# Your DNA, explained

Start with the meaning, explore the biology, and keep the questions that matter to you.



> 91 of 105 selected marker checks read

This is not a whole-genome health assessment.

Hereditary-cancer genes are not interpreted. The marker count describes coverage, not a score for health or the number of actions you need.

sequencing · 25 October 2026 · Sample report

**Sample whole genome A · male · 35×.** A public reference genome (NIST Genome in a Bottle), openly consented for research and public use. Not a real customer.

START HERE

# Your findings

The readings your plan rests on, your strongest results and your medicines result, each in the words of the chapter that explains it. Open any chapter for the rest.

## Blood sugar and liver

TCF7L2 — two T2D-associated alleles: a raised tendency at this one marker, not a diagnosis or a combined genetic score (rs7903146: TT). Eat: Fibre-rich, lower-glycaemic meals and activity lower diabetes risk. That holds with two copies too: they raise the odds, not the verdict. Your measured fasting glucose or HbA1c says more than this marker.

PNPLA3 — one 148M allele; a modestly higher liver-fat tendency, larger with more body fat (not a diagnosis) (rs738409: CG). Eat: Cutting alcohol and added sugar/fructose is the highest-leverage step. This variant raises the liver-fat tendency, and it is strongly diet-modifiable.

## Gut and digestion

MCM6 — Lactase non-persistent genotype — adult lactose intolerance is more likely; symptoms vary (rs4988235: GG). Eat: Hard cheese, yoghurt or lactose-free milk usually sit well. You likely produce little lactase in adulthood; ordinary milk may cause bloating, a lactase enzyme helps, and dairy isn't required (plenty of non-dairy calcium sources).

FUT2 — a non-secretor (no ABO antigens secreted into mucus or saliva). In cohorts non-secretors differ in gut bacteria and B12 levels and resist some norovirus strains — associations, not a diagnosis (rs601338: AA). Eat: Prebiotic fibre and fermented foods support gut bifidobacteria. As a likely non-secretor you tend to carry fewer of them: onion, garlic, oats and legumes, plus yoghurt, kefir, sauerkraut or kimchi, support microbiome diversity. Measure: Serum B12 or active B12 — none on file.

## Training and recovery

ACTN3 — both copies make this fast-muscle-fibre protein. It does not pick a sport or predict your training response (rs1815739: CC). Live: Strength and aerobic training both pay off whatever your ACTN3 result.

## Reassuring results and

## next blood tests

Read and not found: HFE: no C282Y, the main iron-overload variant, at the site read; F5: no Factor V Leiden at the site read; F2: no prothrombin G20210A at the site read; SERPINA1: no alpha-1 antitrypsin Z at the site read. Next blood tests: Lp(a) once in adult life: it is mostly inherited, and a single blood test measures it; tTG-IgA if you have gut symptoms, to check for coeliac disease.

## Medicines

17 of 22 genes read. Results other than “normal”:

Warfarin — VKORC1: you may need a lower dose of warfarin, and of acenocoumarol or phenprocoumon; if you take or are prescribed one, talk to your doctor or pharmacist before changing any medicine (rs9923231 T/T (no single phenotype label)). CYP4F2: part of how a warfarin dose is worked out, together with VKORC1 and CYP2C9; talk to your doctor or pharmacist before changing any medicine (*4/*5 (no single phenotype label)).

Hydralazine and isoniazid — NAT2: you may need a higher hydralazine dose; talk to your doctor or pharmacist before changing any medicine (intermediate metabolizer).

CYP3A5: tacrolimus is dosed for this result by default; talk to your doctor or pharmacist before changing any medicine (poor metabolizer).

Not read: CYP2D6, HLA-A, HLA-B, MT-RNR1, SLCO1B1. A gene we could not read is unknown, not normal.

Preliminary, not a diagnosis. Don't start, stop or change a medicine because of this result. Talk to your doctor or pharmacist; a clinical test must confirm it before any medical decision.

## Your chapter journey

Your plan · Methylation · Food and vitamins · Gut and digestion · Blood sugar and liver · Heart and lipids · Sleep and body clock · Mood and focus · Skin and hair · Training and recovery · Detox and histamine · Immunity · Hormones · Longevity · Senses · Medicines · What whole-genome sequencing could add · Closing

Read in layers: the opening answer, the illustrated explanation, then the detailed observation and source record. Direct observations and statistically inferred calls keep their different labels. A readable marker is not a measurement of how your body works today.

# Your plan

> seven things · three areas

## OBSERVATION SCOPE, IN CONTEXT

Seven things, each backed by a genotype that was actually read and by established or probable evidence. Where a list is empty it says so. We do not dose supplements from a genotype; a supplement is a practitioner’s line and waits for a blood value.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



Genes can have an impact on many areas of your health, and that can be overwhelming. We keep it to a small number of changes you could keep for a year: food you would eat anyway and a habit or two. Where a measured value says more than a gene, the page says so; whether to test is between you and your doctor.

## Blood sugar and liver

Two of the 5 markers read here carry a variant with an effect on established or probable evidence: TCF7L2, PNPLA3. Read on your file: TCF7L2 rs7903146 (TT) — two T2D-associated alleles: a raised tendency at this one marker, not a diagnosis or a combined genetic score. PNPLA3 rs738409 (CG) — one 148M allele; a modestly higher liver-fat tendency, larger with more body fat (not a diagnosis).

| EAT | LIVE | MEASURE |
|---|---|---|
| Fibre-rich, lower-glycaemic meals and activity lower diabetes risk. That holds with two copies too: they raise the odds, not the verdict. Your measured fasting glucose or HbA1c says more than this marker. | Nothing here — no measured genotype in this area carries a rule for it. | An HbA1c — none on file. |
| Cutting alcohol and added sugar/fructose is the highest-leverage step. This variant raises the liver-fat tendency, and it is strongly diet-modifiable. | | |

## Gut and digestion

Three of the 8 markers read here carry a variant with an effect on established or probable evidence: FUT2 (two sites), AOC1. The plan here rests on MCM6. Read on your file: MCM6 rs4988235 (GG) — Lactase non-persistent genotype — adult lactose intolerance is more likely; symptoms vary. FUT2 rs601338 (AA) — a non-secretor (no ABO antigens secreted into mucus or saliva). In cohorts non-secretors differ in gut bacteria and B12 levels and resist some norovirus strains — associations, not a diagnosis. FUT2 rs602662 (AA) — two non-secretor-tag alleles (consistent with non-secretor; read with another FUT2 marker). Studies link this genotype to higher total serum B12, which does not measure your B12 status.

| EAT | LIVE | MEASURE |
|---|---|---|
| Hard cheese, yoghurt or lactose-free milk usually sit well. You likely produce little lactase in adulthood; ordinary milk may cause bloating, a lactase enzyme helps, and dairy isn't required (plenty of non-dairy calcium sources). | Nothing here — no measured genotype in this area carries a rule for it. | Serum B12 or active B12 — none on file. |
| Prebiotic fibre and fermented foods support gut bifidobacteria. As a likely non-secretor you tend to carry fewer of them: onion, garlic, oats and legumes, plus yoghurt, kefir, sauerkraut or kimchi, support microbiome diversity. | | |

## Training and recovery

No marker read here carries a variant with an effect on established or probable evidence. The plan here rests on ACTN3. Read on your file: ACTN3 rs1815739 (CC) — both copies make this fast-muscle-fibre protein. It does not pick a sport or predict your training response.

| EAT | LIVE | MEASURE |
| --- | --- | --- |
| Nothing here — no measured genotype in this area carries a rule for it. | Strength and aerobic training both pay off whatever your ACTN3 result. | Nothing here — no measured genotype in this area carries a rule for it. |

## Interesting, not a plan

CYP1A2 — This variant does NOT predict your baseline caffeine clearance. In non-smokers it shows no measurable effect on caffeine metabolic ratio (Sachse 1999, n=185; Ghotbi 2007, n=194; Djordjevic 2010, n=178). MTHFR is not a general 'detoxification' gene.

## Worth a look

- **ADORA2A** rs5751876 (TT) — linked in two studies to more jitteriness after caffeine, less so in regular drinkers. Not a sleep result: sleep studies point to the other allele. (Sleep and body clock)
- **HERC2** rs12913832 (GG) — two blue-eye alleles: with this genotype eyes are most often blue, and brown is rare. Other genes also shape eye colour. (Skin and hair)
- **PTPN22** rs2476601 (AG) — one 620W allele; a modestly raised tendency to type 1 diabetes and rheumatoid arthritis (not a diagnosis). (Immunity)
- **CTLA4** rs3087243 (GA) — one G allele, the more common allele; a small autoimmune-risk tendency in cohorts, not a diagnosis. It went with slightly more autoimmune thyroid disease and type 1 diabetes. (Immunity)
- **SRD5A2** rs523349 (GC) — one 89L allele; a modestly lower 5α-reductase activity (small effect). (Hormones)

## Why this many

A rule fires only on a genotype that was read, and only where the evidence is established or probable. Areas with variants but no rule yet are described in the story and left off the plan on purpose.

# What can folate-related DNA markers tell me?

> 5 of 6 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

5 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**What your MTHFR results mean for folate**

**Your finding**

MTHFR — one 677T allele. Research links this variant to enzyme properties and folate-related biomarkers; it does not measure your enzyme activity (rs1801133: GA).

MTHFR — context only. This call does not measure folate processing (rs1801131: TT).

**What this means for you**

These observations do not establish folate deficiency.

People with common MTHFR variants can process folic acid.

**What is missing**

No dated folate, B12 or homocysteine result is supplied here. This report cannot assess your nutrient status from these DNA observations.

**Your next step**

Review the foods and supplements you already use.

If you have relevant blood results, add their dates, units and laboratory ranges to the discussion.

Before testing or changing a supplement, ask which specific question it would answer.

Keep any existing clinician advice.

**The useful next question**

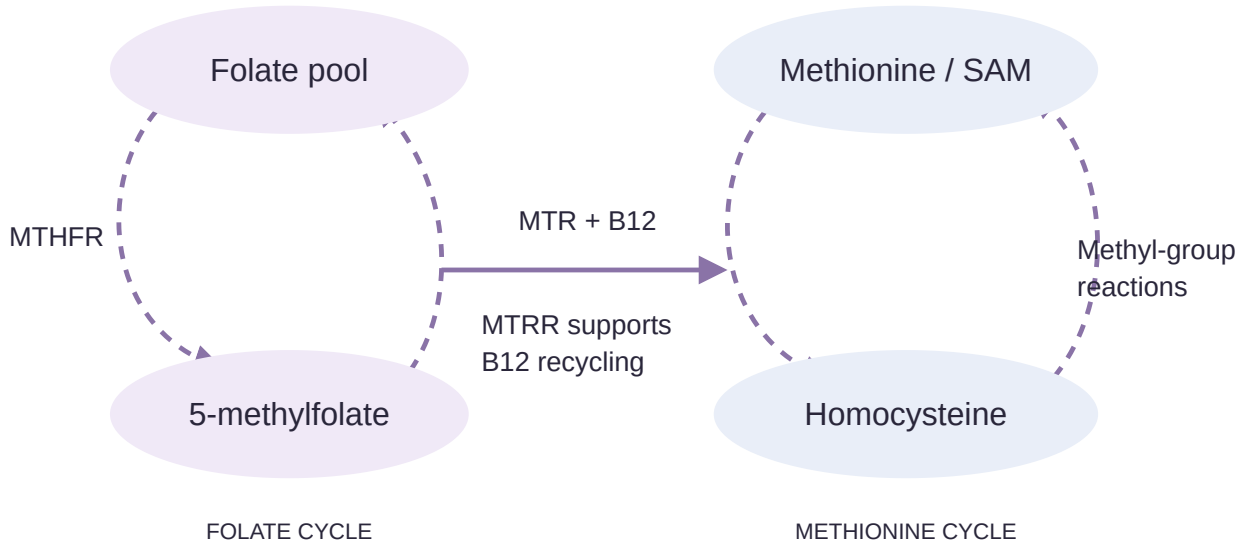
Folate, B12, any existing homocysteine result, diet and your actual question.



Keeping an appropriate routine or making no change is a valid outcome.

| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| MTHFR · rs1801133 | GA | Sequencing |
| MTHFR · rs1801131 | TT | Sequencing |
| MTRR · rs1801394 | AA | Sequencing |

The first 3 of 5 markers read in this chapter. Each is explained below.




Schematic · not a measurement of your pathway speed, nutrients or epigenetic methylation

Read the loops as connected reactions. Your selected observations are listed below the map with their scope. No colour indicates impairment or a supplement requirement.

Methylation is the transfer of a small chemical unit called a methyl group. The body uses this chemistry in many different reactions. When wellness reports say “methylation,” they often mean the connected handling of folate, methionine and related compounds. That is broader than one gene, and it is not one function that can be given a speed from two letters.

MTHFR supplies instructions for an enzyme that converts one form of folate into 5-methyltetrahydrofolate, often shortened to 5-MTHF. This form helps a multistep process recycle homocysteine into methionine. Methionine is used to make proteins and other compounds. That is the relevant biological connection behind the MTHFR markers included in this report. [8]

## Why B12 appears in the same conversation

Vitamin B12 is a cofactor for methionine synthase, an enzyme in the conversion of homocysteine to methionine. It also has roles in blood-cell formation and the nervous system. A discussion about folate status can therefore need B12 context as well; the same word “methylation” should not hide these different roles. [10]

## Methylation chemistry and DNA methylation

DNA methylation concerns chemical marks on DNA that can affect gene activity. Reading inherited variants in MTHFR does not measure those marks. An inherited variant report and an epigenetic measurement are different products. This report contains the former. [12]

Research and guidance: [8] The MTHFR enzyme (MedlinePlus Genetics); [9] Folate: food, function and supplementation (NIH Office of Dietary Supplements); [10] Vitamin B12: function and assessment (NIH Office of Dietary Supplements); [12] What epigenetics measures (MedlinePlus Genetics). These explain biology or guidance; they do not verify a private genotype.

## Why common-variant research is relevant

Researchers can compare folate and homocysteine concentrations between groups with different MTHFR genotypes. In a randomized folic-acid trial involving 932 northern Chinese women of childbearing age, genotype was associated with folate and homocysteine concentrations, but did not significantly change the response to folic-acid dose. That supports explaining a biological relationship; it does not supply your blood values or a supplement prescription. The participants, genotype groups and outcomes matter when applying the finding. [11]

## Why a marker name and the reported letters can differ

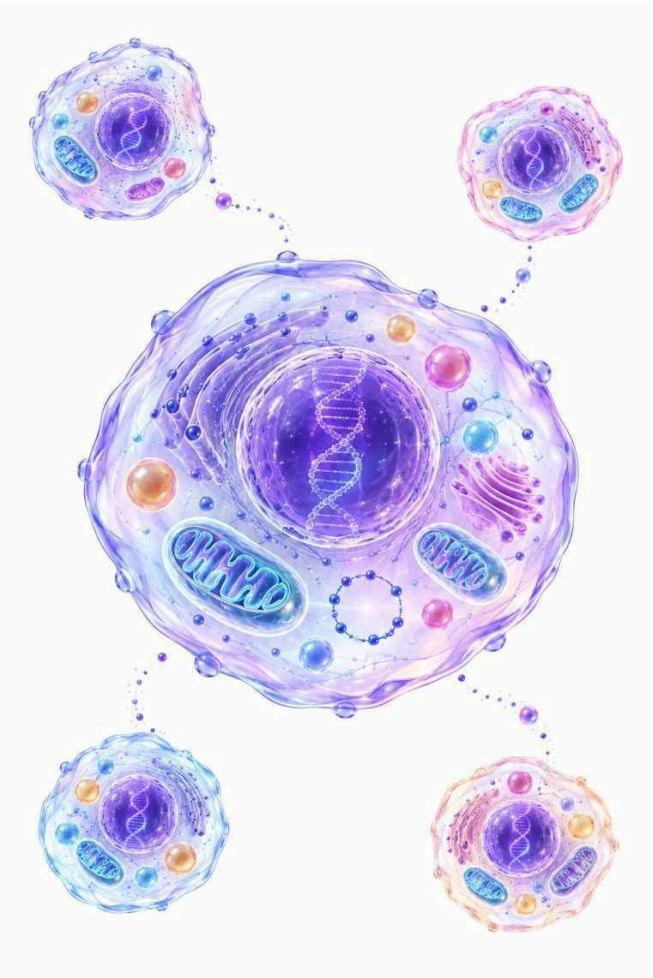
A marker can be named on a different DNA strand from the letters shown in a file. The identifier and the reference build help establish which site was read. Use the interpretation beside the actual observation rather than comparing the letters with a variant name. Missing strand or assay details remain unresolved.

## Can selected sites establish a combined effect?

Selected sites do not establish which chromosome carries which change, or provide a validated combined functional score. We therefore do not label the combination as severe, assign a cumulative percentage, or call it a complete methylation assessment. The missing detail should be resolved in the record, not replaced with a confident story.

## What a homocysteine result adds

Homocysteine can provide a biochemical observation related to this network. It is not a direct methylation-speed test. B12 status, folate and kidney function can influence it, so an elevated value needs interpretation rather than a single-gene explanation. [10] A result is most useful when the report also retains its date, units and the laboratory’s reference interval.



**From a nutrient to a working cell**
Folate and B12 participate in connected chemistry. Food, absorption and an actual nutrient measurement give the inherited observation its context.

## A worked example of useful context

**Illustrative situation:** someone reads “MTHFR” and worries that a fortified breakfast cereal is unsuitable. They feel well and have no abnormal nutrient result. The report should explain the variant and the food question, rather than treating an absent laboratory value as evidence of deficiency. The immediate decision can be to keep a tolerated food and understand the label.

If the same person later has a clinically identified B12 issue, the useful discussion changes. That measured finding has its own assessment and treatment context. The inherited MTHFR observation stays the same, but it is no longer the whole conversation.

Research and guidance: [10] Vitamin B12: function and assessment (NIH Office of Dietary Supplements). These explain biology or guidance; they do not verify a private genotype.

## Start with food you already eat

Folate occurs in foods such as leafy greens, beans, peas and asparagus. Fortified products may supply folic acid; availability varies between products and countries, so read the label. [9] If you want a practical food step, choose one familiar meal where adding a food you enjoy makes your diet more varied. This is an intake decision, not a genetic treatment.

## Folic acid and methylfolate: answer the actual question

CDC guidance says people with common MTHFR variants can process folate, including folic acid. It does not support avoiding folic acid simply because of these common variants. [4] That answers the “am I unable to use it?” concern. It does not decide every possible supplement choice.

NIH notes that research on supplementation for people with the 677 variant is inconclusive, and that some of them might benefit from 5-MTHF. The same fact sheet adds that CDC recommends folic acid, not 5-MTHF, for people who could become pregnant, including those with this variant. [9] CDC’s reason is that folic acid is the only form of folate shown to help prevent neural tube defects. [4] The useful question is why a supplement is being considered and which outcome it is intended to address. This report does not select a form or dose from selected DNA observations.

| A USEFUL CHOICE | A CHOICE WITHOUT A CLEAR BASIS HERE |
|---|---|
| Review your current diet and any existing supplement labels. | Add several methylated products because the report names MTHFR. |
| If testing is clinically relevant, ask which specific measurement would answer the question. | Order a broad “methylation panel” solely to follow a gene list. |
| Bring an abnormal result to a professional with its date, units and medication context. | Assume MTHFR explains fatigue, mood, every symptom or every abnormal blood value. |
| Keep established nutrient or pregnancy-related advice unless your clinician changes it. | Stop a recommended supplement or avoid folic acid because of an online warning about common variants. |

## Before buying a supplement

Ask: **What problem is this intended to address? What evidence suggests I have that problem? How will I know the intervention helped?** If the only answer is “my gene is highlighted,” the report has not provided a sufficient decision. Large folate intakes can correct the anaemia associated with B12 deficiency without addressing its neurological effects, which is one reason to consider B12 context before escalating supplements. [9]

## Two questions to take away

**Do I need to fix my methylation?** This file does not establish a broken system to fix. **Can the result still be useful?** Yes: it explains a common biological variation, puts supplement claims in perspective and helps you formulate a specific nutrient question.

Research and guidance: [4] MTHFR and folic acid (CDC); [9] Folate: food, function and supplementation (NIH Office of Dietary Supplements). These explain biology or guidance; they do not verify a private genotype.

> No relevant dated laboratory measurement is supplied in this chapter. This describes the report inputs; it does not mean a test was never performed.

# Selected markers and their interpretation

## MTHFR rs1801133

### GA · Sequencing

MTHFR — one 677T allele. Research links this variant to enzyme properties and folate-related biomarkers; it does not measure your enzyme activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A call at this site does not diagnose a deficiency or measure pathway activity. Missing data remain unknown.

Observation: GRCh38 · 1:11796321. Selected site only. Interpretation: single-marker reading; association evidence: established.
Association source: Frosst 1995 Nat Genet (PMID 7647779); Crider 2011 Am J Clin Nutr (PMID 21508090); CDC MTHFR & folic acid guidance (cdc.gov/folic-acid/data-research/mthfr). Research attribution is separate from the private observation.

## MTHFR rs1801131

### TT · Sequencing

MTHFR — context only. This call does not measure folate processing.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A call at this site does not diagnose a deficiency or measure pathway activity. Missing data remain unknown.

Observation: GRCh38 · 1:11794419. Selected site only. Interpretation: single-marker reading; association evidence: emerging.
Association source: Weisberg 1998 Mol Genet Metab (PMID 9719624); van der Put 1998 Am J Hum Genet (PMID 9545395); CDC MTHFR & folic acid guidance (cdc.gov/folic-acid/data-research/mthfr). Research attribution is separate from the private observation.

## MTRR rs1801394

### AA · Sequencing

MTRR — context only. This call does not establish B12 deficiency, reduced B12 recycling, high homocysteine or extra choline need.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A call at this site does not diagnose a deficiency or measure pathway activity. Missing data remain unknown.

Observation: GRCh38 · 5:7870860. Selected site only. Interpretation: single-marker reading; association evidence: emerging.
Association source: Olteanu 2002 Biochemistry (PMID 12416982); Gaughan 2001 Atherosclerosis (PMID 11472746); Ganz 2016 FASEB J (PMID 27342765). Research attribution is separate from the private observation.

## MTR rs1805087

### AA · Sequencing

MTR — context only. This common-site call does not measure B12 need or enzyme activity, and does not screen for rare inherited disorders.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A call at this site does not diagnose a deficiency or measure pathway activity. Missing data remain unknown.

Observation: GRCh38 · 1:236885200. Selected site only. Interpretation: single-marker reading; association evidence: emerging.
Association source: Harmon 1999 Genet Epidemiol (PMID 10520212); Ma 1999 Cancer Epidemiol Biomarkers Prev (PMID 10498402). Research attribution is separate from the private observation.

## COMT rs4680

### AA · Sequencing

COMT — biological context only. This call does not predict your personality, stress response or methyl-group requirement.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This report does not use COMT to select nutrients, supplements or caffeine advice.

Observation: GRCh38 · 22:19963748. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Chen 2004 Am J Hum Genet (PMID 15457404).
Research attribution is separate from the private observation.

## CBS rs234706

### Not read

Why it was not read: no call in this sequencing file passed the quality check.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# Which inherited food clues are useful to me?

21 of 21 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

21 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Foods and portions you tolerate, a relevant symptom pattern and any existing nutrient result.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| MCM6/LCT · rs4988235 | GG | Sequencing |
| ALDH2 · rs671 | GG | Sequencing |
| ADH1B · rs1229984 | CC | Sequencing |

The first 3 of 21 markers read in this chapter. Each is explained below.

Lactose is the sugar in milk. Lactase is an enzyme in the small intestine that helps break lactose into smaller sugars so they can be absorbed. Some inherited regulatory variants support continued lactase production in adulthood. Without the relevant persistence variants, production commonly declines. DNA near LCT, within the neighbouring MCM6 gene, helps regulate this difference. [1]

If lactose is not fully digested and absorbed, it can reach the colon, where bacteria help break it down and produce fluid and gas. That can contribute to digestive symptoms. [13] Lactose malabsorption and symptomatic intolerance are different: some people can consume lactose without symptoms. [27] Where a readable inherited observation is available, relate it to your experience before assuming every dairy food must be excluded.

## Why a lactase marker is narrower than “milk is bad for me”

An inherited lactase marker concerns milk-sugar processing. Milk-protein allergy is a different condition with a different cause. [13] If an observation is available, it can help make a question more specific: “Could lactose in this product be relevant?” Symptoms after a dairy product still need their own context.

Research and guidance: [1] Lactose: the inherited mechanism (MedlinePlus Genetics); [36] Lactose intolerance: symptoms and assessment (NIDDK). These explain biology or guidance; they do not verify a private genotype.

| YOUR EXPERIENCE | WHAT IT SUGGESTS DOING NEXT |
|---|---|
| Milk and other dairy are comfortable | Keep the result as background. The report has not given you a reason to remove tolerated foods. |
| A usual milk drink repeatedly causes discomfort | Consider whether a lactose-free version of that product would be a useful comparison. |
| Some dairy is comfortable and some is not | Record the product and portion. A single label such as “dairy intolerant” may hide the pattern you need to understand. |
| Symptoms happen with many foods or do not fit this pattern | Keep the genetic clue in perspective. Discuss persistent symptoms rather than making progressively wider exclusions. |

## A concrete everyday example

**Illustrative situation:** you are comfortable eating hard cheese, but a large milk-based drink sometimes causes discomfort. The useful report does not tell you to abandon both. It helps you separate the product and portion from the broad food category. A comparison with the lactose-free version of the drink may answer a more relevant question than buying a new diet programme.

If that comparison makes no clear difference, it is useful information. It means the simple explanation did not answer the question in that setting. It does not invalidate the inherited observation, and it does not prove that you need to exclude more foods.



**Keep food choices familiar**
A finding becomes useful when it fits foods you enjoy, the portion you eat and your actual experience.

## A practical next step

If you have mild, familiar discomfort after a usual milk product, first note the product, amount and symptoms for a few ordinary occasions. If appropriate, substitute a lactose-free version while keeping the rest of the routine similar. Record whether the experience changes. This is an observation exercise, not a clinical diagnosis or a challenge to provoke symptoms.

| WHAT HAPPENS | WHAT YOU CAN CONCLUDE |
|---|---|
| The substituted product is more comfortable | That is useful for choosing the product. It supports the comparison in your routine; it does not prove a genetic cause. |

| WHAT HAPPENS | WHAT YOU CAN CONCLUDE |
|---|---|
| There is no consistent difference | Do not force the lactose explanation. Reconsider the question and discuss persistent symptoms if needed. |
| You already tolerate both products | There is no practical problem established by this comparison. Keep the result for reference. |

## Keeping nutrition practical

Hard cheese and yoghurt may be easier to tolerate than some other dairy products. Lactose-free dairy can also be an option. If dairy is reduced or excluded, maintaining calcium and vitamin D intake is a separate nutritional question. NIDDK describes food choices and alternatives in that context. [2]

## Questions readers commonly have

**Should I remove all dairy?** Your actual tolerance guides the useful choice. **Does lactose-free mean sugar-free?** It describes lactose handling; read the product's nutrition label for its contents. **Do I need to buy a genetic diet?** This result does not establish that need. **What if milk allergy is suspected?** Do not use a home food comparison to assess it; seek appropriate advice.

**What to avoid.** Avoid excluding several food groups at once to "follow your genes." You would lose the ability to tell which change helped, and may remove foods you tolerate. Significant or persistent symptoms need their own assessment.

Research and guidance: [2] Food choices with lactose intolerance (NIDDK); [36] Lactose intolerance: symptoms and assessment (NIDDK). These explain biology or guidance; they do not verify a private genotype.

> No relevant dated laboratory measurement is supplied in this chapter. This describes the report inputs; it does not mean a test was never performed.

# Selected markers and their interpretation

## MCM6/LCT rs4988235

### GG · Sequencing

MCM6/LCT — Lactase non-persistent genotype — adult lactose intolerance is more likely; symptoms vary.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 2:135851076. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Enattah 2002 Nat Genet; ClinVar VCV000007685 (ncbi.nlm.nih.gov/clinvar/variation/7685). Research attribution is separate from the private observation.

## ALDH2 rs671

### GG · Sequencing

ALDH2 — no ALDH2*2 allele at this site; that does not rule out flushing from other causes.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 12:111803962. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Brooks 2009 PLoS Med; ClinPGx (PharmGKB) ALDH2 (clinpgx.org/gene/PA24696). Research attribution is separate from the private observation.

## ADH1B rs1229984

### CC · Sequencing

ADH1B — no fast ADH1B*2 allele at this site, which does not measure how fast you clear alcohol.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 4:99318162. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Edenberg 2007 Alcohol Res Health (PMID 17718394); Bierut 2012 Mol Psychiatry (PMID 21968928); ClinPGx (PharmGKB) ADH1B (clinpgx.org/gene/PA24571). Research attribution is separate from the private observation.

## FTO rs9939609

### TT · Sequencing

FTO — no adiposity-linked allele at this variant, which does not measure your body weight.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 16:53786615. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Frayling 2007 Science; Kilpeläinen 2011 PLoS Med. Research attribution is separate from the private observation.

## FTO rs1421085

### TT · Sequencing

FTO — no adiposity-linked allele at the causal FTO variant, which does not measure your body weight.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 16:53767042. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Claussnitzer 2015 NEJM (causal ARID5B→IRX3/5 mechanism); Frayling 2007 Science. Research attribution is separate from the private observation.

## MTHFR rs1801133

### GA · Sequencing

MTHFR — one 677T allele. Research links this variant to enzyme properties and folate-related biomarkers; it does not measure your enzyme activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

MTHFR is not a general 'detoxification' gene

Observation: GRCh38 · 1:11796321. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Frosst 1995 Nat Genet (PMID 7647779); Crider 2011 Am J Clin Nutr (PMID 21508090); CDC MTHFR & folic acid guidance (cdc.gov/folic-acid/data-research/mthfr). Research attribution is separate from the private observation.

## MTHFR rs1801131

### TT · Sequencing

MTHFR — context only. This call does not measure folate processing.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

CDC: there isn't enough evidence that A1298C alone significantly affects how the body processes folate

Observation: GRCh38 · 1:11794419. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Weisberg 1998 Mol Genet Metab (PMID 9719624); van der Put 1998 Am J Hum Genet (PMID 9545395); CDC MTHFR & folic acid guidance (cdc.gov/folic-acid/data-research/mthfr). Research attribution is separate from the private observation.

## FUT2 rs602662

### AA · Sequencing

FUT2 — two non-secretor-tag alleles (consistent with non-secretor; read with another FUT2 marker). Studies link this genotype to higher total serum B12, which does not measure your B12 status.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

Secretor status shifts B12 and the gut microbiome but is not a B12-deficiency diagnosis

Observation: GRCh38 · 19:48703728. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Hazra 2008 Nat Genet; Tanaka 2009. Research attribution is separate from the private observation.

## BCMO1 rs12934922

### TT · Sequencing

BCMO1 — two 267S alleles, which do not measure your beta-carotene conversion. The cited study found lower conversion in carriers of a second variant (379V), which this report does not read.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A BCMO1 variant is not vitamin-A deficiency on a mixed diet

Observation: GRCh38 · 16:81268089. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Leung 2009 FASEB J (PMID 19103647). Research attribution is separate from the private observation.

## TAS2R38 rs713598

### CC · Sequencing

TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:141973545. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Kim 2003 Science (PMID 12595690); Bufe 2005 Curr Biol (PMID 15723792). Research attribution is separate from the private observation.

## TAS2R38 rs1726866

### AA · Sequencing

TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:141972905. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Kim 2003 Science (PMID 12595690); Bufe 2005 Curr Biol (PMID 15723792). Research attribution is separate from the private observation.

## TAS2R38 rs10246939

### TT · Sequencing

TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:141972804. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Kim 2003 Science (PMID 12595690); Bufe 2005 Curr Biol (PMID 15723792). Research attribution is separate from the private observation.

## HFE rs1800562

### GG · Sequencing

HFE — No HFE variant at this marker.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 6:26092913. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: EASL 2022 haemochromatosis guideline, J Hepatol (PMID 35662478); ClinVar VCV000000009 (ncbi.nlm.nih.gov/clinvar/variation/9). Research attribution is separate from the private observation.

## HFE rs1799945

### CC · Sequencing

HFE — No HFE variant at this marker.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 6:26090951. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: EASL 2022 haemochromatosis guideline, J Hepatol (PMID 35662478); ClinVar VCV000000010 (ncbi.nlm.nih.gov/clinvar/variation/10). Research attribution is separate from the private observation.

## HFE rs1800730

### AA · Sequencing

HFE — No HFE variant at this marker.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 6:26090957. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Mura 1999 Blood (PMID 10194428); Pedersen 2009 Ann Hematol (PMID 19159930); ClinVar VCV000000011 (ncbi.nlm.nih.gov/clinvar/variation/11). Research attribution is separate from the private observation.

## COMT rs4680

### AA · Sequencing

COMT — biological context only. This call does not predict your personality, stress response or methyl-group requirement.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

COMT personality labels are a pop-science oversimplification — this report assigns no temperament from COMT

Observation: GRCh38 · 22:19963748. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Chen 2004 Am J Hum Genet (PMID 15457404).
Research attribution is separate from the private observation.

## COMT rs4633

### TT · Sequencing

COMT — observed site only; no genotype or personal activity inferred at another COMT marker.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A linked marker is not an observed rs4680 call or a measured haplotype.

Observation: GRCh38 · 22:19962712. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Nackley 2006 Science (PMID 17185601).
Research attribution is separate from the private observation.

## VDR rs2228570

### AG · Sequencing

VDR — FokI: one copy each of the alleles for the shorter and the longer form of the receptor. Cell studies disagree on whether the two forms differ in activity. A 25-OH-D blood test, not this marker, shows your vitamin D status.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

VDR genotype does not set your vitamin-D level — a 25-OH-D blood test does

Observation: GRCh38 · 12:47879112. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Arai 1997 J Bone Miner Res (PMID 9169350); Gross 1998 J Bone Miner Res (PMID 9797477); Uitterlinden 2004 Gene (PMID 15315818). Research attribution is separate from the private observation.

## VDR rs1544410

### CC · Sequencing

VDR — BsmI: no variant allele at this marker, which does not measure your vitamin D status.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 12:47846052. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Uitterlinden 2004 Gene (PMID 15315818); Uitterlinden 2006 Ann Intern Med (PMID 16908916); Morrison 1994 Nature (PMID 8161378; corrected 1997). Research attribution is separate from the private observation.

## VDR rs731236

### AA · Sequencing

VDR — TaqI: no variant allele at this tag, which does not measure your vitamin D status.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 12:47844974. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Uitterlinden 2004 Gene (PMID 15315818); Uitterlinden 2006 Ann Intern Med (PMID 16908916). Research attribution is separate from the private observation.

## VDR rs7975232

### AA · Sequencing

VDR — ApaI: two copies of an intronic tag allele with no established effect of its own. A 25-OH-D blood test, not this marker, shows your vitamin D status.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

ApaI is an intronic marker with no established standalone function — part of the VDR haplotype only

Observation: GRCh38 · 12:47845054. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Uitterlinden 2004 Gene (PMID 15315818); Uitterlinden 2006 Ann Intern Med (PMID 16908916). Research attribution is separate from the private observation.

# What can DNA explain about digestion?

> 8 of 10 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

8 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
What happens, the food/portion involved, timing, medicines and any existing assessment.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| MCM6 · rs4988235 | GG | Sequencing |
| FUT2 · rs601338 | AA | Sequencing |
| FUT2 · rs602662 | AA | Sequencing |

The first 3 of 8 markers read in this chapter. Each is explained below.



**One symptom, several possible questions**
Digestive experience, selected inherited mechanisms and current measurements answer different questions. Keep each question attached to its own evidence.

## Start with the experience you want to understand

Record what happens, the food or portion involved, the timing and whether the pattern repeats. A report of selected DNA sites cannot identify the cause of a digestive symptom. A missing marker is an unresolved input, and a readable marker is not a diagnosis.

## Lactose, coeliac disease and other digestive questions

The food chapter explains one inherited lactase mechanism. Lactose intolerance and milk allergy are different questions. Coeliac assessment is also separate: the selected markers in this wellness edition do not supply a clinical exclusion or

diagnosis. Symptoms and any existing assessment should guide the question brought to care. [13, 27, 19]

## What an association can contribute

Some selected gut-related markers concern nutrient handling or susceptibility studied in populations. They do not measure absorption today, the organisms living in the gut, inflammation or the cause of discomfort. The observation rows below state which sites were read and where an interpretation stops.

## Read measurements in their original context

An existing B12 result is a dated blood measurement, rather than a direct test of digestive function. Keep its units, laboratory reference and reason for testing with it. If a result is supplied, it does not establish why a value changed or which intervention is needed. [10]

> **Keep the question specific**
> A useful note might ask what an existing assessment already established, what remains uncertain and whether the symptom pattern needs further discussion. This report supplies no instruction to remove a food group or start a supplement.

## Your practical record

- The symptom or food question I want to clarify
- Food, portion, timing and usual experience
- Existing assessment or result, with date and source
- What is still unknown
- The reply received and the reason for any choice, including no change

> No relevant dated laboratory measurement is supplied in this chapter. This describes the report inputs; it does not mean a test was never performed.

# Selected markers and their interpretation

## MCM6 rs4988235

### GG · Sequencing

MCM6 — Lactase non-persistent genotype — adult lactose intolerance is more likely; symptoms vary.

**Eat:** Hard cheese, yoghurt or lactose-free milk usually sit well. You likely produce little lactase in adulthood; ordinary milk may cause bloating, a lactase enzyme helps, and dairy isn't required (plenty of non-dairy calcium sources).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 2:135851076. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Enattah 2002 Nat Genet; Bersaglieri 2004 Am J Hum Genet. Research attribution is separate from the private observation.

## FUT2 rs601338

### AA · Sequencing

FUT2 — a non-secretor (no ABO antigens secreted into mucus or saliva). In cohorts non-secretors differ in gut bacteria and B12 levels and resist some norovirus strains — associations, not a diagnosis.

**Eat:** Prebiotic fibre and fermented foods support gut bifidobacteria. As a likely non-secretor you tend to carry fewer of them: onion, garlic, oats and legumes, plus yoghurt, kefir, sauerkraut or kimchi, support microbiome diversity.

**Measure:** Serum B12 or active B12 — none on file.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This site reads W143X, the common inactive FUT2 allele in many populations. Other inactive or weak alleles are not read here (in East and Southeast Asia a different one, I129F, is the common one), so a result without two W143X copies is consistent with being a secretor and does not establish it.

Observation: GRCh38 · 19:48703417. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Kelly 1995 J Biol Chem; Wacklin 2011 PLoS One (microbiome); Thorven 2005 J Virol (norovirus); Soejima 2021 Sci Rep (other inactive alleles; PMCID PMC7862633). Research attribution is separate from the private observation.

## FUT2 rs602662

### AA · Sequencing

FUT2 — two non-secretor-tag alleles (consistent with non-secretor; read with another FUT2 marker). Studies link this genotype to higher total serum B12, which does not measure your B12 status.

**Measure:** Serum B12 or active B12 — none on file.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A secretor tag SNP — the primary secretor call is rs601338 (W143X). Read the two together; a tag is a linkage proxy, not the functional variant.

Observation: GRCh38 · 19:48703728. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: McGovern 2010 Hum Mol Genet; Tong 2014 (FUT2 secretor tag). Research attribution is separate from the private observation.

## AOC1 rs10156191

### CT · Sequencing

AOC1 — one 16Met allele, which does not measure your DAO activity. In one study it went with lower DAO activity in blood, not with histamine intolerance itself.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:150856517. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Maintz 2011 Allergy (PMID 21488903); Ayuso 2007 Pharmacogenet Genomics (PMID 17700358). Research attribution is separate from the private observation.

## AOC1 rs1049793

### CC · Sequencing

AOC1 — no 645Asp allele at this site, which does not measure your DAO activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

C and G are each other's complement, so a file written on the opposite strand could be read the wrong way round.

Observation: GRCh38 · 7:150860577. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Ayuso 2007 Pharmacogenet Genomics (PMID 17700358). Research attribution is separate from the private observation.

## HLA-DQA1 rs2187668

### CC · Sequencing

HLA-DQA1 — no DQ2.5 tag allele, but that does not exclude the risk type, and one tag SNP cannot show your HLA-DQ type; HLA-DQ2.5 is the main inherited risk type for celiac disease. Only a dedicated HLA-DQ test, not this marker, can show your HLA-DQ type.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This SNP only TAGS the HLA alleles, and the HLA genes themselves are not typed from this file — treat any HLA-DQ2.5 call here as LOW-CONFIDENCE / effectively a blind spot. A dedicated HLA-DQ typing test is needed to confirm; a negative tag does NOT exclude the risk haplotype (its absence is unknown, never a clinical negative).

Observation: GRCh38 · 6:32638107. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Monsuur 2008 PLoS One (rs2187668 as DQ2.5 tag; PMID 18509540); Romanos 2009 Gastroenterology; Erlich 2008 Diabetes (PMID 18252895). Research attribution is separate from the private observation.

## NOD2 rs2066844

### CC · Sequencing

NOD2 — no 702W allele at this site, which does not rule out Crohn's disease or another NOD2 risk allele.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 16:50712015. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Hugot 2001 Nature (PMID 11385576); Ogura 2001 Nature; Cuthbert 2002 Gastroenterology (PMID 11910337); Bonen 2003 Gastroenterology (PMID 12512038). Research attribution is separate from the private observation.

## NOD2 rs2066845

### GG · Sequencing

NOD2 — no 908R allele at this site, which does not rule out Crohn's disease or another NOD2 risk allele.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

C/G is palindromic, so a file written on the opposite strand could be read the wrong way round. Read with R702W (rs2066844); the third common NOD2 risk allele, the frameshift 3020insC, is not read here.

Observation: GRCh38 · 16:50722629. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Hugot 2001 Nature (PMID 11385576); Ogura 2001 Nature; Cuthbert 2002 Gastroenterology (PMID 11910337); Bonen 2003 Gastroenterology (PMID 12512038). Research attribution is separate from the private observation.

## HLA-DQB1 rs7454108

### Not read

Why it was not read: no call in this sequencing file passed the quality check.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

## HLA-DQB1 rs7775228

### Not read

Why it was not read: no call in this sequencing file passed the quality check.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# What is inherited, and what needs a measurement?

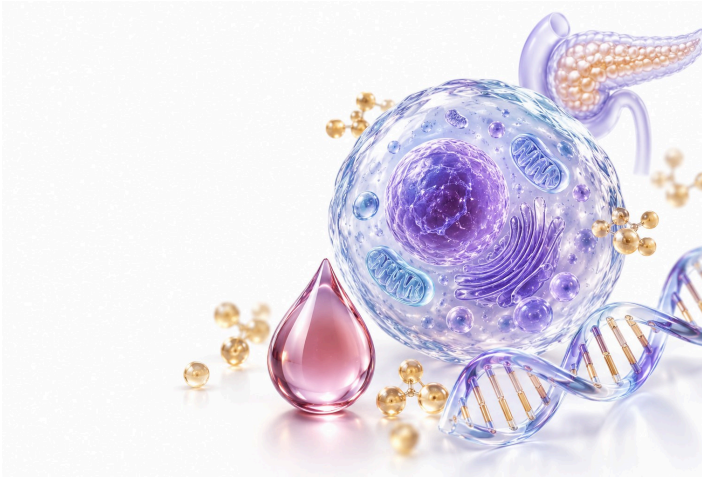
> 5 of 5 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

5 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
A dated glucose or HbA1c record, its conditions, family history and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| TCF7L2 · rs7903146 | TT | Sequencing |
| PNPLA3 · rs738409 | CG | Sequencing |
| FTO · rs9939609 | TT | Sequencing |

The first 3 of 5 markers read in this chapter. Each is explained below.

Researchers identify common genetic associations by comparing people with different outcomes. TCF7L2 is a well-studied diabetes-associated region. A supported observation at this region can provide context about inherited susceptibility; its individual meaning depends on the actual call and the evidence supplied. It is not a measurement of your blood glucose or a statement that diabetes will happen.

## What the prevention study can tell us

The Diabetes Prevention Program analysis studied TCF7L2 in people who already had impaired glucose tolerance. It connected genotype with progression and insulin-related measurements in that population. [3] The study helps explain why the finding is relevant, but its participants' absolute risks should not be pasted into a report for a generally healthy person. The study did not establish a statistically significant interaction between genotype and the intervention. It does not identify which intervention will work best for you.

## The difference between an association and your probability

An association describes a difference observed between study groups. A personal probability needs a defined outcome and time period, relevant population data and a model that includes the

necessary context. This edition has none of those ingredients for a calibrated probability. That is why it gives you a meaningful explanation instead of a percentage that looks precise but answers the wrong question.

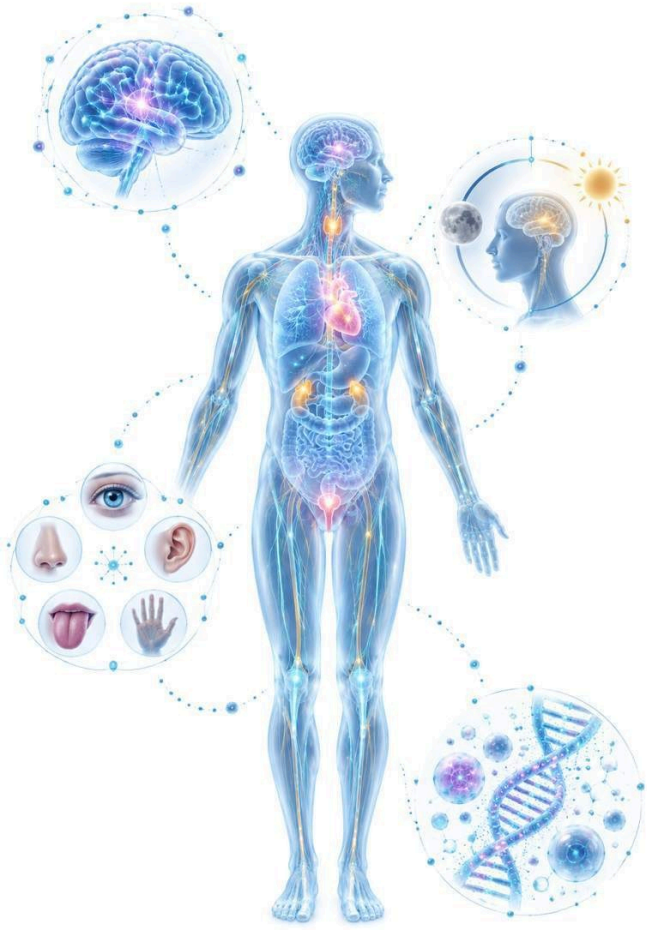
## A finding without blame

A body-weight-associated marker does not measure willpower, explain every weight change or tell you which diet you must follow. A useful plan needs to fit your actual circumstances. The purpose of this chapter is to clarify a health question, not assign responsibility to a gene or to the reader.

Research and guidance: [3] TCF7L2 and diabetes prevention (Florez et al., NEJM 2006). These explain biology or guidance; they do not verify a private genotype.

| INFORMATION TO ADD | THE QUESTION IT HELPS ANSWER |
|---|---|
| Family history and your medical history | How does the common marker fit into the broader assessment? |
| A1C/HbA1c, date and units, if available | What has blood glucose been doing over recent months? |
| A fasting glucose result, with date and conditions, if available | What was measured at that particular time? |
| Previous results and any existing care plan | Is there a change, or a question already being followed? |

A1C reflects average blood glucose over roughly the preceding three months. A fasting plasma glucose test measures a point in time. Clinicians use blood tests, interpreted in context and sometimes repeated, to assess diabetes or prediabetes. [14, 15] These measurements answer a different question from inherited susceptibility. Conditions affecting red-cell lifespan, recent blood loss or transfusion, and some haemoglobin variants can affect A1C interpretation. A result that disagrees with other measurements needs its original assay and clinical context. [15]



**Inherited clues, observed patterns**
The inherited record is one part of the picture. A dated blood result and your history answer different questions.

## Two examples of interpretation in context

**Illustrative situation A:** you feel well and have a recent clinically reviewed blood result. The inherited finding does not invalidate that result or establish a diagnosis. Keep it as background for the appropriate screening schedule.

**Illustrative situation B:** your family history concerns you and you cannot find a recent result. The useful next step is a specific routine-care question: “Am I up to date with appropriate glucose screening?” The marker adds context, but it does not by itself justify an intensive monitoring programme.

## For an everyday habit

If you want to improve a routine, choose a general health step that you can actually maintain, such as including a fibre-rich food in a meal or making regular activity easier to fit into the week. These are general examples. A TCF7L2 observation does not select a diet or exercise prescription.

| A USEFUL TARGET | HOW TO KEEP IT UNDERSTANDABLE |
|---|---|
| Make a usual meal more varied | Choose a food you like. Describe the change in ordinary language rather than claiming it “switches off” a variant. |
| Make regular movement easier to maintain | Choose an appropriate activity and a realistic routine. Fit it around your current abilities and relevant medical advice. |

| A USEFUL TARGET | HOW TO KEEP IT UNDERSTANDABLE |
| --- | --- |
| Clarify your measurement history | Find the date and units of an existing result before assuming no result exists or ordering a new one. |

## What to avoid

Avoid treating the marker as a diagnosis, converting an association into your personal probability, or buying a strict “TCF7L2 diet” from this finding alone. If you already receive diabetes care, continue your care plan and discuss changes with your team rather than redesigning treatment from a consumer-file report.

## Questions readers commonly have

**Does a common genotype elsewhere mean I am protected?** It does not give overall clearance. **Will healthy habits remove the variant?** The inherited letters remain; a useful habit can still matter. **Do I need a CGM because this is highlighted?** This finding alone does not establish that requirement. Ask which measurement would answer your actual question.

> No relevant dated laboratory measurement is supplied in this chapter. This describes the report inputs; it does not mean a test was never performed.

# Selected markers and their interpretation

## TCF7L2 rs7903146

### TT · Sequencing

TCF7L2 — two T2D-associated alleles: a raised tendency at this one marker, not a diagnosis or a combined genetic score.

**Eat:** Fibre-rich, lower-glycaemic meals and activity lower diabetes risk. That holds with two copies too: they raise the odds, not the verdict. Your measured fasting glucose or HbA1c says more than this marker.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 10:112998590. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Grant 2006 Nat Genet (PMID 16415884); Morris 2012 Nat Genet (DIAGRAM, PMID 22885922); Saxena 2006 Diabetes (PMID 17003358). Research attribution is separate from the private observation.

## PNPLA3 rs738409

### CG · Sequencing

PNPLA3 — one 148M allele; a modestly higher liver-fat tendency, larger with more body fat (not a diagnosis).

**Eat:** Cutting alcohol and added sugar/fructose is the highest-leverage step. This variant raises the liver-fat tendency, and it is strongly diet-modifiable.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 22:43928847. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Romeo 2008 Nat Genet (PMID 18820647); Rotman 2010 Hepatology (PMID 20684021); Stender 2017 Nat Genet (PMID 28436986). Research attribution is separate from the private observation.

## FTO rs9939609

### TT · Sequencing

FTO — no adiposity-linked allele at this variant, which does not measure your body weight.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 16:53786615. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Frayling 2007 Science; Kilpeläinen 2011 PLoS Med. Research attribution is separate from the private observation.

## MC4R rs17782313

### TT · Sequencing

MC4R — no adiposity-linked allele at this marker, which does not measure your body weight.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 18:60183864. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Loos 2008 Nat Genet (PMID 18454148); Qi 2008 Hum Mol Genet (PMID 18697794). Research attribution is separate from the private observation.

## TM6SF2 rs58542926

### CC · Sequencing

TM6SF2 — no 167K allele at this site, which does not rule out fatty liver.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 19:19268740. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Kozlitina 2014 Nat Genet (PMID 24531328). Research attribution is separate from the private observation.

# How do inherited clues fit my lipid picture?

5 of 5 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

5 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Dated lipid results in their original units, family history and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| APOE · rs429358 | TT | Sequencing |
| LPA · rs10455872 | AA | Sequencing |
| LPA · rs3798220 | TT | Sequencing |

The first 3 of 5 markers read in this chapter. Each is explained below.

## Your lipid picture has several parts

Inherited information and blood measurements answer different questions. A genetic finding may help explain a tendency; a laboratory result records a value at a particular time and under particular conditions. Neither alone provides a complete assessment of heart and vascular health. If you already have a lipid report, keep its date, original units and source with this chapter. If you already receive care, use the report within that care.

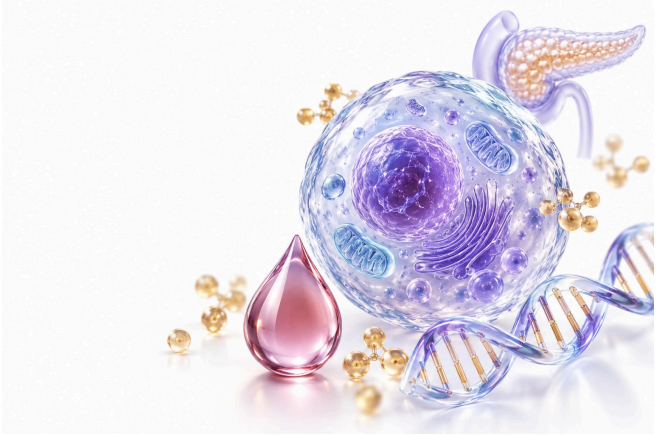
## Cholesterol carried and particle number

LDL cholesterol reports cholesterol carried in LDL particles. HDL cholesterol and triglycerides add other parts of the lipid picture. Non-HDL cholesterol can be calculated from total cholesterol and HDL from the same draw in matching units. ApoB provides information about the number of particles that can contribute to atherosclerosis. These measures overlap but are not interchangeable. A calculation should identify its inputs and units. [30]

## The inherited component of Lp(a)

Lp(a) has a strong inherited component. Keep any previous measurement with its date and method. Its relative stability does not make every old record a measurement of today. Preserve the unit reported by the laboratory: mass and particle-concentration units cannot be exchanged using a universal conversion factor. Do not force records with different methods or units into a trend.

The selected LPA tag variants do not fully assess the complex repeat region and do not measure a blood concentration. Absence of their highlighted alleles does not rule out raised Lp(a). Their informativeness differs across studied populations. A selected common-variant result is not a complete assessment of inherited lipid disorders.



**A connected metabolic picture**
Lipid and glucose records can be discussed together in existing care. Their original dates, conditions and units remain distinct.

## Context gives a measurement meaning

Relevant history can include cardiovascular disease, blood pressure, smoking, diabetes, medicines and family history. A target used in one clinical setting is not a universal definition of a result suitable for every reader. This edition does not calculate an overall cardiovascular risk score, assign a treatment target, or select a medicine or supplement.

## Sensitive findings have a separate scope

APOE isoforms require appropriate information at both defining sites, with uncertainty retained when information is missing, conflicting or phase-ambiguous. APOE also has implications beyond lipids. A clinical assessment of inherited lipid disorders is a separate question and is not excluded by this common-variant report.

## Keep a useful question

Locate any existing lipid results first. Record what was reviewed, what remains missing and what follow-up was agreed. Continuing an appropriate existing plan or deciding that no change is needed can be useful outcomes. Dietary and activity choices can be discussed within your circumstances; these marker observations do not select a special diet, supplement, medicine or dose.

Research and guidance: [42] Lipoprotein(a) interpretation and units (European Atherosclerosis Society consensus). These explain biology or guidance; they do not verify a private genotype.

> No relevant dated laboratory measurement is supplied in this chapter. This describes the report inputs; it does not mean a test was never performed.

# Selected markers and their interpretation

## APOE rs429358

### TT · Sequencing

APOE — no ε4-defining C allele at this SNP (isoform needs another APOE marker too to call fully).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This row reads rs429358 alone; the ε2/ε3/ε4 isoform also depends on rs7412.

Observation: GRCh38 · 19:44908684. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Deelen 2019 Nat Commun; Corder 1993 Science. Research attribution is separate from the private observation.

## LPA rs3798220

### TT · Sequencing

LPA — no high-Lp(a) tag allele (does not rule out high Lp(a) — your measured Lp(a) says more than these markers).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

Lp(a) is largely set by the LPA KIV-2 kringle repeat copy number — not a SNP. rs3798220 only tags high Lp(a); a negative tag is not reassurance. Your measured Lp(a) says more than these markers.

Observation: GRCh38 · 6:160540105. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Luke 2007 Arterioscler Thromb Vasc Biol (PMID 17569884); Clarke 2009 NEJM (PMID 20032323). Research attribution is separate from the private observation.

## LPA rs10455872

### AA · Sequencing

LPA — no high-Lp(a) tag allele (but this does not rule out high Lp(a) — your measured Lp(a) says more than these markers).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

Lp(a) is largely set by the LPA KIV-2 kringle repeat copy number — not a SNP. rs10455872 only tags high Lp(a); a negative tag is not reassurance. Your measured Lp(a) says more than these markers.

Observation: GRCh38 · 6:160589086. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Clarke 2009 NEJM (PMID 20032323); Kamstrup 2009 JAMA (PMID 19509380). Research attribution is separate from the private observation.

## F5 rs6025

### CC · Sequencing

F5 — no Factor V Leiden at this site, which does not test the rest of the clotting system.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

One site of one clotting gene; it does not test the rest of the clotting system. A blood test (activated protein C resistance, then DNA) confirms it.

Observation: GRCh38 · 1:169549811. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Bertina 1994 Nature (PMID 8164741); Vandenbroucke 1994 Lancet (with oral contraceptives); GeneReviews Factor V Leiden Thrombophilia (NBK1368). Research attribution is separate from the private observation.

## F2 rs1799963

### GG · Sequencing

### F2 — no prothrombin G20210A at this site, which does not test the rest of the clotting system.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

One site of one clotting gene; it does not test the rest of the clotting system. A targeted DNA test confirms it.

Observation: GRCh38 · 11:46739505. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Poort 1996 Blood (PMID 8916933); GeneReviews Prothrombin Thrombophilia (NBK1148). Research attribution is separate from the private observation.

# Can these findings help me understand coffee and sleep?

> 4 of 5 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

4 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Caffeine timing, preferred and actual sleep, work constraints and your daytime experience.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| ADORA2A · rs5751876 | TT | Sequencing |
| BHLHE41 · rs121912617 | GG | Sequencing |
| MTNR1B · rs10830963 | CC | Sequencing |

The first 3 of 4 markers read in this chapter. Each is explained below.

## Feeling jittery and sleeping after caffeine

ADORA2A is involved in the response to adenosine, a signal relevant to wakefulness and sleep. Research on caffeine-related anxiety and research on sleep ask different questions. Their findings do not create an established scale that ranks everyone from insensitive to sensitive. Habitual use and study conditions also matter. Your observed experience should keep those outcomes separate.

## Clearance and timing

The CYP1A2 marker concerns enzyme inducibility in studied conditions. [28] It does not measure your current caffeine clearance, establish a personal half-life, or produce an ideal last-coffee time. Preferred timing, actual sleep and the schedule you must follow are separate information.

A cup of coffee can be discussed in at least three ways: how the drink makes you feel, how the body handles caffeine, and how its timing fits with your sleep. Those questions overlap in daily life, but a marker relevant to one should not become a verdict on all three.

| THE QUESTION | WHAT A USEFUL EXPLANATION NEEDS |
|---|---|
| “Do I feel jittery after coffee?” | Your experience, usual intake and the specific evidence for a sensitivity claim. |
| “How quickly do I clear caffeine?” | A supported metabolism interpretation, with the exact variant and relevant context. |
| “Does my last drink affect sleep?” | The timing of intake and a consistent record of your sleep experience. |
| “Am I genetically a night owl?” | Evidence for the specific clock claim, not an isolated unreplicated association. |

## Why the explanation is still useful

You can now distinguish a genetic label from the question you actually want answered. Even when the current markers do not settle the question, the report can help you design a more informative comparison and explain why no personal cut-off time is printed.

Research and guidance: [5] Caffeine, ADORA2A and habitual use (Rogers et al., Neuropsychopharmacology 2010). These explain biology or guidance; they do not verify a private genotype.

| CONTEXT | WHY IT CHANGES THE QUESTION |
|---|---|
| Usual drinks, amount and timing | A late drink and a morning drink may be different practical questions. Tea and other sources also count. |
| How regularly you use caffeine | Habitual use is relevant to interpreting research and to noticing withdrawal when habits change. |
| Bedtime and work pattern | A clock-time suggestion is less useful without knowing when you actually sleep. |
| What you want to improve | Feeling jittery, difficulty falling asleep and low morning alertness need different comparisons. |

## An example of a useful question

**Illustrative situation, independent of genotype:** a person drinks coffee at different times and notices variable sleep. A useful first step is to record the existing routine, sleep opportunity and daytime experience. The record may clarify the question without assuming that a DNA marker explains the pattern.

If you feel comfortable and sleep well with your existing pattern, a genetic association alone does not establish a problem to solve. Understanding that is also a useful result from the report.



**Notice the day as well as the night**
Caffeine timing, sleep opportunity and daytime experience form a record you can understand without assigning a genetic bedtime.

## How to interpret the result

A consistent improvement can support keeping a useful habit. No clear change is also an answer: the comparison did not establish a practical benefit in that setting. Neither outcome proves a genetic mechanism. Avoid repeatedly changing several things to obtain the result you expected from the label.

## What to avoid

Do not increase caffeine to test a genetic sensitivity. If you regularly consume caffeine, an abrupt reduction can itself affect how you feel. Do not use a fixed “genetic cut-off time” that the available evidence has not established. Persistent sleep problems deserve an assessment on their own merits.

## Questions readers commonly have

**Is decaf useful as a comparison?** Check what you are comparing and the product’s contents; the aim is a small routine change. **Can I use a watch?** It may help you keep a consistent record, but device estimates are not a diagnosis. **What if the result is unclear?** Keep the uncertainty and reconsider the question. You do not have to force the gene label to fit.

> **The useful outcome**
> You leave with a clearer description of your own caffeine pattern, and understand which part of that conclusion comes from observation rather than genetics.

## Choose one question and record your day simply

If caffeine or difficult evenings are relevant to your question, record your existing routine, sleep opportunity and actual experience. Keep other changes and missing entries visible. Make no change if you are comfortable with your routine. A diary can help organise observations for a discussion; it does not validate a genetic caffeine cutoff or prescribe a treatment.

Record the last caffeine drink, product and approximate amount, bedtime and wake time, settling or awakenings, and the daytime experience you want to understand. Add shift changes, naps or unusual nights where useful. A missing entry stays missing. A device estimate and your own experience should retain their different source labels.

At review, ask what stayed similar, what changed repeatedly, and what makes the comparison uncertain. A useful change does not prove that a gene caused the experience. An unclear comparison can stay unclear. Do not increase caffeine to test a label; changing habitual intake can introduce withdrawal effects.

If poor sleep continues to affect your day, discuss it on its own merits. Loud snoring, gasping or breathing pauses need a different question from coffee sensitivity. A genetic label or device score does not rule out a sleep problem.

Research and guidance: [37] Sleep diary and assessment (NHLBI); [38] Caffeine anxiety and habitual use (Rogers et al. 2010); [39] Caffeine-related sleep response (Retey et al. 2007). These explain biology or guidance; they do not verify a private genotype.

# Selected markers and their interpretation

## ADORA2A rs5751876

### TT · Sequencing

ADORA2A — linked in two studies to more jitteriness after caffeine, less so in regular drinkers. Not a sleep result: sleep studies point to the other allele.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 22:24441333. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Childs 2008 Neuropsychopharmacology (PMID 18305461); Rogers 2010 Neuropsychopharmacology (PMID 20520601); Rétey 2007 Clin Pharmacol Ther (PMID 17329997); Bodenmann 2012 Br J Pharmacol (PMID 21950736). Research attribution is separate from the private observation.

## BHLHE41 rs121912617

### GG · Sequencing

BHLHE41 — the common genotype; no familial-short-sleep allele (expected).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

A rare allele — the vast majority carry zero copies, so a negative here is expected and uninformative. It did not replicate in UK Biobank carriers. C/G is palindromic, so a file written on the opposite strand could be read the wrong way round. Not a lever for the general population.

Observation: GRCh38 · 12:26122364. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: He 2009 Science (PMID 19679812); Weedon 2022 PLoS Genet (PMID 36137075, population non-replication). Research attribution is separate from the private observation.

## MTNR1B rs10830963

### CC · Sequencing

MTNR1B — the common genotype; no glucose-raising allele from this variant.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

C/G is palindromic, so a file written on the opposite strand could be read the wrong way round. This variant's strong evidence is for fasting glucose, not sleep; its only sleep-side finding is a small, unreplicated melatonin-offset result, and population sleep timing was unaffected. Measured/wearable sleep decides.

Observation: GRCh38 · 11:92975544. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Prokopenko 2009 Nat Genet (PMID 19060907; fasting glucose, G allele); Lane 2016 Diabetes (PMID 26868293; melatonin offset, lab n≈95; no population sleep-timing effect). Research attribution is separate from the private observation.

## CYP1A2 rs762551

### AA · Sequencing

CYP1A2 — the higher-inducibility genotype. Measurable mainly in smokers and heavy coffee drinkers; in non-smokers it has not been shown to change caffeine clearance.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This variant does NOT predict your baseline caffeine clearance. In non-smokers it shows no measurable effect on caffeine metabolic ratio (Sachse 1999, n=185; Ghotbi 2007, n=194; Djordjevic 2010, n=178)

Observation: GRCh38 · 15:74749576. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Sachse 1999 Br J Clin Pharmacol; Ghotbi 2007 Eur J Clin Pharmacol; Djordjevic 2010; Cornelis 2016 Hum Mol Genet (GWAS). Research attribution is separate from the private observation.

## PER3 rs57875989

### Not read

#### Why it was not read: a repeat length, not a single site — needs a dedicated assay.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.

Interpretation: none, the site was not read.

Association source: none, the site was not read. Research attribution is separate from the private observation.

# What does brain biology explain - and where does it stop?

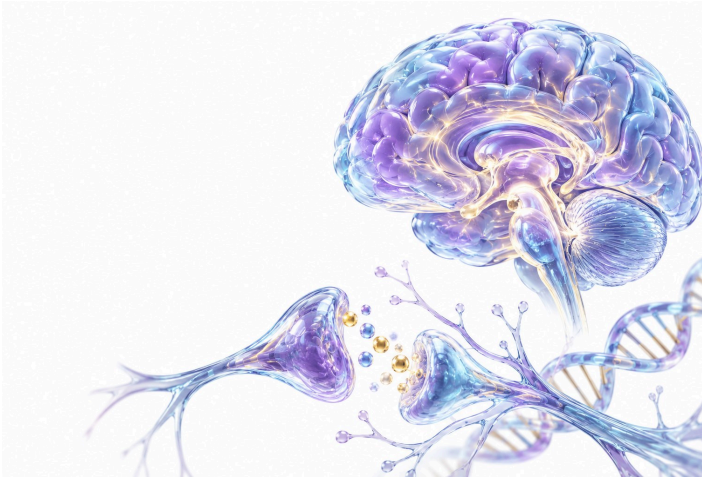
> 3 of 4 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

3 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| COMT · rs4680 | AA | Sequencing |
| MAOA · rs6323 | T (single X copy) | Sequencing |
| BDNF · rs6265 | CC | Sequencing |

COMT provides instructions for an enzyme that helps break down some chemical messengers, including dopamine. Common variation at rs4680 can affect the enzyme's properties. [6] That is why the gene appears in discussions about brain biology. A common-marker observation does not measure your dopamine concentration, or show how the full signalling system behaves during a workday.

> **The main answer**

> The source does not establish a temperament type. It gives limited observations that can be explained, while your actual question about mood or focus needs its own context.

Research and guidance: [6] What COMT does (MedlinePlus Genetics). These explain biology or guidance; they do not verify a private genotype.

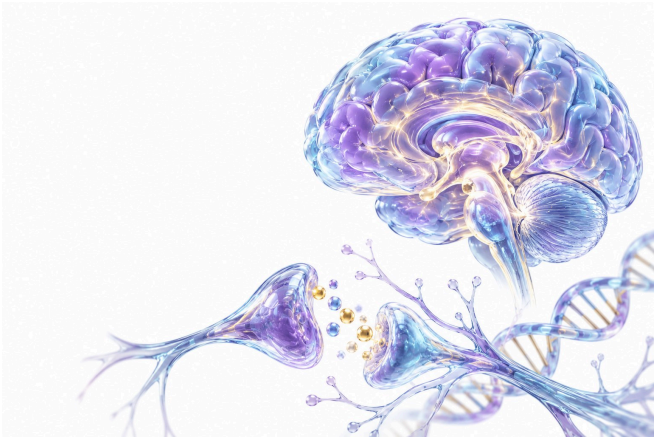
Focus can mean staying with a task, remembering what you read or avoiding distraction. Stress can mean a brief response or a persistent concern. Name the experience before trying to explain it.

| A BROAD QUESTION | A MORE USEFUL QUESTION TO INVESTIGATE |
|---|---|
| “Am I a worrier because of COMT?” | “Which situations repeatedly leave me tense, and what do I notice before and after them?” |
| “Do I have bad focus genes?” | “Does concentration become difficult at a particular time, with particular tasks, or after poor sleep?” |
| “Is my cortisol genetically high?” | “What specific symptoms or clinical question would justify evaluating a stress-related concern?” |
| “Does this explain a mental-health condition?” | “What assessment and history are relevant to the concern I am experiencing?” |

## An everyday example

**Illustrative situation:** concentration is comfortable in the morning but drops during late meetings. A “slow COMT” label would not tell you whether sleep, workload or caffeine timing helps explain it. The recurring situation gives you something concrete to observe.

That does not mean every difficulty is a habit problem. It means the gene label has not established the cause. If the difficulty is persistent, distressing or affects everyday functioning, the useful next step is an appropriate assessment rather than a more elaborate personality interpretation.



**A pathway is not a personality**
Neurotransmitter biology is connected to many systems. A common selected variant does not define how you think or cope.

## For an everyday pattern

Choose one question, such as “When does concentration drop?” Record the time, task, sleep, caffeine timing and a simple note about the experience. This organizes the question; it is not a diagnostic score.

If a recurring setting becomes clear, consider one manageable change that is appropriate for you, such as adjusting when you do a demanding task or making a break easier to take. Evaluate whether it helps. The value comes from a useful fit with your experience, not a claim that the change is a COMT treatment.

## For a persistent concern

Bring a description of the problem, its duration, its effect on daily life and any relevant treatment history. The genetic page can remain background. It should not replace the clinical history or determine a treatment on its own.

| CARRY FORWARD | AVOID FROM THESE OBSERVATIONS ALONE |
|---|---|
| A clearly described question and a pattern you can explain. | A “warrior,” “worrier” or resilience identity assigned from the genotype. |
| A useful assessment if symptoms deserve one. | Assuming fatigue, anxiety or attention difficulties share one genetic explanation. |
| Your existing treatment and professional advice. | Starting or changing psychiatric treatment, hormones or supplements from these markers. |

## Questions readers commonly have

**Does no clear trait prediction mean COMT does nothing?** No. Its biological role is real; a reliable personal prediction is a separate claim. **Could future evidence improve this interpretation?** Yes, if it establishes a relevant claim for the exact observation and intended use. **Should I choose supplements that “balance dopamine”?** This report does not establish that you have an imbalance or that such a product would address it.

## Selected markers and their interpretation

### COMT rs4680

#### AA · Sequencing

COMT — biological context only. This call does not predict your personality, stress response or methyl-group requirement.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This report does not use COMT to select nutrients, supplements or caffeine advice.

Observation: GRCh38 · 22:19963748. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Chen 2004 Am J Hum Genet (PMID 15457404). Research attribution is separate from the private observation.

### MAOA rs6323

#### T (single X copy) · Sequencing

MAOA — the T allele at this tag, which does not measure your MAO-A activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

MAOA is on chromosome X: males carry one copy, so one allele is the whole read, and a call needs the sex the analysis was run with. Not a measure of MAO-A activity, and never a behavioural verdict.

Observation: GRCh38 · X:43731789. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Hotamisligil 1991 Am J Hum Genet (PMID 1678250); Verma 2014 (PMID 24291416); Wang 2018 (PMID 30018578); dbSNP rs6323 (ncbi.nlm.nih.gov/snp/rs6323). Research attribution is separate from the private observation.

## BDNF rs6265

### CC · Sequencing

BDNF — no Met66 allele at this site, which does not measure BDNF release, memory or stress response.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 11:27658369. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Egan 2003 Cell; Hariri 2003 J Neurosci.
Research attribution is separate from the private observation.

## SLC6A4 rs25531

### Not read

Why it was not read: an insertion, deletion or copy-number change — not read here as a single site.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# How does pigment biology fit my sun routine?

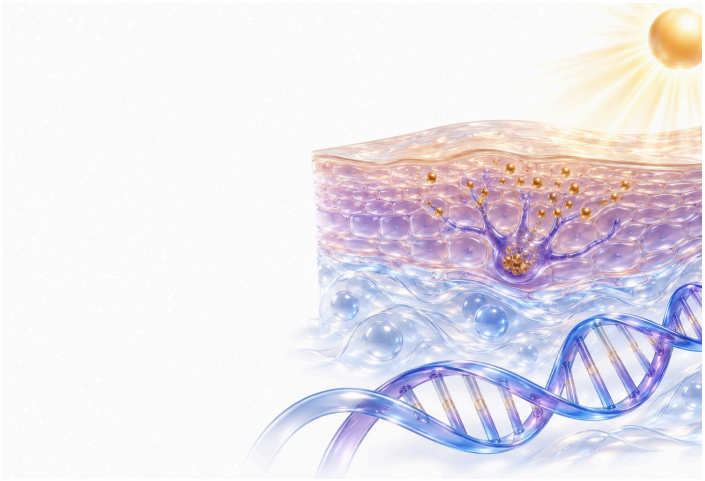
> 5 of 6 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

5 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| HERC2 · rs12913832 | GG | Sequencing |
| MC1R · rs1805007 | CC | Sequencing |
| MC1R · rs1805008 | CC | Sequencing |

The first 3 of 5 markers read in this chapter. Each is explained below.

MC1R encodes a receptor involved in signalling within pigment-producing cells. Its activity helps influence the types of melanin made. Eumelanin provides more protection from ultraviolet radiation than pheomelanin; variants affecting this biology help explain why inherited pigmentation can be relevant to sun response. [16]

## What this can help you understand

The useful insight is that your sun response has a biological context. If you already notice that you burn easily, a pigment-related finding can make that experience easier to understand. If the description does not match your appearance or experience, the report should not insist that a small set of markers is a complete description of your skin.

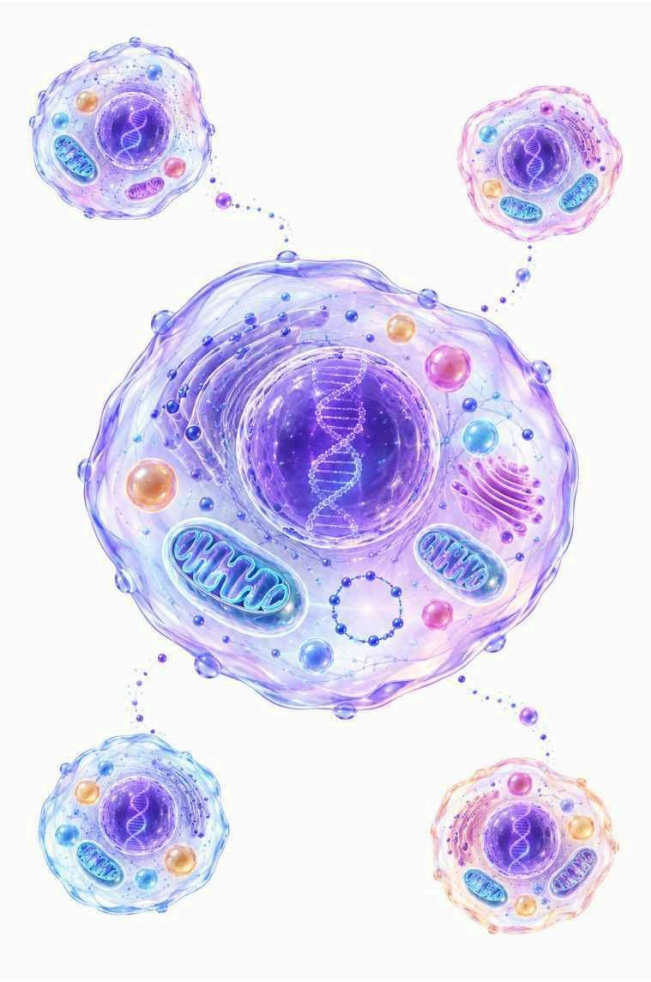
## Why the result is different from an examination

A changing mole is a current physical observation. An inherited pigment-associated result cannot assess it. Equally, a comfortable day in the sun does not establish that exposure caused no harm. These different questions need different evidence.

Research and guidance: [16] Pigmentation and MC1R (MedlinePlus Genetics). These explain biology or guidance; they do not verify a private genotype.

CDC recommends practical sun protection such as shade, protective clothing, sunglasses and broad-spectrum sunscreen. [7] These measures are general guidance; the report does not establish a special sunscreen product or a dose of protection from your genotype.

| YOUR SETTING | A PRACTICAL QUESTION |
|---|---|
| Walking, cycling or commuting outdoors | Which exposed areas and times are easy to overlook in this routine? |
| Long outdoor sessions or holidays | How will protection remain available and appropriate throughout the time outside? |
| You already burn easily | Does your usual routine provide practical protection before that happens? |
| You notice a new or changing skin concern | What assessment does that observation need, regardless of this DNA result? |



**Protection is an everyday practice**
Your actual response to sun and a suitable protection routine matter alongside pigmentation biology.

## An everyday example

**Illustrative situation:** you remember sunscreen on holiday but spend regular time outdoors during the week. The useful report helps you connect protection with those ordinary days. The habit does not need a genetic score to be sensible; the pigment explanation gives context for why sun response deserves attention.

## What to avoid

Avoid using a common marker result as reassurance that you are protected, or treating a pigment finding as a skin examination. Do not select vitamin or antioxidant products merely because the report includes a skin section. A useful decision follows the exposure or concern you actually have.

## Questions readers commonly have

**Does this explain my exact skin colour?** The selected observations are not a complete pigmentation model. **Should I seek an examination because a gene is listed?** The relevant reason is your clinical history, actual concern and appropriate advice. **Can I keep my current routine?** Yes, if it is appropriate; the aim is a usable review of protection, not change for its own sake.

> **The useful outcome**

> You understand the pigment biology and can name one practical protection question for your routine. Keep physical skin concerns separate from the genetic explanation.

Research and guidance: [7] Sun protection (CDC). These explain biology or guidance; they do not verify a private genotype.

## Selected markers and their interpretation

### HERC2 rs12913832

#### GG · Sequencing

HERC2 — two blue-eye alleles: with this genotype eyes are most often blue, and brown is rare. Other genes also shape eye colour.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 15:28120472. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Eiberg 2008 Hum Genet; Sturm 2008 Am J Hum Genet. Research attribution is separate from the private observation.

### MC1R rs1805007

#### CC · Sequencing

MC1R — no 151C allele at this MC1R red-hair variant (read with another MC1R marker).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 16:89919709. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Valverde 1995 Nat Genet (PMID 7581459); Box 1997 Hum Mol Genet (PMID 9302268). Research attribution is separate from the private observation.

### MC1R rs1805008

#### CC · Sequencing

MC1R — no 160W allele at this MC1R red-hair variant (read with another MC1R marker).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 16:89919736. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Valverde 1995 Nat Genet (PMID 7581459); Box 1997 Hum Mol Genet (PMID 9302268). Research attribution is separate from the private observation.

### SLC45A2 rs16891982

#### GG · Sequencing

SLC45A2 — no 374Leu allele at this site, which does not measure how your skin responds to the sun. Other genes also shape pigmentation.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

C and G are each other's complement, so a file written on the opposite strand could be read the wrong way round. One of several pigmentation genes; it does not give a skin type.

Observation: GRCh38 · 5:33951588. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Graf 2005 Hum Mutat (PMID 15714523); Soejima 2007 Int J Legal Med. Research attribution is separate from the private observation.

## FLG rs61816761

### GG · Sequencing

FLG — no copies here, but this report reads only a few FLG variants, so that does not rule out others.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

FLG has several loss-of-function alleles. R501X (rs61816761) is a nonsense SNP this report reads, but the other common European one, 2282del4, is a 4bp deletion (indel) that is not read from this file — so a negative R501X here does not rule out FLG barrier loss (its absence via the indel is unknown, never a negative).

Observation: GRCh38 · 1:152313385. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Palmer 2006 Nat Genet (PMID 16550169); Smith 2006 Nat Genet (PMID 16444271). Research attribution is separate from the private observation.

## MMP1 rs1799750

### Not read

Why it was not read: an insertion, deletion or copy-number change — not read here as a single site.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# What can I learn without putting myself in a sport category?

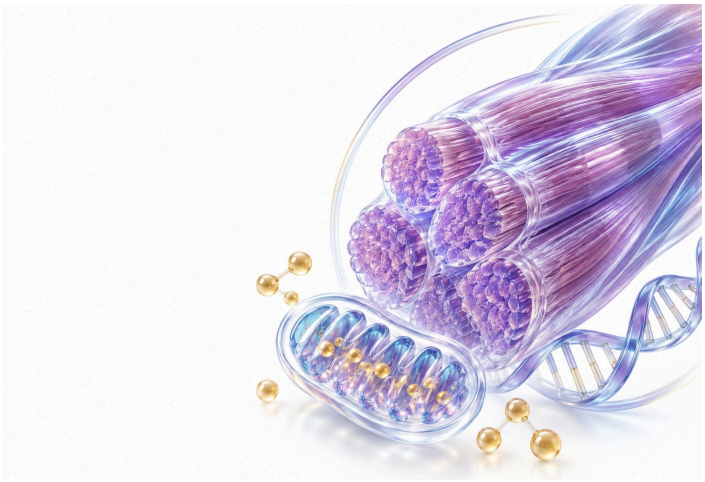
> 5 of 6 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

5 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your goal, actual activity, recovery, symptoms and any sourced fitness estimate.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| ACTN3 · rs1815739 | CC | Sequencing |
| PPARGC1A · rs8192678 | CT | Sequencing |
| COL1A1 · rs1800012 | CC | Sequencing |

The first 3 of 5 markers read in this chapter. Each is explained below.

## Muscle biology is interesting. Your response is observed.

ACTN3 contributes to a protein found in fast muscle fibres. ACE participates in signalling relevant to blood pressure and other functions. [29] Studies have compared these and other variants in groups of athletes. A group association does not identify your ideal sport, assign a power or endurance identity, or select a training programme. Technique, previous training, opportunity, motivation, sleep and your present health all matter.

The PPARGC1A Gly482Ser marker has been studied in endurance athletes and in exercise interventions. The early elite-athlete comparison was not a measurement of an individual training response. Later findings depend on population, outcome and programme. This edition does not predict your gains from the marker or assign you a lower potential.

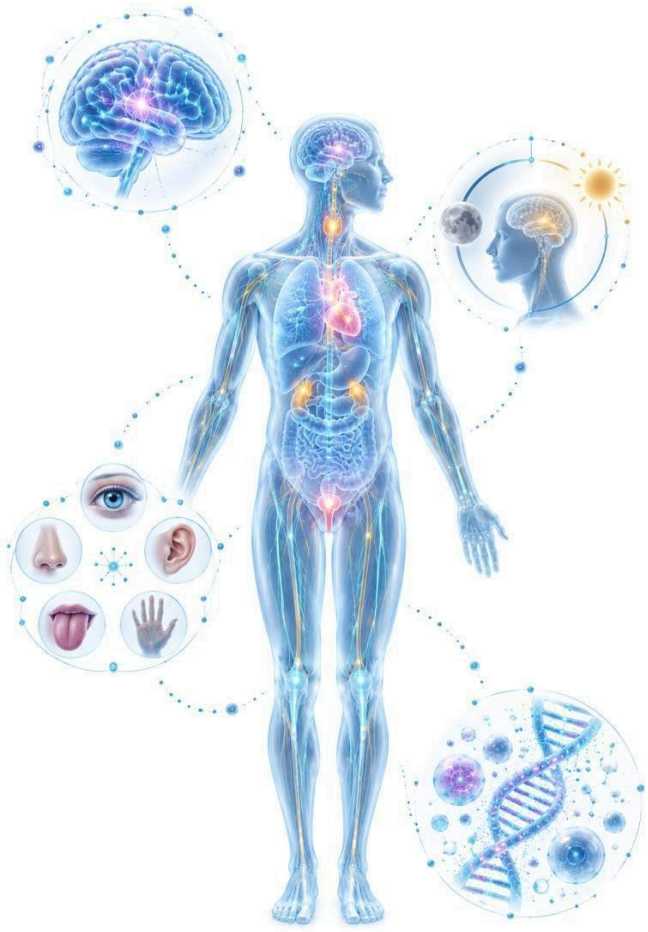
## A selected AMPD1 observation has a narrow scope

AMPD1 encodes an enzyme involved in muscle energy metabolism. The selected Q12X site is one variant in that gene. A result without that allele does not establish normal enzyme activity or exclude

every other AMPD1 variant. A single copy is usually not associated with symptoms of the recessive deficiency; even two copies require appropriate clinical context. It does not explain an exercise symptom by itself. [25]

## Tendons respond to load and recovery

COL5A1 has been studied in relation to particular tendon and ligament outcomes. The cited Australian and South African case-control study concerned chronic Achilles tendinopathy. It does not support a general personal injury forecast. If a readable observation is supplied, keep it separate from a general injury ranking. A tendon symptom should be understood through its location, duration, load history and an appropriate assessment.



**Start with a goal you can observe**
Keep your actual activity and recovery with the question. A common marker does not select an ideal sport.

## Start with the activity you want to sustain

Choose a goal that makes sense in your life: comfortable walking, maintaining strength, enjoying a sport, or returning to a previous routine. Keep an appropriate plan that already works. A genotype does not establish a reason to replace it. If you are building a routine, record what you actually do and whether it fits, rather than trying to prove a gene label.

A useful training record includes the activity, date, duration or load when known, how it felt and any relevant symptom. Missing days remain missing; they are not recorded as inactivity. Progress can mean better consistency, easier daily tasks or learning which activity you enjoy. You do not need to buy a device to make those observations.

## When a fitness estimate is available

A device estimate of aerobic fitness should carry its source, date, unit and method. Compare like with like when possible. A single estimate is not a diagnosis, and values from different devices or conditions may not be comparable. This report does not turn a gene into a measured VO2 value. A laboratory exercise test is a different kind of evidence from an estimate on a watch.

## Comfortable activity and symptoms ask different questions

If you feel well and an appropriate routine works, no change is a valid outcome. If symptoms repeatedly limit activity, describe the symptoms and seek suitable advice. Do not use a reassuring genotype to dismiss them or a candidate association to explain them. The question becomes what is happening during activity, not which genetic category you belong to.

Research and guidance: [17] AMPD1 function and scope (MedlinePlus Genetics); [40] PPARGC1A elite-athlete comparison (Lucia et al. 2005); [41] COL5A1 and Achilles tendinopathy (September et al. 2009). These explain biology or guidance; they do not verify a private genotype.

# Selected markers and their interpretation

## ACTN3 rs1815739

### CC · Sequencing

ACTN3 — both copies make this fast-muscle-fibre protein. It does not pick a sport or predict your training response.

**Live:** Strength and aerobic training both pay off whatever your ACTN3 result.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 11:66560624. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Yang 2003 Am J Hum Genet; MacArthur 2007 Nat Genet. Research attribution is separate from the private observation.

## PPARGC1A rs8192678

### CT · Sequencing

PPARGC1A — one Ser allele. Studied in athlete status and some training adaptations; it does not forecast your aerobic gains.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 4:23814039. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Lucia 2005 J Appl Physiol (PMID 15705733; athlete status); Steinbacher 2015 PLoS One (PMID 25886402; training adaptations); Petr 2018 Int J Mol Sci (review). Research attribution is separate from the private observation.

## COL1A1 rs1800012

### CC · Sequencing

COL1A1 — No Sp1 allele at this site. Its injury studies are inconsistent; it does not establish your injury risk.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 17:50200388. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Collins 2009 Br J Sports Med (review); Posthumus 2009 Am J Sports Med. Research attribution is separate from the private observation.

## COL5A1 rs12722

### CT · Sequencing

COL5A1 — one C allele. Its tendon and ligament studies disagree; it does not set your flexibility or injury risk.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 9:134842570. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: September 2009 Br J Sports Med (PMID 18443036); Miyamoto-Mikami 2019 BMC Med Genet (PMID 31791263); Collins 2009 Br J Sports Med (review). Research attribution is separate from the private observation.

## AMPD1 rs17602729

### GG · Sequencing

AMPD1 — not seen at this site, which does not rule out other variants or measure enzyme activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 1:114693436. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Rico-Sanz 2003 Physiol Genomics; Ginevičienė 2014 BMC Genet. Research attribution is separate from the private observation.

## ACE rs4646994

### Not read

Why it was not read: an insertion, deletion or copy-number change — not read here as a single site.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# Does a pathway finding explain how I feel?

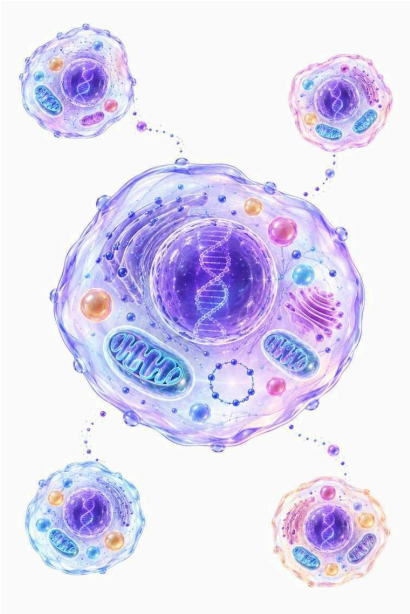
> 10 of 11 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

10 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| COMT · rs4680 | AA | Sequencing |
| MAOA · rs6323 | T (single X copy) | Sequencing |
| GSTP1 · rs1695 | AA | Sequencing |

The first 3 of 10 markers read in this chapter. Each is explained below.

## Clearance, histamine and the temptation of a single explanation

The liver helps process substances in the body. [35] Different reactions involve different enzymes and pathways. A common variant can be relevant to one reaction without describing a whole-body detox capacity. Present-or-deleted genes and complex structural changes require appropriate assays and analysis; a quiet selected-marker report is not a clean bill of health.

Histamine has roles in immune signalling, the gut and other tissues. Similar symptoms can have different causes. A gene label does not diagnose histamine intolerance or identify an elimination diet. Keep symptoms, food exposure, medicines and timing separate from an inherited observation. Avoid removing several food groups or starting a supplement programme to repair a pathway illustration.

If you have no current concern, the chapter can remain educational. If you have a recurring problem, describe its pattern and the question you want answered. What happens, when does it happen, and what else changed? That record is often more informative than collecting more candidate markers. A useful next step may be clarification or appropriate existing care.

# Selected markers and their interpretation

## COMT rs4680

### AA · Sequencing

COMT — biological context only. This call does not predict your personality, stress response or methyl-group requirement.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This report does not use COMT to select nutrients, supplements or caffeine advice.

Observation: GRCh38 · 22:19963748. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Chen 2004 Am J Hum Genet (PMID 15457404). Research attribution is separate from the private observation.

## MAOA rs6323

### T (single X copy) · Sequencing

MAOA — the T allele at this tag, which does not measure your MAO-A activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

MAOA is on chromosome X: males carry one copy, so one allele is the whole read, and a call needs the sex the analysis was run with. Not a measure of MAO-A activity, and never a behavioural verdict.

Observation: GRCh38 · X:43731789. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Hotamisligil 1991 Am J Hum Genet (PMID 1678250); Verma 2014 (PMID 24291416); Wang 2018 (PMID 30018578); dbSNP rs6323 (ncbi.nlm.nih.gov/snp/rs6323). Research attribution is separate from the private observation.

## GSTP1 rs1695

### AA · Sequencing

GSTP1 — no 105Val allele at this site, which does not measure your detoxification.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 11:67585218. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Ali-Osman 1997 J Biol Chem (PMID 9092542); GST review Hayes 2005 Annu Rev Pharmacol Toxicol. Research attribution is separate from the private observation.

## NOS3 rs1799983

### TG · Sequencing

NOS3 — one 298Asp allele, which does not measure your nitric oxide. Cell studies disagree on whether it changes the enzyme's output.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:150999023. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Casas 2004 Circulation (PMID 15007011); Casas 2006 Am J Epidemiol (PMID 17018701); Hingorani 1999 Circulation (PMID 10510054); Joshi 2007 FASEB J (PMID 17449720); McDonald 2004 Pharmacogenetics (PMID 15608562); Fairchild 2001 J Biol Chem (PMID 11331296). Research attribution is separate from the private observation.

## PEMT rs7946

### TT · Sequencing

PEMT — context only. This call does not measure liver phosphatidylcholine production, or establish extra dietary choline need.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

The rs7946 marker was not associated with depletion susceptibility in da Costa 2006; findings for another PEMT site cannot be transferred here.

Observation: GRCh38 · 17:17506246. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Song 2005 FASEB J (PMID 16051693); da Costa 2006 FASEB J (PMID 16816108); Ganz 2017 Int J Mol Sci (PMID 28134761). Research attribution is separate from the private observation.

## AOC1 rs10156191

### CT · Sequencing

AOC1 — one 16Met allele, which does not measure your DAO activity. In one study it went with lower DAO activity in blood, not with histamine intolerance itself.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:150856517. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Maintz 2011 Allergy (PMID 21488903); Ayuso 2007 Pharmacogenet Genomics (PMID 17700358). Research attribution is separate from the private observation.

## AOC1 rs1049793

### CC · Sequencing

AOC1 — no 645Asp allele at this site, which does not measure your DAO activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

C and G are each other's complement, so a file written on the opposite strand could be read the wrong way round.

Observation: GRCh38 · 7:150860577. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Ayuso 2007 Pharmacogenet Genomics (PMID 17700358). Research attribution is separate from the private observation.

## SOD2 rs4880

### AA · Sequencing

SOD2 — two Val16 alleles, which does not measure your antioxidant defence. In a laboratory assay the Val form entered mitochondria less well; people were not measured.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 6:159692840. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Sutton 2003 Pharmacogenetics (PMID 12618592); Bresciani 2015 Adv Clin Chem (review). Research attribution is separate from the private observation.

## GPX1 rs1050450

### GA · Sequencing

GPX1 — one 198Leu allele, which does not measure your antioxidant defence. One study found the enzyme 5% less active per copy; another found no difference.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

The two cited studies disagree on whether the enzyme is less active. One enzyme's activity is not a measure of antioxidant defence.

Observation: GRCh38 · 3:49357401. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Ravn-Haren 2006 Carcinogenesis (PMID 16287877); Jablonska 2009 Eur J Nutr (PMID 19415410). Research attribution is separate from the private observation.

## NQO1 rs1800566

### GG · Sequencing

NQO1 — no 187Ser allele at this site, which does not measure NQO1 activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 16:69711242. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Traver 1997 Br J Cancer (PMID 9000600); Siegel 1999 Pharmacogenetics (PMID 10208650); Ross 2000 Chem Biol Interact (NQO1 review). Research attribution is separate from the private observation.

## GSTM1 GSTM1 null (whole-gene deletion)

### Not read

Why it was not read: an insertion, deletion or copy-number change — not read here as a single site.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# What is the difference between a clue and current inflammation?

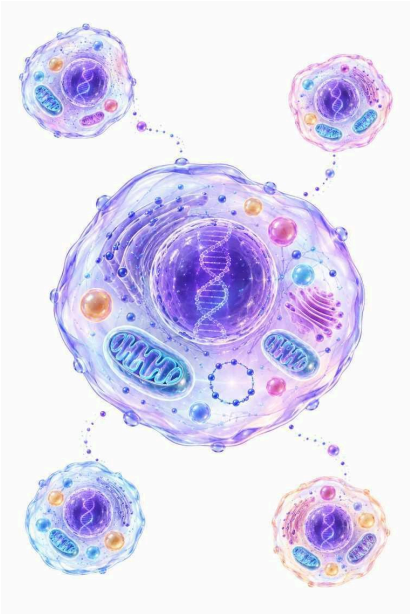
> 5 of 8 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

5 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| PTPN22 · rs2476601 | AG | Sequencing |
| CTLA4 · rs3087243 | GA | Sequencing |
| HLA-DQA1 · rs2187668 | CC | Sequencing |

The first 3 of 5 markers read in this chapter. Each is explained below.

## A response system, not an inflammation score

The immune system uses signals to recognise threats, recruit cells and end a response. [31] Common variants have been studied in relation to immune traits and disease susceptibility. The selected observations here do not measure your present inflammation, immune competence or likelihood of every immune condition.

A dated blood measurement, an infection history, a symptom pattern and a genetic association are different evidence. A value should retain its test name, unit, date and source. A marker does not turn a missing measurement into an abnormal one. More sequencing does not automatically explain fatigue or recurrent symptoms.

If you feel well, this chapter does not establish a need for an immune test bundle or an anti-inflammatory supplement. If an ongoing symptom or an existing care question matters, use the report to

organise the relevant history and records. Keep an appropriate care plan rather than treating each candidate association as a new task.

> No relevant dated laboratory measurement is supplied in this chapter. This describes the report inputs; it does not mean a test was never performed.

## Selected markers and their interpretation

### PTPN22 rs2476601

#### AG · Sequencing

PTPN22 — one 620W allele; a modestly raised tendency to type 1 diabetes and rheumatoid arthritis (not a diagnosis).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 1:113834946. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Bottini 2004 Nat Genet (PMID 15004560); Begovich 2004 Am J Hum Genet (PMID 15208781); Vang 2005 Nat Genet (PMID 16273109). Research attribution is separate from the private observation.

### CTLA4 rs3087243

#### GA · Sequencing

CTLA4 — one G allele, the more common allele; a small autoimmune-risk tendency in cohorts, not a diagnosis. It went with slightly more autoimmune thyroid disease and type 1 diabetes.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 2:203874196. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Ueda 2003 Nature (PMID 12724780); for the gene's other sites, Kavvoura 2005 Am J Epidemiol (meta-analysis, PMID 15961581). Research attribution is separate from the private observation.

### HLA-DQA1 rs2187668

#### CC · Sequencing

HLA-DQA1 — no DQ2.5 tag allele, but that does not exclude the risk type, and one tag SNP cannot show your HLA-DQ type; HLA-DQ2.5 is the main inherited risk type for celiac disease. Only a dedicated HLA-DQ test, not this marker, can show your HLA-DQ type.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This SNP only TAGS the HLA alleles, and the HLA genes themselves are not typed from this file — treat any HLA-DQ2.5 call here as LOW-CONFIDENCE / effectively a blind spot. A dedicated HLA-DQ typing test is needed to confirm; a negative tag does NOT exclude the risk haplotype (its absence is unknown, never a clinical negative).

Observation: GRCh38 · 6:32638107. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Monsuur 2008 PLoS One (rs2187668 as DQ2.5 tag; PMID 18509540); Romanos 2009 Gastroenterology; Erlich 2008 Diabetes (PMID 18252895). Research attribution is separate from the private observation.

## IL6 rs1800795

### CG · Sequencing

IL6 — one -174C allele, which does not measure your inflammation. The first study found lower IL-6 in blood with C, and 17 studies combined found no link.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

Studies disagree on whether this site changes IL-6 in blood; a measured hs-CRP or IL-6 says more. C and G are each other's complement, so a file written on the opposite strand could be read the wrong way round.

Observation: GRCh38 · 7:22727026. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Fishman 1998 J Clin Invest (PMID 9769329); Huth 2009 Ann Med (pooled analysis of 17 studies, PMID 18752089). Research attribution is separate from the private observation.

## TNF rs1800629

### GG · Sequencing

TNF — no -308A allele at this site, which does not measure your inflammatory tone.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 6:31575254. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Wilson 1997 PNAS; Bayley 2004 Genes Immun (review); Mekinian 2011 (PMID 21768061). Research attribution is separate from the private observation.

## HLA-DQB1 rs7454108

### Not read

Why it was not read: no call in this sequencing file passed the quality check.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

## HLA-DQB1 rs7775228

### Not read

Why it was not read: no call in this sequencing file passed the quality check.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

## HLA-B rs116488202

### Not read

Why it was not read: no call in this sequencing file passed the quality check.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# Can a common variant tell me my hormone levels?

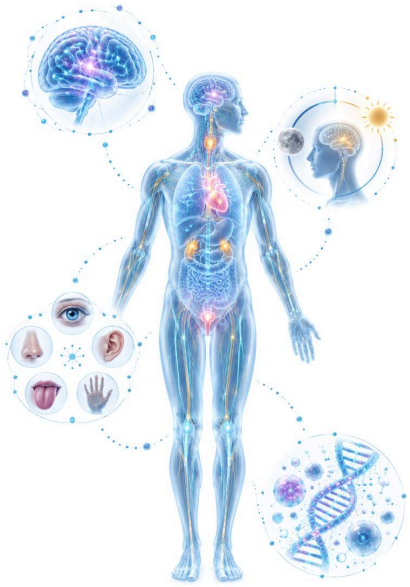
> 6 of 7 selected markers read.

OBSERVATION SCOPE, IN CONTEXT

6 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| SRD5A2 · rs523349 | GC | Sequencing |
| CYP19A1 · rs10046 | GG | Sequencing |
| CYP19A1 · rs700518 | TT | Sequencing |

The first 3 of 6 markers read in this chapter. Each is explained below.

## Messages that depend on timing and context

Hormones carry signals between tissues. Their production, transport, receptors and clearance involve many biological steps. [32] A common marker in one component does not measure your hormone levels or establish a hormonal imbalance. A pathway drawing is a map of the biology, not a picture of your current activity.

Symptoms, life stage, menstrual or treatment context where relevant, collection timing and laboratory methods can change how a measurement is interpreted. Do not infer those details from a genetic profile or from another person’s record. An available measurement should keep its original date, unit, method and clinical context.

The practical route begins with your own question. If no concern is present, no change is appropriate. If you already have care or a dated result, locate it

and record what remains unresolved. This edition does not choose a hormone product, dose or testing schedule from a common variant.

## Selected markers and their interpretation

### SRD5A2 rs523349

#### GC · Sequencing

SRD5A2 — one 89L allele; a modestly lower 5α-reductase activity (small effect).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

C/G is palindromic, so a file written on the opposite strand could be read the wrong way round. The effect on hormone levels in blood is small; a measured testosterone or DHT says more.

Observation: GRCh38 · 2:31580636. Selected site only.
Interpretation: single-marker reading; association evidence: probable.
Association source: Makridakis 1997 Cancer Res (PMID 9067262). Research attribution is separate from the private observation.

### CYP19A1 rs10046

#### GG · Sequencing

CYP19A1 — no A allele at this tag, which does not measure your aromatase activity or your estradiol.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 15:51210789. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Dunning 2004 J Natl Cancer Inst (PMID 15199113); Haiman 2007 Cancer Res (PMID 17325027). Research attribution is separate from the private observation.

### CYP19A1 rs700518

#### TT · Sequencing

CYP19A1 — no C allele at this tag, which does not measure your aromatase activity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 15:51236915. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Ma 2005 Cancer Res (PMID 16322257); Haiman 2007 Cancer Res (PMID 17325027). Research attribution is separate from the private observation.

### ESR1 rs2234693

#### TT · Sequencing

ESR1 — no 'P' allele at this tag, which does not measure your receptor sensitivity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

Read the ESR1 PvuII (rs2234693) and XbaI (rs9340799) tags as a haplotype pair, not singly — the cohort findings are small and inconsistent, and say nothing about one person's receptor sensitivity.

Observation: GRCh38 · 6:151842200. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Herrington 2002 N Engl J Med; Schuit 2004 JAMA (ESR1 haplotype); Kjaergaard 2007 (PMID 17309937). Research attribution is separate from the private observation.

## ESR1 rs9340799

### AA · Sequencing

ESR1 — no 'X' allele at this tag, which does not measure your receptor sensitivity.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

Read the ESR1 PvuII (rs2234693) and XbaI (rs9340799) tags as a haplotype pair, not singly — the cohort findings are small and inconsistent, and say nothing about one person's receptor sensitivity.

Observation: GRCh38 · 6:151842246. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Herrington 2002 N Engl J Med; Schuit 2004 JAMA (ESR1 haplotype); Kjaergaard 2007 (PMID 17309937). Research attribution is separate from the private observation.

## SHBG rs6259

### GG · Sequencing

SHBG — no 356N allele at this site, which does not measure your SHBG.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 17:7633209. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Cousin 2004 J Clin Endocrinol Metab (PMID 14764814); Dunning 2004 J Natl Cancer Inst (PMID 15199113); Coviello 2012 PLoS Genet (PMID 22829776); Prescott 2012 PLoS One (doi 10.1371/journal.pone.0037815). Research attribution is separate from the private observation.

## AR AR CAG repeat (polyQ length)

### Not read

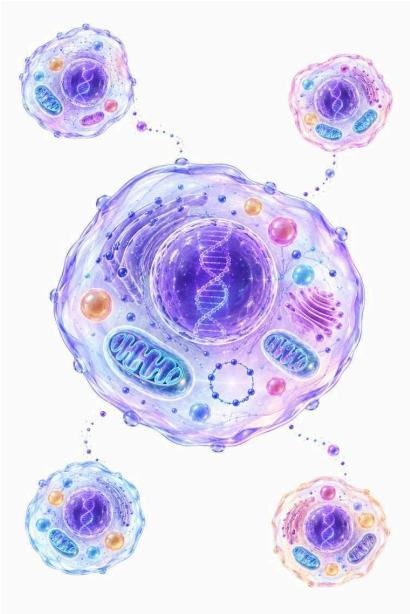
Why it was not read: a repeat length, not a single site — needs a dedicated assay.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# What can a longevity association actually tell me?

> 4 of 4 selected markers read.



OBSERVATION SCOPE, IN CONTEXT

4 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.

| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| APOE · rs429358 | TT | Sequencing |
| FOXO3 · rs2802292 | GT | Sequencing |
| KLOTHO · rs9536314 | TT | Sequencing |

The first 3 of 4 markers read in this chapter. Each is explained below.

## An association is not a lifespan forecast

Researchers compare genetic patterns in people who live to advanced ages and in other groups. Some common variants appear more often in one group. Survival, environment, ancestry and how the study selected participants all affect interpretation. This edition does not calculate how long you will live or assign a biological age. [33]

A longevity-associated allele is not a guarantee, and a different allele is not a sentence. Where a readable observation is supplied, the letters are useful as an explanation of research, not as a reason to buy a rejuvenation programme. Nor does a common result establish that disease-related questions have been assessed.

Use this chapter to separate curiosity from decisions. Ordinary goals such as maintaining comfortable activity, sleep and appropriate preventive care can stand on their own reasons. A candidate longevity marker does not select a supplement, intervention or personal schedule.

# Selected markers and their interpretation

## APOE rs429358

### TT · Sequencing

APOE — no ε4-defining C allele at this SNP (isoform needs another APOE marker too to call fully).

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

This row reads rs429358 alone; the ε2/ε3/ε4 isoform also depends on rs7412.

Observation: GRCh38 · 19:44908684. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Deelen 2019 Nat Commun; Corder 1993 Science. Research attribution is separate from the private observation.

## FOXO3 rs2802292

### GT · Sequencing

FOXO3 — one longevity-associated allele: more common among long-lived people in the cited cohorts; an odds nudge, not a guarantee.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 6:108587315. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Willcox 2008 PNAS (PMID 18765803); Flachsbart 2009 PNAS (PMID 19196970). Research attribution is separate from the private observation.

## KLOTHO rs9536314

### TT · Sequencing

KLOTHO — non-carrier (no KL-VS tag allele) at this marker.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

The early reports describe a benefit with one copy and none with two, so 'more is better' does not apply; later cohorts found neither. This site tags the KL-VS haplotype and does not show its second change (C370S) or phase.

Observation: GRCh38 · 13:33054001. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Arking 2002 PNAS (PMID 11792841); Dubal 2014 Cell Rep (PMID 24813892); Wu 2020 Sci Rep (PMID 32581247); Amin 2022 J Gerontol A Biol Sci Med Sci (PMID 34893828). Research attribution is separate from the private observation.

## SERPINA1 rs28929474

### CC · Sequencing

SERPINA1 — no Z variant at this site, which does not rule out the gene's rarer variants.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

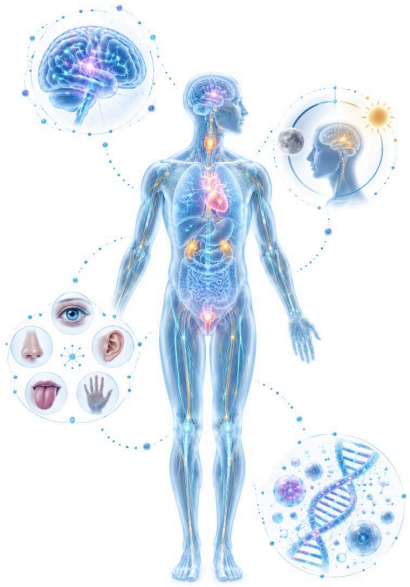
▼ Detailed source limitation

One site (Z) of one gene: it cannot see the gene's other deficiency variants, S among them. A blood test of the alpha-1 antitrypsin level, with its type, confirms it.

Observation: GRCh38 · 14:94378610. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: ATS/ERS statement on alpha-1 antitrypsin deficiency 2003 (PMID 14522813); Strnad 2020 NEJM (PMID 32268028). Research attribution is separate from the private observation.

# Could biology explain an everyday preference?

5 of 7 selected markers read.



OBSERVATION SCOPE, IN CONTEXT

5 selected observations are available for this topic. Read each interpretation beside its own marker and method.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.

| SELECTED FINDING | LETTERS | OBSERVED HOW |
|---|---|---|
| TAS2R38 · rs713598 | CC | Sequencing |
| TAS2R38 · rs1726866 | AA | Sequencing |
| TAS2R38 · rs10246939 | TT | Sequencing |

The first 3 of 5 markers read in this chapter. Each is explained below.

## Small inherited differences in everyday experience

Taste and smell involve receptors and many downstream processes. [34] Some selected variants help explain why people perceive certain bitter compounds or odours differently. A result concerns the particular trait studied; it is not a ranking of the quality of your senses.

Your actual preferences remain useful information. Food experience depends on learning, culture, preparation and context as well as biology. A bitter-taste association does not establish that you dislike every vegetable or need a particular diet. You can use preparation and variety to explore foods you enjoy without trying to fit a genetic category.

New or persistent changes in taste, smell, hearing or vision ask a different question from an inherited preference. A common result does not explain a new symptom or rule out a condition. Keep the timing and circumstances of the change distinct from the genetic observation.

# Selected markers and their interpretation

## TAS2R38 rs713598

### CC · Sequencing

TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:141973545. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Kim 2003 Science (PMID 12595690); Bufe 2005 Curr Biol (PMID 15723792; rarer AAI/AAV/PVI haplotypes ~40% of PAV response); Mennella 2010 BMC Genet (PMID 20594349; haplotype frequencies). Research attribution is separate from the private observation.

## TAS2R38 rs1726866

### AA · Sequencing

TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:141972905. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Kim 2003 Science (PMID 12595690); Bufe 2005 Curr Biol (PMID 15723792). Research attribution is separate from the private observation.

## TAS2R38 rs10246939

### TT · Sequencing

TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

Observation: GRCh38 · 7:141972804. Selected site only.
Interpretation: single-marker reading; association evidence: established.
Association source: Kim 2003 Science (PMID 12595690); Bufe 2005 Curr Biol (PMID 15723792). Research attribution is separate from the private observation.

## OPRM1 rs1799971

### AG · Sequencing

OPRM1 — one 118G allele, with an effect too small for guidelines to act on. Surgery studies saw slightly higher opioid needs in carriers.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

About opioid medicines, not how you feel pain; guidelines judge the effect too small to act on.

Observation: GRCh38 · 6:154039662. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Walter & Lötsch 2009 Pain (PMID 19683391, meta-analysis); Walter 2013 Pharmacogenomics (PMID 24236490); Hwang 2014 Anesthesiology (PMID 25102313); CPIC opioid guideline, Crews 2021 (PMID 33387367; no OPRM1 recommendation); Bond 1998 PNAS (PMID 9689128; receptor binding in cells). Research attribution is separate from the private observation.

## SCN9A rs6746030

### GG · Sequencing

SCN9A — the common genotype; no 1150W allele.

Scope: One supplied sequence observation; this finding does not assess every variant in the gene.

▼ Detailed source limitation

Small studies with mixed replication; no genome-wide study has found it.

Observation: GRCh38 · 2:166242648. Selected site only.
Interpretation: single-marker reading; association evidence: emerging.
Association source: Reimann 2010 PNAS (PMID 20212137); Valdes 2011 Arthritis Care Res (PMID 21031562); no association: Holliday 2012 Mol Pain (PMID 23006801). Research attribution is separate from the private observation.

## CYP2D6 CYP2D6 activity (star alleles + copy number)

### Not read

Why it was not read: an insertion, deletion or copy-number change — not read here as a single site.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

## OR6A2 rs72921001

### Not read

Why it was not read: no call in this sequencing file passed the quality check.

Scope: No usable call is supplied for this selected site; absence is unresolved.

Observation: no call recorded for this site. Selected site only.
Interpretation: none, the site was not read.
Association source: none, the site was not read. Research attribution is separate from the private observation.

# What is available for a prescribing conversation?

> 17 of 22 genes read · the sequencing gVCF

OBSERVATION SCOPE, IN CONTEXT

17 of 22 genes read. Results other than “normal”:

Warfarin — VKORC1: you may need a lower dose of warfarin, and of acenocoumarol or phenprocoumon; if you take or are prescribed one, talk to your doctor or pharmacist before changing any medicine (rs9923231 T/T (no single phenotype label)). CYP4F2: part of how a warfarin dose is worked out, together with VKORC1 and CYP2C9; talk to your doctor or pharmacist before changing any medicine (*4/*5 (no single phenotype label)).

Hydralazine and isoniazid — NAT2: you may need a higher hydralazine dose; talk to your doctor or pharmacist before changing any medicine (intermediate metabolizer).

CYP3A5: tacrolimus is dosed for this result by default; talk to your doctor or pharmacist before changing any medicine (poor metabolizer).

Not read: CYP2D6, HLA-A, HLA-B, MT-RNR1, SLCO1B1. A gene we could not read is unknown, not normal.

Preliminary, not a diagnosis. Don't start, stop or change a medicine because of this result. Talk to your doctor or pharmacist; a clinical test must confirm it before any medical decision.



**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.

# How your genes may affect some medicines

These results are read from your DNA file using published pharmacogenomic guidelines. They are preliminary: a clinical pharmacogenetic test must confirm a result before it is used for any medical decision. They are not a diagnosis, they do not tell you whether to take a medicine or how much, and they are no substitute for seeing your doctor.

**Each gene below can affect how your body handles some medicines.** Preliminary, not a diagnosis. Don't start, stop or change a medicine because of this result. Talk to your doctor or pharmacist; a clinical test must confirm it before any medical decision.

| GENE | RESULT | DIPLOTYPE |
|---|---|---|
| ABCG2 | Normal Function | rs2231142 G/G |
| CACNA1S | Uncertain Susceptibility | Reference/Reference |
| CYP2B6 | Normal Metabolizer | *2/*5 |
| CYP2C19 | Normal Metabolizer | *1/*1 |
| CYP2C9 | Normal Metabolizer | *1/*1 |
| CYP3A4 | Normal Metabolizer | *1/*1 |
| CYP3A5 | Poor Metabolizer | *3/*3 |
| CYP4F2 | *4/*5 (no single phenotype label) | |
| DPYD | Normal Metabolizer | Reference/Reference |
| G6PD | Normal | B (reference) |
| IFNL3 | rs12979860 C/C (no single phenotype label) | |
| NAT2 | Intermediate Metabolizer | *4/*6 |
| NUDT15 | Normal Metabolizer | *1/*1 |
| RYR1 | Uncertain Susceptibility | Reference/Reference |
| TPMT | Normal Metabolizer | *1/*1 |
| UGT1A1 | Normal Metabolizer | *1/*1 |
| VKORC1 | rs9923231 T/T (no single phenotype label) | |

**ABCG2.** Medicines it concerns: rosuvastatin. This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**CACNA1S.** Medicines it concerns: volatile anaesthetics (desflurane, isoflurane, sevoflurane) and succinylcholine. An uncertain result does not rule out malignant hyperthermia: tell an anaesthetist

about any family history of a bad reaction to anaesthesia; talk to your doctor or pharmacist before changing any medicine.

**CYP2B6.** Medicines it concerns: efavirenz (an HIV medicine) and sertraline. This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**CYP2C19.** Medicines it concerns: clopidogrel; the antidepressants citalopram, escitalopram, sertraline and amitriptyline; the stomach-acid medicines omeprazole, lansoprazole and pantoprazole; voriconazole. This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**CYP2C9.** Medicines it concerns: warfarin, phenytoin, fluvastatin, and the painkillers celecoxib, ibuprofen, meloxicam and piroxicam. This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**CYP3A5.** Medicines it concerns: tacrolimus (a transplant medicine). Tacrolimus is dosed for this result by default; talk to your doctor or pharmacist before changing any medicine.

**CYP4F2.** Medicines it concerns: warfarin. Part of how a warfarin dose is worked out, together with VKORC1 and CYP2C9; talk to your doctor or pharmacist before changing any medicine.

**DPYD.** Medicines it concerns: fluorouracil (5-FU) and capecitabine, chemotherapy medicines. Four variants were read, and rarer ones were not: this result does not replace the DPYD test before treatment with fluorouracil or capecitabine; talk to your doctor or pharmacist before changing any medicine.

**G6PD.** Medicines it concerns: rasburicase, pegloticase, primaquine, tafenoquine, dapsone and methylene blue; nitrofurantoin with caution. This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**IFNL3.** Medicines it concerns: peginterferon alfa for hepatitis C, now rarely used.

**NAT2.** Medicines it concerns: hydralazine and isoniazid (a tuberculosis medicine). You may need a higher hydralazine dose; talk to your doctor or pharmacist before changing any medicine.

**NUDT15.** Medicines it concerns: azathioprine, mercaptopurine and thioguanine. This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**RYR1.** Medicines it concerns: volatile anaesthetics (desflurane, isoflurane, sevoflurane) and succinylcholine. An uncertain result does not rule out malignant hyperthermia: tell an anaesthetist about any family history of a bad reaction to anaesthesia; talk to your doctor or pharmacist before changing any medicine.

**TPMT.** Medicines it concerns: azathioprine, mercaptopurine and thioguanine. This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**UGT1A1.** Medicines it concerns: atazanavir (an HIV medicine) and irinotecan (a chemotherapy medicine). This result does not change how they are usually prescribed; talk to your doctor or pharmacist before changing any medicine.

**VKORC1.** Medicines it concerns: warfarin, acenocoumarol and phenprocoumon. You may need a lower dose of warfarin, and of acenocoumarol or phenprocoumon; if you take or are prescribed one, talk to your doctor or pharmacist before changing any medicine.

**Not read.** Not called from a sequencing file here: it needs its own caller: CYP2D6, HLA-A, HLA-B, MT-RNR1 · Held: a position that could change this result was not read: SLCO1B1

**CYP2D6.** Medicines it concerns: codeine and tramadol, tamoxifen, metoprolol, ondansetron, and many antidepressants and antipsychotics. If you take or are prescribed codeine, tramadol, tamoxifen

or an antidepressant, a clinical CYP2D6 test can answer what this file cannot; talk to your doctor or pharmacist before changing any medicine.

**HLA-A.** Medicines it concerns: carbamazepine. A clinical HLA test answers this before carbamazepine where guidelines ask for one; talk to your doctor or pharmacist before changing any medicine.

**HLA-B.** Medicines it concerns: abacavir, allopurinol, carbamazepine, oxcarbazepine and phenytoin. Before abacavir, allopurinol, carbamazepine, oxcarbazepine or phenytoin, a clinical HLA test answers this where guidelines ask for one; talk to your doctor or pharmacist before changing any medicine.

**MT-RNR1.** Medicines it concerns: aminoglycoside antibiotics such as gentamicin, tobramycin and amikacin. Before an aminoglycoside antibiotic, a clinical test answers what this file cannot; talk to your doctor or pharmacist before changing any medicine.

**SLCO1B1.** Medicines it concerns: statins, simvastatin most of all. Talk to your doctor or pharmacist before changing any medicine.

- Keep taking your medicines as prescribed. Never start, stop or change a medicine because of this report; talk to your doctor or pharmacist first.
- Some genes are not called from a sequencing file here: CYP2D6, HLA-A, HLA-B and MT-RNR1. A gene we could not read is unknown, not normal.
- A “normal” result means we found none of the variants we can read. It does not rule out rarer variants that change how you respond.
- Other medicines, age, and kidney or liver function also change how a medicine works.

## A conversation aid for existing care

Medicine response can involve enzymes, transporters, the medicine itself and many features of current care. Pharmacogenomic interpretation needs an appropriate, sufficiently complete assay and a validated gene-level analysis. A single selected variant is not automatically a diplotype, metaboliser category or prescribing instruction.

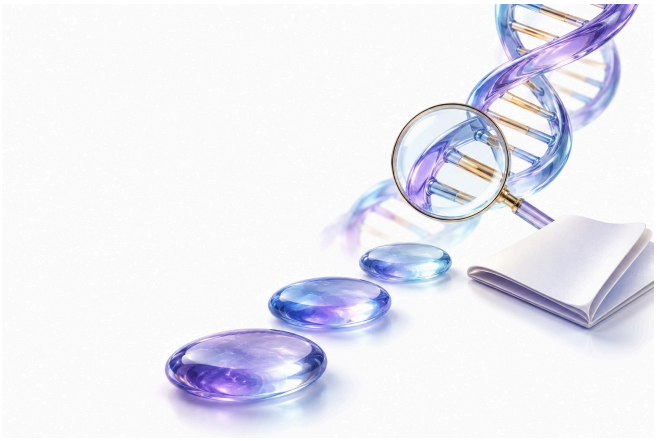
The current result status is shown at the opening of this chapter. If an earlier medicines analysis is out of date or did not finish, it is not shown, and nothing is put in its place. Unknown and normal are different answers.

If you take a medicine and have a relevant question, record the medicine and the question for the person who prescribes. Keep the original result, laboratory, date and method if a clinical test exists. Do not start, stop or change a medicine from this report. Complex genes such as CYP2D6 and HLA typing need analyses that a selected-marker file does not provide. Clinical confirmation and current prescribing guidance belong within care.

## What a metaboliser label means

Some medicines need conversion into an active substance; for others, the relevant question concerns clearance or transport. A slow or fast label cannot describe every relationship. Intermediate and poor metaboliser are predicted-function categories for a named gene. They do not describe the general health of your liver. A normal category within a particular assay does not guarantee effectiveness, no side effects or complete assessment of that gene.

FDA distinguishes relationships supported by therapeutic-management evidence from other safety/response and pharmacokinetic associations. An entry in its table does not automatically mean a genetic test is recommended before every prescription. The actual medicine, other treatments, health information and monitoring remain part of the decision.



**Keep the question with the evidence**
A useful prescribing conversation starts with the medicine, the reason for taking it and the current clinical record.

## Gene names, observations and assumptions

Star-allele names summarise an interpretation of genetic observations. A name alone does not show which defining positions were read, their quality, phase, copy-number assessment or the assay scope. Different candidate pairs can sometimes support the same functional category without resolving an exact pair. The technical interpretation and its support should travel together.

A position assumed to be reference because it is absent is different from an observed reference genotype. A software match does not make missing evidence measured. A variants-only sequencing file usually omits reference positions; its omissions do not establish normal function. Reference-supporting data and specialised analysis may be needed for the relevant gene and variant type.

## A medicine-specific question

A useful question is: is medicine-gene information relevant to this current or planned prescription, what existing evidence is suitable, and would it change a decision in my care? Record the evidence actually reviewed, the professional response, the reason for the outcome and any agreed review point. The outcome can be keeping an appropriate plan.

Research and guidance: [18] Different pharmacogenetic evidence scopes (FDA); [43] Missing, reference and assay limitations (PharmCAT FAQ). These explain biology or guidance; they do not verify a private genotype.

# Would a broader DNA record answer my question?

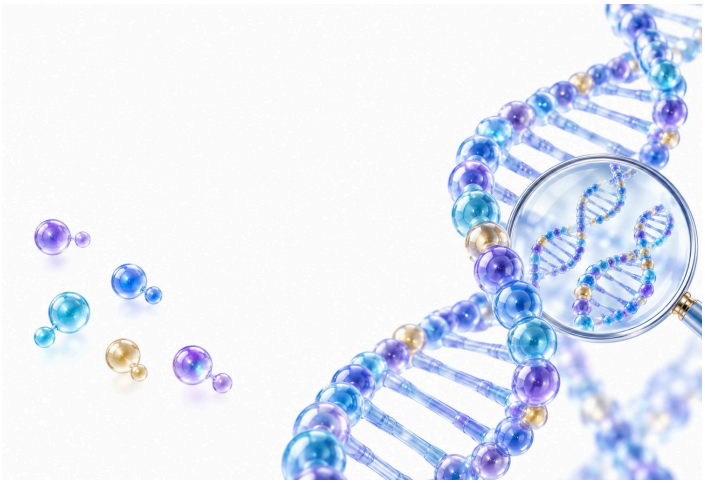
> A comparison of methods, not a claim about a future personal result.

OBSERVATION SCOPE, IN CONTEXT

Broader sequencing can add observations. It does not guarantee an answer to a particular health question.

**The useful next question**
The question you want answered, supported analysis, assay quality and interpretation scope.

Keeping an appropriate routine or making no change is a valid outcome.



A SNP array measures selected, known positions. Imputation statistically infers additional genotypes using observed data and haplotype patterns in a reference population. [26] Whole-genome sequencing reads DNA across a much broader span, including coding and non-coding regions. It can support finding variants beyond those chosen in advance. This describes the underlying record, before deciding what any observation means. [20, 21]

**If WGS already exists**
The next opportunity may be to use more of the existing sequence record and verify its quality. Buying another sequencing test is a different decision from improving the analysis of a file already available.

Research and guidance: [20] Exome and genome sequencing (MedlinePlus Genetics); [21] Human genomic variation and how it is detected (NHGRI). These explain biology or guidance; they do not verify a private genotype.

| YOUR QUESTION | WHAT WGS COULD CONTRIBUTE | WHAT MUST ACCOMPANY IT |
|---|---|---|
| A variant is unavailable or only inferred | A direct sequence observation may add evidence about that position. | Adequate reads, valid variant calling and the exact assay scope. |
| A clinical concern or family-history pattern | A broader search for rare variants or variants outside a selected panel. | Clinical interpretation tied to the question, appropriate confirmation and counselling. [22] |

| YOUR QUESTION | WHAT WGS COULD CONTRIBUTE | WHAT MUST ACCOMPANY IT |
|---|---|---|
| A specific medicine question | Broader data may support a validated analysis of a relevant gene. | An appropriately validated analysis, current medicine information and professional review or confirmation. The FDA association table does not by itself select a prescription. [18] |
| Larger DNA changes or difficult regions | Some analyses can examine insertions, deletions or structural variants. | The laboratory must state which variant types and regions are reliable. Routine sequencing does not resolve every region. [21, 22] |
| Future reinterpretation | A retained broader record can be revisited as evidence develops. | Usable files, reference information, quality records and an appropriate reanalysis process. [20, 22] |

## Methylation is a good example of the distinction

WGS may provide more inherited observations across the enzymes shown on the map. It would still not establish the current activity of the whole network, your folate or B12 status, or the cause of a symptom. Sequence data and a measurement of DNA methylation are also different kinds of information. The relevant question determines which measurement would add something useful. [12, 20]

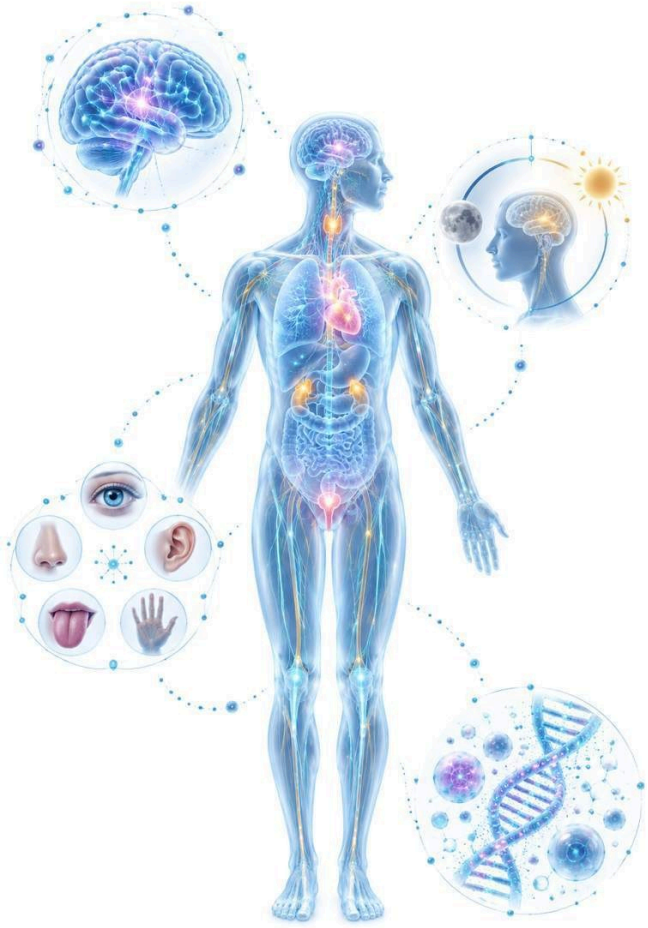
## More observations can also mean more uncertainty

Some additional variants have no established clinical significance. A variant of uncertain significance means the connection with a health condition is unresolved; it should remain described that way. A long list of rare changes is not, by itself, a more useful explanation. [24]

Research and guidance: [12] What epigenetics measures (MedlinePlus Genetics); [18] Pharmacogenetic associations in context (FDA); [20] Exome and genome sequencing (MedlinePlus Genetics); [21] Human genomic variation and how it is detected (NHGRI); [22] Interpreting genomic reports: limitations and follow-up (NHGRI / clinical education toolkit); [23] Genetic testing: uses and interpretation (NHGRI); [24] Variants of uncertain significance (NHGRI). These explain biology or guidance; they do not verify a private genotype.

| WHAT YOU WANT TO KNOW | A USEFUL NEXT STEP |
|---|---|
| How are common MTHFR markers interpreted? | Read the pathway and context. If a readable observation is available, its explanation does not automatically require more sequencing. |
| What is my current folate, B12 or blood-sugar status? | Relevant history and measurements address this question. WGS does not supply your current blood values. |
| Could a result affect a particular prescription? | Use the supported gene-drug guideline with a professional and confirm relevant preliminary results. |
| Could a clinical or family pattern have an inherited explanation? | Clinical genetics can help select the appropriate assay for a clinical question. WGS is one possible option; the assay and interpretation scope matter. [20, 22, 23] |

| WHAT YOU WANT TO KNOW | A USEFUL NEXT STEP |
| --- | --- |
| I already have WGS, but the report feels narrow | Check the available files, quality and scope of analysis. Better explanation and a supported broader analysis may be the missing pieces. |



**Broader observation needs a defined question**
A larger assay expands what can be observed. Its quality and supported interpretation determine which questions it can answer.

## Questions for a sequencing or interpretation provider

- Which observations are directly measured and which are inferred?
- Which regions and variant types are assessed reliably, and what is outside the assay scope?
- Which findings are educational, and which are intended for clinical use?
- What confirmation and professional follow-up would a relevant result need?
- Will I receive usable data files and quality information, and can they be reinterpreted later?

WGS adds value when its broader record helps answer a supported, relevant question. For an everyday wellness question, clearer explanation and the right current context can be more useful than a larger list of DNA letters.

Research and guidance: [22] Interpreting genomic reports: limitations and follow-up (NHGRI / clinical education toolkit); [23] Genetic testing: uses and interpretation (NHGRI). These explain biology or guidance; they do not verify a private genotype.

# The first steps

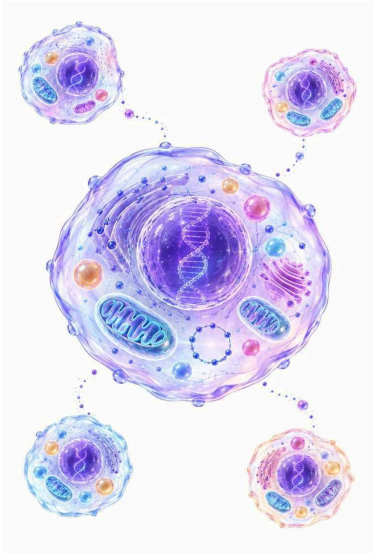
> Longevity and wellness information, never a diagnosis.

OBSERVATION SCOPE, IN CONTEXT

Having read your story, we hope a few things that were vague before now have a name. Pick the two food or habit changes that are easiest this month.

**The useful next question**
Your actual question, symptoms if relevant, dated records and existing care.

Keeping an appropriate routine or making no change is a valid outcome.



## What we read, and how

- **Your raw file.** The calls your sequencing laboratory made from your genome — 5.4 million indexed.
- **Checked, not filled in.** Calls the laboratory's own caller rejected are left out, and a reference position is read from the gVCF only where the reads passed a quality check. Nothing is estimated from a panel, and nothing leaves for a consumer service.
- **Annotated.** The medicine panel (CPIC) was run on the sequencing gVCF.
- **Read.** Every result carries its evidence: what was found, how ready the answer is, and what is not included.

## Words on the page

- **rsID** — the public number of one site in the genome, like a house number; look it up anywhere.
- **allele** — one of the two letters you carry at a site; the effect allele is the one the evidence is about.
- **evidence** — established · probable · emerging — how well replicated the finding is, never how big.

## Bring this to a practitioner

Nothing in this report is a diagnosis, and a practitioner may weigh a measured value very differently from a genotype — that is the right order of things.

## When this report changes

Your genes do not change; what we can read and what we know does. Your online report always shows the current reading; a PDF you downloaded keeps the reading from the day it was made. If a better reading of your file becomes possible — a supported sequencing result, a targeted genotype or copy-number test — the “not read” rows are the first to fill in.

Longevity and wellness information, never medicine or a diagnosis. A result here is a tendency read from common variation; a measured value always outranks it. To delete your file, report or account, email support@indaga.ai.

COMPLETE SELECTED-MARKER INVENTORY

# What was included

This directory records the selected wellness markers once each. It is not an analysis of every variant in the source file. Unread, withheld and outside this edition are distinct from a negative result.

> 75 of 87 distinct selected sites read. A site can inform more than one chapter.

| MARKER | OBSERVATION | MEANING AND SCOPE |
|---|---|---|
| MTHFR rs1801133 | GA Sequencing | MTHFR — one 677T allele. Research links this variant to enzyme properties and folate-related biomarkers; it does not measure your enzyme activity. |
| MTHFR rs1801131 | TT Sequencing | MTHFR — context only. This call does not measure folate processing. |
| MTRR rs1801394 | AA Sequencing | MTRR — context only. This call does not establish B12 deficiency, reduced B12 recycling, high homocysteine or extra choline need. |
| MTR rs1805087 | AA Sequencing | MTR — context only. This common-site call does not measure B12 need or enzyme activity, and does not screen for rare inherited disorders. |
| CBS rs234706 | Not read | Why it was not read: no call in this sequencing file passed the quality check. |
| COMT rs4680 | AA Sequencing | COMT — biological context only. This call does not predict your personality, stress response or methyl-group requirement. |
| MAOA rs6323 | T (single X copy) Sequencing | MAOA — the T allele at this tag, which does not measure your MAO-A activity. |
| GSTP1 rs1695 | AA Sequencing | GSTP1 — no 105Val allele at this site, which does not measure your detoxification. |
| GSTM1 GSTM1 null (whole-gene deletion) | Not read | Why it was not read: an insertion, deletion or copy-number change — not read here as a single site. |
| NOS3 rs1799983 | TG Sequencing | NOS3 — one 298Asp allele, which does not measure your nitric oxide. Cell studies disagree on whether it changes the enzyme's output. |
| PEMT rs7946 | TT Sequencing | PEMT — context only. This call does not measure liver phosphatidylcholine production, or establish extra dietary choline need. |
| AOC1 rs10156191 | CT Sequencing | AOC1 — one 16Met allele, which does not measure your DAO activity. In one study it went with lower DAO activity in blood, not with histamine intolerance itself. |
| AOC1 rs1049793 | CC Sequencing | AOC1 — no 645Asp allele at this site, which does not measure your DAO activity. |
| SOD2 rs4880 | AA Sequencing | SOD2 — two Val16 alleles, which does not measure your antioxidant defence. In a laboratory assay the Val form entered mitochondria less well; people were not measured. |

| MARKER | OBSERVATION | MEANING AND SCOPE |
|---|---|---|
| GPX1<br>rs1050450 | GA<br>Sequencing | GPX1 — one 198Leu allele, which does not measure your antioxidant defence. One study found the enzyme 5% less active per copy; another found no difference. |
| NQO1<br>rs1800566 | GG<br>Sequencing | NQO1 — no 187Ser allele at this site, which does not measure NQO1 activity. |
| HLA-DQA1<br>rs2187668 | CC<br>Sequencing | HLA-DQA1 — no DQ2.5 tag allele, but that does not exclude the risk type, and one tag SNP cannot show your HLA-DQ type; HLA-DQ2.5 is the main inherited risk type for celiac disease. Only a dedicated HLA-DQ test, not this marker, can show your HLA-DQ type. |
| HLA-DQB1<br>rs7454108 | Not read | Why it was not read: no call in this sequencing file passed the quality check. |
| HLA-DQB1<br>rs7775228 | Not read | Why it was not read: no call in this sequencing file passed the quality check. |
| HLA-B<br>rs116488202 | Not read | Why it was not read: no call in this sequencing file passed the quality check. |
| PTPN22<br>rs2476601 | AG<br>Sequencing | PTPN22 — one 620W allele; a modestly raised tendency to type 1 diabetes and rheumatoid arthritis (not a diagnosis). |
| CTLA4<br>rs3087243 | GA<br>Sequencing | CTLA4 — one G allele, the more common allele; a small autoimmune-risk tendency in cohorts, not a diagnosis. It went with slightly more autoimmune thyroid disease and type 1 diabetes. |
| IL6<br>rs1800795 | CG<br>Sequencing | IL6 — one -174C allele, which does not measure your inflammation. The first study found lower IL-6 in blood with C, and 17 studies combined found no link. |
| TNF<br>rs1800629 | GG<br>Sequencing | TNF — no -308A allele at this site, which does not measure your inflammatory tone. |
| APOE<br>rs429358 | TT<br>Sequencing | APOE — no ε4-defining C allele at this SNP (isoform needs another APOE marker too to call fully). |
| FOXO3<br>rs2802292 | GT<br>Sequencing | FOXO3 — one longevity-associated allele: more common among long-lived people in the cited cohorts; an odds nudge, not a guarantee. |
| KLOTHO<br>rs9536314 | TT<br>Sequencing | KLOTHO — non-carrier (no KL-VS tag allele) at this marker. |
| SERPINA1<br>rs28929474 | CC<br>Sequencing | SERPINA1 — no Z variant at this site, which does not rule out the gene's rarer variants. |
| AR<br>AR CAG repeat (polyQ length) | Not read | Why it was not read: a repeat length, not a single site — needs a dedicated assay. |
| SRD5A2<br>rs523349 | GC<br>Sequencing | SRD5A2 — one 89L allele; a modestly lower 5α-reductase activity (small effect). |
| CYP19A1<br>rs10046 | GG<br>Sequencing | CYP19A1 — no A allele at this tag, which does not measure your aromatase activity or your estradiol. |
| CYP19A1<br>rs700518 | TT<br>Sequencing | CYP19A1 — no C allele at this tag, which does not measure your aromatase activity. |

| MARKER | OBSERVATION | MEANING AND SCOPE |
|---|---|---|
| ESR1<br>rs2234693 | TT<br>Sequencing | ESR1 — no 'P' allele at this tag, which does not measure your receptor sensitivity. |
| ESR1<br>rs9340799 | AA<br>Sequencing | ESR1 — no 'X' allele at this tag, which does not measure your receptor sensitivity. |
| SHBG<br>rs6259 | GG<br>Sequencing | SHBG — no 356N allele at this site, which does not measure your SHBG. |
| SLC6A4<br>rs25531 | Not read | Why it was not read: an insertion, deletion or copy-number change — not read here as a single site. |
| BDNF<br>rs6265 | CC<br>Sequencing | BDNF — no Met66 allele at this site, which does not measure BDNF release, memory or stress response. |
| PER3<br>rs57875989 | Not read | Why it was not read: a repeat length, not a single site — needs a dedicated assay. |
| BHLHE41<br>rs121912617 | GG<br>Sequencing | BHLHE41 — the common genotype; no familial-short-sleep allele (expected). |
| MTNR1B<br>rs10830963 | CC<br>Sequencing | MTNR1B — the common genotype; no glucose-raising allele from this variant. |
| CYP2D6<br>CYP2D6 activity (star alleles + copy number) | Not read | Why it was not read: an insertion, deletion or copy-number change — not read here as a single site. |
| TAS2R38<br>rs713598 | CC<br>Sequencing | TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster. |
| TAS2R38<br>rs1726866 | AA<br>Sequencing | TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster. |
| TAS2R38<br>rs10246939 | TT<br>Sequencing | TAS2R38 — No copy of the taster variant here; read with the other two TAS2R38 markers, the diplotype is AVI/AVI — a bitter non-taster. |
| OR6A2<br>rs72921001 | Not read | Why it was not read: no call in this sequencing file passed the quality check. |
| OPRM1<br>rs1799971 | AG<br>Sequencing | OPRM1 — one 118G allele, with an effect too small for guidelines to act on. Surgery studies saw slightly higher opioid needs in carriers. |
| SCN9A<br>rs6746030 | GG<br>Sequencing | SCN9A — the common genotype; no 1150W allele. |
| MC1R<br>rs1805007 | CC<br>Sequencing | MC1R — no 151C allele at this MC1R red-hair variant (read with another MC1R marker). |
| MC1R<br>rs1805008 | CC<br>Sequencing | MC1R — no 160W allele at this MC1R red-hair variant (read with another MC1R marker). |
| SLC45A2<br>rs16891982 | GG<br>Sequencing | SLC45A2 — no 374Leu allele at this site, which does not measure how your skin responds to the sun. Other genes also shape pigmentation. |

| MARKER | OBSERVATION | MEANING AND SCOPE |
|---|---|---|
| HERC2<br>rs12913832 | GG<br>Sequencing | HERC2 — two blue-eye alleles: with this genotype eyes are most often blue, and brown is rare. Other genes also shape eye colour. |
| FLG<br>rs61816761 | GG<br>Sequencing | FLG — no copies here, but this report reads only a few FLG variants, so that does not rule out others. |
| MMP1<br>rs1799750 | Not read | Why it was not read: an insertion, deletion or copy-number change — not read here as a single site. |
| LPA<br>rs10455872 | AA<br>Sequencing | LPA — no high-Lp(a) tag allele (but this does not rule out high Lp(a) — your measured Lp(a) says more than these markers). |
| LPA<br>rs3798220 | TT<br>Sequencing | LPA — no high-Lp(a) tag allele (does not rule out high Lp(a) — your measured Lp(a) says more than these markers). |
| F5<br>rs6025 | CC<br>Sequencing | F5 — no Factor V Leiden at this site, which does not test the rest of the clotting system. |
| F2<br>rs1799963 | GG<br>Sequencing | F2 — no prothrombin G20210A at this site, which does not test the rest of the clotting system. |
| TCF7L2<br>rs7903146 | TT<br>Sequencing | TCF7L2 — two T2D-associated alleles: a raised tendency at this one marker, not a diagnosis or a combined genetic score. |
| FTO<br>rs9939609 | TT<br>Sequencing | FTO — no adiposity-linked allele at this variant, which does not measure your body weight. |
| MC4R<br>rs17782313 | TT<br>Sequencing | MC4R — no adiposity-linked allele at this marker, which does not measure your body weight. |
| PNPLA3<br>rs738409 | CG<br>Sequencing | PNPLA3 — one 148M allele; a modestly higher liver-fat tendency, larger with more body fat (not a diagnosis). |
| TM6SF2<br>rs58542926 | CC<br>Sequencing | TM6SF2 — no 167K allele at this site, which does not rule out fatty liver. |
| MCM6<br>rs4988235 | GG<br>Sequencing | MCM6 — Lactase non-persistent genotype — adult lactose intolerance is more likely; symptoms vary. |
| ALDH2<br>rs671 | GG<br>Sequencing | ALDH2 — no ALDH2*2 allele at this site; that does not rule out flushing from other causes. |
| ADH1B<br>rs1229984 | CC<br>Sequencing | ADH1B — no fast ADH1B*2 allele at this site, which does not measure how fast you clear alcohol. |
| CYP1A2<br>rs762551 | AA<br>Sequencing | CYP1A2 — the higher-inducibility genotype. Measurable mainly in smokers and heavy coffee drinkers; in non-smokers it has not been shown to change caffeine clearance. |
| ADORA2A<br>rs5751876 | TT<br>Sequencing | ADORA2A — linked in two studies to more jitteriness after caffeine, less so in regular drinkers. Not a sleep result: sleep studies point to the other allele. |
| FTO<br>rs1421085 | TT<br>Sequencing | FTO — no adiposity-linked allele at the causal FTO variant, which does not measure your body weight. |

| MARKER | OBSERVATION | MEANING AND SCOPE |
|---|---|---|
| FUT2<br>rs602662 | AA<br>Sequencing | FUT2 — two non-secretor-tag alleles (consistent with non-secretor; read with another FUT2 marker). Studies link this genotype to higher total serum B12, which does not measure your B12 status. |
| BCMO1<br>rs12934922 | TT<br>Sequencing | BCMO1 — two 267S alleles, which do not measure your beta-carotene conversion. The cited study found lower conversion in carriers of a second variant (379V), which this report does not read. |
| HFE<br>rs1800562 | GG<br>Sequencing | HFE — No HFE variant at this marker. |
| HFE<br>rs1799945 | CC<br>Sequencing | HFE — No HFE variant at this marker. |
| HFE<br>rs1800730 | AA<br>Sequencing | HFE — No HFE variant at this marker. |
| COMT<br>rs4633 | TT<br>Sequencing | COMT — observed site only; no genotype or personal activity inferred at another COMT marker. |
| VDR<br>rs2228570 | AG<br>Sequencing | VDR — FokI: one copy each of the alleles for the shorter and the longer form of the receptor. Cell studies disagree on whether the two forms differ in activity. A 25-OH-D blood test, not this marker, shows your vitamin D status. |
| VDR<br>rs1544410 | CC<br>Sequencing | VDR — BsmI: no variant allele at this marker, which does not measure your vitamin D status. |
| VDR<br>rs731236 | AA<br>Sequencing | VDR — TaqI: no variant allele at this tag, which does not measure your vitamin D status. |
| VDR<br>rs7975232 | AA<br>Sequencing | VDR — ApaI: two copies of an intronic tag allele with no established effect of its own. A 25-OH-D blood test, not this marker, shows your vitamin D status. |
| FUT2<br>rs601338 | AA<br>Sequencing | FUT2 — a non-secretor (no ABO antigens secreted into mucus or saliva). In cohorts non-secretors differ in gut bacteria and B12 levels and resist some norovirus strains — associations, not a diagnosis. |
| NOD2<br>rs2066844 | CC<br>Sequencing | NOD2 — no 702W allele at this site, which does not rule out Crohn's disease or another NOD2 risk allele. |
| NOD2<br>rs2066845 | GG<br>Sequencing | NOD2 — no 908R allele at this site, which does not rule out Crohn's disease or another NOD2 risk allele. |
| ACTN3<br>rs1815739 | CC<br>Sequencing | ACTN3 — both copies make this fast-muscle-fibre protein. It does not pick a sport or predict your training response. |
| ACE<br>rs4646994 | Not read | Why it was not read: an insertion, deletion or copy-number change — not read here as a single site. |
| PPARGC1A<br>rs8192678 | CT<br>Sequencing | PPARGC1A — one Ser allele. Studied in athlete status and some training adaptations; it does not forecast your aerobic gains. |
| COL1A1<br>rs1800012 | CC<br>Sequencing | COL1A1 — No Sp1 allele at this site. Its injury studies are inconsistent; it does not establish your injury risk. |
| COL5A1<br>rs12722 | CT<br>Sequencing | COL5A1 — one C allele. Its tendon and ligament studies disagree; it does not set your flexibility or injury risk. |

| MARKER | OBSERVATION | MEANING AND SCOPE |
|---|---|---|
| AMPD1 rs17602729 | GG Sequencing | AMPD1 — not seen at this site, which does not rule out other variants or measure enzyme activity. |

# Where the explanation comes from

The personal observation record and the published research answer different questions. Published sources cannot confirm who a DNA sample belongs to or verify a private genotype. Observation method, build and quality appear beside the supplied findings. An unrecorded provenance detail remains unrecorded.


1. Lactose: the inherited mechanism — MedlinePlus Genetics
2. Food choices with lactose intolerance — NIDDK
3. TCF7L2 and diabetes prevention — Florez et al., NEJM 2006
4. MTHFR and folic acid — CDC
5. Caffeine, ADORA2A and habitual use — Rogers et al., Neuropsychopharmacology 2010
6. What COMT does — MedlinePlus Genetics
7. Sun protection — CDC
8. The MTHFR enzyme — MedlinePlus Genetics
9. Folate: food, function and supplementation — NIH Office of Dietary Supplements
10. Vitamin B12: function and assessment — NIH Office of Dietary Supplements
11. MTHFR genotype and folate response — Crider et al., AJCN 2011; randomized trial
12. Epigenetics and the epigenome — MedlinePlus Genetics
13. Lactose intolerance: symptoms and causes — NIDDK
14. Blood tests and diabetes — NIDDK
15. A1C: meaning and limitations — NIDDK
16. Pigmentation and MC1R — MedlinePlus Genetics
17. The AMPD1 enzyme — MedlinePlus Genetics
18. Pharmacogenetic associations in context — FDA
19. Diagnosis of coeliac disease — NIDDK
20. Exome and genome sequencing — MedlinePlus Genetics
21. Human genomic variation and how it is detected — NHGRI
22. Interpreting genomic reports: limitations and follow-up — CSER Consortium clinical education toolkit, hosted by NHGRI
23. Genetic testing: uses and interpretation — NHGRI
24. Variants of uncertain significance — NHGRI
25. AMP deaminase deficiency and inheritance — MedlinePlus Genetics
26. Genotype imputation from reference haplotypes — Howie, Marchini and Stephens, G3 2011
27. Lactose malabsorption and symptoms — NIDDK
28. CYP1A2 inducibility in smokers and non-smokers — Sachse et al., British Journal of Clinical Pharmacology 1999
29. Genetics and athletic performance — MedlinePlus Genetics
30. ApoB, LDL and non-HDL: different measurements — National Lipid Association expert clinical consensus 2024
31. How the immune response works — MedlinePlus Medical Encyclopedia
32. Hormones and endocrine signalling — MedlinePlus
33. Genetics and longevity — MedlinePlus Genetics
34. Taste biology and changes in taste — NIDCD
35. The liver and processing substances — MedlinePlus
36. Lactose intolerance: symptoms and assessment — NIDDK
37. Sleep diary and assessment — NHLBI
38. Caffeine anxiety and habitual use — Rogers et al. 2010
39. Caffeine-related sleep response — Retey et al. 2007
40. PPARGC1A elite-athlete comparison — Lucia et al. 2005
41. COL5A1 and Achilles tendinopathy — September et al. 2009
42. Lipoprotein(a) interpretation and units — European Atherosclerosis Society consensus
43. Missing, reference and assay limitations — PharmCAT FAQ


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