Y-chromosome haplogroup H1a1d2c1 was identified by analysing Y-chromosome markers in your DNA - the same chromosome passed through the direct father-to-son line across the largely non-recombining Y chromosome.

BROAD PHYLOGEOGRAPHIC ROUTE · HAPLOGROUP H1A1D2C1

TRACED TO · H1





PATERNAL LINEAGE · Y-DNA

EVOLUTIONARY PATH → YOUR HAPLOGROUP

Y-chromosomal Adam → H → H1 → H1a → H1a1 → H1a1d → H1a1d2 → H1a1d2c → H1a1d2c1

WHAT HAPLOGROUP H1A1D2C1 MEANS

Haplogroup H1a1d2c1 is a phylogenetic placement for one direct paternal line. The array supports the displayed branch at the marker resolution shown above. It does not establish a personal origin, migration route, archaeological culture, language, community, or the ancestry of the rest of the genome.

NOTABLE FACTS

- This haplogroup describes one direct uniparental line, not the rest of the genome.
- It does not determine community, caste, religion, language, culture, or nationality.

HELIXLINE

THE SCIENCE & FAMILY TREE OF YOUR PATERNAL LINEAGE

YOUR DIRECT PATERNAL LINE · PHYLOGENETIC PLACEMENT

The largely non-recombining region of the Y chromosome is normally inherited through one direct father-to-son line and accumulates variants over generations. The branch sequence below is the array-supported phylogenetic path for this sample. It is not a complete paternal biography, a dated personal lineage, a geographic origin, or a migration route.

H1a1d2c1	94/100	Confirmed	Direct Y markers	3
YOUR HAPLOGROUP	POSITIONS CONFIRMING THIS BRANCH	MOST SPECIFIC MARKER	EVIDENCE TYPE	NEARBY BRANCHES

YOUR LINEAGE PATH

Each displayed node is a branching label on the Y-chromosome phylogeny; only the tested direct paternal line is represented.

- Y-chromosomal Adam
- H
- H1
- H1a
- H1a1
- H1a1d
- H1a1d2
- H1a1d2c
- **H1a1d2c1**

SISTER & NEARBY PATERNAL BRANCHES

Close relatives of **H1a1d2c1** on the DNA family tree. A direct sister shares the immediate parent branch; a nearby cousin branches off one level further back.

H1a1d2c2	DIRECT SISTER BRANCH	H1a1d2c → H1a1d2c2
H1a1d2a	NEARBY COUSIN BRANCH	H1a1d2 → H1a1d2a
H1a1d2b	NEARBY COUSIN BRANCH	H1a1d2 → H1a1d2b

Tree source: local ISOGG/ybrowse hg19 snapshot. These are your place on the DNA family tree - not people who live near you or share your surname.

WHAT IS THE Y-CHROMOSOME?

A Y chromosome, when present, is normally inherited through the direct paternal line. Its largely non-recombining region contains markers that can support phylogenetic placement; it represents only one lineage and not the rest of the genome.

Inheritance	Paternal-only (males)
Genome size	~57 Mb
Mutation rate (Y-STR)	Not inferred here
Common ancestor	Not a sample estimate
Number of genes	~50 (NRY portion)
Recombination	Pseudoautosomal only

HOW DEEP THIS PATERNAL CALL GOES

SUPPORTED BY THIS ARRAY

This branch is supported by 94 out of 100 defining DNA positions we checked, including the single most specific marker for this branch. It shows where your one direct paternal line sits on the Y-DNA family tree.

WHAT IT DOES NOT DETERMINE

A Y-haplogroup does not define your state, community, language, caste or total ancestry. Most ancestors are outside this single father-to-son line; autosomal chapters describe the broader genome.

PHYLOGENETIC SCOPE OF H1A1D2C1

Not inferred

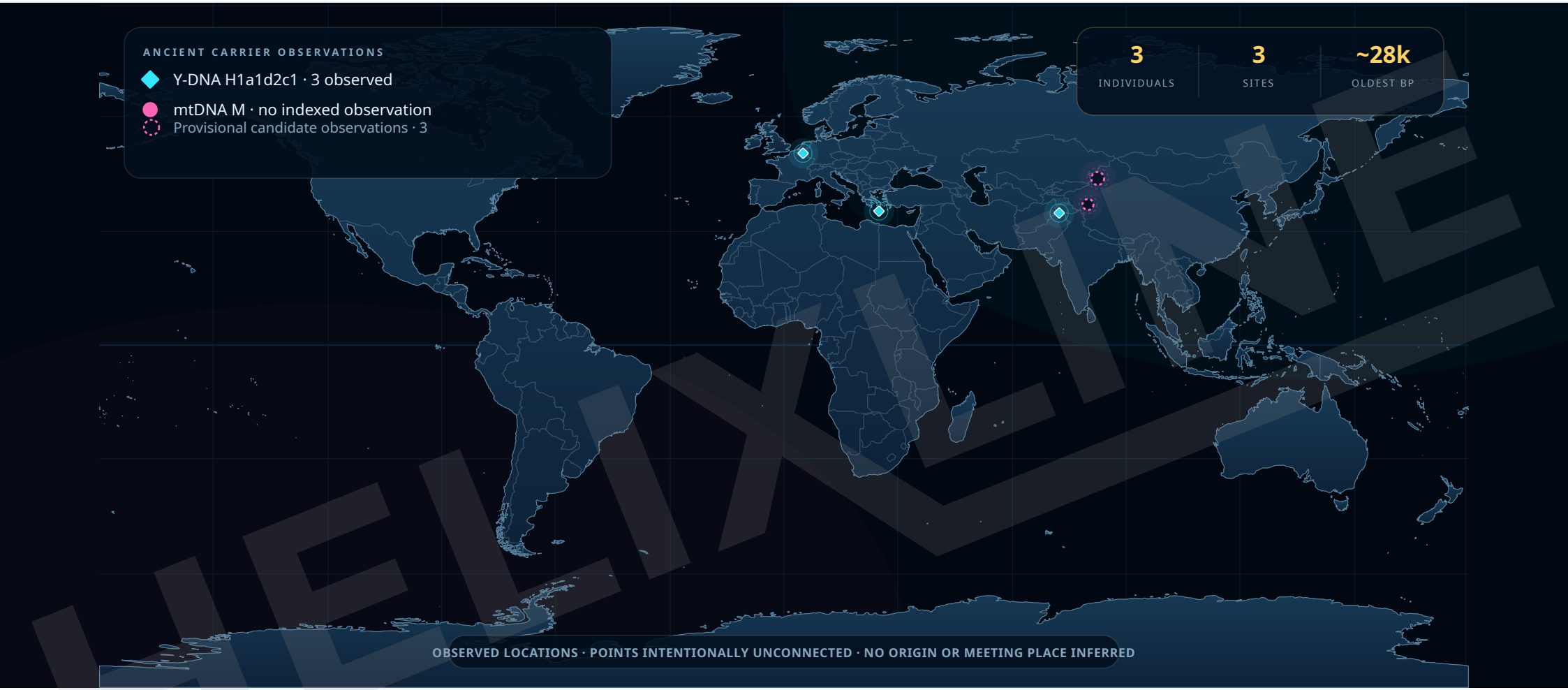
The displayed label is a phylogenetic branch name. This array result does not infer the branch's geographic origin, age, ethnolinguistic identity, or worldwide frequency.

HELIXLINE

ANCIENT CARRIER OBSERVATION CONTEXT

MT-DNA M

Y-DNA H1a1d2c1



HELIXLINE

MATERNAL · MT-DNA

M

0 INDIVIDUALS · 0 EXCAVATION SITES · - OLDEST (YEARS AGO)

COEXISTENCE IN YOU

These two lineages are shown together simply because you carry both - not because they crossed paths at a mapped location.

PATERNAL · Y-DNA

H1a1d2c1

3 INDIVIDUALS · 3 EXCAVATION SITES · ~27.8k OLDEST (YEARS AGO)

HOW TO READ THIS MAP

Each symbol represents one or more ancient people found at that location whose DNA belongs to the same genetic branch shown on this page. The symbols aren't connected to each other - ancient DNA survives only in scattered pockets, so what's plotted here reflects gaps in the archaeological record, not proven ancestors, origins, journey stops, or a place where lineages met. The map shows where this branch has turned up historically; it is not a confirmed route travelled by your own family.

Hollow rose symbols match a more specific branch, M3, that we're still confirming - treat this deeper placement as tentative, not final.

WHY SOME MAPS ARE SPARSE

Ancient DNA survives unevenly across climates and regions. Few or zero points describe the surviving record-not the age, importance, or geographic origin of your lineage.

DNA RELATIVES

PRIVACY-SAFE MATCHING AVAILABLE

Choose relative matching in your secure account. Relationship bands appear only when validated overlap and kinship thresholds are met.

YOUR BROAD GENETIC CLUSTER

South India - broad best fit

Independent ancestry-model result; not inferred from the relative shown beside it.

NEAREST CONSENTED COHORT SAMPLE

Ready when you opt in

Your privacy choice is active; use the account QR above whenever you would like matching enabled.

Relative matching is ready when you opt in

Your current privacy choice keeps living-relative comparison off. You can enable the optional search from your secure account whenever you would like to look for opted-in matches.

ABOUT DNA RELATIVE MATCHING

This screen compares the submitted genotype array with opted-in cohort samples using KING-robust kinship. It can support a close-relative degree band when the SNP-overlap gate passes. It does not calculate IBD segments, shared centimorgans, a unique pedigree relationship, parent of origin, or community affinity.

HOW DNA RELATIVE MATCHING WORKS

KING genotype comparison
The production endpoint compares autosomal genotypes with opted-in cohort samples using the KING-robust kinship coefficient. It does not call IBD segments.

Shared cM not calculated
Shared centimorgans are not calculated by this KING-only screen. The reference table below contains broad educational averages, not this sample's measured values.

Degree-band classification
Thresholds convert KING kinship into duplicate, first-, second-, or third-degree bands. The screen does not choose one exact relationship within a band.

Community affinity not inferred
Mean cohort kinship is not a validated ancestry or community classifier and is not reported.

ILLUSTRATIVE PEDIGREE AVERAGES · NOT THIS RESULT

RELATIONSHIP	SHARED CM	DNA %
Parent / Child	~3,400 cM	~50% DNA
Full Sibling	2,200-3,400 cM	~50% DNA
1st Cousin	~850 cM	~12.5% DNA
1st Cousin Once Removed	~425 cM	~6.25% DNA
2nd Cousin	~212 cM	~3.125% DNA
3rd Cousin	~53 cM	~0.78% DNA
4th Cousin	~13 cM	~0.2% DNA

SCOPE OF THE KINSHIP SCREEN

⚡ KINSHIP FACTS

- A degree band includes multiple possible pedigrees; the screen does not choose one exact relationship.
- Population structure and endogamy can affect kinship interpretation; real-family, ancestry-stratified validation is still required.
- IBD segment lengths and shared cM are outside this KING-only endpoint and are not calculated.
- Fresh validation classified 299 of 300 synthetic close-pedigree pairs into the correct degree band (99.7%; Wilson 95% CI 98.1-99.9%). Complex and real endogamous pedigrees still require validation.

CLOSE RELATIVES (3RD DEGREE +)

Relative matching is ready when you opt in

No opted-in cohort member met the validated close-relative threshold in this search. Your match list can grow as more relatives join.

0
MATCHES

Endogamy score (F-ROH) = 2.82% - genetically **moderately endogamous (typical of an endogamous jati)**. High community endogamy means many distant relatives share background ancestry; close genealogical relatives still need to be in our cohort.

Measured with plink1.9 homozyg: minimum segment **1000 kb** and **100 SNPs**, sliding window 50 SNPs, plus density and gap filters that exclude centromeric artefacts. 36 segments totalling 81.3 Mb. **F-ROH depends strongly on these settings** - a shorter minimum segment gives a substantially higher figure - so comparing against another tool requires matching the parameters above.

Grow your match list - encourage your relatives to test with Helixline. As more opted-in relatives join, the searchable cohort can contain more qualifying matches.

HELIXLINE

THE COMPLETE NARRATIVE

FROM AFRICA TO YOU - IN ONE PAGE

Around **70,000 years ago**, a small group of humans left Africa. These were the ancestors of everyone alive outside Africa today. Over millennia they moved through the Middle East and into Central Asia, splitting into bands that would eventually populate every continent.

Along the way, **they met other human species**. Your DNA carries **1.7% Neanderthal** heritage - gene variants inherited through population mixing with Neanderthals in the Near East and western Asia roughly 50,000-60,000 years ago. Some of these ancient variants are linked to immune and metabolic traits across human populations; this percentage alone does not predict either trait in you.

Eventually, ancestral population movements reached the Indian subcontinent. Your validated broad genetic result is **South Asian**. South Asian genomes carry shared history related to early subcontinental foragers, Iranian-related ancient populations and Steppe-related pastoralists. The report's Ancient Populations chapter compares your DNA with those reference groups directly. It shows how closely you rank against each group, kept separate from the percentage-mixture estimates elsewhere in the report. Your displayed mixture percentages come from a statistical ancestry-modelling method (qpAdm) that tested whether a specific combination of ancient source populations fits your DNA, and passed that test within an estimated margin of error. These numbers are relative to the specific ancient comparison groups used, not a literal tie to a named ancient culture - a good statistical fit does not mean it is the only possible explanation. Minor secondary signals in our underlying ancestry model are not the same as recent European ancestry, and are not described that way here.

Among the state references tested, **Maharashtra** ranked first in this exploratory comparison. This is a reference-panel ranking, not a determination of state of origin, community or identity. Your direct maternal line is placed at haplogroup **M** - as specific as your chip data can reliably support. Your direct paternal Y-DNA line is placed at haplogroup **H1a1d2c1** from the observed Y markers.

Each chapter asks a different genetic question and has its own evidence standard.

Broad ancestry, local chromosome-painting views, ancient affinity and uniparental lineages should be interpreted without treating one metric as another.

Your report distinguishes measured results, reference context and withheld claims so you can see exactly what the data supports.

VALIDATED METHODOLOGY

1000G + AADR	1,253	Genotyping array	99.76%
Reference resources used; each result is validated separately	People with known ancestry, used only to test accuracy	Direct array genotypes, plus quality-checked extra data	How often our model's top guess was correct

READ THE EVIDENCE IN LAYERS

"Start with the broad South Asian classification. Use chromosome painting as a supporting visual layer, the ancient-similarity comparison (outgroup f3) for how closely you match ancient reference groups, and mtDNA/Y-DNA for one direct maternal or paternal line.

None of these alone determines a community, culture, nationality, or exact ancient-source percentage."

DATA QUALITY & REFERENCE COHORT

HELIXLINE

QUALITY CHECK PASSED

99.45%

GENOTYPE CALL RATE · PASSED

Helixline minimum: ≥97% · excellent at ≥99%

612,854

high-confidence genotype calls used from your chip-array data

616,214

assayed markers reviewed before endpoint-specific filtering

INPUT PROFILE High-density genotyping array, GRCh37 VCF · directly observed calls remain the stable evidence record for future refreshes

PREDICTED ABO BLOOD GROUP A · research only; Rh not inferred; confirm by serology

01 · YOUR DIRECT DNA DATA

Measured on a high-density genotyping array

Your directly measured genotype file is the primary evidence record. High-quality observed genotypes support your broad ancestry result, direct lineage markers, your endogamy measurement (runs of homozygosity) and other results that only need this array data.

The raw DNA does not change. Future reports may interpret it differently as reference panels, labels and validated models improve.

DATA QUALITY AND IMPUTATION

HIGH CONFIDENCE

612,854 submitted markers · statistical imputation available for 22 autosomes where an endpoint permits it

Fills statistical gaps, with limits

For results that passed their own accuracy checks, the pipeline can statistically fill in DNA positions your chip didn't directly read, using nearby positions it did read plus a large reference database. Each result only uses filled-in positions that clear its own quality bar.

Imputed is not directly measured. It can improve how much of your genome we can use, but it cannot by itself validate a state, community, physical trait or ancient-source model.

HELIXLINE REFERENCE RESOURCE

7,000+

DE-IDENTIFIED PARTICIPANTS IN THE BROADER PROPRIETARY COHORT

How state and community models use the cohort

The broader cohort supports research, model development and reference-panel improvement. It is **not** treated as one single pool used for everything. For each state, region or community result, only consented records that pass strict quality, labelling-accuracy, ancestry-relevance and relatedness checks are used.

Endpoint-specific evidence
The current broad-region benchmark used 31 eligible samples with canonical cleanroom holdout.

Your report label
State/community candidates are directional or low-confidence unless a page explicitly states that its exact endpoint passed validation.

YOUR PROFILE · YOUR REPORT HISTORY

Current report

Keep this PDF as the dated interpretation generated from your current chip-array genotype file.

Raw-data access

Your profile is the access point for the raw genotype-data files and report downloads provided with your account.

Refresh after 6 months

Return to your profile after six months to request a refreshed ancestry report using the latest eligible panel and model release.

How confidence labels protect interpretation: **High** means strong validation only for that specific result and level of detail-never 100% certainty. **Moderate** means a measured result with a meaningful margin of error. **Low** means exploratory context that must not be treated as identity, genealogy or an actionable conclusion. **Exploratory preview** is displayed only as clearly separated research context and is never used to support the main ancestry conclusion.

Why a refreshed result may change: an updated reference dataset can add more people like you, correct labels, or shift which fine-grained match is the best fit. A change reflects improved comparison evidence; it does not mean your DNA changed. Method validation sets the maximum confidence; sample quality can only lower it.

METHODS, REFERENCES AND LIMITATIONS

ANCESTRY TECHNICAL APPENDIX
HIGH CONFIDENCE
Model names, parameters and marker counts documented for independent review

HIGH CONFIDENCE
Methods, scientific references, limitations and update policy documented

CUSTOMER-SAFE SCIENTIFIC LEDGER

Modern ancestry

Frozen supervised K=6 coordinate model; 179,903 model markers and 1,459 reference anchors. Resolution is selected for the product, not per customer.

ADMIXTURE framework · Alexander et al. 2009

Ancient reference affinity

outgroup f3(Mbuti; target, reference) with 5 cM block jackknife across 111,649 direct markers. AADR groups and a labelled proxy are comparisons, not ancestors.

f-statistics · Patterson et al. 2012; Narasimhan et al. 2019

Source decomposition

ADMIXTOOLS 2 qpAdm vs AADR v66 ancient allele frequencies; fits accepted only at $p \geq 0.05$. Checked against standard ADMIXTOOLS output. Proportions are model fits, not counted ancestors.

qpAdm · Haak et al. 2015; ADMIXTOOLS 2 · Maier et al. 2023

Chromosome painting

A clearly labelled FLARE_v0.6 exploratory preview summarizes 193 segments across all 22 autosomes. 89.7% passed the preview's posterior threshold; exact tract boundaries remain approximate and the K=6 composition page remains the primary broad ancestry result.

Local ancestry · Browning et al. 2023

ANALYSIS RELEASE
General ancestry / high-density array input · v25-HL

REFERENCE RELEASE
HLX-2026.08-stage2-v2

IN-HOUSE MODELS
Region and community classifiers are Helixline-trained, not third-party, and abstain rather than guess.

CODE
72435e802092f2e905b49104f02c5e9e84a0f6d0

YOUR ANCESTRY REPORT LIFECYCLE

One DNA file, a versioned interpretation that can improve

01 · SOURCE
Original array genotype file
Your directly measured genotype record.

02 · QUALITY
Sample QC passed
99.45% call rate and endpoint filters.

03 · REPORT
Confidence-labelled PDF
Results tied to the current panel release.

04 · REFRESH
Updated after 6 months
Requested from your profile with the latest eligible release.

Usually no new sample is needed for a report refresh. The same quality-passing genotype file can be re-analysed. A new specimen or denser assay is relevant only when a feature specifically requires DNA coverage the current array does not provide.

REFRESH RE-CHECKS · BROAD ANCESTRY · STATE & COMMUNITY · ANCIENT SIMILARITY · CHROMOSOME PAINTING · NEW VALIDATED ENDPOINTS

Current snapshot: pipeline v25-HL · reference release HLX-2026.08-stage2-v2. The 7,000+ figure describes the broader proprietary resource, not the eligible sample count or validation performance of every endpoint. Endpoint-specific confidence labels in the report remain authoritative.

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DNA PROFILE CARD

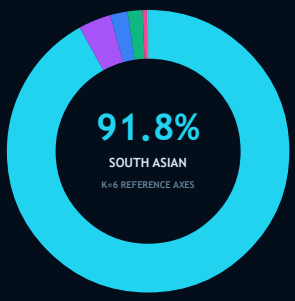
YOUR ANCESTRY SUMMARY

Demo User

SOUTH ASIAN

High-confidence broad ancestry classification

YOUR K=6 GENETIC PROFILE



South Asian	91.8%
West Asian · trace	3.7%
European-labelled K=6 axis · trace	2.0%
African · trace	1.7%

GENOTYPE DATA QUALITY · HIGH

99.45% call rate

616,214 directly genotyped markers · quality gate passed.

STRONGEST ANCIENT AFFINITY · HIGH FOR RANKING

Indus Periphery

Raw outgroup-f3 reference ranking, not a direct ancestor.

YOUR ANCIENT REFERENCE-AFFINITY RANKING

Direct-marker outgroup-f3 ranking · reference similarity only · not ancestry percentages or parts of a 100% mixture

#1 STRONGEST TESTED AFFINITY
Indus Periphery

0.07669 RAW OUTGROUP-f3

#2	Steppe Middle/Late Bronze Age	0.07578
#3	Iran Neolithic	0.07503
#4	AASI-related proxy (Onge)	0.07410

POPULATION TIMING · PUBLISHED CONTEXT
~1,900-4,200 years ago

Population context - not your measured date. Published ANI-ASI mixture window across 73 Indian groups (Moorjani et al. 2013). It is population history shown for context, not a measurement from your DNA and not a midpoint selected for you. No personal admixture date was estimated for this sample.

DIRECT LINES & GENOME SIGNALS

YOUR TWO DIRECT LINES

MATERNAL · mtDNA
M
one direct maternal line

PATERNAL · Y-DNA
H1a1d2c1
one direct paternal line

GENOME-WIDE MEASURES

ARCHAIC · MODERATE
1.7 ± 0.3%
f4-ratio · jackknife SE

CHROMOSOME PAINTING · PREVIEW
22 / 22
89.7% posterior-passing · research only

RUNS OF HOMOZYGOSITY
2.82%
36 segments · 81.3 Mb

PIGMENTATION MARKERS
38/41
marker evidence · no phenotype call

This ancestry summary is one view of your biological history; genetic results do not define your identity, culture, language or community. It prioritizes released broad ancestry, rank-only ancient affinity, measured direct-line placements and calibrated genome features. Low-confidence state/community calls and detailed model-dependent qpAdm proportions are intentionally excluded from this share card.

PROFILE EVIDENCE AT A GLANCE

VALIDATED RESULTS · LABELLED RESEARCH CONTEXT

99.76%	99.45%	111,456	22 / 22
BROAD CLASS	ARRAY CALL RATE	ANCIENT SNPs	CHROMOSOME PAINTING · PREVIEW
1253 held-out samples	Quality gate passed	Direct f3 ranking	89.7% posterior-passing · research only

Each number applies only to the endpoint and resolution named directly beneath it.

YOUR ORIGINS, DECODED

Your line. Your story.

Thank you for trusting us.

FOLLOW US • SHARE YOUR STORY

HELIXLINE

PERSONALIZED HEALTH GENETICS

HEALTH REPORT



DECODE

NAME **Demo User** · SEX **Male** · ISSUED **October 2026**

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WHAT THIS REPORT CONTAINS

PART 01 — Health Snapshot
A one-page overview of your trait, polygenic, and carrier findings, plus a glossary of the terms you will see.

PART 02 — Carrier Screening
Selected recessive-carrier, inherited-risk and low-penetrance variants, with each result category stated separately.

PART 03 — Polygenic Risk Scores
PGS-Catalog disease scores standardised against our Indian reference cohort - one full page per disease, with percentile + provenance.

PART 04 — Trait Pages
A divider page introducing the section, a snapshot table of all traits, then one full page per trait grouped by theme.

APPENDIX — Methodology & Limits
Data sources, scoring formulas, references, validation, and the limitations of every result above.

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PLEASE READ BEFORE USING THIS REPORT

Not a medical diagnosis. Every score is a statistical estimate - genetics nudges your odds, it does not decide them. Risk levels are computed from peer-reviewed effect sizes and standardised against our Indian reference cohort. · **Talk to a clinician before any change.** Do not start, stop or alter medication or screening based on this report alone. · **Privacy:** your raw DNA is processed only with explicit consent and never sold. · **Not for insurers or employers.** Do not share this report with third parties without physician review.

A SNAPSHOT OF YOUR GENETIC HEALTH

HELIXLINE
SCAN TO OPEN YOUR ACCOUNT
Sign in to view your reports

IF YOU READ NOTHING ELSE - YOUR TOP 3 TAKEAWAYS

1 DISCUSS WITH DOCTOR

- **Major Depressive Disorder** - 93rd percentile PRS
- **LDL Cholesterol Levels** - limited marker association
- **Psoriasis Susceptibility** - limited marker association

2 GENERAL HEALTH HABITS

- **Do not derive a care plan from these DNA results**
- **Follow broadly applicable healthy habits** within your abilities and clinician advice
- **Use symptoms and standard measurements** not a marker band, to guide care

3 PLAN ROUTINE CARE

- **Keep usual preventive care** for your age, history, symptoms, and local guidelines
- **Ask whether confirmatory testing is needed** before a finding affects care
- **Do not change screening or supplements** from this report alone

49 traits analysed • **12 higher-priority marker associations**

4 diseases scored • **1 elevated**

11 variants successfully screened • **0 flagged finding(s)**
1 further variant could not be assessed - not a negative result

MARKER-BASED HEALTH TRAITS	LOW CONFIDENCE	49 trait entries · 49 measured · 0 not measurable · 22 measured result(s) above the typical range	direct+imputed
POLYGENIC SCORES	EXPLORATORY	4 score(s), shown as a population percentile	direct+imputed
CARRIER SCREENING	LOW CONFIDENCE	11/12 panel variants screened · 0 carrier result(s) · 1 variant(s) not screened (those conditions are not reported either way)	direct+imputed

WHAT THIS REPORT TELLS YOU

We analysed your DNA through **three lenses** - genotype-associated **traits**, quality-qualified **polygenic scores**, and **selected clinically relevant variants**. The variant panel includes different inheritance and penetrance categories; it is not comprehensive carrier screening. This is not a diagnosis.

PRIORITY DISCUSSION POINTS

Findings worth flagging in a conversation with your doctor. These are signals to discuss - **not** diagnoses.

PRS — **Major Depressive Disorder** - **93rd percentile** · rank only; not an individual probability or care threshold

TRAIT — **LDL Cholesterol Levels** - **Association detected in this limited marker panel** · Multi-marker panel · Association detected in this limited marker panel

TRAIT — **Psoriasis Susceptibility** - **Association detected in this limited marker panel** · C/T · Association detected in this limited marker panel

+ 10 further findings flagged in this report. Your complete set of findings appears later in this report, in the trait summary and the category pages.

This report contains three different kinds of genetic information. They do not have the same meaning or level of certainty. Your main discussion points are shown above; the complete set appears later in this report.

Relative genetic rank
Your position in a named reference distribution - not a personal probability.

Marker association
A limited association from the markers assessed - not a personal disease probability.

Named-variant screen
Only the listed variants were assessed - a negative does not rule out the condition.

A QUICK WORD ON THE TERMS YOU WILL SEE

Percentile
Your rank among 100 South Asians lined up from lowest to highest. 80th = higher than 80% of them.

Association band
A qualitative marker or panel category. It is not an individual probability, diagnosis, or treatment threshold.

For wellness and educational use only. This report does not diagnose disease and is not a substitute for medical care. Discuss any flagged finding with a qualified clinician before changing medication, diet, or screening routine. Genetic findings reflect inherited predisposition - not certainty.

SELECTED VARIANTS

02

A limited marker panel covering recessive carrier, X-linked, susceptibility and low-penetrance findings. These categories are reported separately and are not interchangeable.

HELIXLINE

1 WHY IT MATTERS

This panel includes different biological categories. A recessive carrier result can inform reproductive counselling; an inherited-risk or low-penetrance result instead describes susceptibility and is not carrier status or a diagnosis.

2 WHAT WE TEST

Named, quality-qualified variants prioritised for South Asian relevance. Each result applies only to the **specific screened marker**; conditions requiring CNV, repeat or broader sequencing are marked not assessed.

3 HOW TO READ IT

Each row shows the named marker and whether it was detected. **Not detected is not the same as not a carrier** for the whole gene or condition. Positive findings require clinical confirmation.

WHAT YOU'LL SEE NEXT

A table of every listed condition, its result category, named variant, population context, and interpretation limits.

SELECTED CLINICALLY RELEVANT VARIANT SCREENING

HELIXLINE

LIMITED NAMED-VARIANT CARRIER SCREEN

This panel includes recessive carrier variants plus a small number of inherited risk or low-penetrance variants. These categories are not interchangeable. A carrier finding is most useful **before pregnancy**; a risk-variant finding describes susceptibility, not carrier status or a diagnosis. Only quality-qualified calls receive a result.

12	11	1
VARIANTS LISTED	INTERPRETED	NOT SCREENED

1 panel variants were not interpreted. They were absent or below the required imputation-quality threshold; this is not a negative result. See the detail pages for each withheld marker.

NAMED VARIANT NOT DETECTED

For each condition below: This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

- ✓ **Sickle Cell Disease** — HBB
- ✓ **G6PD Deficiency** — G6PD
- ✓ **Galactosemia (Classic)** — GALT
- ✓ **Thrombophilia (Inherited)** — F5
- ✓ **Hereditary Haemochromatosis** — HFE
- ✓ **Phenylketonuria (PKU)** — PAH
- ✓ **Beta Thalassemia** — HBB
- ✓ **Wilson Disease** — ATP7B
- ✓ **Hereditary Hearing Loss (DFNB1)** — GJB2
- ✓ **Thrombophilia (Inherited)** — F2
- ✓ **HFE H63D Variant (low penetrance)** — HFE

For informational purposes only - not medical advice. "Not detected" applies only to the named variant and does not exclude the condition or other disease-associated variants. Confirm positive findings clinically and consult a qualified genetic counsellor or clinician before reproductive or medical decisions.

HELIXLINE

EVERY VARIANT WE SCREENED

Sickle Cell Disease

Misshapen red blood cells that block circulation.

`HBB · rs334 (p.Glu7Val (HbS))`

AUTOSOMAL RECESSIVE

ClinVar VCV000015333.178 · verified 2026-07-25

YOUR GENOTYPE

T/T

GRCh37 genomic-forward pathogenic allele: **A**

GRCh37 chr11:5248232
VCF T>A · direct array call

India: Up to 35% in some tribal populations (Gond, Bhil, Madia)

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

Beta Thalassemia

Reduced haemoglobin, causing lifelong anaemia.

`HBB · rs33915217 (IVS1-5 (G>C))`

AUTOSOMAL RECESSIVE

ClinVar VCV000015447.139 · verified 2026-07-25

YOUR GENOTYPE

C/C

GRCh37 genomic-forward pathogenic allele: **G**

GRCh37 chr11:5248155
VCF C>G · direct array call

India: 3-4% (Indian general population)

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

G6PD Deficiency

Enzyme shortage that can trigger anaemia from certain foods and drugs.

`G6PD · rs5030868 (Mediterranean (p.Ser188Phe))`

X-LINKED RECESSIVE

ClinVar VCV000100057.129 · verified 2026-07-25

YOUR GENOTYPE

G/G

GRCh37 genomic-forward pathogenic allele: **A**

GRCh37 chrX:153762634
VCF G>A · direct array call

India: 5-10% males in some Indian groups

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

Wilson Disease

Copper builds up and damages the liver and brain.

`ATP7B · rs76151636 (p.His1069Gln)`

AUTOSOMAL RECESSIVE

ClinVar VCV000003848.120 · verified 2026-07-25

YOUR GENOTYPE

G/G

GRCh37 genomic-forward pathogenic allele: **T**

GRCh37 chr13:52518281
VCF G>T · direct array call

India: ~1 in 90 (Indian general population)

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

HELIXLINE

EVERY VARIANT WE SCREENED

Galactosemia (Classic)

The body cannot break down galactose (milk sugar).

`GALT · rs75391579 (p.Gln188Arg)`

AUTOSOMAL RECESSIVE

ClinVar VCV000003614.139 · verified 2026-07-25

YOUR GENOTYPE

A/A

GRCh37 genomic-forward pathogenic allele: **G**

GRCh37 chr9:34648167
VCF A>G · direct array call

India: ~1 in 150 (estimated South Asian)

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

Hereditary Hearing Loss (DFNB1)

Inherited hearing loss present from birth.

`GJB2 · rs72474224 (p.Val37Ile (c.109G>A))`

AUTOSOMAL RECESSIVE

ClinVar VCV000017023.156 · verified 2026-07-25

YOUR GENOTYPE

C/C

GRCh37 genomic-forward pathogenic allele: **T**

GRCh37 chr13:20763612
VCF C>T · direct array call

India: ~2–4% East/South Asian populations

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

Thrombophilia (Inherited)

A raised tendency to form abnormal blood clots.

`F5 · rs6025 (p.Arg506Gln (Factor V Leiden))`

AUTOSOMAL DOMINANT (VARIABLE PENETRANCE)

ClinVar VCV000000642.133 · verified 2026-08-06

YOUR GENOTYPE

C/C

GRCh37 genomic-forward pathogenic allele: **T**

GRCh37 chr1:169519049
VCF T>C · direct array call

India: ~1–2% (Indian general population)

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

Thrombophilia (Inherited)

A raised tendency to form abnormal blood clots.

`F2 · rs1799963 (c.*97G>A (Prothrombin G20210A))`

AUTOSOMAL DOMINANT (VARIABLE PENETRANCE)

ClinVar VCV000013310.154 · verified 2026-07-25

YOUR GENOTYPE

G/G

GRCh37 genomic-forward pathogenic allele: **A**

GRCh37 chr11:46761055
VCF G>A · direct array call

India: ~0.5% (Indian general population)

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

HELIXLINE

EVERY VARIANT WE SCREENED

Hereditary Haemochromatosis

Iron overload that can damage organs over time.

`HFE · rs1800562 (p.Cys282Tyr)`

AUTOSOMAL RECESSIVE

ClinVar VCV000000009.144 · verified 2026-07-25

YOUR GENOTYPE

G/G

GRCh37 genomic-forward pathogenic allele: **A**

GRCh37 chr6:26093141
VCF G>A · direct array call

India: Very rare in South Asians (~0.2% allele frequency, ~0.4% heterozygous carriers per 1000G SAS, n=489). Common in Northern Europeans (~6% allele frequency, ~12% heterozygous).

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

HFE H63D Variant (low penetrance)

A common HFE variant. On its own it is not associated with iron overload.

`HFE · rs1799945 (p.His63Asp)`

AUTOSOMAL RECESSIVE (LOW PENETRANCE)

ClinVar VCV000000010.138 · verified 2026-07-25

YOUR GENOTYPE

C/C

GRCh37 genomic-forward pathogenic allele: **G**

GRCh37 chr6:26091179
VCF C>G · direct array call

India: ~15% heterozygous (H63D allele ~8%: LASI-DAD 8.4%, Moorjani 2025; gnomAD SAS 7.8%)

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

Phenylketonuria (PKU)

The body cannot process the amino acid phenylalanine.

`PAH · rs5030858 (p.Arg408Trp (c.1222C>T))`

AUTOSOMAL RECESSIVE

ClinVar VCV000000577.149 · verified 2026-07-25

YOUR GENOTYPE

G/G

GRCh37 genomic-forward pathogenic allele: **A**

GRCh37 chr12:103234271
VCF G>A · direct array call

India: ~1 in 100 (European general); lower in South Asian

NAMED VARIANT NOT DETECTED

This named variant was not detected. This does not rule out the condition or other disease-causing variants in the same gene.

Cystic Fibrosis

Thick mucus affecting the lungs and digestion.

`CFTR · rs113993960 (p.Phe508del (ΔF508))`

AUTOSOMAL RECESSIVE

ClinVar VCV000007105.208 · verified 2026-07-25

YOUR GENOTYPE

–

GRCh37 genomic-forward pathogenic allele: **D**

Alleles unavailable · not screened

India: 1 in 40 (Indian general population)

NOT SCREENED

Variant not on array This is not a negative result.

HELIXLINE

CONDITIONS THIS TEST DOES NOT ASSESS

FOUR CONDITIONS WERE NOT ASSESSED

Carrier screening often includes the four conditions below. This report does not assess any of them. Each one is caused by a whole gene copy being missing or duplicated, or by a short piece of DNA repeating far too many times - changes a genotyping array cannot read, whatever the quality of your sample. We did not test you for these four, so nothing in this report says anything about them, in either direction.

NOT ASSESSED BY THIS PANEL

Alpha Thalassemia

HBA1

NOT ASSESSED

Caused by one or more copies of the alpha-globin genes being missing, rather than by a single letter of DNA being changed.

To assess it: A targeted copy-number test (gap-PCR or MLPA) can assess it.

Spinal Muscular Atrophy

SMN1

NOT ASSESSED

Caused by a missing copy of the SMN1 gene, sitting beside an almost identical neighbouring gene an array cannot tell it from.

To assess it: An SMN1 copy-number test (MLPA or ddPCR) can assess it.

Fragile X Syndrome

FMR1

NOT ASSESSED

Caused by a short piece of DNA repeating far too many times. An array reads single letters, not how often a sequence repeats.

To assess it: A repeat-sizing test (TP-PCR, or a Southern blot) can assess it.

Duchenne / Becker Muscular Dystrophy

DMD

NOT ASSESSED

Most often caused by whole sections of the DMD gene being missing or duplicated, rather than by a single letter being changed.

To assess it: MLPA or exon-level sequencing can assess it.

This is not a result. It does not imply a finding in either direction. These four conditions were not tested here, so your carrier status for them is unknown.

If you are planning a pregnancy, or any of these conditions runs in your family, the test named beside each condition is the one that can answer it. A clinician or genetic counsellor can arrange the right one and read it alongside your family history.

HELIXLINE

POLYGENIC RISK SCORES

03

Most common diseases emerge from many small genetic effects acting together. A polygenic risk score combines quality-passing variants and ranks the score against its versioned, marker-matched South Asian reference artifact.

1 WHAT A PRS IS

A **weighted sum** of quality-passing dosages from the named scoring artifact, ranked against a versioned South Asian reference. The report separates markers observed in your DNA from any small missing tail completed at the reference-panel mean; a percentile requires at least 95% of reference markers to be observed.

2 WHAT IT TELLS YOU

Where you sit in the inherited-risk distribution: **top 5%**, **top 20%**, **middle 60%**, or **bottom 20%**. A high score is a meaningful inherited push - not a verdict.

3 WHY ANCESTRY MATTERS

Effect sizes and predictive performance vary across populations. Each disease page names its scoring source and ancestry evidence. Ranking against a South Asian reference does **not** fully correct portability or calibration bias.

WHAT YOU'LL SEE NEXT

A summary card of the PRSs available for this sample, then one full-page deep-dive per score - with percentile, marker coverage, provenance, limitations, and preventive-care context.

YOUR RELATIVE GENETIC RANK - AT A GLANCE

HELIXLINE

WHAT IS A POLYGENIC RISK SCORE?

Common conditions such as type 2 diabetes, hypertension, asthma, and major depressive disorder are influenced by many variants. A Polygenic Risk Score (PRS) combines them into one number, then ranks you against a reference population. A score at the **80th percentile** means your inherited genetic loading is higher than 80 of 100 people - it does **not** mean you will develop the condition.

Below Average	Average	Above Average	High
< 20th pctl	20th - 80th pctl	80th - 95th pctl	above 95th pctl

What this percentile is: This percentile shows your relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk. **Limitations:** PRS accuracy varies by trait. A South Asian comparison distribution changes who the score is ranked against; it does **not** by itself establish transportability, calibration, or clinical validity. A percentile is shown only when the source has published evaluation evidence for the customer's broad genetic ancestry and the reference population matches it. Treat results as **research-informed relative ranks**, not clinical predictions.

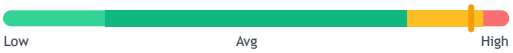
Major Depressive Disorder

PGS000138

UPPER RELATIVE RANK

93rd percentile

the Helixline South Asian reference panel



20,563 markers observed ★★★

Asthma

PGS004724

MIDDLE REFERENCE RANGE

68th percentile

the Helixline South Asian reference panel



1,989,661 markers observed ★★★

Type 2 Diabetes

PGS005377

LOWER RELATIVE RANK

10th percentile

the Helixline South Asian reference panel



1,164,968 markers observed ★★★

SCORES NOT REPORTED FOR YOU · MEASURED SCORES & OTHER LIMITATIONS

NOT MEASURABLE

Hypertension

This score could not be produced for you. No percentile, risk category or average value has been substituted.

No percentile, risk category or average value has been substituted.

For informational purposes only - not medical advice and not a clinical diagnosis. Polygenic risk scores were developed primarily in European cohorts and have reduced predictive performance in South Asians. Consult a qualified clinician before making any health decisions based on these scores.

MAJOR DEPRESSIVE DISORDER

HELIXLINE

YOUR GENETIC LOADING

UPPER RELATIVE RANK

<20th | 20-80th | 80-95th | 95th+

Your score is at the **93rd percentile** of the Helixline South Asian reference panel - **Upper relative rank**. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

GENETIC RANK

93rd percentile

rank within the Helixline South Asian reference panel

lower | middle | upper

PGS CATALOG ID

PGS000138

Percentile and relative genetic loading only. No individual lifetime-risk percentage is inferred.

ABOUT THE MAJOR DEPRESSIVE DISORDER PRS

This calculation used **20,563 quality-passing variants** from a marker-matched subset of the **PGS000138** PGS Catalog scoring file. The published catalog score contains **22,272 variants**; 20,563 were present in the versioned scoring/reference subset used for this run. Each observed variant carries its published per-allele weight; your weighted allele sum is standardised against **the versioned South Asian reference cohort** using the same 20,563 markers - so the percentile reflects your rank on the exact score computed for you.

★★★ HIGH MARKER COVERAGE · 20,563 USED / 22,272 CATALOG (92.3% CATALOG COVERAGE; 100% OF SCREENED PASSED QC)

500+ quality-passing markers used. Clinical validity still depends on ancestry, external validation and calibration.

YOUR GENETIC RANK AMONG 100 SOUTH ASIANS

93rd percentile



◆ marks your position - **93rd percentile** within the Helixline South Asian reference panel. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

SCIENTIFIC PROVENANCE

PEER-REVIEWED SOURCE

SCORING SOURCE

Cai N et al. (2020) Nat Genet 52:1046. PMID:32231276

ANCESTRY VALIDATION

European (UK Biobank); cross-ancestry PRS transfer for MDD is limited

20,563	20,563	1.52	μ=0.33
MARKERS SCREENED	MARKERS USED	Z-SCORE	INDIAN REFERENCE

WHAT THIS MEANS FOR YOU

Your score ranks at the 93rd percentile in the coverage-matched Indian/South Asian reference cohort (elevated relative category). This is an exploratory relative position in the named reference cohort. The score's discovery evidence has not been independently validated for the broad genetic ancestry inferred from this sample, so the percentile and category must not be interpreted as an absolute disease probability, diagnosis, or treatment threshold.

WHAT TO DO WITH THIS SCORE

Upper relative rank in the Helixline South Asian reference panel. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

HOW TO USE THIS RESULT

- ✓ Do not change screening, medication, or supplements based on this PRS rank alone.
- ✓ Interpret the score with age, family history, ancestry, biomarkers, and a validated clinical risk model.
- ✓ Use current local clinical guidelines; confirm any medically important result with an appropriate clinical test.

For informational purposes only - not medical advice. Consult a qualified clinician before making any health decisions based on genetic findings.

ASTHMA

HELIXLINE

YOUR GENETIC LOADING

MIDDLE REFERENCE RANGE



Your score is at the **68th percentile** of the Helixline South Asian reference panel - **Middle reference range**. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

GENETIC RANK

68th percentile

rank within the Helixline South Asian reference panel



PGS CATALOG ID

PGS004724

Percentile and relative genetic loading only. No individual lifetime-risk percentage is inferred.

ABOUT THE ASTHMA PRS

This calculation used **1,989,661 quality-passing variants** from a marker-matched subset of the **PGS004724** PGS Catalog scoring file. The published catalog score contains **2,303,253 variants**; 1,989,661 were present in the versioned scoring/reference subset used for this run. Each observed variant carries its published per-allele weight; your weighted allele sum is standardised against **the versioned South Asian reference cohort** using the same 1,989,661 markers - so the percentile reflects your rank on the exact score computed for you.

★★★ **HIGH MARKER COVERAGE · 1,989,661 USED / 2,303,253 CATALOG (86.4% CATALOG COVERAGE; 100% OF SCREENED PASSED QC)**
500+ quality-passing markers used. Clinical validity still depends on ancestry, external validation and calibration.

YOUR GENETIC RANK AMONG 100 SOUTH ASIANS

68th percentile



◆ marks your position - **68th percentile** within the Helixline South Asian reference panel. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

SCIENTIFIC PROVENANCE

PEER-REVIEWED SOURCE

SCORING SOURCE
Truong B et al. (2024) Cell Genom 4:100523. Integrative PRS improves prediction accuracy of complex traits. PMID:38508198. DOI:10.1016/j.xgen.2024.100523

ANCESTRY VALIDATION
100% South Asian development + 100% South Asian evaluation (PRSmix, Cell Genom 2024; harmonized to GRCh37 by EBI)

1,989,661	1,989,661	0.46	μ=1.75
MARKERS SCREENED	MARKERS USED	Z-SCORE	INDIAN REFERENCE

WHAT THIS MEANS FOR YOU

Your score ranks at the 68th percentile in the coverage-matched Indian/South Asian reference cohort (average relative category).

WHAT TO DO WITH THIS SCORE
Middle reference range in the Helixline South Asian reference panel. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

HOW TO USE THIS RESULT

- ✓ Do not change screening, medication, or supplements based on this PRS rank alone.
- ✓ Interpret the score with age, family history, ancestry, biomarkers, and a validated clinical risk model.
- ✓ Use current local clinical guidelines; confirm any medically important result with an appropriate clinical test.

For informational purposes only - not medical advice. Consult a qualified clinician before making any health decisions based on genetic findings.

TYPE 2 DIABETES

HELIXLINE

YOUR GENETIC LOADING

LOWER RELATIVE RANK

<20th | 20-80th | 80-95th | 95th+

Your score is at the **10th percentile** of the Helixline South Asian reference panel - **Lower relative rank**. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

GENETIC RANK

10th percentile

rank within the Helixline South Asian reference panel

lower | middle | upper

PGS CATALOG ID

PGS005377

Percentile and relative genetic loading only. No individual lifetime-risk percentage is inferred.

ABOUT THE TYPE 2 DIABETES PRS

This calculation used **1,164,968 quality-passing variants** from a marker-matched subset of the **PGS005377** PGS Catalog scoring file. The published catalog score contains **1,327,177 variants**; 1,164,968 were present in the versioned scoring/reference subset used for this run. Each observed variant carries its published per-allele weight; your weighted allele sum is standardised against **the versioned South Asian reference cohort** using the same 1,164,968 markers - so the percentile reflects your rank on the exact score computed for you.

★★★ HIGH MARKER COVERAGE · 1,164,968 USED / 1,327,177 CATALOG (87.8% CATALOG COVERAGE; 100% OF SCREENED PASSED QC)

500+ quality-passing markers used. Clinical validity still depends on ancestry, external validation and calibration.

YOUR GENETIC RANK AMONG 100 SOUTH ASIANS

10th percentile



◆ marks your position - **10th percentile** within the Helixline South Asian reference panel. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

SCIENTIFIC PROVENANCE

PEER-REVIEWED SOURCE

SCORING SOURCE

Huerta-Chagoya A et al. (2026) Lancet Diabetes Endocrinol. Multi-ancestry PRS for prediction of T2D and complications. PMID:42061389. DOI:10.1016/S2213-8587(25)00374-9

ANCESTRY VALIDATION

100% South Asian GWAS + 100% South Asian development + 100% South Asian evaluation (PRS-CSx)

1,164,968	1,164,968	-1.25	μ=0.73
MARKERS SCREENED	MARKERS USED	Z-SCORE	INDIAN REFERENCE

WHAT THIS MEANS FOR YOU

Your score ranks at the 10th percentile in the coverage-matched Indian/South Asian reference cohort (low relative category).

WHAT TO DO WITH THIS SCORE

Lower relative rank in the Helixline South Asian reference panel. This is a relative position in the named reference distribution. It is not a diagnosis or an estimate of your individual absolute disease risk.

HOW TO USE THIS RESULT

- ✓ Do not change screening, medication, or supplements based on this PRS rank alone.
- ✓ Interpret the score with age, family history, ancestry, biomarkers, and a validated clinical risk model.
- ✓ Use current local clinical guidelines; confirm any medically important result with an appropriate clinical test.

For informational purposes only - not medical advice. Consult a qualified clinician before making any health decisions based on genetic findings.

HYPERTENSION

NOT MEASURABLE

Hypertension

This score could not be produced for you. No percentile, risk category or average value has been substituted.

Reason: **no percentile could be produced for this score** QC state: blocking.

No percentile, band, risk category or average value has been substituted for this score.

WHAT THIS TECHNICAL STATUS MEANS

The model remains listed so the report accounts for every contracted score. No midpoint or “average” value was inserted. A future report may become measurable after a versioned scoring asset and matching reference distribution pass the release checks.

WHAT THIS MEANS

Do not interpret this missing result as lower, average, or higher genetic rank. Continue routine care using symptoms, medical and family history, and standard clinical measurements; retain this report for possible reanalysis after a validated replacement score and matching reference distribution become available.

HELIXLINE

HELIXLINE

YOUR TRAITS

04

49 traits measured across 9 health categories - every finding in one place before the detailed pages.

01 WHAT A TRAIT IS

A trait is a single biological characteristic - like cholesterol response, lactose tolerance, or vitamin D handling - shaped by one or a few well-studied genes. We score traits across 9 themes from cardiometabolic to wellness.

02 HOW WE SCORE

Each result uses the marker model defined for that trait: a directly observed genotype or a limited multi-marker panel. These trait associations are not presented as calibrated polygenic risk scores.

03 HOW TO READ

Each row shows the observed genotype or panel and a qualitative association band. Single- and few-marker traits do not receive phenotype percentiles. Detailed cards and conservative health context follow.

HELIXLINE

WHAT YOU'LL SEE NEXT

A summary table of every trait - then one detailed page per trait grouped by theme: cardiometabolic, nutrition, cancer, neuro, autoimmune, fitness, reproductive, bone, wellness.

SUMMARY OF YOUR TRAITS

HELIXLINE

QUICK SCAN

Every trait measured, grouped by category and sorted by association band. **GENOTYPE / PANEL** shows the observed call; single- and few-marker traits do not have calibrated phenotype percentiles. **WHAT IT MEANS** summarises each finding. These are associations, not diagnoses, and do not set care.

#	TRAIT & GENES	RISK BAND	GENOTYPE / %	WHAT IT MEANS
	CARDIOMETABOLIC HEALTH			
1	**Genetic Tendency Toward Lower HDL-C** CETP + LIPC + LPL TIER A	MORE RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	In this limited panel, 5 risk-associated allele copies were observed across the assessed markers (CETP + LIPC + LPL). This is not an estimate of personal disease probability.
2	**Total Cholesterol Tendency** CELSR2 + PPP1R3B + APOA1 + LDLR + APOE TIER B	MORE RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	A cluster of variants associated with higher average total cholesterol. Worth a fasting lipid panel if you have not had one recently.
3	**Coronary Artery Disease & Atherosclerosis Risk** CDKN2B-AS1 + LDLR TIER B	MORE RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	A higher burden of CAD-associated alleles. Discuss overall cardiovascular risk with a clinician this variant panel is one input among many.
4	**LDL Cholesterol Levels** APOE TIER A	ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL	Multi-marker panel GENOTYPE	APOE ε3/ε4 heterozygote - modestly elevated LDL (~10-15 mg/dL over ε3/ε3). Earlier lipid intervention warranted
5	**Elevated Triglycerides (APOC3 Variant)** APOC3 TIER A	ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL	C/G GENOTYPE	One copy of the variant C allele (SstI 3238G in mRNA). Slightly elevated APOC3 transcript stability → modestly higher fasting and post-prandial triglycerides. Cardiometabolic risk modestly amplified.
6	**Type 2 Diabetes Genetic Risk** TCF7L2 + MNTR1B + KCNQ1 + IRS1 + IGF2BP2 + SLC30A8 + CDKN2B + CDKAL1 + HHEX + GCKR + MC4R + FTO TIER B	ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL	Multi-marker panel GENOTYPE	More T2D risk alleles than average. Weight, activity and diet have a large effect on whether this genetic tendency ever expresses.
7	**Triglyceride Levels** APOA5 + APOC3 + LPL + GCKR TIER A	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	Typical TG panel.
8	**Lipoprotein(a) - Cardiovascular Marker** LPA TIER A	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	A/A GENOTYPE	Two copies of the common A allele. Lp(a) is expected to be near population baseline. No additional cardiovascular risk amplification from this locus.
9	**Gout & Uric Acid Risk** ABCG2 TIER A	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	G/G GENOTYPE	G/G at ABCG2 rs2231142 - reference (higher urate excretion). Baseline hyperuricaemia risk.
10	**Hypothyroidism Genetic Tendency** FOXE1 + SH2B3 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	A typical thyroid-susceptibility variant profile.

ASSOCIATION BANDS Qualitative categories from the observed genotype/panel; not phenotype percentiles or diagnoses.

For informational purposes only - not medical advice and not a clinical diagnosis. Trait risks here are population-level signals; lifestyle, family history, and clinical biomarkers all carry more individual predictive weight. Consult a qualified clinician before making any health decisions based on these findings.

#	TRAIT & GENES	RISK BAND	GENOTYPE / %	WHAT IT MEANS
	CARDIOMETABOLIC HEALTH			
11	**Hyperthyroidism Genetic Tendency** CTLA4 + LOC101930098 + LOC105378120 TIER B	FEWER RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	Fewer autoimmune-thyroid susceptibility alleles.
	CANCER RISK			
12	**Lung Cancer & Nicotine Dependence** CHRNA3 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	G/G GENOTYPE	G/G at CHRNA3 rs1051730 - reference genotype. Indian tobacco exposure is often via bidis, hookah, and gutkha - all elevate lung cancer risk. Even non-smokers exposed to household biomass smoke or urban air pollution have measurable lung cancer risk.
	NEUROLOGICAL HEALTH			
13	**Caffeine Metabolism** CYP1A2 TIER A	VARIANT PRESENT	C/A GENOTYPE	One copy of the A allele at CYP1A2 - intermediate caffeine metabolism. Moderate consumers (2-3 cups/day) usually tolerate fine
14	**Sleep Duration Tendency** ABCC9 TIER B	VARIANT PRESENT	G/A GENOTYPE	One copy of the A allele at ABCC9 - slight shift toward shorter natural sleep duration (~10-20 min less per night on average). Maintain good sleep hygiene total hours matter for cognitive performance regardless of genotype.
15	**Migraine Predisposition** TRPM8 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	T/C GENOTYPE	Heterozygous at TRPM8. Lifestyle triggers (sleep, hydration, dietary triggers) dominate manifestation.
16	**Alcohol Dependence & Metabolism Tendency** ADH1B + PKNOX2 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	A typical alcohol-metabolism variant profile.
17	**Narcolepsy Genetic Susceptibility** TCRA TIER B	FEWER RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	Does not carry the TCRA susceptibility allele.
	AUTOIMMUNE & INFLAMMATION			
18	**Psoriasis Susceptibility** HLA-Cw6 TIER A	ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL	C/T GENOTYPE	One copy of the T risk allele at HLA-C - modestly elevated lifetime psoriasis risk. Most carriers never develop psoriasis trigger factors (stress, infection, smoking) interact with the genetic background.
19	**Rheumatoid Arthritis Risk** HLA-DRB1 + PTPN22 + STAT4 TIER A	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	Single low-effect risk allele. Typical population RA risk.
20	**Chronic Urticaria (Hives) Susceptibility** LILRA3 + IGHG1/2 + TDGF1 + HLA-G + PTPN22 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	A typical immune-reactivity variant profile.

For informational purposes only - not medical advice and not a clinical diagnosis. Trait risks here are population-level signals; lifestyle, family history, and clinical biomarkers all carry more individual predictive weight. Consult a qualified clinician before making any health decisions based on these findings.

#	TRAIT & GENES	RISK BAND	GENOTYPE / %	WHAT IT MEANS
	AUTOIMMUNE & INFLAMMATION			
21	**Oxidative Stress & Antioxidant Capacity** SOD2 + CAT + GPX1 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	A typical antioxidant-enzyme profile.
22	**Celiac Disease Risk** HLA-DQA1 1 of 2 declared markers assessed TIER A	FEWER RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	No common HLA-DQ2.5/DQ8 tagging combination was identified by this two-marker proxy. This lowers genetic susceptibility but is not full HLA typing and does not exclude celiac disease.
	NUTRITION & METABOLISM			
23	**Lactose Intolerance** MCM6 / LCT TIER A	VARIANT PRESENT	G/G GENOTYPE	Two copies of the lactase non-persistence allele (G). You are likely to experience lactose intolerance symptoms (bloating, cramps, diarrhoea) after consuming significant amounts of lactose. This is the typical genotype across South Asian populations (~75-90%).
24	**Iron Deficiency Anaemia Susceptibility** TMPRSS6 + TF TIER A	MORE RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	Multi-locus high-risk iron-deficiency panel.
25	**Dietary Protein Response Tendency** FTO TIER B	MORE RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	Profile associated with a stronger-than-average response to higher-protein intake for appetite control and body composition.
26	**Omega-3 Fatty Acid Metabolism** FADS1 TIER A	VARIANT NOT PRESENT	C/C GENOTYPE	C/C at FADS1 rs174546 - slower ALA → EPA/DHA conversion. Most South Asians are C-carriers.
27	**Folate Processing** MTHFR TIER A	VARIANT NOT PRESENT	Multi-marker panel GENOTYPE	Single MTHFR variant carrier (C677T or A1298C heterozygote). Enzyme activity ~65-80% of normal. Adequate green leafy vegetables (methi, palak) + B12 sufficiency is enough.
28	**Body Composition & Obesity Predisposition** UCP1 + ADRB3 + FTO + MC4R + SH2B1 + TMEM18 + NEGR1 + KCTD15 + TFAP2B + QPCTL + NUDT3, RPS10-NUDT3 + LINC01741 - SEC16B + ZC3H4 + CADM2 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	A typical spread of body-composition variants.
29	**Gut Sensitivity & IBS Genetic Tendency** IL10 + TLR9 + SI + HTR3E + KLB + IL4 + TNF + GRID2IP + TNFSF15 + CDH1 TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	A typical inflammatory and gut-barrier variant profile.
30	**Susceptibility to Lower Vitamin D Levels** GC 1 of 3 declared markers assessed TIER A	FEWER RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	No highlighted association was found in the assessed markers. This does not establish a lower personal probability or rule out the condition.

For informational purposes only - not medical advice and not a clinical diagnosis. Trait risks here are population-level signals; lifestyle, family history, and clinical biomarkers all carry more individual predictive weight. Consult a qualified clinician before making any health decisions based on these findings.

#	TRAIT & GENES	RISK BAND	GENOTYPE / %	WHAT IT MEANS
NUTRITION & METABOLISM				
31	**Salt Sensitivity & Blood Pressure Tendency** AGT + NPPA + ADD1 TIER B	FEWER RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	Less salt-sensitive variant profile.
FITNESS & PERFORMANCE				
32	**Muscle Recovery** IL6 TIER B	VARIANT PRESENT	G/G GENOTYPE	G/G at IL6 - highest IL-6 response, potentially slower recovery from intense exercise. Prioritise sleep, anti-inflammatory foods (haldi/turmeric, adrak/ginger), and adequate protein for recovery.
33	**Muscle Power & Strength Potential** IGF2 + MSTN TIER B	VARIANT PRESENT	Multi-marker panel GENOTYPE	A power-oriented natural profile - you may find explosive and strength-focused training especially rewarding.
34	**Connective Tissue & Injury Resilience** COL5A1 + COL1A1 TIER B	VARIANT PRESENT	Multi-marker panel GENOTYPE	A collagen profile associated with higher reported soft-tissue and ligament injury rates. Prioritise gradual load progression, eccentric strengthening and recovery.
35	**Power vs Endurance Profile** ACTN3 TIER A	VARIANT NOT PRESENT	T/T GENOTYPE	T/T at ACTN3 - X/X (loss-of-function homozygote). Slight endurance advantage. ~10% of South Asians. Doesn't preclude power sports but marginally favours endurance running / cycling / swimming.
36	**VO2 Max Trainability** PPARGC1A TIER B	VARIANT NOT PRESENT	C/C GENOTYPE	No copies of the T allele at PPARGC1A rs8192678 - population-baseline VO2 max trainability. Cardio training produces typical improvement curves.
REPRODUCTIVE HEALTH				
37	**Testosterone & Male Hormonal Balance Tendency** SHBG TIER B	MORE RISK-ASSOCIATED MARKERS OBSERVED	Multi-marker panel GENOTYPE	Variant profile associated with lower free-testosterone availability.
BONE & JOINT HEALTH				
38	**Joint & Disc Degeneration Risk** COL6A4P1 + NCOA3 + COL2A1 TIER B	ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL	Multi-marker panel GENOTYPE	Association detected in the assessed marker panel. This is not an estimate of personal disease probability.
39	**Bone Mineral Density & Osteoporosis Risk** SP7 + LRP5 + COL1A1 + FAM3C TIER B	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	A typical bone-density variant profile.
WELLNESS & TRAITS				
40	**Bitter Taste Perception** TAS2R38 TIER A	VARIANT PRESENT	G/C GENOTYPE	G/C at TAS2R38 rs713598 is associated with intermediate bitter perception. A full PAV/AVI haplotype requires additional markers and was not measured.

For informational purposes only - not medical advice and not a clinical diagnosis. Trait risks here are population-level signals; lifestyle, family history, and clinical biomarkers all carry more individual predictive weight. Consult a qualified clinician before making any health decisions based on these findings.

#	TRAIT & GENES	RISK BAND	GENOTYPE / %	WHAT IT MEANS
	WELLNESS & TRAITS			
41	**Eye Colour Pigmentation** OCA2 / HERC2 TIER A	VARIANT PRESENT	G/A GENOTYPE	One copy of the G allele at OCA2/HERC2 - intermediate iris pigmentation hazel or light brown eyes possible. The variant is uncommon in Indians (mostly seen in admixed individuals).
42	**Glaucoma Predisposition (Open-Angle)** CDKN2B-AS1 TIER A	ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL	T/T GENOTYPE	T/T at CDKN2B-AS1 (9p21) is associated with increased POAG susceptibility in population studies.
43	**Skin Ageing & Sun Sensitivity** ASIP + AHR + ELN + FOXC1 + SLC2A4 TIER B	VARIANT PRESENT	Multi-marker panel GENOTYPE	Some variants associated with greater sun sensitivity or earlier visible ageing.
44	**Age-Related Cataract Risk** EPHA2 + CRYAA TIER B	VARIANT PRESENT	Multi-marker panel GENOTYPE	Variant present in the assessed markers (EPHA2 + CRYAA).
45	**Hair Texture** TCHH TIER A	VARIANT NOT PRESENT	A/A GENOTYPE	A/A at TCHH rs11803731 - wavy/curly hair tendency (more common in Indian populations). No clinical implications.
46	**Skin UV Sensitivity** MC1R TIER B	VARIANT NOT PRESENT	C/C GENOTYPE	C/C at MC1R rs1805007 - wild-type MC1R standard melanin response to UV. Sun-tan response normal.
47	**Hair Greying Onset** IRF4 TIER A	VARIANT NOT PRESENT	C/C GENOTYPE	Two copies of the common C allele. Typical age-of-onset for greying pigmentation maintained longer.
48	**Age-Related Vision Loss (Macular Degeneration)** CFH + ARMS2 TIER A	NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED	Multi-marker panel GENOTYPE	Single risk allele - modestly elevated AMD risk.
49	**Sleep Quality & Disturbance Tendency** ADORA2A + CLOCK + DRD2 + TNF + PER2 TIER B	VARIANT NOT PRESENT	Multi-marker panel GENOTYPE	A typical sleep-regulation variant profile.

For informational purposes only - not medical advice and not a clinical diagnosis. Trait risks here are population-level signals; lifestyle, family history, and clinical biomarkers all carry more individual predictive weight. Consult a qualified clinician before making any health decisions based on these findings.

TRAIT PAGES

04

Higher-signal findings receive a dedicated page; typical and informational findings are grouped densely without dropping any qualified result.

HELIXLINE

1 WHAT IT IS

A **trait** is a single biological characteristic - like cholesterol response, lactose tolerance, or vitamin D handling - shaped by one or a few well-studied genes. We score traits across 9 themes.

2 HOW WE SCORE

Each result uses the marker model defined for that trait: a directly observed genotype or a limited multi-marker panel. These associations are not presented as calibrated polygenic risk scores.

3 HOW TO READ THE PAGES

Every trait page shows the observed genotype or multi-marker panel result, the published effect direction, what the association means biologically, and conservative health context. Single-marker traits do not receive invented percentiles.

WHAT YOU'LL SEE NEXT

Dedicated pages for higher-signal findings, followed by complete compact pages for every remaining qualified trait across all health themes.

CARDIOMETABOLIC HEALTH

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

Heart, blood vessels, glucose, lipids - the foundation of long-term wellness.

Your cardiovascular and metabolic genetics govern how your heart pumps, how your arteries age, and how your body handles glucose and fat. Variants in these genes interact silently with diet, exercise, and stress for decades before clinical risk surfaces - making early awareness your most powerful tool. The traits here cover the five leading causes of premature death in South Asian populations.

11	6	4	1
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
11 trait entries · 11 measured · 6 measured result(s) above the typical range			

YOUR GENETIC STORY IN THIS SECTION

This limited panel found **3 higher-priority marker associations** (Genetic Tendency Toward Lower HDL-C, Total Cholesterol Tendency, Coronary Artery Disease & Atherosclerosis Risk) and **3 additional associations** (LDL Cholesterol Levels, Elevated Triglycerides (APOC3 Variant), Type 2 Diabetes Genetic Risk) in this category. Cardiometabolic disease burden varies across populations and is strongly influenced by clinical measurements, family history, environment, and lifestyle. The limited marker associations in this section do not quantify personal disease probability or prove that risks compound. Fewer risk-associated markers were observed at Hyperthyroidism Genetic Tendency. This limited panel does not rule out the condition or account for non-genetic risk factors.

TRAITS IN THIS SECTION

11 TOTAL

1 LDL Cholesterol Levels

APOE ε3/ε4 heterozygote - modestly elevated LDL (~10-15 mg/dL over ε3/ε3). Earlier lipid intervention warranted

APOE TIER A

ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL

Multi-marker panel

2 Triglyceride Levels

Typical TG panel.

APOA5 + APOC3 + LPL + GCKR TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

Multi-marker panel

3 Lipoprotein(a) - Cardiovascular Marker

Two copies of the common A allele. Lp(a) is expected to be near population baseline. No additional cardiovascular risk amplification from this locus.

LPA TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

A/A genotype

4 Elevated Triglycerides (APOC3 Variant)

One copy of the variant C allele (SstI 3238G in mRNA). Slightly elevated APOC3 transcript stability → modestly higher fasting and post-prandial triglycerides. Cardiometabolic risk modestly amplified.

APOC3 TIER A

ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL

C/G genotype

5 Gout & Uric Acid Risk

G/G at ABCG2 rs2231142 - reference (higher urate excretion). Baseline hyperuricaemia risk.

ABCG2 TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

G/G genotype

6 Genetic Tendency Toward Lower HDL-C

In this limited panel, 5 risk-associated allele copies were observed across the assessed markers (CETP + LIPC + LPL). This is not an estimate of personal disease probability.

CETP + LIPC + LPL TIER A

MORE RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

7 Type 2 Diabetes Genetic Risk

More T2D risk alleles than average. Weight, activity and diet have a large effect on whether this genetic tendency ever expresses.

TCF7L2 + MNTR1B + KCNQ1 + IRS1 + IGF2BP2 + SLC30A8 + CDKN2B + CDKAL1 + HHEX + GCKR + MC4R + FTO TIER B

ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL

Multi-marker panel

8 Total Cholesterol Tendency

A cluster of variants associated with higher average total cholesterol. Worth a fasting lipid panel if you have not had one recently.

CELSR2 + PPP1R3B + APOA1 + LDLR + APOE TIER B

MORE RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

9 Coronary Artery Disease & Atherosclerosis Risk

A higher burden of CAD-associated alleles. Discuss overall cardiovascular risk with a clinician this variant panel is one input among many.

CDKN2B-AS1 + LDLR TIER B

MORE RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

10 Hyperthyroidism Genetic Tendency

Fewer autoimmune-thyroid susceptibility alleles.

CTLA4 + LOC101930098 + LOC105378120 TIER B

FEWER RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

11 Hypothyroidism Genetic Tendency

A typical thyroid-susceptibility variant profile.

FOXE1 + SH2B3 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

Multi-marker panel

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

LDL CHOLESTEROL LEVELS

APOE ε3/ε4 (heterozygous elevated)

HELIXLINE

YOUR RESULT

Association detected in this limited marker panel

Genotype association · no calibrated percentile

GENE PANEL

APOE

TIER A · SA VALIDATED

Markers analysed · rs429358 · rs7412

ABOUT THIS TRAIT

LDL (low-density lipoprotein) is the 'bad cholesterol' - it deposits cholesterol in artery walls, driving atherosclerosis. Genetic variation at APOE (ε2/ε3/ε4 haplotype) is one of the strongest determinants of baseline LDL levels. In South Asians, ε4 carriers typically have 10-15 mg/dL higher LDL than ε3/ε3, and ε4/ε4 homozygotes 25-40 mg/dL higher. This matters more for Indians because SA-typical atherogenic small-dense LDL, high Lp(a), and low HDL amplify the LDL burden into premature coronary disease - a hallmark of the Indian cardiac phenotype.

YOUR GENETIC PROFILE

RESULT TYPE
Multi-marker panel

ASSOCIATION BAND
Association detected in this limited marker panel

SA RISK-ALLELE FREQ
8.7%
1000G SAS (n=489)

WHAT THIS MEANS FOR YOU

APOE ε3/ε4 heterozygote - modestly elevated LDL (~10-15 mg/dL over ε3/ε3). Earlier lipid intervention warranted

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 20686565 *Marker association; clinical utility and transferability may be limited.*

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL
Multi-marker panel
rs429358 · rs7412

EVIDENCE CONTEXT
South-Asian replicated
Rajkumar 2011 (INDICO cohort), Bhattacharyya 2019 - APOE haplotype replicated as major LDL determinant in Indians.

CONFIRMATION BOUNDARY
Not diagnostic
A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION
~28% urban Indians have LDL >130 mg/dL (ICMR-CARRS, ICMR-INDIAB); APOE ε4 carrier frequency ~10% in South Asians.

SOUTH-ASIAN CONTEXT
~28% urban South Asians have LDL requiring intervention

HELIXLINE

ELEVATED TRIGLYCERIDES (APOC3 VARIANT)

Modestly elevated triglyceride drive

HELIXLINE

YOUR RESULT

Association detected in this limited marker panel

Genotype association · no calibrated percentile

GENE

APOC3

TIER A · SA VALIDATED

Markers analysed · rs5128

ABOUT THIS TRAIT

APOC3 (apolipoprotein C-III) inhibits lipoprotein lipase - the enzyme that clears triglycerides from blood. The SstI variant (rs5128) common in South Asians raises APOC3 transcript stability, indirectly raising fasting and post-prandial triglycerides. This variant is a secondary TG modifier compared to the primary APOA5/LPL/GCKR panel; it adds meaningful information for individuals with borderline TG results.

YOUR GENETIC PROFILE

GENOTYPE	RISK-ALLELE COPIES	SA RISK-ALLELE FREQ
C/G	1 of 2	66.2%
		1000G SAS (n=489)

WHAT THIS MEANS FOR YOU

One copy of the variant C allele (SstI 3238G in mRNA). Slightly elevated APOC3 transcript stability → modestly higher fasting and post-prandial triglycerides. Cardiometabolic risk modestly amplified.

Observed genotype **C/G**. The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 25928461 *Marker association; clinical utility and transferability may be limited.*

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL

C/G

The genomic C allele shown here is the same variant the literature calls the APOC3 SstI "3238G" allele; APOC3 lies on the minus strand, so the two notations describe one allele.

rs5128

EVIDENCE CONTEXT

South-Asian replicated

APOC3 rs5128 (SstI) replicated in Indian dyslipidaemia cohorts; interacts strongly with high-carb + high-fat Indian dietary patterns.

CONFIRMATION BOUNDARY

Not diagnostic

A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION

~40% urban Indians have TG >150 mg/dL. The risk allele reported here (genomic C, called the SstI "3238G" allele in the mRNA notation used by much of the literature) is common in South Asians; its measured frequency is shown with this result.

SOUTH-ASIAN CONTEXT

APOC3 G-carriers show 10-20% higher fasting TG on average

GENETIC TENDENCY TOWARD LOWER HDL-C

High-risk HDL panel (multi-locus)

HELIXLINE

YOUR RESULT

More risk-associated markers observed

Genotype association · no calibrated percentile

GENE PANEL

CETP + LIPC + LPL

TIER A · SA VALIDATED

Markers analysed · rs708272 · rs1800588 · rs328

ABOUT THIS TRAIT

HDL (high-density lipoprotein) is the 'good cholesterol' - it clears cholesterol from artery walls back to the liver. Low HDL is the signature Indian dyslipidaemia pattern, present in 60-70% of urban Indians even at normal LDL. The CETP + LIPC + LPL variants combined modulate HDL levels; low-HDL genotypes are common in South Asians. This matters because low HDL predicts cardiovascular events independently of LDL, especially in the SA phenotype where atherogenic small-dense LDL and elevated Lp(a) coexist.

YOUR GENETIC PROFILE

RESULT TYPE
Multi-marker panel

ASSOCIATION BAND
More risk-associated markers observed

SA RISK-ALLELE FREQ
55.2%
1000G SAS (n=489)

WHAT THIS MEANS FOR YOU

In this limited panel, 5 risk-associated allele copies were observed across the assessed markers (CETP + LIPC + LPL). This is not an estimate of personal disease probability.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 20686565 *Marker association; clinical utility and transferability may be limited.*

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL
Multi-marker panel
rs708272 · rs1800588 · rs328

EVIDENCE CONTEXT
South-Asian replicated
CETP Taq1B (Bhanushali 2010, 2013), LIPC and LPL variants replicated in Indian metabolic-syndrome cohorts.

CONFIRMATION BOUNDARY
Not diagnostic
A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION
60-70% urban Indians have low HDL (<40 mg/dL male, <50 female) - the highest global rate (ICMR-CARRS).

SOUTH-ASIAN CONTEXT
~65% urban South Asian adults have suboptimal HDL

HELIXLINE

TYPE 2 DIABETES GENETIC RISK

Above-average T2D genetic load

HELIXLINE

YOUR RESULT

Association detected in this limited marker panel

Genotype association · no calibrated percentile

GENE PANEL

TCF7L2 + MNTR1B + KCNQ1 + IRS1 + IGF2BP2 + SLC30A8 + CDKN2B + CDKAL1 + HHEX + GCKR + MC4R + FTO

TIER B · REPLICATED

Markers analysed · **16** - all listed in Your evidence record below

ABOUT THIS TRAIT

Type 2 diabetes is strongly polygenic: dozens of common variants each nudge insulin secretion, insulin sensitivity or beta-cell reserve. This panel reads well-replicated loci including TCF7L2 (the largest common-variant effect on T2D), FTO, MC4R, CDKAL1, IGF2BP2 and KCNQ1. South Asians develop T2D at younger ages and lower BMI than European populations, so genetic load sits on top of an already-elevated background risk. Genotype is not diagnosis: fasting glucose and HbA1c remain the tests that matter.

YOUR GENETIC PROFILE

RESULT TYPE

Multi-marker panel

ASSOCIATION BAND

Association detected in this limited marker panel

WHAT THIS MEANS FOR YOU

More T2D risk alleles than average. Weight, activity and diet have a large effect on whether this genetic tendency ever expresses.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 16415884 *Marker association; clinical utility and transferability may be limited.*

Evidence context (continued): The marker panel and per-genotype effect directions come from a curated research genotype-scoring model, so this citation supports the lead association rather than every marker in the panel. All GRCh37 coordinates were independently verified against the 1000G Phase 3 reference (2026-07-31).

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL

Multi-marker panel

rs12255372 · rs10830963 · rs1387153 · rs7903146 · rs2237892 · rs2237897 · rs2943641 · rs4402960 · rs13266634 · rs10811661 · rs7756992 · rs5015480 · rs780094 · rs17782313 · rs9939609 · rs8050136

EVIDENCE CONTEXT

Transferability limited

Primary-locus citation: TCF7L2 (Grant 2006, Nat Genet) (PMID:16415884), verified against PubMed.

CONFIRMATION BOUNDARY

Not diagnostic

A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION

~11% of Indian adults have diabetes; another ~15% have prediabetes (ICMR-INDIAB).

SOUTH-ASIAN CONTEXT

Elevated baseline T2D risk across South Asian populations

TOTAL CHOLESTEROL TENDENCY

Markedly higher cholesterol tendency

HELIXLINE

YOUR RESULT

More risk-associated markers observed

Genotype association · no calibrated percentile

GENE PANEL

`CELSR2 + PPP1R3B + APOA1 + LDLR + APOE`

TIER B · REPLICATED

Markers analysed · **6** - all listed in Your evidence record below

ABOUT THIS TRAIT

Total cholesterol reflects the combined pool of LDL, HDL and remnant lipoproteins. This panel reads common variants near APOE, APOA1, LDLR, CELSR2 and PPP1R3B that shift lifelong average cholesterol. Effects are modest individually but additive, and they act on top of diet, activity and body composition - a lipid profile remains the only way to know your actual numbers.

YOUR GENETIC PROFILE

RESULT TYPE
Multi-marker panel

ASSOCIATION BAND
More risk-associated markers observed

WHAT THIS MEANS FOR YOU

A cluster of variants associated with higher average total cholesterol. Worth a fasting lipid panel if you have not had one recently.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 20686565 *Marker association; clinical utility and transferability may be limited.*

Evidence context (continued): The marker panel and per-genotype effect directions come from a curated research genotype-scoring model, so this citation supports the lead association rather than every marker in the panel. All GRCh37 coordinates were independently verified against the 1000G Phase 3 reference (2026-07-31).

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL
Multi-marker panel
rs646776 · rs9987289 · rs964184 · rs6511720 · rs629301 · rs4420638

EVIDENCE CONTEXT
Transferability limited
Primary-locus citation: blood-lipid loci (Teslovich 2010, Nature) (PMID:20686565), verified against PubMed.

CONFIRMATION BOUNDARY
Not diagnostic
A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION
~25-30% of urban Indian adults have raised total cholesterol.

SOUTH-ASIAN CONTEXT
Dyslipidaemia is common and often under-detected in Indian adults

HELIXLINE

CORONARY ARTERY DISEASE & ATHEROSCLEROSIS RISK

HELIXLINE

High CAD variant load

YOUR RESULT

More risk-associated markers observed

Genotype association · no calibrated percentile

GENE PANEL

CDKN2B-AS1 + LDLR

TIER B · REPLICATED

Markers analysed · rs10757274 · rs1122608 · rs2228671

ABOUT THIS TRAIT

The 9p21 locus (CDKN2B-AS1/ANRIL) is the most consistently replicated common-variant association with coronary artery disease across world populations, and acts largely independently of cholesterol. Paired here with LDLR variants that influence LDL handling. Common variants shift population-level odds; they do not diagnose disease and do not override blood pressure, lipids, smoking status or family history.

YOUR GENETIC PROFILE

RESULT TYPE

Multi-marker panel

ASSOCIATION BAND

More risk-associated markers observed

WHAT THIS MEANS FOR YOU

A higher burden of CAD-associated alleles. Discuss overall cardiovascular risk with a clinician this variant panel is one input among many.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 17478681 *Marker association; clinical utility and transferability may be limited.*

Evidence context (continued): The marker panel and per-genotype effect directions come from a curated research genotype-scoring model, so this citation supports the lead association rather than every marker in the panel. All GRCh37 coordinates were independently verified against the 1000G Phase 3 reference (2026-07-31).

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL

Multi-marker panel

rs10757274 · rs1122608 · rs2228671

EVIDENCE CONTEXT

Transferability limited

Primary-locus citation: 9p21 (McPherson 2007, Science) (PMID:17478681), verified against PubMed.

CONFIRMATION BOUNDARY

Not diagnostic

A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION

CAD affects Indians ~10 years earlier on average than European populations.

SOUTH-ASIAN CONTEXT

Premature CAD is a leading cause of adult mortality in India

HELIXLINE

CANCER PREDISPOSITION

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

Common-variant predisposition for cancers most relevant to Indian cohorts.

Common variants can be associated with cancer susceptibility at population level, but this limited marker panel is not a validated personal cancer-risk model or an early-detection test. Age, family history, exposures, symptoms and established screening guidance remain the basis for clinical care.

1	0	1	0
TRAITS 1 trait entries · 1 measured	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS

TRAITS IN THIS SECTION

1 TOTAL

1

Lung Cancer & Nicotine Dependence

G/G at CHRNA3 rs1051730 - reference genotype. Indian tobacco exposure is often via bidis, hookah, and gutkha - all elevate lung cancer risk. Even non-smokers exposed to household biomass smoke or urban air pollution have measurable lung cancer risk.

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

G/G genotype

Lung cancer is the leading cause of cancer death worldwide. The CHRNA3 rs1051730 variant is associated with heavier smoking behaviour and modestly higher lung cancer risk in smokers. In India, tobacco exposure is often via bidis, hookah, gutkha, and khaini - all with distinct carcinogen profiles vs Western cigarettes. Non-smokers exposed to household biomass smoke or urban air pollution (Delhi PM2.5 often 200+) also carry measurable lung cancer risk. Complete tobacco cessation eliminates most of this genetic-plus-environmental risk over 10-15 years.

CHRNA3 TIER B

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

Higher-signal findings receive dedicated detail pages; all other qualified traits remain in the personalized compact section.

TAILORED TO THIS SAMPLE
Only quality-qualified direct or imputed calls enter the findings above.

NO SILENT SUBSTITUTION
An unrelated marker or population average is never used to fill a missing result.

CLINICAL BOUNDARY
These associations support discussion; they do not diagnose disease or set treatment.

NEUROLOGICAL & COGNITIVE

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

Brain, mood, sleep - the genetics of cognition and emotion.

Neurological and psychiatric traits are complex and often highly polygenic. The selected-marker associations shown here are research context, not diagnoses, calibrated probabilities, or predictions of an individual outcome.

5	0	4	1
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
5 trait entries · 5 measured · 2 measured result(s) above the typical range			

YOUR GENETIC STORY IN THIS SECTION

Fewer risk-associated markers were observed at Narcolepsy Genetic Susceptibility. This limited panel does not rule out the condition or account for non-genetic risk factors.

TRAITS IN THIS SECTION

5 TOTAL

1 **Caffeine Metabolism**
One copy of the A allele at CYP1A2 - intermediate caffeine metabolism. Moderate consumers (2-3 cups/day) usually tolerate fine
CYP1A2 TIER A
VARIANT PRESENT
C/A genotype

2 **Sleep Duration Tendency**
One copy of the A allele at ABCC9 - slight shift toward shorter natural sleep duration (~10-20 min less per night on average). Maintain good sleep hygiene total hours matter for cognitive performance regardless of genotype.
ABCC9 TIER B
VARIANT PRESENT
G/A genotype

3 **Migraine Predisposition**
Heterozygous at TRPM8. Lifestyle triggers (sleep, hydration, dietary triggers) dominate manifestation.
TRPM8 TIER B
NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
T/C genotype

4 **Narcolepsy Genetic Susceptibility**
Does not carry the TCRA susceptibility allele.
TCRA TIER B
FEWER RISK-ASSOCIATED MARKERS OBSERVED
Multi-marker panel

5 **Alcohol Dependence & Metabolism Tendency**
A typical alcohol-metabolism variant profile.
ADH1B + PKNOX2 TIER B
NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
Multi-marker panel

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

Higher-signal findings receive dedicated detail pages; all other qualified traits remain in the personalized compact section.

HELIXLINE

AUTOIMMUNE & INFLAMMATION

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

How your immune system tolerates self vs reacts to environment.

Immune-related variants can be associated with population-level susceptibility, but they do not determine whether an individual will develop an autoimmune condition. This limited marker panel is not a diagnostic or clinical risk assessment.

5	1	3	1
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
5 trait entries · 5 measured · 1 measured result(s) above the typical range			

YOUR GENETIC STORY IN THIS SECTION

This limited panel found **1 marker association** in this category: **Psoriasis Susceptibility**. HLA-linked autoimmune traits share a common inflammation axis. Environmental triggers (infection, gut, stress) matter as much as HLA - genetic risk without a trigger typically stays silent. Fewer risk-associated markers were observed at Celiac Disease Risk. This limited panel does not rule out the condition or account for non-genetic risk factors.

TRAITS IN THIS SECTION

5 TOTAL

1 **Celiac Disease Risk**
No common HLA-DQ2.5/DQ8 tagging combination was identified by this two-marker proxy. This lowers genetic susceptibility but is not full HLA typing and does not exclude celiac disease.
HLA-DQA1 · 1 of 2 declared markers assessed · TIER A
FEWER RISK-ASSOCIATED MARKERS OBSERVED
Multi-marker panel

2 **Rheumatoid Arthritis Risk**
Single low-effect risk allele. Typical population RA risk.
HLA-DRB1 + PTPN22 + STAT4 · TIER A
NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
Multi-marker panel

3 **Psoriasis Susceptibility**
One copy of the T risk allele at HLA-C - modestly elevated lifetime psoriasis risk. Most carriers never develop psoriasis trigger factors (stress, infection, smoking) interact with the genetic background.
HLA-Cw6 · TIER A
ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL
C/T genotype

4 **Chronic Urticaria (Hives) Susceptibility**
A typical immune-reactivity variant profile.
LILRA3 + IGHG1/2 + TDGF1 + HLA-G + PTPN22 · TIER B
NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
Multi-marker panel

5 **Oxidative Stress & Antioxidant Capacity**
A typical antioxidant-enzyme profile.
SOD2 + CAT + GPX1 · TIER B
NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
Multi-marker panel

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

Higher-signal findings receive dedicated detail pages; all other qualified traits remain in the personalized compact section.

PSORIASIS SUSCEPTIBILITY

HLA-C heterozygous risk

HELIXLINE

YOUR RESULT

Association detected in this limited marker panel

Genotype association · no calibrated percentile

GENE

HLA-Cw6

TIER A · SA VALIDATED 4.66× per-allele or

Markers analysed · rs10484554

ABOUT THIS TRAIT

Psoriasis is a chronic immune-mediated skin disease causing scaly, inflamed plaques on scalp, elbows, knees, and other sites. HLA-Cw6 is the strongest genetic driver - carriers have 10-15× baseline risk. In India, psoriasis prevalence is 0.4-2.8%, higher in the North than the South. Triggers include streptococcal throat infection, stress, cold-dry winters (North India), and certain medications. Modern treatments (biologics, targeted small molecules) can control even severe disease.

YOUR GENETIC PROFILE

GENOTYPE	RISK-ALLELE COPIES	SA RISK-ALLELE FREQ
C/T	1 of 2	19.6%
		1000G SAS (n=489)

WHAT THIS MEANS FOR YOU

One copy of the T risk allele at HLA-C - modestly elevated lifetime psoriasis risk. Most carriers never develop psoriasis trigger factors (stress, infection, smoking) interact with the genetic background.

Observed genotype C/T. The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 19169254 *Marker association; clinical utility and transferability may be limited.*

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL
C/T
rs10484554

EVIDENCE CONTEXT
South-Asian replicated
HLA-Cw6 (rs10484554) replicated in Indian psoriasis cohorts (PGIMER Chandigarh).

CONFIRMATION BOUNDARY
Not diagnostic
A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION
0.4-2.8% Indian adults have psoriasis; higher in northern populations (Dogra 2013 Br J Dermatol).

SOUTH-ASIAN CONTEXT
~1.5% lifetime psoriasis risk in South Asians

NUTRITION & METABOLISM

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

How your body absorbs, processes and uses food and nutrients.

Nutritional genomics examines research associations between selected variants and traits such as nutrient metabolism or food tolerance. This limited marker panel does not measure nutrient status or prescribe an optimal diet; symptoms and standard clinical measurements remain more informative for care.

9	2	5	2
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
9 trait entries · 9 measured · 3 measured result(s) above the typical range			

YOUR GENETIC STORY IN THIS SECTION

This limited panel found **2 higher-priority marker associations** in this category: **Iron Deficiency Anaemia Susceptibility, Dietary Protein Response Tendency**. Vegetarian diets common in India interact with these findings - e.g. B12 status depends on FUT2 AND diet, not either alone. Annual biomarkers beat intermittent big changes. Fewer risk-associated markers were observed at Susceptibility to Lower Vitamin D Levels, Salt Sensitivity & Blood Pressure Tendency. This limited panel does not rule out the condition or account for non-genetic risk factors.

TRAITS IN THIS SECTION

9 TOTAL

1 Lactose Intolerance

VARIANT PRESENT

G/G genotype

Two copies of the lactase non-persistence allele (G). You are likely to experience lactose intolerance symptoms (bloating, cramps, diarrhoea) after consuming significant amounts of lactose. This is the typical genotype across South Asian populations (~75-90%).

MCM6 / LCT TIER A

2 Omega-3 Fatty Acid Metabolism

VARIANT NOT PRESENT

C/C genotype

C/C at FADS1 rs174546 - slower ALA → EPA/DHA conversion. Most South Asians are C-carriers.

FADS1 TIER A

3 Iron Deficiency Anaemia Susceptibility

MORE RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

Multi-locus high-risk iron-deficiency panel.

TMPRSS6 + TF TIER A

4 Folate Processing

VARIANT NOT PRESENT

Multi-marker panel

Single MTHFR variant carrier (C677T or A1298C heterozygote). Enzyme activity ~65-80% of normal. Adequate green leafy vegetables (methi, palak) + B12 sufficiency is enough.

MTHFR TIER A

5 Susceptibility to Lower Vitamin D Levels

FEWER RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

No highlighted association was found in the assessed markers. This does not establish a lower personal probability or rule out the condition.

GC 1 of 3 declared markers assessed TIER A

6 Body Composition & Obesity Predisposition

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

Multi-marker panel

A typical spread of body-composition variants.

UCP1 + ADRB3 + FTO + MC4R + SH2B1 + TMEM18 + NEGR1 + KCTD15 + TFAP2B + QPCTL + NUDT3, RPS10-NUDT3 + LINC01741 - SEC16B + ZC3H4 + CADM2 TIER B

7 Gut Sensitivity & IBS Genetic Tendency

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

Multi-marker panel

A typical inflammatory and gut-barrier variant profile.

IL10 + TLR9 + SI + HTR3E + KLB + IL4 + TNF + GRID2IP + TNFSF15 + CDH1 TIER B

8 Salt Sensitivity & Blood Pressure Tendency

FEWER RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

Less salt-sensitive variant profile.

AGT + NPPA + ADD1 TIER B

9 Dietary Protein Response Tendency

MORE RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

Profile associated with a stronger-than-average response to higher-protein intake for appetite control and body composition.

FTO TIER B

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

IRON DEFICIENCY ANAEMIA SUSCEPTIBILITY

High-risk iron-deficiency panel

HELIXLINE

YOUR RESULT

More risk-associated markers observed

Genotype association · no calibrated percentile

GENE PANEL

TMPRSS6 + TF

TIER A · SA VALIDATED

Markers analysed · rs855791 · rs3811647

ABOUT THIS TRAIT

Iron deficiency anaemia (IDA) is a state where reduced iron stores impair haemoglobin synthesis, causing fatigue, pallor, and reduced cognitive/physical performance. India has the world's highest anaemia burden - 57% women and 25% men (NFHS-5, 2021) - driven by menstrual losses, pregnancy, vegetarian diets low in heme iron, and poor iron absorption from plant sources. TMPRSS6 regulates hepcidin (the iron-absorption master switch); TF encodes transferrin (iron transport).

YOUR GENETIC PROFILE

RESULT TYPE
Multi-marker panel

ASSOCIATION BAND
More risk-associated markers observed

SA RISK-ALLELE FREQ
54.3%
1000G SAS (n=489)

WHAT THIS MEANS FOR YOU

Multi-locus high-risk iron-deficiency panel.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 33054130 *Marker association; clinical utility and transferability may be limited.*

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL
Multi-marker panel
rs855791 · rs3811647

EVIDENCE CONTEXT
South-Asian replicated
TMPRSS6 V736A (Sarkate 2016), TF rs3811647 (Benyamin 2014 SA sub-analysis) - replicated in Indian anaemia cohorts.

CONFIRMATION BOUNDARY
Not diagnostic
A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION
57% Indian women and 25% men are anaemic (NFHS-5, 2021) - the highest global rate.

SOUTH-ASIAN CONTEXT
~57% South Asian women have anaemia (Hb <12 g/dL)

DIETARY PROTEIN RESPONSE TENDENCY

Strong protein response

HELIXLINE

YOUR RESULT

More risk-associated markers observed

Genotype association · no calibrated percentile

GENE PANEL

FTO

TIER B · REPLICATED

Markers analysed · rs9939609 · rs1558902

ABOUT THIS TRAIT

FTO variants influence satiety signalling and appetite, and several trials report that higher-protein diets produce greater weight and body-composition improvement in carriers of the risk alleles. This trait reads the FTO locus specifically for that diet-response signal - the same gene appears in the body-composition panel because FTO genuinely has pleiotropic effects on both.

YOUR GENETIC PROFILE

RESULT TYPE

Multi-marker panel

ASSOCIATION BAND

More risk-associated markers observed

WHAT THIS MEANS FOR YOU

Profile associated with a stronger-than-average response to higher-protein intake for appetite control and body composition.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 22891219 *Marker association; clinical utility and transferability may be limited.*

Evidence context (continued): The marker panel and per-genotype effect directions come from a curated research genotype-scoring model, so this citation supports the lead association rather than every marker in the panel. All GRCh37 coordinates were independently verified against the 1000G Phase 3 reference (2026-07-31).

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL

Multi-marker panel

rs9939609 · rs1558902

EVIDENCE CONTEXT

Transferability limited

Primary-locus citation: FTO x diet, POUNDS LOST (Zhang 2012, Diabetes) (PMID:22891219), verified against PubMed.

CONFIRMATION BOUNDARY

Not diagnostic

A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION

FTO risk alleles are common across South Asian populations.

SOUTH-ASIAN CONTEXT

Protein intake is often below optimal in predominantly cereal-based Indian diets

FITNESS & PERFORMANCE

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

Athletic phenotype, training response and injury susceptibility.

Research has linked selected variants with muscle, oxygen-use and connective-tissue traits at population level. These associations do not predict an individual training response, performance ceiling, injury, or recovery plan.

5	0	5	0
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
5 trait entries · 5 measured · 3 measured result(s) above the typical range			

TRAITS IN THIS SECTION

5 TOTAL

1 Power vs Endurance Profile

T/T at ACTN3 - X/X (loss-of-function homozygote). Slight endurance advantage. ~10% of South Asians. Doesn't preclude power sports but marginally favours endurance running / cycling / swimming.

ACTN3 TIER A

VARIANT NOT PRESENT
T/T genotype

2 VO2 Max Trainability

No copies of the T allele at PPARGC1A rs8192678 - population-baseline VO2 max trainability. Cardio training produces typical improvement curves.

PPARGC1A TIER B

VARIANT NOT PRESENT
C/C genotype

3 Muscle Recovery

G/G at IL6 - highest IL-6 response, potentially slower recovery from intense exercise. Prioritise sleep, anti-inflammatory foods (haldi/turmeric, adrak/ginger), and adequate protein for recovery.

IL6 TIER B

VARIANT PRESENT
G/G genotype

4 Muscle Power & Strength Potential

A power-oriented natural profile - you may find explosive and strength-focused training especially rewarding.

IGF2 + MSTN TIER B

VARIANT PRESENT
Multi-marker panel

5 Connective Tissue & Injury Resilience

A collagen profile associated with higher reported soft-tissue and ligament injury rates. Prioritise gradual load progression, eccentric strengthening and recovery.

COL5A1 + COL1A1 TIER B

VARIANT PRESENT
Multi-marker panel

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

Higher-signal findings receive dedicated detail pages; all other qualified traits remain in the personalized compact section.

REPRODUCTIVE HEALTH

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

Hormonal traits relevant to androgen balance and long-term health.

Reproductive and hormonal genomics covers the regulation of sex-hormone levels, binding-protein availability and the downstream effects on muscle mass, bone density, energy and fertility. These variants interact strongly with age, body composition, sleep and metabolic health, all of which move hormone levels far more than genotype does. Blood testing remains the only way to establish your actual hormonal status.

1	1	0	0
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
1 trait entries · 1 measured · 1 measured result(s) above the typical range			

YOUR GENETIC STORY IN THIS SECTION

This limited panel found **1 higher-priority marker association** in this category: **Testosterone & Male Hormonal Balance Tendency**.

TRAITS IN THIS SECTION

1 TOTAL

1

Testosterone & Male Hormonal Balance Tendency

Variant profile associated with lower free-testosterone availability.

MORE RISK-ASSOCIATED MARKERS OBSERVED

Multi-marker panel

Most circulating testosterone is bound to sex hormone-binding globulin (SHBG); only the unbound fraction is biologically active. SHBG variants shift total and free testosterone levels, and are associated with metabolic syndrome risk in men. These variants influence the set-point - they do not measure your hormone levels, which requires a morning serum testosterone test.

SHBG TIER B

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT

These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE

Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES

Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED

Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

Higher-signal findings receive dedicated detail pages; all other qualified traits remain in the personalized compact section.

TAILORED TO THIS SAMPLE

Only quality-qualified direct or imputed calls enter the findings above.

NO SILENT SUBSTITUTION

An unrelated marker or population average is never used to fill a missing result.

CLINICAL BOUNDARY

These associations support discussion; they do not diagnose disease or set treatment.

TESTOSTERONE & MALE HORMONAL BALANCE TENDENCY

Markedly lower free-testosterone availability

HELIXLINE

YOUR RESULT

More risk-associated markers observed

Genotype association · no calibrated percentile

GENE PANEL

SHBG

TIER B · REPLICATED

Markers analysed · rs12150660 · rs6258

ABOUT THIS TRAIT

Most circulating testosterone is bound to sex hormone-binding globulin (SHBG); only the unbound fraction is biologically active. SHBG variants shift total and free testosterone levels, and are associated with metabolic syndrome risk in men. These variants influence the set-point - they do not measure your hormone levels, which requires a morning serum testosterone test.

YOUR GENETIC PROFILE

RESULT TYPE
Multi-marker panel

ASSOCIATION BAND
More risk-associated markers observed

WHAT THIS MEANS FOR YOU

Variant profile associated with lower free-testosterone availability.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 21998597 *Marker association; clinical utility and transferability may be limited.*

Evidence context (continued): The marker panel and per-genotype effect directions come from a curated research genotype-scoring model, so this citation supports the lead association rather than every marker in the panel. All GRCh37 coordinates were independently verified against the 1000G Phase 3 reference (2026-07-31).

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL
Multi-marker panel
rs12150660 · rs6258

EVIDENCE CONTEXT
Transferability limited
Primary-locus citation: serum testosterone / SHBG (Ohlsson 2011, PLoS Genet) (PMID:21998597), verified against PubMed.

CONFIRMATION BOUNDARY
Not diagnostic
A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION
SHBG variants influencing testosterone are common across populations.

SOUTH-ASIAN CONTEXT
Metabolic syndrome is a common driver of low testosterone in Indian men

BONE & JOINT HEALTH

LOW CONFIDENCE

HELIXLINE

WHY THIS SECTION MATTERS

Bone density, joint health and lifelong skeletal resilience.

Selected variants have research associations with bone density, vitamin metabolism and connective-tissue traits. This panel does not measure bone density, diagnose osteoporosis, or determine supplementation, exercise, or imaging decisions.

2	1	1	0
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
2 trait entries · 2 measured · 1 measured result(s) above the typical range			

YOUR GENETIC STORY IN THIS SECTION

This limited panel found **1 marker association** in this category: **Joint & Disc Degeneration Risk**.

TRAITS IN THIS SECTION

2 TOTAL

1 Bone Mineral Density & Osteoporosis Risk

A typical bone-density variant profile.

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED

Multi-marker panel

Peak bone mass and later bone loss are influenced by COL1A1 (the main structural protein of bone), LRP5 (Wnt signalling, a central bone-formation pathway), SP7/osterix (osteoblast differentiation) and FAM3C. These variants shift lifetime bone-density trajectory. They do not measure your bone density - a DEXA scan does - and calcium, vitamin D, resistance training and not smoking remain the dominant modifiable factors.

`SP7 + LRP5 + COL1A1 + FAM3C` TIER B

2 Joint & Disc Degeneration Risk

Association detected in the assessed marker panel. This is not an estimate of personal disease probability.

ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL

Multi-marker panel

Osteoarthritis and intervertebral disc degeneration share cartilage-biology genetics. COL2A1 encodes the main collagen of cartilage and the disc's nucleus pulposus, NCOA3 is associated with hip osteoarthritis, and the COL6A4P1/3p24 region with knee osteoarthritis. These are susceptibility markers; joint loading history, body weight and injury history are stronger determinants.

`COL6A4P1 + NCOA3 + COL2A1` TIER B

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

Higher-signal findings receive dedicated detail pages; all other qualified traits remain in the personalized compact section.

TAILORED TO THIS SAMPLE
Only quality-qualified direct or imputed calls enter the findings above.

NO SILENT SUBSTITUTION
An unrelated marker or population average is never used to fill a missing result.

CLINICAL BOUNDARY
These associations support discussion; they do not diagnose disease or set treatment.

JOINT & DISC DEGENERATION RISK

Raised degeneration susceptibility

HELIXLINE

YOUR RESULT

Association detected in this limited marker panel

Genotype association · no calibrated percentile

GENE PANEL

COL6A4P1 + NCOA3 + COL2A1

TIER B · REPLICATED

Markers analysed · rs7639618 · rs6094710 · rs2276454

ABOUT THIS TRAIT

Osteoarthritis and intervertebral disc degeneration share cartilage-biology genetics. COL2A1 encodes the main collagen of cartilage and the disc's nucleus pulposus, NCOA3 is associated with hip osteoarthritis, and the COL6A4P1/3p24 region with knee osteoarthritis. These are susceptibility markers; joint loading history, body weight and injury history are stronger determinants.

YOUR GENETIC PROFILE

RESULT TYPE

Multi-marker panel

ASSOCIATION BAND

Association detected in this limited marker panel

WHAT THIS MEANS FOR YOU

Association detected in the assessed marker panel. This is not an estimate of personal disease probability.

The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 23989986 *Marker association; clinical utility and transferability may be limited.*

Evidence context (continued): The marker panel and per-genotype effect directions come from a curated research genotype-scoring model, so this citation supports the lead association rather than every marker in the panel. All GRCh37 coordinates were independently verified against the 1000G Phase 3 reference (2026-07-31).

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL

Multi-marker panel

rs7639618 · rs6094710 · rs2276454

EVIDENCE CONTEXT

Transferability limited

Primary-locus citation: osteoarthritis GWAS meta-analysis (Evangelou 2014, Ann Rheum Dis) (PMID:23989986), verified against PubMed.

CONFIRMATION BOUNDARY

Not diagnostic

A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION

Knee osteoarthritis affects ~28% of Indian adults over 45 - among the highest reported rates globally.

SOUTH-ASIAN CONTEXT

Knee osteoarthritis is notably prevalent in Indian adults

WELLNESS & APPEARANCE

LOW CONFIDENCE

HELIXLINE

WELLNESS & INFORMATIONAL TRAITS These findings are for educational interest only and carry no clinical implications.

WHY THIS SECTION MATTERS

Visible & sensory traits - how your DNA shapes everyday experience.

Wellness and sensory traits reflect the remarkable diversity encoded in the human genome - from bitter taste perception and sleep chronotype to skin characteristics and earwax type. These findings carry no clinical implications and are presented for personal curiosity and educational interest. They illustrate how ancestry shapes lived experience in ways that are visible, tangible, and uniquely yours.

10	1	9	0
TRAITS	HIGHER-PRIORITY ASSOCIATIONS	TYPICAL	FEWER ASSOCIATED MARKERS
10 trait entries · 10 measured · 5 measured result(s) above the typical range			

YOUR GENETIC STORY IN THIS SECTION

This limited panel found **1 marker association** in this category: **Glaucoma Predisposition (Open-Angle)**.

TRAITS IN THIS SECTION

10 TOTAL

1. **Bitter Taste Perception** — VARIANT PRESENT — G/C genotype
   G/C at TAS2R38 rs713598 is associated with intermediate bitter perception. A full PAV/AVI haplotype requires additional markers and was not measured.
   TAS2R38 TIER A

2. **Hair Texture** — VARIANT NOT PRESENT — A/A genotype
   A/A at TCHH rs11803731 - wavy/curly hair tendency (more common in Indian populations). No clinical implications.
   TCHH TIER A

3. **Eye Colour Pigmentation** — VARIANT PRESENT — G/A genotype
   One copy of the G allele at OCA2/HERC2 - intermediate iris pigmentation hazel or light brown eyes possible. The variant is uncommon in Indians (mostly seen in admixed individuals).
   OCA2 / HERC2 TIER A

4. **Skin UV Sensitivity** — VARIANT NOT PRESENT — C/C genotype
   C/C at MC1R rs1805007 - wild-type MC1R standard melanin response to UV. Sun-tan response normal.
   MC1R TIER B

5. **Hair Greying Onset** — VARIANT NOT PRESENT — C/C genotype
   Two copies of the common C allele. Typical age-of-onset for greying pigmentation maintained longer.
   IRF4 TIER A

6. **Age-Related Vision Loss (Macular Degeneration)** — NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED — Multi-marker panel
   Single risk allele - modestly elevated AMD risk.
   CFH + ARMS2 TIER A

7. **Glaucoma Predisposition (Open-Angle)** — ASSOCIATION DETECTED IN THIS LIMITED MARKER PANEL — T/T genotype
   T/T at CDKN2B-AS1 (9p21) is associated with increased POAG susceptibility in population studies.
   CDKN2B-AS1 TIER A

8. **Sleep Quality & Disturbance Tendency** — VARIANT NOT PRESENT — Multi-marker panel
   A typical sleep-regulation variant profile.
   ADORA2A + CLOCK + DRD2 + TNF + PER2 TIER B

9. **Skin Ageing & Sun Sensitivity** — VARIANT PRESENT — Multi-marker panel
   Some variants associated with greater sun sensitivity or earlier visible ageing.
   ASIP + AHR + ELN + FOXC1 + SLC2A4 TIER B

10. **Age-Related Cataract Risk** — VARIANT PRESENT — Multi-marker panel
    Variant present in the assessed markers (EPHA2 + CRYAA).
    EPHA2 + CRYAA TIER B

LIFESTYLE LEVERS FOR THIS CATEGORY

Applies to every trait in this section

CONTEXT
These marker associations are not a clinical risk assessment and do not set a personal care plan.

ROUTINE CARE
Continue preventive care appropriate to your age, history, symptoms, and local clinical guidelines.

NO DNA-ONLY CHANGES
Do not change medication, supplements, diet, or screening solely because of these findings.

CONFIRM IF NEEDED
Use standard clinical tests or validated confirmatory genetics when a finding could affect care.

GLAUCOMA PREDISPOSITION (OPEN-ANGLE)

CDKN2B-AS1 (9p21) increased-susceptibility association

HELIXLINE

YOUR RESULT

Association detected in this limited marker panel

Genotype association · no calibrated percentile

GENE

CDKN2B-AS1

TIER A · SA VALIDATED

Markers analysed · rs2157719

ABOUT THIS TRAIT

Primary open-angle glaucoma (POAG) is a slowly progressive optic-nerve disease usually associated with elevated intraocular pressure. Left untreated it causes irreversible peripheral-vision loss and eventually blindness. In India, POAG prevalence is ~3.5% adults 40+ - higher than the global average due to under-diagnosis and later presentation. Angle-closure glaucoma is also more common in South Asians (smaller anterior chamber depth). Early detection via tonometry + optic-nerve assessment can prevent blindness through eye-drop therapy.

YOUR GENETIC PROFILE

GENOTYPE	RISK-ALLELE COPIES	SA RISK-ALLELE FREQ
T/T	2 of 2	72.6%
		1000G SAS (n=489)

WHAT THIS MEANS FOR YOU

T/T at CDKN2B-AS1 (9p21) is associated with increased POAG susceptibility in population studies.

Observed genotype T/T. The limited catalog model assigned the qualitative association band shown above. No calibrated personal probability, absolute risk, diagnosis, screening schedule, or treatment recommendation was calculated.

QUESTIONS TO DISCUSS WITH A CLINICIAN

EVIDENCE-LIMITED RESULT

- ✓ If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.
- ✓ Do not change medication, supplements, diet, or screening based on this marker result.
- ✓ Use symptoms, family history, examination, and standard clinical tests to guide care.

PMID: 21532571 *Marker association; clinical utility and transferability may be limited.*

Evidence context (continued): A common tag SNP does not diagnose glaucoma or set an examination schedule; routine ophthalmic screening guidance applies to everyone.

Informational - not medical advice. Consult a qualified clinician before making health decisions based on genetic findings.

YOUR EVIDENCE RECORD

MEASURED SIGNAL	EVIDENCE CONTEXT	CONFIRMATION BOUNDARY
T/T	South-Asian replicated	Not diagnostic
rs2157719	CDKN2B-AS1 (ANRIL, 9p21.3) rs2157719 is a replicated primary open-angle glaucoma locus.	A clinical decision requires the appropriate validated lab test and a qualified clinician.

HOW COMMON IS THIS

IN THE POPULATION	SOUTH-ASIAN CONTEXT
~3.5% Indian adults 40+ have POAG (Vellore Eye Study, Aravind).	~3.5% South Asian adults 40+ have POAG

TYPICAL & INFORMATIONAL FINDINGS

6 findings - no highlighted association in the assessed markers

HELIXLINE

No highlighted association was found in the markers assessed. This limited panel does not rule out the condition or account for non-genetic risk factors.

13 Triglyceride Levels

CARDIOMETABOLIC · APOA5 + APOC3 + LPL + GCKR TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

Typical TG panel.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

14 Lipoprotein(a) - Cardiovascular Marker

CARDIOMETABOLIC · LPA TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

Two copies of the common A allele. Lp(a) is expected to be near population baseline. No additional cardiovascular risk amplification from this locus.

YOUR RESULT
A/A · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

15 Gout & Uric Acid Risk

CARDIOMETABOLIC · ABCG2 TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

G/G at ABCG2 rs2231142 - reference (higher urate excretion). Baseline hyperuricaemia risk.

YOUR RESULT
G/G · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

16 Hyperthyroidism Genetic Tendency

CARDIOMETABOLIC · CTLA4 + LOC101930098 + LOC105378120 TIER B

FEWER RISK-ASSOCIATED MARKERS OBSERVED
GENOTYPE ASSOCIATION

Fewer autoimmune-thyroid susceptibility alleles.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

17 Hypothyroidism Genetic Tendency

CARDIOMETABOLIC · FOXE1 + SH2B3 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

A typical thyroid-susceptibility variant profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

18 Lung Cancer & Nicotine Dependence

CANCER · CHRNA3 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

G/G at CHRNA3 rs1051730 - reference genotype. Indian tobacco exposure is often via bidis, hookah, and gutkha - all elevate lung cancer risk. Even non-smokers exposed to household biomass smoke or urban air pollution have measurable lung cancer risk.

YOUR RESULT
G/G · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

No highlighted association under this limited catalog rule is not an absence of genetic or clinical risk.

TYPICAL FINDINGS (CONTINUED)

6 findings - no highlighted association in the assessed markers

HELIXLINE

No highlighted association was found in the markers assessed. This limited panel does not rule out the condition or account for non-genetic risk factors.

19 Caffeine Metabolism

BRAIN & NEUROLOGY · CYP1A2 TIER A

VARIANT PRESENT
GENOTYPE ASSOCIATION

One copy of the A allele at CYP1A2 - intermediate caffeine metabolism. Moderate consumers (2-3 cups/day) usually tolerate fine

YOUR RESULT
C/A · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

20 Sleep Duration Tendency

BRAIN & NEUROLOGY · ABCC9 TIER B

VARIANT PRESENT
GENOTYPE ASSOCIATION

One copy of the A allele at ABCC9 - slight shift toward shorter natural sleep duration (~10-20 min less per night on average). Maintain good sleep hygiene total hours matter for cognitive performance regardless of genotype.

YOUR RESULT
G/A · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

21 Migraine Predisposition

BRAIN & NEUROLOGY · TRPM8 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

Heterozygous at TRPM8. Lifestyle triggers (sleep, hydration, dietary triggers) dominate manifestation.

YOUR RESULT
T/C · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

22 Narcolepsy Genetic Susceptibility

BRAIN & NEUROLOGY · TCRA TIER B

FEWER RISK-ASSOCIATED MARKERS OBSERVED
GENOTYPE ASSOCIATION

Does not carry the TCRA susceptibility allele.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

23 Alcohol Dependence & Metabolism Tendency

BRAIN & NEUROLOGY · ADH1B + PKNOX2 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

A typical alcohol-metabolism variant profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

24 Celiac Disease Risk

AUTOIMMUNE · HLA-DQA1 1 of 2 declared markers assessed TIER A

FEWER RISK-ASSOCIATED MARKERS OBSERVED
GENOTYPE ASSOCIATION

No common HLA-DQ2.5/DQ8 tagging combination was identified by this two-marker proxy. This lowers genetic susceptibility but is not full HLA typing and does not exclude celiac disease.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

HELIXLINE

No highlighted association under this limited catalog rule is not an absence of genetic or clinical risk.

TYPICAL FINDINGS (CONTINUED)

5 findings - no highlighted association in the assessed markers

HELIXLINE

No highlighted association was found in the markers assessed. This limited panel does not rule out the condition or account for non-genetic risk factors.

25 Rheumatoid Arthritis Risk

AUTOIMMUNE · HLA-DRB1 + PTPN22 + STAT4 TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

Single low-effect risk allele. Typical population RA risk.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

26 Chronic Urticaria (Hives) Susceptibility

AUTOIMMUNE · LILRA3 + IGHG1/2 + TDGF1 + HLA-G + PTPN22 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

A typical immune-reactivity variant profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

27 Oxidative Stress & Antioxidant Capacity

AUTOIMMUNE · SOD2 + CAT + GPX1 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

A typical antioxidant-enzyme profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

28 Lactose Intolerance

NUTRITION · MCM6 / LCT TIER A

VARIANT PRESENT
GENOTYPE ASSOCIATION

Two copies of the lactase non-persistence allele (G). You are likely to experience lactose intolerance symptoms (bloating, cramps, diarrhoea) after consuming significant amounts of lactose. This is the typical genotype across South Asian populations (~75-90%).

YOUR RESULT
G/G · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

29 Omega-3 Fatty Acid Metabolism

NUTRITION · FADS1 TIER A

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

C/C at FADS1 rs174546 - slower ALA → EPA/DHA conversion. Most South Asians are C-carriers.

YOUR RESULT
C/C · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

HELIXLINE

No highlighted association under this limited catalog rule is not an absence of genetic or clinical risk.

TYPICAL FINDINGS (CONTINUED)

5 findings - no highlighted association in the assessed markers

HELIXLINE

No highlighted association was found in the markers assessed. This limited panel does not rule out the condition or account for non-genetic risk factors.

30 Folate Processing

NUTRITION · MTHFR TIER A

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

Single MTHFR variant carrier (C677T or A1298C heterozygote). Enzyme activity ~65-80% of normal. Adequate green leafy vegetables (methi, palak) + B12 sufficiency is enough.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

31 Susceptibility to Lower Vitamin D Levels

NUTRITION · GC 1 of 3 declared markers assessed TIER A

FEWER RISK-ASSOCIATED MARKERS OBSERVED
GENOTYPE ASSOCIATION

No highlighted association was found in the assessed markers. This does not establish a lower personal probability or rule out the condition.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

32 Body Composition & Obesity Predisposition

NUTRITION · UCP1 + ADRB3 + FTO + MC4R + SH2B1 + TMEM18 + NEGR1 + KCTD15 + TFAP2B + QPCTL + NUDT3, RPS10-NUDT3 + LINC01741 - SEC16B + ZC3H4 + CADM2 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

A typical spread of body-composition variants.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

33 Gut Sensitivity & IBS Genetic Tendency

NUTRITION · IL10 + TLR9 + SI + HTR3E + KLB + IL4 + TNF + GRID2IP + TNFSF15 + CDH1 TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

A typical inflammatory and gut-barrier variant profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

34 Salt Sensitivity & Blood Pressure Tendency

NUTRITION · AGT + NPPA + ADD1 TIER B

FEWER RISK-ASSOCIATED MARKERS OBSERVED
GENOTYPE ASSOCIATION

Less salt-sensitive variant profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

HELIXLINE

No highlighted association under this limited catalog rule is not an absence of genetic or clinical risk.

TYPICAL FINDINGS (CONTINUED)

5 findings - no highlighted association in the assessed markers

HELIXLINE

No highlighted association was found in the markers assessed. This limited panel does not rule out the condition or account for non-genetic risk factors.

35 Power vs Endurance Profile

FITNESS · ACTN3 TIER A

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

T/T at ACTN3 - X/X (loss-of-function homozygote). Slight endurance advantage. ~10% of South Asians. Doesn't preclude power sports but marginally favours endurance running / cycling / swimming.

YOUR RESULT
T/T · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

36 VO2 Max Trainability

FITNESS · PPARGC1A TIER B

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

No copies of the T allele at PPARGC1A rs8192678 - population-baseline VO2 max trainability. Cardio training produces typical improvement curves.

YOUR RESULT
C/C · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

37 Muscle Recovery

FITNESS · IL6 TIER B

VARIANT PRESENT
GENOTYPE ASSOCIATION

G/G at IL6 - highest IL-6 response, potentially slower recovery from intense exercise. Prioritise sleep, anti-inflammatory foods (haldi/turmeric, adrak/ginger), and adequate protein for recovery.

YOUR RESULT
G/G · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

38 Muscle Power & Strength Potential

FITNESS · IGF2 + MSTN TIER B

VARIANT PRESENT
GENOTYPE ASSOCIATION

A power-oriented natural profile - you may find explosive and strength-focused training especially rewarding.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

39 Connective Tissue & Injury Resilience

FITNESS · COL5A1 + COL1A1 TIER B

VARIANT PRESENT
GENOTYPE ASSOCIATION

A collagen profile associated with higher reported soft-tissue and ligament injury rates. Prioritise gradual load progression, eccentric strengthening and recovery.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

HELIXLINE

No highlighted association under this limited catalog rule is not an absence of genetic or clinical risk.

TYPICAL FINDINGS (CONTINUED)

5 findings - no highlighted association in the assessed markers

HELIXLINE

No highlighted association was found in the markers assessed. This limited panel does not rule out the condition or account for non-genetic risk factors.

40 Bone Mineral Density & Osteoporosis Risk

BONE & JOINT · SP7 + LRP5 + COL1A1 + FAM3C TIER B

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

A typical bone-density variant profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

41 Bitter Taste Perception

WELLNESS · TAS2R38 TIER A

VARIANT PRESENT
GENOTYPE ASSOCIATION

G/C at TAS2R38 rs713598 is associated with intermediate bitter perception. A full PAV/AVI haplotype requires additional markers and was not measured.

YOUR RESULT
G/C · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

42 Hair Texture

WELLNESS · TCHH TIER A

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

A/A at TCHH rs11803731 - wavy/curly hair tendency (more common in Indian populations). No clinical implications.

YOUR RESULT
A/A · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

43 Eye Colour Pigmentation

WELLNESS · OCA2 / HERC2 TIER A

VARIANT PRESENT
GENOTYPE ASSOCIATION

One copy of the G allele at OCA2/HERC2 - intermediate iris pigmentation hazel or light brown eyes possible. The variant is uncommon in Indians (mostly seen in admixed individuals).

YOUR RESULT
G/A · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

44 Skin UV Sensitivity

WELLNESS · MC1R TIER B

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

C/C at MC1R rs1805007 - wild-type MC1R standard melanin response to UV. Sun-tan response normal.

YOUR RESULT
C/C · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

HELIXLINE

No highlighted association under this limited catalog rule is not an absence of genetic or clinical risk.

TYPICAL FINDINGS (CONTINUED)

5 findings - no highlighted association in the assessed markers

HELIXLINE

No highlighted association was found in the markers assessed. This limited panel does not rule out the condition or account for non-genetic risk factors.

45 Hair Greying Onset

WELLNESS · IRF4 TIER A

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

Two copies of the common C allele. Typical age-of-onset for greying pigmentation maintained longer.

YOUR RESULT
C/C · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

46 Age-Related Vision Loss (Macular Degeneration)

WELLNESS · CFH + ARMS2 TIER A

NO HIGHLIGHTED ASSOCIATION IN THE MARKERS ASSESSED
GENOTYPE ASSOCIATION

Single risk allele - modestly elevated AMD risk.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

47 Sleep Quality & Disturbance Tendency

WELLNESS · ADORA2A + CLOCK + DRD2 + TNF + PER2 TIER B

VARIANT NOT PRESENT
GENOTYPE ASSOCIATION

A typical sleep-regulation variant profile.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

48 Skin Ageing & Sun Sensitivity

WELLNESS · ASIP + AHR + ELN + FOXC1 + SLC2A4 TIER B

VARIANT PRESENT
GENOTYPE ASSOCIATION

Some variants associated with greater sun sensitivity or earlier visible ageing.

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

49 Age-Related Cataract Risk

WELLNESS · EPHA2 + CRYAA TIER B

VARIANT PRESENT
GENOTYPE ASSOCIATION

Variant present in the assessed markers (EPHA2 + CRYAA).

YOUR RESULT
Multi-marker panel · association band shown above

SUGGESTED NEXT STEP
If this result is relevant to your symptoms, family history, or routine laboratory findings, discuss appropriate clinical evaluation with a qualified healthcare professional.

HELIXLINE

No highlighted association under this limited catalog rule is not an absence of genetic or clinical risk.

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Type the printed HTTPS address into your browser, sign in, and select Genetic Health under Reports.

PRIVACY

Do not share report access or screenshots unless you intend the recipient to see your health information.

HOW WE ANALYSED YOUR DNA

HELIXLINE

HELIXLINE PRECISION RISK SCORING ENGINE

Your microarray data was evaluated with versioned marker panels, explicit quality gates, and score-specific reference artifacts.

MARKER COVERAGE AND PROVENANCE — HIGH CONFIDENCE — 49 trait entries · 49 measured · 0 not measurable · 12 carrier condition(s) · 4 polygenic score(s) · uses imputed genotypes — direct+imputed

CLINICIAN-FACING LIMITATIONS — HIGH CONFIDENCE — consumer genomics, not clinical diagnosis — context

ASSOCIATION

TRAIT INSIGHTS

Published genotype or limited-panel associations with conservative interpretation and no invented phenotype percentile.

QUALITY GATED

POLYGENIC RISK

Quality-qualified scoring artifacts ranked against versioned, marker-matched South Asian references.

MARKER LIMITED

SELECTED VARIANTS

Named ClinVar-linked variants screened directly; negative findings apply only to those markers.

EVIDENCE TIERS - HOW WE GRADE EVERY FINDING

TIER A · SA VALIDATED

South Asian Validated

Association replicated in ≥1 South Asian cohort (e.g. INDICO, IndiGen, UK Biobank SAS arm). Highest confidence for Indian populations.

TIER B · REPLICATED

Multi-Ethnic Replicated

Replicated in ≥2 GWAS across multiple populations; included in the PGS Catalog. Effect sizes may differ modestly in South Asians.

TIER C · EMERGING

Emerging / Preliminary

Promising association from a single large study; marker coverage may be limited in your imputed data. Treat as a directional signal only.

QUALITY CONTROL STANDARDS

Genome Build
GRCh37 (hg19) - all variant calls, carrier positions, and PRS scores

Imputation Quality
Marker-specific quality gates; missing or inadequate calls are withheld, never assumed normal

Reference Panel
Versioned score-specific South Asian reference artifact; size and provenance shown on each PRS

Scoring Engine
Published score weights or curated genotype rules; no absolute-risk conversion without validated calibration

IMPORTANT LIMITATIONS

Not a diagnostic test.
Findings are statistical associations - not diagnoses. A single variant does not determine your health outcome.

Common variants only.
Rare familial conditions require clinical-grade sequencing. Significant family history warrants a clinical genetics referral.

Lifestyle dominates.
For most common conditions, lifestyle and environment outweigh genetic predisposition. Genetics informs - it does not dictate.

KEY SCIENTIFIC REFERENCES

Buniello A et al. 2019 - NHGRI-EBI GWAS Catalog. *Nucleic Acids Res*

Lambert SA et al. 2021 - PGS Catalog for reproducibility. *Nature Genetics*

Landrum MJ et al. 2018 - ClinVar variant interpretations. *Nucleic Acids Res*

Martin AR et al. 2019 - PRS and health disparities. *Nature Genetics*

Khera AV et al. 2018 - Genome-wide polygenic scores. *Nature Genetics*

EXPLORE MORE WITH YOUR GENETIC DATA

HELIXLINE

Your existing raw data may support future reanalysis, depending on marker coverage, assay quality, and the requirements of future methods. Some newer methods need markers or sequencing depth this assay does not provide.

WHAT WE SHARE WITH YOU

Array Genotype VCF `.vcf.gz`
Your raw genotype calls in VCF format - unmodified, portable ground truth from your chip run.

Tabix Index `.tbi`
Lets any tool jump instantly to a genomic region without scanning the full file.

Imputed VCF `.vcf.gz`
Imputed records from the versioned reference workflow; the exact count and reference provenance are recorded in the technical manifest. Go to your **profile → Raw Data** section to download the file when it is included in your delivery package.

HOW TO ANALYSE YOUR DATA - STEP BY STEP

1

Download Your Files

Log in to your Helixline account and download your `.vcf.gz` and `.tbi` files. Store them securely - this is your lifelong genetic record.

2

Visualise Your Genome

Open your VCF in **IGV** (free, Broad Institute) to browse any gene or region visually. Search your gene of interest and inspect your exact calls.

`igv.org → Open File`

3

Query Public Databases

Look up any variant across open databases:

- **ClinVar** Clinical significance
- **gnomAD** Population frequency
- **PGS Catalog** Polygenic scores
- **PharmGKB** Drug response

4

Re-Analyse Periodically

New GWAS and clinical findings publish every week. Run Ensembl VEP or SnpEff on your same VCF to get fresh annotations - no new test needed.

↻ Every 12-24 months

KEY BIOLOGICAL DATABASES

ClinVar
clinvar.ncbi.nlm.nih.gov
Clinical variant classifications - pathogenic, likely-pathogenic, VUS. Best first stop for any variant you want to interpret medically.

gnomAD
gnomad.broadinstitute.org
Population frequencies across 125,000+ exomes and genomes. A common variant (MAF >1%) in gnomAD is rarely clinically significant on its own.

PGS Catalog
pgscatalog.org
Repository of polygenic score weights. Download SAS-validated models and apply them to your imputed VCF with PLINK2 score.

PharmGKB
pharmgkb.org
Drug-gene interaction database. Look up your CYP2C19, SLCO1B1 or DPYD genotype to flag drug dosing considerations before any new prescription.

OMIM
omim.org
Gold-standard catalog of Mendelian diseases and their causative genes. Use it to understand any rare variant found in your VCF.

dbSNP
ncbi.nlm.nih.gov/snp
NCBI reference for every known variant - rs IDs, allele frequencies, linked publications. Useful for cross-referencing raw variant identifiers.

BIOINFORMATICS TOOLS & USE CASES

bcftools
Filter, subset, and query variants. Use bcftools view to extract a gene region; bcftools stats for QC summary.
`brew install bcftools`

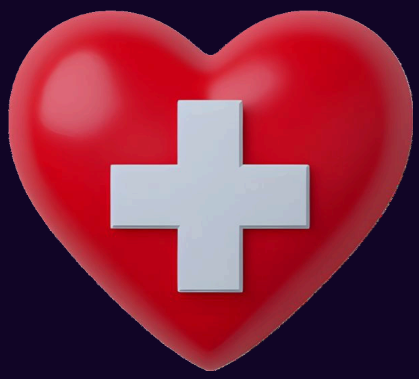
Ensembl VEP
Annotate variants with gene consequence, SIFT/PolyPhen scores, population frequency, and ClinVar status in one run.
`ensembl.org/Tools/VEP`

SnpEff
Java annotator for functional impact - missense, stop-gain, splice-site. Outputs HTML summary with variant counts by effect class.
`pcingola.github.io/SnpEff`

PLINK2
Compute polygenic risk scores (--score), run basic GWAS, calculate IBD and kinship. Native VCF input supported.
`plink2.readthedocs.io`

IGV
Browser-based genome viewer. Load your VCF + BAM to visually confirm any variant - inspect read support, phasing, and nearby indels.
`igv.org`

Michigan Imputation
Cloud imputation with selectable reference panels. Output marker count and suitability vary by input, panel, build and server QC; review the service documentation before use.
`imputationserver.sph.umich.edu`



YOUR GENES, DECODED

Your blueprint. Your power.

Thank you for trusting us.

FOLLOW US • SHARE YOUR STORY

HELIXLINE

PHARMACOGENOMIC ANALYSIS

PHARMACO GENOMICS



DECODE

NAME **Demo User** · SEX **Male** · ISSUED **October 2026**

HELIXLINE PGX ENGINE V3.3 · VERSIONED CPIC / CLINPGX EVIDENCE · GRCH37

TABLE OF CONTENTS

HELIXLINE

WHAT THIS REPORT CONTAINS

INTRO — How to Read + Drug Summary

Start here. Four rules for reading this report, then a one-page personalised summary of every drug finding.

60s Guide · Traffic Lights · Drug Snapshot

PART 01 — Your Findings

Your quality-qualified pharmacogene results, current source-linked medicine associations, and any clinically reviewed alerts.

27 Genes · Critical Alerts · Family Sharing

PART 02 — Specialty Index

All your drugs grouped by medical specialty so Cardiologists, Psychiatrists, Oncologists and other specialists can navigate directly to their section.

Cardiology · Psychiatry · Oncology · +6 more

PART 03 — Drug Detail Pages

Every contracted gene-medicine pair is listed with its result state and reason. Source-review pairs receive a detail page; unresolved and research-context pairs appear in grouped tables. Dosing guidance appears only when clinically approved.

Result State · Reason & Limits · Source Link

APPENDIX — Methodology & Glossary

How your genes were typed - imputation method, HaplotypeLock version, PharmGuard QC results, known limitations, and a plain-English glossary of every term.

Engine v3.3 · PharmGuard QC · Glossary

INTRO

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PART 01 · YOUR FINDINGS

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PART 02 · MEDICINE GUIDE

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PART 03 · CONTEXT & APPENDIX

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HELIXLINE

PLEASE READ BEFORE USING THIS REPORT

Decision support, not a prescription. Evidence sources include CPIC, DPWG and regulatory material; a dose statement appears only when its wording has passed clinical release review. · **Carry this report to medication encounters** - especially for cardiology, oncology, psychiatry and pain management. Check "Critical Alerts" to see whether this report authorized any alert; an empty alert page is not an all-clear. · **Privacy:** your raw DNA is processed only with explicit consent and never sold.

YOUR REPORT IN 60 SECONDS

WHAT THIS REPORT IS

An informational map of which listed gene-medicine relationships this SNP-array result could support, and what to discuss with the clinician who prescribes for you.

Four rules govern everything on the pages that follow. Read them before you read any result.

01

The report examines the gene-medicine pairs listed in it.

That list is the whole scope of the test. A medicine that is not listed has not been examined here, and nothing in this report is a finding about it.

02

Some genes can be resolved from this array and others cannot.

This test reads a fixed set of DNA positions. Where a gene could not be resolved to a reportable result, the report names the gene and gives the reason instead of an uncertain answer.

03

No medicine or dose should be changed from this report alone.

Take it to the clinician who prescribes for you. Your diagnosis, your other medicines, your organ function and your history all sit alongside genetics in any prescribing decision.

04

“No approved action” does not mean a medicine is safe for you.

A result showing no gene-based action applies only to the gene-medicine evidence assessed here. It does not establish that a medicine is safe, appropriate or effective for you, and it must not be generalised to untested medicines.

WHERE THE DETAIL LIVES

Result states and evidence tiers

What each result state on a drug page means, and how strong the evidence behind it is. Full definitions are in the methodology appendix.

What this test cannot see

Gene copy number, interactions with your other medicines, and the non-genetic factors that shape drug response. Full explanations are in the methodology appendix.

Words used on the drug pages

Every technical term on a drug page, in plain English. Full definitions are in the glossary.

Your PGx Results at a Glance

A plain-language map of the gene results this assay could support and the medicines linked to them.

HELIXLINE

Demo User

35 gene-medicine pairs have an interpretable evidence record.

"Interpretable" means the assay supported a named evidence category; it does not automatically mean a medication action. 22 require confirmation, 10 are source-linked association-only, and 3 provide research context. None authorizes a medicine or dose change unless separately counted as a clinically reviewed action below. Results were available for 7 genes: 3 were resolved within the tested marker set and 4 produced partial, non-unique findings. The remaining 20 genes could not be reliably resolved from this assay. All 131 contracted pairs across 105 medicines remain accounted for.

How to use this: This is an informational SNP-array screening report, not a diagnostic PGx test or a prescription. **No personalized dose or medicine-change instruction is printed in this report.** Find the exact medicine, check the gene-result certainty, and show that row to the prescriber. Do not start, stop, switch or change a dose from this report alone.

0	22	32	96 / 131
CLINICALLY REVIEWED ACTIONS-USE ONLY WITH PRESCRIBER	PROVISIONAL PAIRS REQUIRING CONFIRMATION	SOURCE-LINKED PAIRS, INCLUDING PROVISIONAL	PAIRS FULLY RETAINED IN LIMITATIONS

HIGH CONFIDENCE SCOPE ACCOUNTING: 131 contracted gene-medicine pairs across 105 unique medicines; 35 have interpretable evidence. **Complete pair-state accounting:** 0 approved medication actions; 22 pairs for source review; 10 association-only pairs; 3 research-only pairs; 96 pairs not resolvable from this assay; 0 pairs in other assessed states = 131 contracted pairs.

THE GENE RESULTS AVAILABLE IN THIS REPORT · ALL 7 SHOWN

ABCG2

RESOLVED · TESTED MARKERS · DIRECT

Normal Function

`*1/*1`

Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context.

CACNA1S

RESOLVED · TESTED MARKERS · DIRECT

No assayed MH-associated variant detected

`Negative`

Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context.

CYP2C19

PROVISIONAL · CONFIRM · DIRECT

Intermediate Metabolizer (provisional; assayed variant set)

`*2 detected (one copy); remaining allele background not fully characterised`

Confirm with a validated clinical PGx assay before prescribing use.

CYP3A5

PROVISIONAL · CONFIRM · DIRECT

Intermediate Expressor (provisional; assayed variant set)

`*3 detected (one copy); remaining allele background not fully characterised`

Confirm with a validated clinical PGx assay before prescribing use.

NAT2

PROVISIONAL · CONFIRM · PROXY

rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established

`rs1495741 A/A proxy observation`

Confirm the proxy-based result with a validated clinical PGx assay before prescribing use.

SLCO1B1

PROVISIONAL · CONFIRM · DIRECT

Normal Function (provisional; assayed variant set)

`*1B detected (one copy); remaining allele background not fully characterised`

Confirm with a validated clinical PGx assay before prescribing use.

VKORC1

RESOLVED · TESTED MARKERS · DIRECT

Standard

`-1639 G/G (rs9923231)`

Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context.

Medication evidence · What the 35 interpreted pairs mean

33 medicines are represented by assay-supported canonical pairs - for example Allopurinol, Amitriptyline, Atorvastatin, Citalopram. This count comes only from the canonical PGx inventory below, not from a separate feature summary.

22	**Provisional variant-based interpretations.** A real marker was observed, but confirmation is advised before prescribing use.
10	**Source-linked gene-medicine relationships.** Review the cited source if the medicine is relevant; no dose instruction is authorized here.
3	**Research-context relationships.** Retained for completeness; not clinical guidance or a prediction that a medicine will work.
0	**Validated dose or medicine changes printed in this report.** The treating prescriber still makes the decision.

What about the other 96 pairs?

They remain in the complete accounting, grouped by unresolved gene. They are not negative or normal results. A validated clinical assay may be needed if treatment depends on one of those genes.

See "What This Array Could Not Resolve."

WHEN A LISTED MEDICINE IS RELEVANT

1 · Find the exact pair
Use the specialty index or alphabetical medicine pages. Match both medicine and gene.

2 · Read the source badge
DIRECT, IMPUTED or PROXY tells you how the result was obtained; then check the confirmation instruction.

3 · Use the clinician handoff
Bring the result-first handoff with current medicines, diagnosis, organ function, age and treatment history.

125 matching annotation rows

among 328 readable reportable PharmGKB markers. A matching annotation row can describe a reference or effect genotype and may not encode effect direction; these are source links, not 125 independent personal findings.

Two questions-answered separately

First ask how confidently the gene was measured. Then ask what level of medicine guidance exists. A strong answer to one question does not automatically strengthen the other.

1 · How certain is the gene result?

ANALYTICAL RESULT

Resolved within the tested marker set

Every marker required by this caller passed its quality rules and supported one result without an unresolved phase question. It does not mean the entire gene was sequenced.

ANALYTICAL RESULT

Provisional or partial

A proxy marker, non-unique diplotype, incomplete phase or another defined limitation remains. The observation is useful, but laboratory confirmation is advised before prescribing use.

ANALYTICAL RESULT

Not determined

The assay could not support a prescribing-grade phenotype. A limited screen may still be described, but it is never converted into “normal,” “negative” or an all-clear.

2 · What does the medicine evidence support?

CLINICAL EVIDENCE

Guideline or label context

A versioned CPIC, DPWG or regulatory source discusses the gene-medicine relationship. A customer-specific dose statement is printed only after its result and wording pass release review.

CLINICAL EVIDENCE

Published relationship

A source-linked relationship exists, but this report does not turn it into a dose, a medicine choice, or a prediction that the medicine will work or be safe.

CLINICAL EVIDENCE

Research context

The relationship is retained for scientific completeness. It is not presented as clinical guidance and should not drive treatment.

Where the genotype came from

Direct genotyping

The DNA position was measured on the array. “Direct” describes the measurement-not completeness. Important variants outside the array can remain unseen.

Quality-qualified imputation

The genotype was statistically inferred and included only when cohort accuracy (DR2/R2 ≥ 0.8) and this sample's genotype probability (GP ≥ 0.90) both passed. It is labelled wherever it contributes; imputed phase is not treated as observed clinical phase.

Use all four checks before discussing a medicine

1 · Exact medicine
Find the specific medicine and gene pair-not a marker annotation alone.

2 · Gene certainty
Exact, provisional or not determined for this assay.

3 · Evidence level
Guideline/label, published relationship or research context.

4 · Current context
Indication, dose, other medicines, organ function, age and monitoring.

This report: 3 gene results resolved within the tested marker set, 4 provisional results, and 20 not-determined genes. These categories describe analytical certainty; they do not independently prescribe a medicine or dose.

HELIXLINE

Clinical PGx Handoff

A compact map of the reportable gene results, medicine pairs to review first, analytical provenance and confirmation requirements. The complete evidence and limitations remain in the pages that follow.

Informational SNP-array screening; not a diagnostic PGx result or prescription. This report contains no locally authorized personalized dose or medicine-change instruction. Match the exact medicine and gene, verify direct, imputed or proxy provenance, and apply the current guideline with indication, interactions, organ function, age and treatment history. No change should be made from this page alone.

7	35	0	96
GENES WITH AN ANALYTICAL RESULT	INTERPRETABLE GENE-MEDICINE PAIRS	LOCALLY REVIEWED MEDICINE ACTIONS	PAIRS RETAINED IN LIMITATIONS

MEDICINE PAIRS TO REVIEW FIRST · SHOWING 6 OF 32

MEDICINE + GENE	RESULT SOURCE	PERSONAL RESULT	CLINICAL NEXT STEP
Amitriptyline CYP2C19	DIRECT CPIC	**Intermediate Metabolizer (provisional; assayed variant set)** *2 detected (one copy); remaining allele background not fully characterised	**CONFIRM BEFORE PRESCRIBING USE** Confirm with a validated clinical PGx assay before prescribing use.
Atorvastatin SLCO1B1	DIRECT CPIC	**Normal Function (provisional; assayed variant set)** *1B detected (one copy); remaining allele background not fully characterised	**CONFIRM BEFORE PRESCRIBING USE** Confirm with a validated clinical PGx assay before prescribing use.
Citalopram CYP2C19	DIRECT CPIC	**Intermediate Metabolizer (provisional; assayed variant set)** *2 detected (one copy); remaining allele background not fully characterised	**CONFIRM BEFORE PRESCRIBING USE** Confirm with a validated clinical PGx assay before prescribing use.
Clomipramine CYP2C19	DIRECT ClinPGx/DPWG annotation	**Intermediate Metabolizer (provisional; assayed variant set)** *2 detected (one copy); remaining allele background not fully characterised	**CONFIRM BEFORE PRESCRIBING USE** Confirm with a validated clinical PGx assay before prescribing use.
Clopidogrel CYP2C19	DIRECT CPIC	**Intermediate Metabolizer (provisional; assayed variant set)** *2 detected (one copy); remaining allele background not fully characterised	**CONFIRM BEFORE PRESCRIBING USE** Confirm with a validated clinical PGx assay before prescribing use.
Dexlansoprazole CYP2C19	DIRECT ClinPGx/DPWG annotation	**Intermediate Metabolizer (provisional; assayed variant set)** *2 detected (one copy); remaining allele background not fully characterised	**CONFIRM BEFORE PRESCRIBING USE** Confirm with a validated clinical PGx assay before prescribing use.

REPORTABLE PHARMACOGENE RESULTS · SHOWING 6 OF 7

NAT2 PROXY
rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established
rs1495741 A/A proxy observation
Confirm the proxy-based result with a validated clinical PGx assay before prescribing use.

CYP2C19 DIRECT
Intermediate Metabolizer (provisional; assayed variant set)
*2 detected (one copy); remaining allele background not fully characterised
Confirm with a validated clinical PGx assay before prescribing use.

CYP3A5 DIRECT
Intermediate Expressor (provisional; assayed variant set)
*3 detected (one copy); remaining allele background not fully characterised
Confirm with a validated clinical PGx assay before prescribing use.

SLCO1B1 DIRECT
Normal Function (provisional; assayed variant set)
*1B detected (one copy); remaining allele background not fully characterised
Confirm with a validated clinical PGx assay before prescribing use.

ABCG2 DIRECT
Normal Function
*1/*1
Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context.

CACNA1S DIRECT
No assayed MH-associated variant detected
Negative
Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context.

CURRENT MEDICINES AND SUPPLEMENTS

Medicine · strength · dose · frequency · indication

CONTEXT TO VERIFY

- ☐ Kidney function
- ☐ Liver function
- ☐ Interactions
- ☐ Allergies/reactions
- ☐ Pregnancy/age
- ☐ Indication/history

QUESTIONS FOR THE PRESCRIBING VISIT

1. My CYP2C19 result is Intermediate Metabolizer (provisional; assayed variant set) - does that change anything about Amitriptyline for me?
2. My CYP2C19 result is Intermediate Metabolizer (provisional; assayed variant set) - does that change anything about Citalopram for me?
3. My CYP2C19 result is Intermediate Metabolizer (provisional; assayed variant set) - does that change anything about Clomipramine for me?

REPORT PROVENANCE
Platform: Helixline Genotyping Array · Build: GRCh37 · Run: 4ec75a348a5d
Imputation reference: 1000 Genomes Project Phase 3 South Asian reference panel (489 samples; GRCh37)
Scan for the secure account and the latest regenerated report; a static PDF does not update when evidence changes.

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Your PGx Pocket Reference

Print or save this page. Cut around the dashed outline for a paper copy, and show it to a prescriber or pharmacist when a medicine is being considered. **Scan the QR to sign in and open the complete report.**

HELIXLINE PHARMACOGENOMICS · WALLET CARD

PGx Source Reference

Demo User

Sample ID 9XC7

SCAN TO OPEN YOUR ACCOUNT

PRIORITY MEDICATIONS - FLAG TO PRESCRIBER

No genotype-based medication or dose change is authorized from this report. Partial findings require confirmation before prescribing decisions. The full report lists every contracted gene-medicine pair with its own state and reason. Show this card so the prescriber can confirm a result before prescribing.

PRIORITY GENE CALLS - 0 SHOWN

GENE	DIPLOTYPE	PHENOTYPE

◆ = atypical interpreted phenotype. Medication implications exist only where the full report provides a source-linked gene-medicine row. This pocket card is not the complete panel; normal and withheld genes are documented in the full report.

4 proxy or otherwise non-definitive gene call(s) are described in the full report and intentionally omitted from this standalone card.

INSTRUCTIONS TO PRESCRIBER

1. Cross-check the medication against the complete digital report; the table above shows priority calls only.
2. If a highlighted gene affects the medication, see the corresponding drug detail page in the full report.
3. Recommendations follow CPIC / DPWG / FDA labels - verify with your institution's PGx guidance.

Standard clinical prescribing, contraindications, allergies, drug interactions, organ function and monitoring are outside this card's genetic conclusion and continue to apply in full.

Secure account · full report + updates
https://app.helixline.in/dashboard/reports

SHOW TO PRESCRIBER

HELIXLINE

YOUR DNA × YOUR MEDICATIONS

WHAT YOUR PHARMACOGENES DO

Your liver enzymes, drug transporters and immune-recognition genes shape **how many medicines are processed**. Variants in **high-evidence pharmacogenes** can alter efficacy, exposure, or adverse-reaction risk. This panel contains **131 gene-medicine pairs** covering **105 unique medicines**, assessed against CPIC, DPWG and FDA guidance. The report resolved **7 of 27 genes** from this assay; the remaining **20 genes** are shown with the reason a prescribing-grade result was not available. Direct calls had to pass genotype QC; imputed calls additionally required DR2/R2 ≥ 0.8 and this sample's GP ≥ 0.90.

WHAT THIS PANEL COVERS

27	genes in this panel
7	genes with a result from this assay
20	genes without a prescribing-grade result
131	gene-medicine pairs
105	unique medicines
0	medication changes authorized from this report

Results were available for 7 genes: 3 were resolved within the tested marker set and 4 produced partial, non-unique findings. The remaining 20 genes could not be reliably resolved from this assay.

No medication or dose change is authorized from this report.

METABOLIZER GENES
CYP450 family - how fast or slow your liver clears drugs.

TRANSPORTER GENES
SLCO1B1, ABCG2 - how drugs move into and out of cells.

SAFETY GENES
HLA alleles - immune-mediated drug reaction risk.

GENES	MEDICINES	PROVISIONAL GUIDANCE	REVIEWED CHANGES
7	105	32	0
resolved of 27 analysed	across 131 gene-medicine pairs	confirmation / review	dose or medicine

20 genes did not produce a prescribing-grade result from this assay: CYP1A2, CYP2A6, CYP2B6, CYP2C8, CYP2C9, CYP2D6, CYP4F2, DPYD, G6PD, HLA-A*31:01, HLA-B*15:02, HLA-B*57:01, HLA-B*58:01, IFNL3, MT-RNR1, NUDT15, OPRM1, RYR1, TPMT, UGT1A1. Reasons vary by gene and can include missing or low-quality markers, assay limitations, or readable markers that did not yield a validated phenotype/diplotype. No normal-response assumption was made. Each gene is listed with its own reason in the inventory.

YOUR GENE PANEL

Quality-qualified phenotype calls; medicine action is not inferred from this matrix

CYP2C19	Intermediate Metabolizer (provisional; assayed variant set)	CYP3A5	Intermediate Expressor (provisional; assayed variant set)	ABCG2	Normal Function
CACNA1S	No assayed MH-associated variant detected	NAT2	rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established	SLCO1B1	Normal Function (provisional; assayed variant set)
VKORC1	Standard				

What your assayed gene results mean

These pages expand results and scientifically useful limited screens. “Resolved within the tested marker set” does not mean full-gene sequencing or universal medicine safety.

CYP2C19

Provisional result - confirmation advised

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

YOUR RESULT

Intermediate Metabolizer (provisional; assayed variant set)

DISPLAYED DIPLOTYPE / OBSERVATION

`*2 detected (one copy); remaining allele background not fully characterised`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

25 / 35 catalogued variant positions

CALLER MARKERS

`rs4244285` G/A · direct
`rs4986893` G/G · direct
`rs12248560` C/C · direct

HOW TO INTERPRET IT

A phenotype of Intermediate Metabolizer (provisional; assayed variant set), from the defining markers that were readable.

CONTRACTED MEDICINES LINKED TO THIS GENE

Amitriptyline · Citalopram · Clomipramine · Clopidogrel · Dexlansoprazole · Doxepin · Escitalopram · Imipramine · Lansoprazole · Omeprazole · Pantoprazole · Sertraline · Trimipramine · Voriconazole

Residual limitation: A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

CYP3A5

Provisional result - confirmation advised

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

YOUR RESULT

Intermediate Expressor (provisional; assayed variant set)

DISPLAYED DIPLOTYPE / OBSERVATION

`*3 detected (one copy); remaining allele background not fully characterised`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

5 / 7 catalogued variant positions

CALLER MARKERS

`rs776746` C/T · direct
`rs10264272` C/C · direct
`rs41303343` I/I · direct

HOW TO INTERPRET IT

A phenotype of Intermediate Expressor (provisional; assayed variant set), from the defining markers that were readable.

CONTRACTED MEDICINES LINKED TO THIS GENE

Tacrolimus

Residual limitation: A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

ABCG2

Resolved within the tested marker set

A transporter that moves several medicines across cell membranes and can influence drug exposure.

YOUR RESULT

Normal Function

DISPLAYED DIPLOTYPE / OBSERVATION

`*1/*1`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

1 / 1 catalogued variant positions

CALLER MARKERS

`rs2231142` G/G · direct

HOW TO INTERPRET IT

A phenotype of Normal Function from a uniquely supported *1/*1.

CONTRACTED MEDICINES LINKED TO THIS GENE

Allopurinol · Atorvastatin · Rosuvastatin

Residual limitation: Resolved within the tested marker set: one diplotype is supported by every defining marker this assay could read.

HELIXLINE

What your assayed gene results mean

These pages expand results and scientifically useful limited screens. “Resolved within the tested marker set” does not mean full-gene sequencing or universal medicine safety.

CACNA1S

A skeletal-muscle calcium-channel gene. A defined subset of variants is associated with malignant-hyperthermia susceptibility.

Complete for the defined assay subset - malignant hyperthermia susceptibility is not excluded

YOUR RESULT

No assayed MH-associated variant detected

DISPLAYED DIPLOTYPE / OBSERVATION

Negative

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

2 / 2 catalogued variant positions

CALLER MARKERS

rs772226819 G/G · direct

rs1800559 C/C · direct

HOW TO INTERPRET IT

A phenotype of No assayed MH-associated variant detected from a uniquely supported Negative.

CONTRACTED MEDICINES LINKED TO THIS GENE

Desflurane · Enflurane · Halothane · Isoflurane · Methoxyflurane · Sevoflurane · Succinylcholine

Residual limitation: This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

CYP2C9

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

Limited screen - phenotype not determined

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

Not uniquely resolved

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

44 / 83 catalogued variant positions

CALLER MARKERS

rs1799853 C/C · direct

rs1057910 A/A · direct

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 2 of 2 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

Celecoxib · Flurbiprofen · Fluvastatin · Fosphenytoin · Glimepiride · Glipizide · Glyburide · Ibuprofen · Lornoxicam · Meloxicam · Phenytoin · Piroxicam · Siponimod · Tenoxicam · Tolbutamide · Warfarin

Residual limitation: This gene could not be resolved from this DNA file, so no phenotype-based medication conclusion is drawn from it.

CYP4F2

An enzyme in the vitamin-K oxidation pathway that contributes one part of warfarin dose variability.

Limited screen - phenotype not determined

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

Not uniquely resolved

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

5 / 20 catalogued variant positions

CALLER MARKERS

rs2108622 C/C · direct

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 1 of 1 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

Warfarin

Residual limitation: This gene could not be resolved from this DNA file, so no phenotype-based medication conclusion is drawn from it.

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DPYD

Limited screen - phenotype not determined

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

Not uniquely resolved

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

76 / 83 catalogued variant positions

CALLER MARKERS

rs3918290 C/C · direct
rs55886062 A/A · direct
rs67376798 T/T · direct
rs75017182 G/G · direct

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 4 of 4 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

Capecitabine · Fluorouracil (5-FU)

Residual limitation: This gene could not be resolved from this DNA file, so no phenotype-based medication conclusion is drawn from it.

G6PD

Limited screen - phenotype not determined

An enzyme that protects red blood cells from oxidative stress; deficient activity can increase haemolysis risk with certain medicines.

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

Not uniquely resolved

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

59 / 173 catalogued variant positions

CALLER MARKERS

rs1050828 C/C · direct
rs1050829 T/T · direct
rs5030868 G/G · direct
rs72554665 C/C · direct

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 4 of 4 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

Chloroquine · Dapsone · Methylene blue · Nitrofurantoin · Pegloticase · Primaquine · Rasburicase · Tafenoquine · Toluidine blue

Residual limitation: G6PD is X-linked and this sample is genetically male, so there is a single copy. A variant is present or absent, never a carrier state.

MT-RNR1

Limited screen - phenotype not determined

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

Not uniquely resolved

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

8 / 22 catalogued variant positions

CALLER MARKERS

m.1555A>G A/A

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 1 of 2 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

Amikacin · Dibekacin · Gentamicin · Kanamycin · Neomycin · Netilmicin · Paromomycin · Plazomicin · Ribostamycin · Streptomycin · Tobramycin

Residual limitation: MT-RNR1 is mitochondrial: variants are haploid and inherited maternally, so diploid genotype and diplotype semantics do not apply.

HELIXLINE

What your assayed gene results mean

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NAT2

Provisional result - confirmation advised

An acetylation enzyme involved in processing medicines including isoniazid and hydralazine.

YOUR RESULT

rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established

DISPLAYED DIPLOTYPE / OBSERVATION

`rs1495741 A/A proxy observation`

MEASUREMENT BASIS

directly genotyped proxy/tag marker; target allele or phenotype not directly typed

WHAT WAS ASSESSABLE

24 / 47 catalogued variant positions

CALLER MARKERS

`rs1495741` A/A · proxy

HOW TO INTERPRET IT

A phenotype of rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established, from the defining markers that were readable.

CONTRACTED MEDICINES LINKED TO THIS GENE

Hydralazine · Isoniazid (INH)

Residual limitation: This result is inferred from a linked proxy marker rather than direct typing of the allele itself, and requires laboratory confirmation before it is used in prescribing.

NUDT15

Limited screen - phenotype not determined

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

`Not uniquely resolved`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

3 / 20 catalogued variant positions

CALLER MARKERS

`rs116855232` C/C · direct

`rs186364861` G/G · direct

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 2 of 2 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

6-Mercaptopurine · Azathioprine · Thioguanine

Residual limitation: This gene could not be resolved from this DNA file, so no phenotype-based medication conclusion is drawn from it.

RYR1

Limited screen - phenotype not determined

A skeletal-muscle calcium-release gene in which specific variants are associated with malignant-hyperthermia susceptibility.

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

`Not uniquely resolved`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

93 / 320 catalogued variant positions

CALLER MARKERS

`rs118192172` C/C · direct

`rs118192175` C/C · direct

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 2 of 2 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

Desflurane · Enflurane · Halothane · Isoflurane · Methoxyflurane · Sevoflurane · Succinylcholine

Residual limitation: This assay reads 2 of the 2 assayed marker positions for RYR1. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

HELIXLINE

What your assayed gene results mean

These pages expand results and scientifically useful limited screens. “Resolved within the tested marker set” does not mean full-gene sequencing or universal medicine safety.

SLCO1B1

Provisional result - confirmation advised

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

YOUR RESULT

Normal Function (provisional; assayed variant set)

DISPLAYED DIPLOTYPE / OBSERVATION

`*1B detected (one copy); remaining allele background not fully characterised`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

24 / 35 catalogued variant positions

CALLER MARKERS

rs4149056 T/T · direct
rs2306283 A/G · direct

HOW TO INTERPRET IT

A phenotype of Normal Function (provisional; assayed variant set), from the defining markers that were readable.

CONTRACTED MEDICINES LINKED TO THIS GENE

Atorvastatin · Fluvastatin · Lovastatin · Pitavastatin · Pravastatin · Rosuvastatin · Simvastatin

Residual limitation: A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

TPMT

Limited screen - phenotype not determined

A pharmacogene included because published evidence links variation in it to one or more medicines in the panel.

YOUR RESULT

Phenotype not determined from this assay

DISPLAYED DIPLOTYPE / OBSERVATION

`Not uniquely resolved`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

29 / 45 catalogued variant positions

CALLER MARKERS

rs1800462 C/C · direct
rs1800460 C/C · direct
rs1142345 T/T · direct

HOW TO INTERPRET IT

No reduced-function or risk variant was observed among the 3 of 3 caller-defining markers that passed quality control. This is a limited screening observation, not a normal-function result, because the assay cannot exclude variants it did not assess.

CONTRACTED MEDICINES LINKED TO THIS GENE

6-Mercaptopurine · Azathioprine · Thioguanine

Residual limitation: This gene could not be resolved from this DNA file, so no phenotype-based medication conclusion is drawn from it.

VKORC1

Resolved within the tested marker set

The pharmacological target of warfarin and a major contributor to warfarin dose sensitivity.

YOUR RESULT

Standard

DISPLAYED DIPLOTYPE / OBSERVATION

`-1639 G/G (rs9923231)`

MEASUREMENT BASIS

direct genotyping

WHAT WAS ASSESSABLE

1 / 1 catalogued variant positions

CALLER MARKERS

rs9923231 C/C · direct

HOW TO INTERPRET IT

A phenotype of Standard from a uniquely supported -1639 G/G (rs9923231).

CONTRACTED MEDICINES LINKED TO THIS GENE

Warfarin

Residual limitation: Resolved within the tested marker set: one diplotype is supported by every defining marker this assay could read.

HELIXLINE

GENOTYPE-BASED MEDICATION ALERTS

SHARE THIS PAGE WITH YOUR DOCTOR

This page reports whether your genotype authorizes any change to a medication or its dose. It does not tell you a medicine is safe or appropriate for you - that remains a clinical decision your prescriber makes from your indication, other medicines, organ function and history.

No genotype-based prescribing change is authorized from this report. Standard clinical prescribing, contraindications, and monitoring still apply.

No actionable alerts identified from the resolvable contracted pairs. Genes whose defining markers could not be resolved are reported in the limitations inventory; an unresolved gene is not a negative result. Alerts are raised only for actionable gene-drug pairs.

HLA ALLELE STATUS - IMMUNE DRUG-REACTION GENES

These immune-recognition genes are linked to severe allergic and skin reactions. Array tag SNPs are population-specific proxies and are not reported as HLA allele calls. Confirmatory high-resolution laboratory HLA typing is required before clinical use.

HLA-A*31:01	NOT RESOLVABLE	Carbamazepine	HLA-A*31:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.
HLA-B*15:02	NOT RESOLVABLE	Carbamazepine, Oxcarbazepine, Phenytoin	HLA-B*15:02 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.
HLA-B*57:01	NOT RESOLVABLE	Abacavir (HIV)	HLA-B*57:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.
HLA-B*58:01	NOT RESOLVABLE	Allopurinol (gout)	HLA-B*58:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.

HOW TO USE THESE RESULTS SAFELY

ARRAY TAG MARKERS

A tag SNP is not an HLA allele result. Confirm the named allele with accredited high-resolution HLA typing before any medication decision.

NOT RESOLVABLE

The assay could not establish the allele. This is not a negative result and must not be used as an all-clear.

NO AUTHORIZED CHANGE

Do not start, stop, avoid, or change a dose from this page alone. The treating clinician applies confirmed results in full clinical context.

BRING TO A PRESCRIBING VISIT: this report, the exact medicine and dose, current medicines and supplements, allergies or past reactions, indication, and relevant kidney/liver information. A clinician may request confirmatory testing.

YOUR PGX BY CLINICAL SPECIALTY

HELIXLINE

QUICK NAVIGATION · BY SPECIALTY, AND BY MEDICINE A-Z

Two ways to find a pair: the cards group every contracted pair by therapeutic area, and the index below lists every medicine alphabetically with the gene assessed for it. The number beside a medicine is that pair's place in the 131-pair inventory, printed again on its row in the detail pages and reference tables. Neither index is a prescription or a medicine-safety clearance.

Cardiology

17 CONTRACTED PAIRS

11 SOURCE REVIEW

GENES ABCG2 · CYP2C19 · CYP2C9 · CYP2D6 · CYP4F2 · NAT2 · SLCO1B1 · VKORC1

Psychiatry

26 CONTRACTED PAIRS

8 SOURCE REVIEW

GENES CYP1A2 · CYP2B6 · CYP2C19 · CYP2D6

Pain & Anaesthesia

26 CONTRACTED PAIRS

7 SOURCE REVIEW

GENES CACNA1S · CYP2B6 · CYP2C9 · CYP2D6 · OPRM1 · RYR1

Oncology

12 CONTRACTED PAIRS

NO HIGH-PRIORITY ALERT

GENES CYP2C8 · CYP2D6 · DPYD · G6PD · NUDT15 · TPMT · UGT1A1

Infectious Disease

22 CONTRACTED PAIRS

1 SOURCE REVIEW

GENES CYP2B6 · CYP2C19 · G6PD · HLA-B*57:01 · MT-RNR1 · NAT2 · UGT1A1

Gastroenterology

8 CONTRACTED PAIRS

4 SOURCE REVIEW

GENES CYP2C19 · CYP2D6 · IFNL3

Neurology

8 CONTRACTED PAIRS

NO HIGH-PRIORITY ALERT

GENES CYP2C9 · HLA-A*31:01 · HLA-B*15:02

Immunology & Transplant

4 CONTRACTED PAIRS

1 SOURCE REVIEW

GENES ABCG2 · CYP3A5 · G6PD · HLA-B*58:01

Diabetology

5 CONTRACTED PAIRS

NO HIGH-PRIORITY ALERT

GENES CYP2C8 · CYP2C9

Smoking Cessation

1 CONTRACTED PAIR

NO HIGH-PRIORITY ALERT

GENES CYP2A6

Other

2 CONTRACTED PAIRS

NO HIGH-PRIORITY ALERT

GENES G6PD

MEDICINE INDEX · A TO Z

33 medicines · 35 of 131 contracted pairs

A	Allopurinol	ABCG2	35
	Amitriptyline	CYP2C19	12
	Atorvastatin	SLCO1B1 · ABCG2	4, 33
C	Citalopram	CYP2C19	13
	Clomipramine	CYP2C19	14
	Clopidogrel	CYP2C19	2
D	Desflurane	CACNA1S	20
	Dexlansoprazole	CYP2C19	28
	Doxepin	CYP2C19	15
E	Enflurane	CACNA1S	21
	Escitalopram	CYP2C19	16
F	Fluvastatin	SLCO1B1	5
H	Halothane	CACNA1S	22
	Hydralazine	NAT2	3
I	Imipramine	CYP2C19	17
	Isoflurane	CACNA1S	23
	Isoniazid (INH)	NAT2	34
L	Lansoprazole	CYP2C19	29
	Lovastatin	SLCO1B1	6
M	Methoxyflurane	CACNA1S	24
O	Omeprazole	CYP2C19	30
P	Pantoprazole	CYP2C19	31
	Pitavastatin	SLCO1B1	7
	Pravastatin	SLCO1B1	8
R	Rosuvastatin	ABCG2 · SLCO1B1	1, 9
S	Sertraline	CYP2C19	18
	Sevoflurane	CACNA1S	25
	Simvastatin	SLCO1B1	10
	Succinylcholine	CACNA1S	26
T	Tacrolimus	CYP3A5	32
	Trimipramine	CYP2C19	19
V	Voriconazole	CYP2C19	27
W	Warfarin	VKORC1	11

PHARMACOGENOMICS - DRUG DETAIL SECTION

Cardiology

Quality-qualified gene-medicine associations in this specialty. Exact action wording appears only when it has passed clinical review.

17	0	11	0	1	5
PAIRS IN THIS SECTION	ACTION REQUIRED	SOURCE REVIEW	NO CITABLE GUIDANCE	RESEARCH CONTEXT	GENE NOT RESOLVED

GENE-MEDICINE PAIRS IN THIS SECTION - 17 PAIRS

Atorvastatin	ABCG2	Research context - no clinical guidance
Rosuvastatin	ABCG2	Published relationship - no dose instruction
Clopidogrel	CYP2C19	Partial evidence - confirmation required
Fluvastatin	CYP2C9	Not determined by this assay
Warfarin	CYP2C9	Not determined by this assay
Flecainide	CYP2D6	Not determined by this assay
Metoprolol	CYP2D6	Not determined by this assay
Warfarin	CYP4F2	Not determined by this assay
Hydralazine	NAT2	Published relationship - no dose instruction
Atorvastatin	SLCO1B1	Partial evidence - confirmation required
Fluvastatin	SLCO1B1	Partial evidence - confirmation required
Lovastatin	SLCO1B1	Partial evidence - confirmation required
Pitavastatin	SLCO1B1	Partial evidence - confirmation required
Pravastatin	SLCO1B1	Partial evidence - confirmation required
Rosuvastatin	SLCO1B1	Partial evidence - confirmation required
Simvastatin	SLCO1B1	Partial evidence - confirmation required
Warfarin	VKORC1	Published relationship - no dose instruction

WHAT THESE RESULT STATES MEAN

Resolved within tested marker set	The gene was resolved to a single diplotype and the pair carries current pair-specific guidance.
Partial evidence - confirmation required	Something real was measured, but this assay did not finish the call. Confirmatory testing is required before prescribing decisions.
Published relationship - no dose instruction	The pair has a source-linked relationship, but this report carries no validated dose or medicine change for it.
Research context - no clinical guidance	A published association exists that has not been graded as clinically actionable.
Not determined by this assay	This sample and chip could not resolve the gene, so no prescribing-grade result was produced for this pair.

Pairs with a medicine-specific consideration have their own detail page; the rest are listed with their state in the reference tables. Nothing on these pages is a prescription.

PHARMACOGENOMICS - DRUG DETAIL SECTION

Psychiatry

Quality-qualified gene-medicine associations in this specialty. Exact action wording appears only when it has passed clinical review.

26	0	8	0	0	18
PAIRS IN THIS SECTION	ACTION REQUIRED	SOURCE REVIEW	NO CITABLE GUIDANCE	RESEARCH CONTEXT	GENE NOT RESOLVED

GENE-MEDICINE PAIRS IN THIS SECTION - 26 PAIRS

Clozapine	CYP1A2	Not determined by this assay	
Fluvoxamine	CYP1A2	Not determined by this assay	
Sertraline	CYP2B6	Not determined by this assay	
Amitriptyline	CYP2C19	Partial evidence - confirmation required	
Citalopram	CYP2C19	Partial evidence - confirmation required	
Clomipramine	CYP2C19	Partial evidence - confirmation required	
Doxepin	CYP2C19	Partial evidence - confirmation required	
Escitalopram	CYP2C19	Partial evidence - confirmation required	
Imipramine	CYP2C19	Partial evidence - confirmation required	
Sertraline	CYP2C19	Partial evidence - confirmation required	
Trimipramine	CYP2C19	Partial evidence - confirmation required	
Amitriptyline	CYP2D6	Not determined by this assay	
Aripiprazole	CYP2D6	Not determined by this assay	
Atomoxetine	CYP2D6	Not determined by this assay	
Clomipramine	CYP2D6	Not determined by this assay	
Desipramine	CYP2D6	Not determined by this assay	
Doxepin	CYP2D6	Not determined by this assay	
Fluvoxamine	CYP2D6	Not determined by this assay	
Haloperidol	CYP2D6	Not determined by this assay	
Imipramine	CYP2D6	Not determined by this assay	
Nortriptyline	CYP2D6	Not determined by this assay	
Paroxetine	CYP2D6	Not determined by this assay	
Risperidone	CYP2D6	Not determined by this assay	
Trimipramine	CYP2D6	Not determined by this assay	
Venlafaxine	CYP2D6	Not determined by this assay	
Vortioxetine	CYP2D6	Not determined by this assay	

WHAT THESE RESULT STATES MEAN

Resolved within tested marker set	The gene was resolved to a single diplotype and the pair carries current pair-specific guidance.
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Not determined by this assay	This sample and chip could not resolve the gene, so no prescribing-grade result was produced for this pair.

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PHARMACOGENOMICS - DRUG DETAIL SECTION

Pain & Anaesthesia

Quality-qualified gene-medicine associations in this specialty. Exact action wording appears only when it has passed clinical review.

26	0	7	0	0	19
PAIRS IN THIS SECTION	ACTION REQUIRED	SOURCE REVIEW	NO CITABLE GUIDANCE	RESEARCH CONTEXT	GENE NOT RESOLVED

GENE-MEDICINE PAIRS IN THIS SECTION - 26 PAIRS

Desflurane	CACNA1S	Published relationship - no dose instruction	
Enflurane	CACNA1S	Published relationship - no dose instruction	
Halothane	CACNA1S	Published relationship - no dose instruction	
Isoflurane	CACNA1S	Published relationship - no dose instruction	
Methoxyflurane	CACNA1S	Published relationship - no dose instruction	
Sevoflurane	CACNA1S	Published relationship - no dose instruction	
Succinylcholine	CACNA1S	Published relationship - no dose instruction	
Methadone	CYP2B6	Not determined by this assay	
Celecoxib	CYP2C9	Not determined by this assay	
Flurbiprofen	CYP2C9	Not determined by this assay	
Ibuprofen	CYP2C9	Not determined by this assay	
Lornoxicam	CYP2C9	Not determined by this assay	
Meloxicam	CYP2C9	Not determined by this assay	
Piroxicam	CYP2C9	Not determined by this assay	
Tenoxicam	CYP2C9	Not determined by this assay	
Codeine	CYP2D6	Not determined by this assay	
Hydrocodone	CYP2D6	Not determined by this assay	
Tramadol	CYP2D6	Not determined by this assay	
Morphine	OPRM1	Not determined by this assay	
Desflurane	RYR1	Not determined by this assay	
Enflurane	RYR1	Not determined by this assay	
Halothane	RYR1	Not determined by this assay	
Isoflurane	RYR1	Not determined by this assay	
Methoxyflurane	RYR1	Not determined by this assay	
Sevoflurane	RYR1	Not determined by this assay	
Succinylcholine	RYR1	Not determined by this assay	

WHAT THESE RESULT STATES MEAN

Resolved within tested marker set	The gene was resolved to a single diplotype and the pair carries current pair-specific guidance.
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Published relationship - no dose instruction	The pair has a source-linked relationship, but this report carries no validated dose or medicine change for it.
Research context - no clinical guidance	A published association exists that has not been graded as clinically actionable.
Not determined by this assay	This sample and chip could not resolve the gene, so no prescribing-grade result was produced for this pair.

Pairs with a medicine-specific consideration have their own detail page; the rest are listed with their state in the reference tables. Nothing on these pages is a prescription.

PHARMACOGENOMICS - DRUG DETAIL SECTION

Gastroenterology

Quality-qualified gene-medicine associations in this specialty. Exact action wording appears only when it has passed clinical review.

8	0	4	0	0	4
PAIRS IN THIS SECTION	ACTION REQUIRED	SOURCE REVIEW	NO CITABLE GUIDANCE	RESEARCH CONTEXT	GENE NOT RESOLVED

GENE-MEDICINE PAIRS IN THIS SECTION - 8 PAIRS

Dexlansoprazole	CYP2C19	Partial evidence - confirmation required	
Lansoprazole	CYP2C19	Partial evidence - confirmation required	
Omeprazole	CYP2C19	Partial evidence - confirmation required	
Pantoprazole	CYP2C19	Partial evidence - confirmation required	
Ondansetron	CYP2D6		Not determined by this assay
Tropisetron	CYP2D6		Not determined by this assay
Peginterferon alfa-2a	IFNL3		Not determined by this assay
Peginterferon alfa-2b	IFNL3		Not determined by this assay

WHAT THESE RESULT STATES MEAN

Resolved within tested marker set	The gene was resolved to a single diplotype and the pair carries current pair-specific guidance.
Partial evidence - confirmation required	Something real was measured, but this assay did not finish the call. Confirmatory testing is required before prescribing decisions.
Published relationship - no dose instruction	The pair has a source-linked relationship, but this report carries no validated dose or medicine change for it.
Research context - no clinical guidance	A published association exists that has not been graded as clinically actionable.
Not determined by this assay	This sample and chip could not resolve the gene, so no prescribing-grade result was produced for this pair.

Pairs with a medicine-specific consideration have their own detail page; the rest are listed with their state in the reference tables. Nothing on these pages is a prescription.

Rosuvastatin

HELIXLINE

ABCG2

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Rosuvastatin is also contracted against SLCO1B1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
ABCG2	*1/*1	Normal Function	direct genotyping

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Rosuvastatin** and that the linked result is **ABCG2**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · ABCG2-Rosuvastatin · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-statins-and-slco1b1-abcg2-and-cyp2c9/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Clopidogrel

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Clopidogrel** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Clopidogrel · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/guideline-for-clopidogrel-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Hydralazine

HELIXLINE

NAT2

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
NAT2	rs1495741 A/A proxy observation	rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established	directly genotyped proxy/tag marker; target allele or phenotype not directly typed

EVIDENCE NOT UNIQUELY RESOLVED

This result is inferred from a linked proxy marker rather than direct typing of the allele itself, and requires laboratory confirmation before it is used in prescribing.

Observed: Predicted slow acetylator (single-SNP proxy)

Evidence: CPIC · checked 2026-08-20 · **Result source:** PROXY · **Confirmation:** Confirm the proxy-based result with a validated clinical PGx assay before prescribing use.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Hydralazine** and that the linked result is **NAT2**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · NAT2-Hydralazine · evidence checked 2026-08-20
https://files.cpicpgx.org/data/guideline/publication/hydralazine/2025/40974042.pdf

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Atorvastatin

HELIXLINE

SLCO1B1

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Atorvastatin is also contracted against ABCG2. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
SLCO1B1	*1B detected (one copy); remaining allele background not fully characterised	Normal Function (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*1B

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Atorvastatin** and that the linked result is **SLCO1B1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · SLCO1B1-Atorvastatin · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-statins-and-slco1b1-abcg2-and-cyp2c9/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Fluvastatin

HELIXLINE

SLCO1B1

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Fluvastatin is also contracted against CYP2C9. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
SLCO1B1	*1B detected (one copy); remaining allele background not fully characterised	Normal Function (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*1B

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Fluvastatin** and that the linked result is **SLCO1B1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · SLCO1B1-Fluvastatin · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-statins-and-slco1b1-abcg2-and-cyp2c9/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Lovastatin

HELIXLINE

SLCO1B1

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
SLCO1B1	*1B detected (one copy); remaining allele background not fully characterised	Normal Function (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*1B

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Lovastatin** and that the linked result is **SLCO1B1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · SLCO1B1-Lovastatin · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166264281

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Pitavastatin

HELIXLINE

SLCO1B1

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
SLCO1B1	*1B detected (one copy); remaining allele background not fully characterised	Normal Function (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*1B

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Pitavastatin** and that the linked result is **SLCO1B1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · SLCO1B1-Pitavastatin · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166264281

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Pravastatin

HELIXLINE

SLCO1B1

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
SLCO1B1	*1B detected (one copy); remaining allele background not fully characterised	Normal Function (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*1B

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Pravastatin** and that the linked result is **SLCO1B1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · SLCO1B1-Pravastatin · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-statins-and-slco1b1-abcg2-and-cyp2c9/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Rosuvastatin

HELIXLINE

SLCO1B1

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Rosuvastatin is also contracted against ABCG2. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
SLCO1B1	*1B detected (one copy); remaining allele background not fully characterised	Normal Function (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*1B

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Rosuvastatin** and that the linked result is **SLCO1B1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · SLCO1B1-Rosuvastatin · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-statins-and-slco1b1-abcg2-and-cyp2c9/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Simvastatin

HELIXLINE

SLCO1B1

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
SLCO1B1	*1B detected (one copy); remaining allele background not fully characterised	Normal Function (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*1B

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Simvastatin** and that the linked result is **SLCO1B1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · SLCO1B1-Simvastatin · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-statins-and-slco1b1-abcg2-and-cyp2c9/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Warfarin

HELIXLINE

VKORC1

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Warfarin is also contracted against CYP2C9, CYP4F2. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
VKORC1	-1639 G/G (rs9923231)	Standard	direct genotyping

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Warfarin** and that the linked result is **VKORC1**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · VKORC1-Warfarin · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/guideline-for-warfarin-and-cyp2c9-and-vkorc1/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Amitriptyline

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Amitriptyline is also contracted against CYP2D6. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Amitriptyline** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Amitriptyline · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/guideline-for-tricyclic-antidepressants-and-cyp2d6-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Citalopram

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Citalopram** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Citalopram · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/guideline-for-selective-serotonin-reuptake-inhibitors-and-cyp2d6-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Clomipramine

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL B

YOUR DNA RESULT

Clomipramine is also contracted against CYP2D6. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Clomipramine** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) B · CYP2C19-Clomipramine · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251445

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Doxepin

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL B

YOUR DNA RESULT

Doxepin is also contracted against CYP2D6. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Doxepin** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) B · CYP2C19-Doxepin · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251445

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Escitalopram

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Escitalopram** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Escitalopram · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/guideline-for-selective-serotonin-reuptake-inhibitors-and-cyp2d6-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Imipramine

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL B

YOUR DNA RESULT

Imipramine is also contracted against CYP2D6. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Imipramine** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) B · CYP2C19-Imipramine · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251445

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Sertraline

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Sertraline is also contracted against CYP2B6. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Sertraline** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Sertraline · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/guideline-for-selective-serotonin-reuptake-inhibitors-and-cyp2d6-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Trimipramine

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL B

YOUR DNA RESULT

Trimipramine is also contracted against CYP2D6. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Trimipramine** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) B · CYP2C19-Trimipramine · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251445

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Desflurane

HELIXLINE

CACNA1S

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Desflurane is also contracted against RYR1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CACNA1S	Negative	No assayed MH-associated variant detected	direct genotyping

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context. · **Limits:** This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Desflurane** and that the linked result is **CACNA1S**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CACNA1S-Desflurane · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-volatile-anesthetic-agents-and-malignant-hyperthermia/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Enflurane

HELIXLINE

CACNA1S

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Enflurane is also contracted against RYR1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CACNA1S	Negative	No assayed MH-associated variant detected	direct genotyping

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context. · **Limits:** This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Enflurane** and that the linked result is **CACNA1S**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · CACNA1S-Enflurane · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251460

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Halothane

HELIXLINE

CACNA1S

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Halothane is also contracted against RYR1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CACNA1S	Negative	No assayed MH-associated variant detected	direct genotyping

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context. · **Limits:** This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Halothane** and that the linked result is **CACNA1S**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · CACNA1S-Halothane · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251460

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Isoflurane

HELIXLINE

CACNA1S

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Isoflurane is also contracted against RYR1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CACNA1S	Negative	No assayed MH-associated variant detected	direct genotyping

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context. · **Limits:** This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Isoflurane** and that the linked result is **CACNA1S**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · CACNA1S-Isoflurane · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251460

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Methoxyflurane

HELIXLINE

CACNA1S

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Methoxyflurane is also contracted against RYR1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CACNA1S	Negative	No assayed MH-associated variant detected	direct genotyping

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context. · **Limits:** This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Methoxyflurane** and that the linked result is **CACNA1S**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · CACNA1S-Methoxyflurane · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251460

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Sevoflurane

HELIXLINE

CACNA1S

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Sevoflurane is also contracted against RYR1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CACNA1S	Negative	No assayed MH-associated variant detected	direct genotyping

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context. · **Limits:** This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Sevoflurane** and that the linked result is **CACNA1S**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · CACNA1S-Sevoflurane · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251460

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Succinylcholine

HELIXLINE

CACNA1S

Published relationship - no dose instruction

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

Succinylcholine is also contracted against RYR1. Each gene-medicine pair holds its own state and its own page; this report does not calculate a combined dose from them.

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CACNA1S	Negative	No assayed MH-associated variant detected	direct genotyping

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Resolved within the tested marker set; interpret with the linked guideline and the patient's clinical context. · **Limits:** This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

A source-linked gene-medicine relationship was identified for this result. This report provides no dose or medicine change for the pair; review the linked source with the prescriber if the medicine is being considered. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Association Only, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Succinylcholine** and that the linked result is **CACNA1S**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This result is exact for the caller's assayed markers, not whole-gene sequencing.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CACNA1S-Succinylcholine · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-volatile-anesthetic-agents-and-malignant-hyperthermia/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Voriconazole

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Voriconazole** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Voriconazole · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-voriconazole-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Dexlansoprazole

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL B

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Dexlansoprazole** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) B · CYP2C19-Dexlansoprazole · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251441

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Lansoprazole

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Lansoprazole** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Lansoprazole · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-proton-pump-inhibitors-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Omeprazole

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Omeprazole** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP2C19-Omeprazole · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/cpic-guideline-for-proton-pump-inhibitors-and-cyp2c19/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Pantoprazole

HELIXLINE

CYP2C19

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP2C19	*2 detected (one copy); remaining allele background not fully characterised	Intermediate Metabolizer (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*2

Evidence: ClinPGx/DPWG annotation · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Pantoprazole** and that the linked result is **CYP2C19**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: ClinPGx/DPWG annotation · CPIC-aligned A/B/C/D (ClinPGx-hosted) A · CYP2C19-Pantoprazole · evidence checked 2026-08-20
https://www.clinpgx.org/guideline/PA166251441

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Tacrolimus

HELIXLINE

CYP3A5

Partial evidence - confirmation required

SOURCE REVIEW | CPIC PAIR LEVEL A

YOUR DNA RESULT

GENE	YOUR TWO GENE COPIES	WHAT THEY DO	HOW IT WAS READ
CYP3A5	*3 detected (one copy); remaining allele background not fully characterised	Intermediate Expressor (provisional; assayed variant set)	direct genotyping

EVIDENCE NOT UNIQUELY RESOLVED

A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

Observed: *1/*3

Evidence: CPIC · checked 2026-08-20 · **Result source:** DIRECT · **Confirmation:** Confirm with a validated clinical PGx assay before prescribing use. · **Limits:** A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.

IN PLAIN ENGLISH

There is a source-linked gene-medicine consideration here for your prescriber to review.

Assessed under a documented assay limitation; this pair is not fully resolved from this file. This page reports one gene-medicine pair. It does not establish that the medicine is appropriate, safe or unnecessary for you: your prescriber weighs it with your indication, other medicines, organ function and history.

WHAT YOU SHOULD DO

Do not start, stop, or change this medicine from this report alone. If this medicine is relevant to you, ask your prescriber to review the linked source and your full clinical context.

SOURCE-LINKED CLINICAL CONTEXT

- no validated individual dose statement is included for this pair

No dosing statement is printed for this pair. Its state is Partial, which does not carry approved prescribing wording.

BEFORE A PRESCRIBING DISCUSSION

1 · MATCH THE PAIR

Confirm that the medicine is **Tacrolimus** and that the linked result is **CYP3A5**. A different medicine or gene is a different PGx question.

2 · CHECK CERTAINTY

This is a provisional result. Ask whether a validated confirmatory assay is needed before the source is applied clinically.

3 · ADD CLINICAL CONTEXT

Bring the indication, current dose, all medicines and supplements, allergies, kidney/liver information, age, pregnancy status where relevant, and prior response.

SOURCE GUIDELINES

Open versioned guideline source · checked 2026-08-20

Printed source: CPIC · CPIC A/B/C/D A · CYP3A5-Tacrolimus · evidence checked 2026-08-20
https://cpicpgx.org/guidelines/guideline-for-tacrolimus-and-cyp3a5/

IMPORTANT LIMITS

This report covers genotype only. Liver and kidney function, age, diet, and co-medications independently influence medicine response. PGx does not replace therapeutic drug monitoring or clinical assessment.

Your Gene-Medicine Pairs at a Glance

Every pair here holds the state it actually has, and its badge is that pair's number in the contracted inventory. No row on this page is a prescription.

PERSONALIZED RESULTS · NO VALIDATED DOSE OR MEDICINE CHANGE - 3 gene-medicine pairs summarised for prescriber review. This does not establish a universally safe dose.

Reportable pairs 33-35 of 35 are on this page. The 32 pairs with a detail page are numbered first; the remaining 3 continue across 1 table. 32 + 3 reportable + 96 grouped limitations = 131 contracted pairs, each accounted for exactly once. Every medicine is indexed A-Z on the specialty page.

PAIR · MEDICINE	GENE TESTED	YOUR DIPLOTYPE	RESULT STATE	WHAT THIS ROW SAYS
33 **Atorvastatin** Cardiology	**ABCG2** also assessed: SLCO1B1	*1/*1	Research context - no clinical guidance Normal Function direct genotyping	This relationship is retained as research context. It is not presented as a dose, medicine choice or prediction that the medicine will work. Evidence source: CPIC · cpic-guideline-for-statins-and-slco1b1-abcg2-and-cyp2c9
34 **Isoniazid (INH)** Infectious Disease	**NAT2**	*rs1495741 A/A proxy observation*	Research context - no clinical guidance rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established directly genotyped proxy/tag marker; target allele or phenotype not directly typed	This relationship is retained as research context. It is not presented as a dose, medicine choice or prediction that the medicine will work. Catalog gap: CPIC lists this pair at level C with no published guideline (ClinPGx evidence 1B, 'Informative PGx'). No dosing authority exists to cite.
35 **Allopurinol** Immunology & Transplant	**ABCG2** also assessed: HLA-B*58:01	*1/*1	Research context - no clinical guidance Normal Function direct genotyping	This relationship is retained as research context. It is not presented as a dose, medicine choice or prediction that the medicine will work. Catalog gap: not present in the CPIC pair table (2026-08-01)

HELIXLINE

Factors that can change medicine response

A pharmacogene result is stable, but its clinical importance can change with the medicine, indication, dose and the person's current physiology.

Before a clinician uses this report: bring a current list of prescriptions, over-the-counter medicines and supplements, including dose and reason for use. The account QR remains on the wallet card so the latest report can be opened securely.

Other medicines

Enzyme inhibitors and inducers can make a genetically normal metabolizer behave functionally slower or faster. This is called phenoconversion.

Required input: prescription medicines, over-the-counter products, supplements and recent medication changes.

Liver and kidney function

Clearance can change independently of genotype. Organ impairment may dominate the dose decision even where a gene result is exact.

Required input: recent creatinine/eGFR, liver tests and the prescriber's assessment.

Age and life stage

Children, older adults, pregnancy and breastfeeding can have different dosing, safety and monitoring requirements that are not predicted by the DNA result.

The following life-stage page is populated only when reviewed guidance is applicable.

Smoking, diet and environment

Smoking can induce CYP1A2 activity; diet, alcohol and environmental exposures can also alter drug response. These factors were not inferred from DNA.

Record current status rather than assuming it from genotype or ancestry.

Safe order of interpretation

1. Confirm the exact medicine, indication and current dose.
2. Check whether the gene result is exact, provisional or not determined.
3. Check whether direct genotyping, quality-qualified imputation or a proxy contributed.
4. Read the current guideline or label for that exact gene-medicine-phenotype combination.
5. Apply interactions, organ function, age, history and monitoring before any treatment decision.

Never read "no approved PGx action" as an all-clear. It does not evaluate allergy, contraindications, drug interactions, pregnancy, organ function, diagnosis or medicines outside this report.

HELIXLINE

Pregnancy, Pediatric & Elderly Notes

Some medications on your report have life-stage specific guidance from CPIC or FDA labels. These notes apply when the drug is being considered for you or a family member in a particular life stage - they are additive to the DNA-based recommendation on the main drug page.

Only drugs from *your* panel with explicit life-stage guidance are listed. For a full list of FDA pregnancy categories or Beers-List elderly warnings, consult your clinician or pharmacist.

No result-specific life-stage instruction is printed for this report.

No applicable life-stage guidance. That is a scope statement-not evidence that every listed medicine is suitable at every age or life stage. Life-stage notes appear here only when an actionable gene-medicine row carries reviewed wording.

Pregnancy & breastfeeding

Medicine choice and monitoring can change because of fetal or infant exposure, maternal physiology and the condition being treated. Those questions are not answered by a pharmacogene result alone.

Children & adolescents

Age, weight, developmental pharmacology, formulation and indication can affect treatment independently of genotype. Pediatric prescribing requires an appropriate clinical source.

Older adults

Kidney and liver function, frailty, falls risk, co-morbidities and polypharmacy can outweigh or modify a genetic prediction. Review the complete medication list.

Practical rule: when pregnancy, breastfeeding, pediatric use or older age is relevant, bring this report to the treating clinician or pharmacist together with the exact medicine, dose, indication, current medication list and recent organ-function results.

HELIXLINE

EVERY GENE IN THE PANEL

Every gene this panel contracts to test is listed below - including the ones your DNA file could not resolve, each with the specific reason. **A gene that could not be resolved is never reported as a normal-metabolizer result.** Missing defining markers mean the result is unknown, not that your response is typical.

Marker and allele-catalogue counts for every gene are in the methodology appendix. A "normal" result cannot be fully certified where at least one defining variant was not readable: an untested rare variant and a genuinely typical result look the same on this DNA file, and the gene says so in its own row.

27	7	20	LOW CONFIDENCE
GENES IN PANEL	RESOLVED FROM YOUR FILE	NOT RESOLVABLE · REASON GIVEN	

Gene results: 7/27 genes resolvable from this file

EVERY MEDICINE PAIR THIS PANEL ASSESSES · 131 TOTAL

0	10	3	22	96
ACTIONABLE	ASSOCIATION ONLY	RESEARCH CONTEXT	PARTIAL	NOT RESOLVABLE

532 of 830 catalogued markers were readable from your file (528 measured directly, 4 recovered by quality-qualified imputation). Association-only results state the published association and its source without a dose; research context is not prescribing guidance.

G6PD ploidy safeguard: G6PD is X-linked: this genetically male sample has a hemizygous single-copy context, never a carrier state.

GENE	WHAT THIS ARRAY COULD DETERMINE	WHAT IT COULD NOT DETERMINE	WHAT THIS MEANS FOR THIS REPORT
ABCG2 Normal Function	A phenotype of Normal Function from a uniquely supported *1/*1.	Nothing required for this gene's call was missing.	Medicines contracted against this gene are reported against this result. No dose or medicine change is authorized from it without prescriber review.
CACNA1S No assayed MH-associated variant detected	A phenotype of No assayed MH-associated variant detected from a uniquely supported Negative.	This assay reads 2 of the 2 assayed marker positions for CACNA1S. These positions represent a defined subset rather than the complete set of variants associated with the condition. A result showing no variant detected therefore does NOT exclude malignant-hyperthermia susceptibility, and must not be used to decide that volatile anaesthetics or succinylcholine are safe. Personal and family history, and where indicated in-vitro contracture testing, remain the basis for that decision.	Medicines contracted against this gene are reported against this result. No dose or medicine change is authorized from it without prescriber review.
CYP1A2 NOT RESOLVABLE	No result could be established for this gene from this file.	1/2 defining variants are missing, no-call, or below imputation thresholds (DR2 0.80, GP 0.90); gene withheld (rs2069514=imputed/dr2_below_floor).	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
CYP2A6 NOT RESOLVABLE	No result could be established for this gene from this file.	1/2 defining variants are missing, no-call, or below imputation thresholds (DR2 0.80, GP 0.90); gene withheld (rs1801272=imputed/dr2_below_floor).	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
CYP2B6 NOT RESOLVABLE	20 of the 48 markers that define this gene were readable - not enough for a call.	1/3 defining variants are missing, no-call, or below imputation thresholds (DR2 0.80, GP 0.90); gene withheld (rs2279343=missing/missing_from_vcf).	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
CYP2C19 Intermediate Metabolizer (provisional; assayed variant set)	A phenotype of Intermediate Metabolizer (provisional; assayed variant set), from the defining markers that were readable.	A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.	Medicines contracted against this gene are reported for source review, not as a prescribing conclusion. Confirmatory testing is required before this result is used in a prescribing decision.
CYP2C8 NOT RESOLVABLE	No result could be established for this gene from this file.	Marker coverage passed, but the frozen source snapshot contains only Level-C drug evidence; no customer prescribing result is released.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
CYP2C9 NOT RESOLVABLE	44 of the 83 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
CYP2D6 NOT RESOLVABLE	44 of the 157 markers that define this gene were readable - not enough for a call.	Withheld for this SNP-array assay: CYP2D6 copy number, structural variants, CYP2D6/CYP2D7 hybrid alleles, and a complete phased star-allele definition were not assessed. Confirm with a validated CYP2D6 assay before using this gene for prescribing.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.

IMPORTANT

Genotyping arrays cannot resolve copy number, hybrid alleles or complete haplotype phase for some genes; those genes are listed as not resolvable rather than assumed normal. **Never start, stop, or change a medication based on this report alone.** Discuss these results with your clinician or pharmacist.

EVERY GENE IN THE PANEL (continued)

27 GENES IN PANEL | **7** RESOLVED FROM YOUR FILE | **20** NOT RESOLVABLE · REASON GIVEN | LOW CONFIDENCE

Gene results: 7/27 genes resolvable from this file

GENE	WHAT THIS ARRAY COULD DETERMINE	WHAT IT COULD NOT DETERMINE	WHAT THIS MEANS FOR THIS REPORT
CYP3A5 Intermediate Expressor (provisional; assayed variant set)	A phenotype of Intermediate Expressor (provisional; assayed variant set), from the defining markers that were readable.	A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.	Medicines contracted against this gene are reported for source review, not as a prescribing conclusion. Confirmatory testing is required before this result is used in a prescribing decision.
CYP4F2 NOT RESOLVABLE	5 of the 20 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
DPYD NOT RESOLVABLE	76 of the 83 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
G6PD NOT RESOLVABLE	59 of the 173 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
HLA-A*31:01 NOT RESOLVABLE	No result could be established for this gene from this file.	HLA-A*31:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
HLA-B*15:02 NOT RESOLVABLE	No result could be established for this gene from this file.	HLA-B*15:02 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
HLA-B*57:01 NOT RESOLVABLE	No result could be established for this gene from this file.	HLA-B*57:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
HLA-B*58:01 NOT RESOLVABLE	No result could be established for this gene from this file.	HLA-B*58:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
IFNL3 NOT RESOLVABLE	All 1 defining marker was readable, but no validated phenotype or diplotype could be established.	Marker coverage passed, but the frozen source snapshot contains only Level-C/legacy peginterferon evidence; no customer prescribing result is released.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.

IMPORTANT

Genotyping arrays cannot resolve copy number, hybrid alleles or complete haplotype phase for some genes; those genes are listed as not resolvable rather than assumed normal. **Never start, stop, or change a medication based on this report alone.** Discuss these results with your clinician or pharmacist.

EVERY GENE IN THE PANEL (continued)

27 GENES IN PANEL | **7** RESOLVED FROM YOUR FILE | **20** NOT RESOLVABLE · REASON GIVEN

LOW CONFIDENCE

Gene results: 7/27 genes resolvable from this file

GENE	WHAT THIS ARRAY COULD DETERMINE	WHAT IT COULD NOT DETERMINE	WHAT THIS MEANS FOR THIS REPORT
MT-RNR1 NOT RESOLVABLE	8 of the 22 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
NAT2 rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established	A phenotype of rs1495741 A/A proxy suggests slower acetylation; NAT2 phenotype not established, from the defining markers that were readable.	This result is inferred from a linked proxy marker rather than direct typing of the allele itself, and requires laboratory confirmation before it is used in prescribing.	Medicines contracted against this gene are reported for source review, not as a prescribing conclusion. Confirmatory testing is required before this result is used in a prescribing decision.
NUDT15 NOT RESOLVABLE	3 of the 20 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
OPRM1 NOT RESOLVABLE	No result could be established for this gene from this file.	Marker coverage passed, but the frozen source snapshot contains only Level-C supportive evidence; no customer prescribing result is released.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
RYR1 NOT RESOLVABLE	93 of the 320 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
SLCO1B1 Normal Function (provisional; assayed variant set)	A phenotype of Normal Function (provisional; assayed variant set), from the defining markers that were readable.	A clinically relevant non-reference marker was observed, but this array cannot exclude every alternative allele or establish all required locus features. The observation is reported for source review and confirmation, not as a complete clinical diplotype or a stand-alone prescribing instruction.	Medicines contracted against this gene are reported for source review, not as a prescribing conclusion. Confirmatory testing is required before this result is used in a prescribing decision.
TPMT NOT RESOLVABLE	29 of the 45 markers that define this gene were readable - not enough for a call.	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
UGT1A1 NOT RESOLVABLE	3 of the 4 markers that define this gene were readable - not enough for a call.	A Normal Metabolizer result cannot be asserted on this assay: The clinically used UGT1A1*28 allele is a promoter TA-repeat length. It was not directly sized here; only linked SNPs were read.	Medicines contracted against this gene are listed with the reason instead of a result. An unresolved gene is not a normal result and no typical response has been assumed.
VKORC1 Standard	A phenotype of Standard from a uniquely supported -1639 G/G (rs9923231).	Nothing required for this gene's call was missing.	Medicines contracted against this gene are reported against this result. No dose or medicine change is authorized from it without prescriber review.

IMPORTANT

Genotyping arrays cannot resolve copy number, hybrid alleles or complete haplotype phase for some genes; those genes are listed as not resolvable rather than assumed normal. **Never start, stop, or change a medication based on this report alone.** Discuss these results with your clinician or pharmacist.

WHAT THIS ARRAY COULD NOT RESOLVE

HELIXLINE

UNKNOWN IS NOT NORMAL

The rows below consolidate every unresolved gene once and name all affected medicines. This replaces repetitive medicine-by-medicine "unresolved" pages without removing any contracted pair from the accounting.

20 genes · 96 affected gene-medicine pairs

GENE	WHY IT IS UNRESOLVED	AFFECTED MEDICINES	WHAT THIS MEANS
CYP1A2 2 pairs	1/2 defining variants are missing, no-call, or below imputation thresholds (DR2 0.80, GP 0.90); gene withheld (rs2069514=imputed/dr2_below_floor).	**Clozapine, Fluvoxamine**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
CYP2A6 1 pair	1/2 defining variants are missing, no-call, or below imputation thresholds (DR2 0.80, GP 0.90); gene withheld (rs1801272=imputed/dr2_below_floor).	**Nicotine (NRT)**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
CYP2B6 4 pairs	1/3 defining variants are missing, no-call, or below imputation thresholds (DR2 0.80, GP 0.90); gene withheld (rs2279343=missing/missing_from_vcf).	**Efavirenz, Methadone, Nevirapine, Sertraline**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
CYP2C8 2 pairs	Marker coverage passed, but the frozen source snapshot contains only Level-C drug evidence; no customer prescribing result is released.	**Paclitaxel, Rosiglitazone**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
CYP2C9 16 pairs	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**Celecoxib, Flurbiprofen, Fluvastatin, Fosphenytoin, Glimepiride, Glipizide, Glyburide, Ibuprofen, Lornoxicam, Meloxicam, Phenytoin, Piroxicam, Siponimod, Tenoxicam, Tolbutamide, Warfarin**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
CYP2D6 23 pairs	Withheld for this SNP-array assay: CYP2D6 copy number, structural variants, CYP2D6/CYP2D7 hybrid alleles, and a complete phased star-allele definition were not assessed. Confirm with a validated CYP2D6 assay before using this gene for prescribing.	**Amitriptyline, Aripiprazole, Atomoxetine, Clomipramine, Codeine, Desipramine, Doxepin, Flecainide, Fluvoxamine, Haloperidol, Hydrocodone, Imipramine, Metoprolol, Nortriptyline, Ondansetron, Paroxetine, Risperidone, Tamoxifen, Tramadol, Trimipramine, Tropisetron, Venlafaxine, Vortioxetine**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
CYP4F2 1 pair	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**Warfarin**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.

HELIXLINE

WHAT THIS ARRAY COULD NOT RESOLVE (continued)

GENE	WHY IT IS UNRESOLVED	AFFECTED MEDICINES	WHAT THIS MEANS
DPYD 2 pairs	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**Capecitabine, Fluorouracil (5-FU)**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
G6PD 9 pairs	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**Chloroquine, Dapsone, Methylene blue, Nitrofurantoin, Pegloticase, Primaquine, Rasburicase, Tafenoquine, Toluidine blue**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
HLA-A*31:01 1 pair	HLA-A*31:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	**Carbamazepine**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
HLA-B*15:02 4 pairs	HLA-B*15:02 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	**Carbamazepine, Fosphenytoin, Oxcarbazepine, Phenytoin**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
HLA-B*57:01 1 pair	HLA-B*57:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	**Abacavir**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
HLA-B*58:01 1 pair	HLA-B*58:01 risk alleles are defined at four-digit resolution. This assay carries only tag SNPs, which are population-specific proxies for the allele rather than the allele itself, and a proxy cannot certify presence or absence of a hypersensitivity risk allele. Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.	**Allopurinol**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
IFNL3 2 pairs	Marker coverage passed, but the frozen source snapshot contains only Level-C/legacy peginterferon evidence; no customer prescribing result is released.	**Peginterferon alfa-2a, Peginterferon alfa-2b**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.

HELIXLINE

WHAT THIS ARRAY COULD NOT RESOLVE (continued)

GENE	WHY IT IS UNRESOLVED	AFFECTED MEDICINES	WHAT THIS MEANS
MT-RNR1 11 pairs	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**Amikacin, Dibekacin, Gentamicin, Kanamycin, Neomycin, Netilmicin, Paromomycin, Plazomicin, Ribostamycin, Streptomycin, Tobramycin**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
NUDT15 3 pairs	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**6-Mercaptopurine, Azathioprine, Thioguanine**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
OPRM1 1 pair	Marker coverage passed, but the frozen source snapshot contains only Level-C supportive evidence; no customer prescribing result is released.	**Morphine**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
RYR1 7 pairs	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**Desflurane, Enflurane, Halothane, Isoflurane, Methoxyflurane, Sevoflurane, Succinylcholine**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
TPMT 3 pairs	This result would rest on assigning the reference allele, which is assigned by the absence of variants. Not every variant CPIC uses to define this gene could be read from this file, so an untested variant and a typical result are indistinguishable here.	**6-Mercaptopurine, Azathioprine, Thioguanine**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.
UGT1A1 2 pairs	A Normal Metabolizer result cannot be asserted on this assay: The clinically used UGT1A1*28 allele is a promoter TA-repeat length. It was not directly sized here; only linked SNPs were read.	**Atazanavir, Irinotecan**	No gene-based medication conclusion is issued for these pairs. Use a validated clinical assay if a prescribing decision depends on this gene.

HELIXLINE

YOUR PHARMGKB MARKER EVIDENCE

HELIXLINE

A BROADER, SOURCE-LINKED VIEW OF YOUR PGX DATA

These are annotation rows whose genotype field matches the observed genotype. A matching row may describe a reference genotype or an effect genotype, and the frozen row may not encode effect direction. Treat these as source links, not independent personal findings or medication actions.

125	124	1	328
LEVEL 1A-2B MARKER MATCHES · NOT INDEPENDENT FINDINGS	DIRECTLY MEASURED	QUALITY-QUALIFIED IMPUTED	LEVEL 1A-2B MARKERS READABLE

CLINPGX LEVEL 1A / 1B
155
Guideline-linked or high evidence
Source-linked evidence; a medication action still requires an assay-supported gene result and approved clinical wording.

CLINPGX LEVEL 2A / 2B
36
Replicated association evidence
Shown as association evidence only; it is not dosing or treatment guidance.

CLINPGX LEVEL 3
2672
Preliminary research signal
Counted only when directly measured. Individual rows are not presented as personal findings and imputation is excluded.

Level 3 completeness check: 1686 directly measured markers were readable; 1671 markers matched 2672 preliminary annotation rows touching 482 medicine names. 6 imputed Level 3 markers were excluded.

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs12777823 · GA Measured directly	**Gene not assigned** GUIDELINE OR LABEL Level 1A	**warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs2231142 · GG Measured directly	**ABCG2** GUIDELINE OR LABEL Level 2A, 1A	**allopurinol, rosuvastatin** Efficacy, Metabolism/PK, Toxicity, Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1800559 · CC Measured directly	**CACNA1S** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs772226819 · GG Measured directly	**CACNA1S** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs113993958 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs115545701 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs11971167 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121908751 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**elexacaftor / tezacaftor / ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs121908752 · TT Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121908753 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121908757 · AA Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121909005 · TT Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121909013 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121909020 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121909041 · TT Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs141033578 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs150212784 · TT Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1800076 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**elexacaftor / tezacaftor / ivacaftor, ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1800100 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1800111 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs186045772 · TT Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922525 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs202179988 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs267606723 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs36210737 · TT Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**elexacaftor / tezacaftor / ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs368505753 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs397508256 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs397508442 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs397508513 · AA Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs397508537 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs397508602 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs397508759 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs74503330 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs74551128 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs75039782 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**elexacaftor / tezacaftor / ivacaftor, ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs75527207 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs75541969 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs77409459 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs77834169 · CC Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs78655421 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs80224560 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**elexacaftor / tezacaftor / ivacaftor, ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs80282562 · GG Measured directly	**CFTR** GUIDELINE OR LABEL Level 1A	**ivacaftor** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs3745274 · GT Measured directly	**CYP2A7P1, CYP2B6** GUIDELINE OR LABEL Level 1A, 2A	**efavirenz, nevirapine** Toxicity, Metabolism/PK	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs2108622 · CC Measured directly	**CYP4F2** GUIDELINE OR LABEL Level 2A, 1A	**acenocoumarol, warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs115232898 · TT Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**fluorouracil** Other, Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs148994843 · CC Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**fluorouracil** Other	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs17376848 · AA Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1801158 · CC Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs1801159 · TT Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1801160 · CT Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Toxicity, Other	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1801265 · AA Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1801266 · GG Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**fluorouracil** Other	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1801268 · CC Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**fluorouracil** Other	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs2297595 · TT Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs3918290 · CC Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil, tegafur** Toxicity, Other	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs56038477 · CC Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs67376798 · TT Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil, tegafur** Toxicity, Other	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs72549306 · CC Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**fluorouracil** Other	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs75017182 · GG Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**capecitabine, fluorouracil** Other, Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs78060119 · CC Measured directly	**DPYD** GUIDELINE OR LABEL Level 1A	**fluorouracil** Other, Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121434568 · TT Measured directly	**EGFR** GUIDELINE OR LABEL Level 1A	**gefitinib** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs12979860 · CT Measured directly	**IFNL3, IFNL4** GUIDELINE OR LABEL Level 1A	**boceprevir, peginterferon alfa-2a, peginterferon alfa-2b, ribavirin, telaprevir** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs8099917 · TG Measured directly	**IFNL3, IFNL4** GUIDELINE OR LABEL Level 1B, 2A	**interferons, peginterferon alfa-2a, peginterferon alfa-2b, ribavirin, telaprevir** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs116855232 · CC Measured directly	**NUDT15** GUIDELINE OR LABEL Level 1A	**azathioprine, mercaptopurine** Dosage, Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs111888148 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs112563513 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192122 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192124 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192161 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192162 · AA Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192168 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192172 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs118192175 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192176 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192177 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs118192178 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121918593 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121918594 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121918595 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922747 · TT Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs193922748 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922753 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922768 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922770 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922772 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922802 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922803 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922807 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs193922809 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922816 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922818 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922832 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922843 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922876 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs193922878 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs28933396 · GG Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs28933397 · CC Measured directly	**RYR1** GUIDELINE OR LABEL Level 1A	**desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs4149056 · TT Measured directly	**SLCO1B1** GUIDELINE OR LABEL Level 1A, 2A	**atorvastatin, fluvastatin, hmg coa reductase inhibitors, lovastatin, lovastatin acid, pitavastatin, pravastatin, rosuvastatin, simvastatin, simvastatin acid** Metabolism/PK, Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs2359612 · AG Measured directly	**VKORC1** GUIDELINE OR LABEL Level 1B	**warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs7294 · CT Measured directly	**VKORC1** GUIDELINE OR LABEL Level 1B	**warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs8050894 · CG Measured directly	**VKORC1** GUIDELINE OR LABEL Level 1B	**warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs9923231 · CT Measured directly	**VKORC1** GUIDELINE OR LABEL Level 1A, 2A, 1B	**acenocoumarol, phenprocoumon, warfarin** Dosage, Toxicity, Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs9934438 · GA Measured directly	**VKORC1** GUIDELINE OR LABEL Level 1B, 2A	**acenocoumarol, phenprocoumon, warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs4961 · GG Measured directly	**ADD1** REPLICATED RESEARCH Level 2B	**hydrochlorothiazide** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs1042713 · GA Measured directly	**ADRB2** REPLICATED RESEARCH Level 2A	**salmeterol** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs671 · GG Measured directly	**ALDH2** REPLICATED RESEARCH Level 2B	**ethanol** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs7412 · CC Measured directly	**APOE** REPLICATED RESEARCH Level 2B	**atorvastatin** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs4673993 · TC Measured directly	**ATIC** REPLICATED RESEARCH Level 2B	**methotrexate** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs16969968 · GG Measured directly	**CHRNA3, CHRNA5** REPLICATED RESEARCH Level 2B	**nicotine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1801133 · GG Measured directly	**CLCN6, MTHFR** REPLICATED RESEARCH Level 2A	**methotrexate** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs28399499 · TT Measured directly	**CYP2B6** REPLICATED RESEARCH Level 2A	**nevirapine** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.

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YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs2242480 · CT Quality-qualified imputation	**CYP3A4** REPLICATED RESEARCH Level 2A	**fentanyl, tacrolimus** Metabolism/PK, Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs4646437 · GA Measured directly	**CYP3A4** REPLICATED RESEARCH Level 2A	**tacrolimus** Metabolism/PK	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs121434569 · CC Measured directly	**EGFR** REPLICATED RESEARCH Level 2B	**erlotinib, gefitinib** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1799963 · GG Measured directly	**F2** REPLICATED RESEARCH Level 2B	**hormonal contraceptives for systemic use** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs6025 · CC Measured directly	**F5** REPLICATED RESEARCH Level 2B	**hormonal contraceptives for systemic use** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs7270101 · AA Measured directly	**ITPA** REPLICATED RESEARCH Level 2B	**peginterferon alfa-2b, ribavirin** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1800629 · GG Measured directly	**LST1, LTA, TNF** REPLICATED RESEARCH Level 2B	**etanercept** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

HELIXLINE

YOUR PHARMGKB MARKER EVIDENCE (continued)

MARKER & CALL	GENE & EVIDENCE	LINKED MEDICINES / PHENOTYPE TYPE	SOURCE
rs3812718 · TT Measured directly	**SCN1A** REPLICATED RESEARCH Level 2B	**carbamazepine** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs1051266 · TC Measured directly	**SLC19A1** REPLICATED RESEARCH Level 2A	**methotrexate** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs7853758 · GG Measured directly	**SLC28A3** REPLICATED RESEARCH Level 2B	**anthracyclines and related substances, daunorubicin, doxorubicin** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs10929302 · GA Measured directly	**UGT1A1** REPLICATED RESEARCH Level 2A	**irinotecan** Toxicity	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs2884737 · AA Measured directly	**VKORC1** REPLICATED RESEARCH Level 2A	**warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs61742245 · CC Measured directly	**VKORC1** REPLICATED RESEARCH Level 2A	**warfarin** Dosage	Open PharmGKB source Informational association; no dose or medication change is authorized here.
rs25487 · TT Measured directly	**XRCC1** REPLICATED RESEARCH Level 2B	**Platinum compounds** Efficacy	Open PharmGKB source Informational association; no dose or medication change is authorized here.

HELIXLINE

THE HELIXLINE PGX ENGINE

PGX TECHNICAL APPENDIX

HIGH CONFIDENCE

27 gene(s) documented on build GRCh37

WHAT THE ENGINE DOES

The Helixline PGx Engine reads every addressable CPIC and ClinPGx Level 1A-2B position available in this sample, plus a direct-only Level-3 research completeness layer, preserving **direct versus quality-qualified imputed provenance**. A curated caller then reports results resolved within the tested marker set, provisional variant findings where the complete allele background is not established, and explicit limited screens elsewhere. Level 3 never affects a gene call or medicine action. Published population frequencies are context only; they never change a genotype or force a star-allele call. This report is decision support, not a prescription.

BUILD MANIFEST

HaplotypeLock	v3.3.0
Imputation gate	DR2/R2 ≥ 0.8; sample GP ≥ 0.90
PharmGuard QC	v2.1.0
Synthetic fixture run	2026-07-26
ClinPGx fact index	PharmGKB 2026-07
ClinPGx source archive	2025-07-05
Customer wording	Association-only unless clinically approved

ARRAY + QUALITY-GATED IMPUTATION - COVERAGE & LIMITATIONS

Pharmacogenomic results use **directly measured genotypes first** and may use statistically imputed genotypes only when the marker passes the recorded DR2/R2 and sample-probability thresholds. Every result retains DIRECT, IMPUTED or PROXY provenance; a failed or absent marker is not treated as a typical result. **Clinical confirmation is recommended before any prescribing change.**

PIPELINE: FROM ARRAY TO REVIEW-READY PGX EVIDENCE

01 Array QC — VCF/sample validation and extreme genotype-pattern corruption gate

02 Imputation — Reference-panel imputation; DR2/R2≥0.8 and sample GP≥0.90 per PGx marker

03 Evidence Ledger — 532 of 830 required positions usable for this sample

04 HaplotypeLock — Rule-based limited-variant mapping; incomplete assay capabilities are withheld

05 PharmGuard QC — Automated concordance check against synthetic diplotype truth set

TECHNICAL SPECIFICATIONS & KNOWN LIMITATIONS

ASSAY SPECIFICATIONS

Array platform: **Helixline Genotyping Array**

Genome build: **GRCh37 / hg19**

Imputation: **Beagle 5.5; 1000 Genomes Project Phase 3 South Asian reference panel (489 samples; GRCh37)**

HLA inference: **Tagging-SNP proxy - NOT direct HLA typing (see caveat below)**

CYP2D6: **Withheld - copy number, hybrids, structural variants, and complete clinical diplotype not assessed**

Star-allele definitions: **CPIC allele definitions (populations) 2026-08**

Allele coverage: **All available CPIC/ClinPGx Level 1A-2B positions enter the clinical evidence ledger; Level 3 is direct-only aggregate context. Exact star calls remain limited to validated rule-based definitions. HaplotypeLock is not exhaustive PharmVar haplotype resolution; this is not full-gene sequencing and PharmCAT is not run**

DR2 + GP gate: **An imputed marker must pass DR2/R2 0.8 and this sample's GP 0.90; otherwise the gene is withheld and it is not reported as an uncertain result**

KNOWN LIMITATIONS

HLA	All HLA calls are by tagging-SNP proxy. Must be confirmed by direct HLA typing (SSO/SSP/NGS) before clinical use. Sensitivity for rare alleles is limited.
CYP2D6	Not reported in the General assay. A validated CYP2D6 test that resolves copy number, structural variants, hybrids, and phased star alleles is required before prescribing use.
Rare alleles	Novel or unassessed star alleles are not classified. When the required allele background is incomplete, the result is withheld or explicitly limited; an apparently reference call is not presented as full-gene exclusion.
Structural variants	Large chromosomal rearrangements and gene conversions (e.g. CYP2D6/CYP2D7 hybrids) are not reliably detected from array imputation.
Drug-drug interaction	Phenoconversion by co-medications is not modelled. A CYP2D6 Normal Metabolizer taking a strong inhibitor may function as a Poor Metabolizer.
Non-genetic factors	Age, sex, hepatic/renal function, diet, and environmental exposures all modulate drug response and are outside the scope of this report.

ASSAY COVERAGE BY GENE - MARKERS AND CPIC ALLELE CATALOGUE

MARKERS = gene-defining variants readable from this DNA file / variants defined. **CPIC ALLELES** = star alleles identifiable / star alleles CPIC defines. A dash means no count is recorded for that gene, which is not the same as zero. ⚠ marks a gene where a "normal" result could not be fully certified: at least one defining variant was not readable, so an untested rare variant and a genuinely typical result look alike here. These are assay-capability figures, not results - your own states are printed beside each gene in the gene inventory.

GENE	MARKERS	CPIC ALLELES
ABCG2	1/1	2/2
CACNA1S	2/2	3/3
CYP1A2	–	–
CYP2A6	–	–
CYP2B6	20/48	16/49 ⚠
CYP2C19	25/35	25/39 ⚠
CYP2C8	0/0	0/0 ⚠
CYP2C9	44/83	46/88 ⚠
CYP2D6	44/157	29/184 ⚠
CYP3A5	5/7	5/8 ⚠
CYP4F2	5/20	4/23 ⚠
DPYD	76/83	76/84 ⚠
G6PD	59/173	63/187 ⚠
HLA-A*31:01	–	–
HLA-B*15:02	–	–
HLA-B*57:01	–	–
HLA-B*58:01	–	–
IFNL3	1/1	2/2
MT-RNR1	8/22	9/25 ⚠
NAT2	24/47	30/59 ⚠
NUDT15	3/20	3/22 ⚠
OPRM1	0/0	0/0 ⚠
RYR1	93/320	109/347 ⚠
SLCO1B1	24/35	29/47 ⚠
TPMT	29/45	32/49 ⚠
UGT1A1	3/4	2/9 ⚠
VKORC1	1/1	2/2

THE HELIXLINE PGX ENGINE

PGX TECHNICAL APPENDIX

HIGH CONFIDENCE

27 gene(s) documented on build GRCh37

THIS SAMPLE'S PGX COVERAGE PROFILE

Genotyping platform: Helixline Genotyping Array

Imputation: Beagle 5.5 against 1000 Genomes Project Phase 3 South Asian reference panel (489 samples; GRCh37) - DR2/R2 ≥ 0.8 and sample GP ≥ 0.90

Caller-qualified gene records: 16 / 27

Customer analytical results after resolution rules: 7 / 27

Core evidence coverage: 830 CPIC-defining / ClinPGx Level 1A-2B positions

- **528** typed directly on chip
- **4** imputed (DR2/R2 ≥ 0.8; GP ≥ 0.90)
- **298** missing or below threshold (not interpreted as wild-type)

This report uses a versioned panel of 27 genes and a version-controlled gene-drug catalog. A gene is reported only when its required panel markers and assay capabilities pass the release rules; absent or low-quality required markers cause explicit withholding, not a normal result. Contract PGx gene drug contract 2026-08 · capability snapshot PGx chip capability 2026-08 · pharmgkb / clinpgx (https://www.pharmgkb.org), cc by sa 4.0. whirl carrillo m, et al. clin pharmacol ther. 2021;110(3):563 572.

VARIANTS NOT SCREENED IN THIS REPORT

The following **298 core evidence positions** were not reliably covered for this specific sample (either absent from the chip, dropped during imputation, or below DR2/R2 ≥ 0.8 or sample GP ≥ 0.90). They are limitations, not normal or negative results. If one is relevant to a contemplated medicine, the treating clinician or laboratory should select the locus-appropriate confirmatory assay.

GENE	STAR/VARIANT	RSID	REASON MISSING	DRUG(S) AFFECTED
CYP2B6	CPIC defining position	rs117872433	imputed below the marker-quality floor (DR2/R2=0.0 < 0.8)	bupropion, efavirenz, methadone (+1 more)
NUDT15	CPIC defining position	rs1185228044	not present in this file	mercaptopurine
NUDT15	CPIC defining position	rs1202487323	not present in this file	mercaptopurine
CYP2C9	CPIC defining position	rs1216169538	not present in this file	acenocoumarol, celecoxib, flurbiprofen (+9 more)
SLCO1B1	CPIC defining position	rs1228465562	not present in this file	atorvastatin, fluvastatin, lovastatin (+6 more)
CYP2C9	CPIC defining position	rs1254213342	not present in this file	acenocoumarol, celecoxib, flurbiprofen (+9 more)
TPMT	CPIC defining position	rs1256618794	not present in this file	azathioprine, mercaptopurine, thioguanine
CYP2C9	CPIC defining position	rs1274535931	not present in this file	acenocoumarol, celecoxib, flurbiprofen (+9 more)
CYP2C9	CPIC defining position	rs1289704600	not present in this file	acenocoumarol, celecoxib, flurbiprofen (+9 more)
CYP2C9	CPIC defining position	rs1297714792	not present in this file	acenocoumarol, celecoxib, flurbiprofen (+9 more)
CYP2C9	CPIC defining position	rs1364419386	not present in this file	acenocoumarol, celecoxib, flurbiprofen (+9 more)
NUDT15	CPIC defining position	rs1368252918	not present in this file	mercaptopurine
CYP2C9	CPIC defining position	rs1375956433	not present in this file	acenocoumarol, celecoxib, flurbiprofen (+9 more)
TPMT	CPIC defining position	rs139392616	imputed below the marker-quality floor (DR2/R2=0.0 < 0.8)	azathioprine, mercaptopurine, thioguanine

... and 284 more variants not shown (see clinical appendix).

*HLA allele calls are **not released from array tagging SNPs**. Tag-marker performance is population-dependent; a proxy cannot certify presence or absence or establish the four-digit HLA type required by prescribing guidance. **Confirmatory high-resolution HLA typing from an accredited laboratory is required before this result is used in a prescribing decision, whether it is positive or negative.** An unresolved or negative RYR1/CACNA1S array screen **does NOT exclude malignant-hyperthermia susceptibility**. Use locus-appropriate clinical assessment, full-gene testing and/or IVCT when personal or family history makes this relevant.*

REGULATORY FRAMEWORK & REPORT VERSION

Release status. This is an informational pharmacogenomics screening report for clinician discussion, not a validated diagnostic test. It does not authorize a medicine or dose change. Proxy and partial findings require confirmation with an appropriate clinical test before prescribing use. Applicable consent, privacy and jurisdictional requirements still apply.

Report version. As listed in the build manifest and assay specifications above; automated package checks verify this exact report and its supporting result. **Updates.** This static report does not update automatically. Reissue only after the relevant CPIC/DPWG/FDA and PharmVar changes have been reviewed, approved, versioned, and the report has been regenerated.

HOW THE ANALYTICAL COUNTS CONNECT

16
CALLER-QUALIFIED GENE RECORDS
Required rule-set markers passed the caller's quality gate. This is an internal analytical stage, not automatically a customer result.

7
CUSTOMER ANALYTICAL RESULTS
These remained reportable after phase, reference-call, proxy and complex-locus resolution rules were applied.

4
PROVISIONAL CUSTOMER RESULTS
Useful observations retained with a confirmation requirement; they are not promoted to complete clinical diplotypes.

Why the numbers differ: marker qualification asks whether the defined positions were readable; customer resolution also asks whether the available assay establishes phase, allele background and required locus features. The stricter number is used on result pages. The larger number remains here for complete pipeline accounting.

DIRECT RESULT
The reported marker was measured in the uploaded genotype data. Direct does not mean full-gene sequencing.

IMPUTED RESULT
A statistical genotype passed both cohort DR2/R2 and this sample's GP threshold; the badge and reference panel remain visible.

PROXY RESULT
A tag or surrogate marker supports a provisional observation. Confirm with the locus-appropriate clinical test before prescribing use.

EVERY TERM, IN PLAIN ENGLISH

HELIXLINE

Each term below is defined in one plain sentence - enough to read any drug page with confidence. Helixline-proprietary concepts are marked HL. The precise technical wording behind these terms is kept in full further down this page.

LEGEND

HL Helixline-proprietary

SA South Asian specific

Pharmacogenomics (PGx)
How your DNA affects the way your body handles a medicine.

Gene
A stretch of DNA carrying the instructions for one protein.

Allele
One copy of a gene. You have two of each - one from each parent.

Star allele (e.g. *4, *17)
The standard naming system for gene versions. *1 is the common reference version; other numbers are named variants.

Diplotype
Your two gene copies written together, for example *1/*4.

Phenotype
What your two copies actually *do* - for example Poor, Intermediate, Normal, Rapid or Ultrarapid Metabolizer.

Activity score
A number summarising how much enzyme activity your two copies add up to: 0 is none, 2.0 is typical.

HaplotypeLock HL
Our rule-based method for reading the limited set of variants this assay carries.

PharmGuard QC HL
Our automated quality-control layer, run before any gene is reported.

CPIC
An international expert group that publishes prescribing guidance for gene-medicine pairs.

DPWG
The Dutch expert group that publishes its own pharmacogenomic prescribing guidance.

FDA Label
Pharmacogenomic information printed on a United States drug label. It is regulatory context.

PharmCAT / PharmVar
PharmVar is the official catalogue of star alleles. PharmCAT is a separate external tool, which this report does not run.

Imputation
Estimating DNA letters an array did not measure directly, using a reference population. It is not sequencing.

DR2 / R²
A 0-1 estimate of imputation accuracy across a reference population. We require at least 0.8, but this alone cannot qualify one person's genotype.

GP (genotype probability)
The imputation model's probability for this sample's called genotype. We require at least 0.90 in addition to DR2/R².

South Asian context SA
Published South Asian frequency figures, shown as background only.

Recommendation strength
How firmly CPIC states a recommendation: Strong, Moderate or Optional.

High-risk genetic alert
A medicine that needs confirmatory testing first, because a genetic risk signal was detected.

SJS / TEN / DRESS
Rare, severe immune skin reactions that certain medicines can trigger in carriers of specific HLA types.

TDM (Therapeutic Drug Monitoring)
A clinician measuring drug levels or drug response during treatment.

WHAT THESE EVIDENCE SOURCES DO AND DO NOT ESTABLISH

A CPIC guideline is about a gene and a medicine - not about this DNA file.

CPIC publishes how a prescriber uses a genotype result that is already available and valid. CPIC Level A means the published evidence for that gene-medicine pair is strong enough to change prescribing when such a result exists, and CPIC grades each statement Strong, Moderate or Optional. Naming CPIC beside a pair in this report therefore states that published guidance exists for that pair. **It does not prove that this assay uniquely resolved that genotype for you.** Whether this file could resolve a gene is a separate question, answered by the per-gene coverage table in the engine and methodology appendix and by the state printed beside every gene here. DPWG guidance appears on the same terms, and only where that pair is present in the locked release-evidence snapshot this report was built from.

Neither evidence list is exhaustive.

The FDA table of pharmacogenetic associations and this report's versioned gene-medicine contract are both defined lists. **Neither is a complete inventory of every pharmacogenomic relationship that exists**, and the published literature continues to grow beyond both. A gene or medicine absent from the named Level 1A-2B rows was not clinically interpreted here: that is an absence of assessment, and it must not be read as a finding about the medicine. Preliminary Level-3 literature may still be included only in the aggregate count; that aggregate does not establish medication safety, effectiveness or unsuitability. FDA label information is regulatory context - its presence does not by itself mean that a genotype-specific dose change applies to a given person. Any expansion of the contracted pair list is a separate, separately reviewed product decision.

TECHNICAL DETAIL BEHIND THESE TERMS

Phenotype - HLA genes. For the HLA genes in this panel the phenotype is Positive or Negative for a risk allele, not a metaboliser class.

Activity score. 0 = no activity (Poor Metabolizer), 1.0 = intermediate, 2.0 = normal, above 2.0 = faster than normal. CPIC uses this scale to map diplotype to phenotype for CYP2D6 and CYP2C9; the other genes on this panel do not use it.

Diplotype. One exact pair is printed only where the evidence uniquely supports it. Otherwise the report states the set of pairs still compatible with the evidence, or states that the gene is unresolved.

Therapeutic drug monitoring. Whether monitoring is appropriate depends on the medicine, the indication, the person's clinical context and current clinical guidance. This report does not set a monitoring schedule.

KEY REFERENCES

CPIC / ClinPGx	Versioned guideline, gene-drug and clinical-annotation sources; exact frozen versions are listed in Methodology
PharmVar	Pharmacogene Variation Consortium nomenclature context; the exact caller-definition snapshot is listed in Methodology
Manoharan et al. 2019	CYP2D6 allele frequencies in South Indians (PMID: 31368850)
Khor et al. 2014	HLA-B*15:02 and carbamazepine SJS/TEN risk in Indian population - pooled analysis (PMID: 25266342)
DPWG	Dutch Pharmacogenetics Working Group guidance where registered in the locked pair catalog
Imputation	Michigan Imputation Server - large multi-ancestry reference panel for genotype imputation
Anichavezhi et al. 2012	CYP2C19 *2 and *17 allele frequencies in Indian population (PMID: 21916910)



YOUR GENES, DECODED

Your pharmacogenomic information, for a better-informed conversation with your clinician.

Thank you for trusting us.

FOLLOW US • SHARE YOUR STORY