

Nutrigenomic Summary

B. [Surname] | Panel [number] | Dr. Lauren Cherrier, PharmD, Consulting Pharmacist

INTERPRETATION NOTE -- PLEASE READ THIS FIRST. The findings on this panel are common gene variants, not mutations that cause disease. They do not diagnose low vitamin B12, low folate, mitochondrial disease, trouble with cell cleanup, ADHD, POTS, Ehlers-Danlos syndrome, or how you will respond to any drug. On their own, they should not decide any drug, hormone, diet, or supplement. Your health history, physical exam, and standard lab tests are still what your care is based on.

START HERE

1. Get the bloodwork on the last page before starting any supplement. Nothing on this panel tells us what your levels are right now. Only the labs do.
2. Bring this summary and your supplement bottles to your next visit. Famotidine, atorvastatin, testosterone, and hydroxyzine each interact with something discussed here.
3. If you do start something, start one product at a time. With mast cell activation syndrome, adding two or three at once makes it impossible to tell what caused a reaction.

How to read your results

You have two copies of every gene, one from each parent. A variant is a small spelling change inside a gene. It is not a missing or broken gene. One copy means the change was found on one of your two copies. Two copies means it was found on both. Two copies does not mean the gene is switched off, and for the genes below, carrying two copies is common, not unusual.

Vitamin B12 and how it is carried

TCN2 rs1801198 | TWO COPIES

The B12 carrier protein

TCN2 makes transcobalamin, the protein that carries vitamin B12 from your blood into your cells. Only the B12 attached to this carrier can actually be used, and that is about a fifth to a quarter (20-25 percent) of what a standard test measures.

This is a common variant. Individual studies have conflicted on what it does, but a 2017 meta-analysis of 34 studies found that people with the GG genotype averaged lower holotranscobalamin and higher homocysteine (in participants of European descent). It does not diagnose a B12 transport problem, and it is not the rare inherited transcobalamin deficiency.

What I would do: checking your vitamin B12 status by lab testing is reasonable, given your clinical picture and your famotidine (see below).

MTHFD1 rs1076991 | TWO COPIES

A folate-handling enzyme

MTHFD1 makes an enzyme used in folate handling, part of how your body builds and repairs DNA. This is a common variant. It does not diagnose a folate metabolism disorder, and it does not mean you need any particular form of folate. No specific claim should be attached to this result without confirming the lab's reporting details.

What I would do: nothing on this basis. If folate status ever becomes a question, serum folate and homocysteine can help answer it.

FAMOTIDINE | THE MOST USEFUL PART OF THIS PICTURE

Why your stomach-acid drug matters more than the gene results here

Stomach acid helps free vitamin B12 from food. Long-term acid-suppressing treatment, including famotidine, can lower absorption of food-bound B12 in some people, particularly where other risk factors are present. In a study of more than 200,000 patients, two or more years of H2 blocker use was linked to a higher chance of B12 deficiency. That is a reason to check the level from time to time, not a reason to stop the drug. Do not stop or reduce famotidine on your own.

If deficiency is confirmed, methylcobalamin, hydroxocobalamin, and adenosylcobalamin are all reasonable forms to take by mouth. No oral form skips transcobalamin, since every form uses the same carrier to get into cells. Where absorption itself is the limiting factor, injection is preferred. Your physician should set the form and the dose.

Dopamine and adrenaline -- how they are broken down

COMT rs4680 (Met/Met) | TWO COPIES

The enzyme that clears dopamine, adrenaline, and noradrenaline

COMT makes the enzyme that breaks down dopamine, adrenaline, and noradrenaline. It is most active in the prefrontal cortex, the part of the brain involved in attention and working memory. This variant is linked in lab studies to lower COMT activity.

You will read online that this result causes anxiety. A study of 5,531 adults in the general population found no clear link between this genotype and either anxiety or depression. I would treat it as background information, not as a finding that needs treating. It does not predict a psychiatric diagnosis, your caffeine response, your response to stimulant medication, your anxiety level, or a catecholamine clearance disorder.

On caffeine: caffeine works by blocking adenosine receptors, and it is cleared by a liver enzyme called CYP1A2, not by COMT. Its half-life is around five hours in most adults, so a cup at 4pm still has about a quarter of its dose on board at bedtime. If you have insomnia, palpitations, anxiety, or POTS symptoms, a trial of morning-only caffeine is reasonable to test and judge by how you feel. It is not something your genetics require. Talk to the clinician who manages your POTS first, because some people use caffeine on purpose to support blood pressure and standing tolerance.

On methylated B vitamins: you may read that this COMT result means avoiding methylfolate or methylcobalamin. That advice comes almost entirely from supplement sellers and genetic testing companies, not from clinical trials, and there is no proven gene-based approach here. If you and your physician decide a B vitamin is warranted, start at a low dose and increase slowly, as with any supplement. No supplement is indicated on the basis of this result alone, and decisions about caffeine or stimulant medication should follow how you actually feel, along with your sleep, heart rate, and blood pressure.

Vitamin D

GC rs2282679 | TWO COPIES

The vitamin D carrier protein

GC makes vitamin D binding protein, which carries vitamin D through your bloodstream. Most of the vitamin D in your blood is attached to it at any given moment. This is a common variant, linked in studies to differences in measured total 25-hydroxyvitamin D. It does not prove that less vitamin D reaches your tissues, and it does not mean you need a special formulation.

What I would do: measure 25-hydroxyvitamin D and base any dosing on that result, together with your calcium status, kidney function, diet, and clinical picture. Below 30 ng/mL, supplementation is clearly warranted. Between 30 and 50 ng/mL, a maintenance dose is reasonable and worth discussing. Above that, guidelines disagree -- the Endocrine Society has historically favored a higher target than the Institute of Medicine -- so your physician's preference applies. Vitamin D3 (cholecalciferol) is the usual form. Nothing about this result calls for an oil-based, MCT, or high-dose product, or for added vitamin K2. If you do use a combined D3 and K2 product, confirm with your physician that K2 is appropriate, since it interacts with warfarin and similar blood thinners. Recheck timing should reflect your starting level, the dose, and your physician's plan.

Energy production and cell cleanup

NDUFS8 | TWO COPIES -- ATG5, ATG12, ATG16L1 | ONE COPY EACH

Mitochondria and autophagy genes

NDUFS8 makes one small part of Complex I, the first station your mitochondria use to turn food into usable energy. The ATG genes work together in autophagy, the recycling process cells use to break down and clear out damaged internal parts.

These are common variants. They do not establish mitochondrial disease, a broken energy chain, defective cleanup, chronic oxidative stress, or a need for mitochondrial or autophagy supplements. They are separate from the rare mutations that disable these proteins and cause serious inherited disease. No supplement is indicated on the basis of these results.

What this panel does not explain

Your Ehlers-Danlos syndrome, POTS, and mast cell activation syndrome (MCAS) are clinical diagnoses made by your physicians. None of the genes on this panel cause them, and none can confirm or rule them out. I am stating that plainly because nutrigenomic reports are often written as though B12 and folate genes explain connective tissue problems. They do not. Collagen is built and cross-linked by enzymes that depend on vitamin C, iron, and copper. Vitamin B12 is not one of the ingredients in that process. The one indirect link is worth naming honestly: severe B12 deficiency raises homocysteine, and very high homocysteine can interfere with collagen cross-linking. That is a downstream effect of deficiency, not B12 taking part in collagen production, and it does not make B12 or folate genes an explanation for Ehlers-Danlos syndrome. The genes associated with Ehlers-Danlos syndrome are collagen and matrix genes such as COL5A1, COL5A2, COL3A1, and TNXB, and the hypermobile type has no identified causal gene at all.

Your medications and this panel

Medication	Why it appears here	What to raise
Famotidine	Long-term acid suppression may lower absorption of food-bound vitamin B12 in some people. The most relevant drug on this list.	Ask about periodic vitamin B12 assessment. Do not stop or reduce it on your own.
Atorvastatin	Causes muscle symptoms in a minority of patients. Those symptoms can overlap with your baseline pain, hypermobility, and fatigue, which makes them easy to misattribute in either direction.	Report new, persistent, or significant muscle symptoms rather than absorbing them. Review any supplement, including red yeast rice or quercetin, for interactions and additive effects before using it alongside atorvastatin.
Testosterone	Raises red blood cell production, which your prescribing clinician monitors.	Follow your prescriber's monitoring plan for hemoglobin and hematocrit.
Hydroxyzine	Sedating and anticholinergic. Can dull daytime concentration -- worth separating from any attention symptom you are tracking -- and can raise heart rate, which is relevant with POTS. Magnesium taken at night adds to the sedation.	That added sedation may be useful or unwanted depending on your dosing schedule.

If a supplement is started

- One product at a time, at least seven to fourteen days apart. This is the single most useful rule for someone with MCAS or a history of supplement sensitivity. Adding several at once means a reaction cannot be traced.
- Read the full ingredient list, not just the active ingredient. Dyes, flavorings, gum bases, and fillers are common triggers, and are often the reason a product is not tolerated when the nutrient itself would have been fine.
- Record the product, the dose, and the start date, and photograph each label. If something goes wrong, this is what your MCAS physician will want to see.
- If vitamin C has caused stomach upset before, a buffered form is usually better tolerated. Supplemental L-proline and L-lysine are often sold alongside it for connective tissue, but there is no trial evidence that they help people who are not deficient.
- Nothing here needs to be started urgently. Every recommendation here waits comfortably for your lab results.

Movement and activity

Strength work is one of the most effective things available to you, not something to avoid. Randomized trials in hypermobility spectrum disorder and hypermobile Ehlers-Danlos syndrome have tested progressive and even heavy resistance programs, on the reasoning that stronger muscle and tendon around a lax joint improves stability.

- Work with a physical therapist who knows hypermobility. Program design matters more than the specific exercises, and this is the part that should not be self-directed.
- Build up load gradually and control the range of motion. Stopping short of end range protects the joint capsule. Progressive loading itself is the goal.
- Include proprioceptive and balance work. Joint position sense is commonly reduced in hypermobility and improves with training.
- Limit high-impact and contact activity. This is where caution is warranted, rather than with strength training generally.
- Start POTS-friendly. Recumbent or semi-recumbent work such as a rowing machine, recumbent bike, or swimming is usually better tolerated at first, progressing toward upright activity as tolerance improves.
- Pace by recovery. If you are not back to your baseline within a few hours, reduce the duration or intensity next time rather than pushing through.

Laboratory testing to discuss

Test	Purpose
Vitamin B12 with MMA	Evaluates for vitamin B12 deficiency or functional deficiency where clinically indicated. Methylmalonic acid (MMA) should be interpreted alongside kidney function.
Homocysteine	May support assessment of vitamin B12, folate, vitamin B6, kidney, and thyroid contributors. It is not specific for a genetic methylation disorder.
Serum folate	Establishes current folate status if that becomes a question.
25-hydroxyvitamin D	Establishes vitamin D status and guides supplementation where indicated. Recheck timing should reflect your baseline level, the dose, and your physician's plan.
CBC with hemoglobin and hematocrit	Monitors for a rise in red cell count during testosterone therapy, consistent with your prescribing clinician's monitoring plan. Red cell indices also shift early in folate and B12 deficiency.
Ferritin and iron studies	Worth considering for fatigue, anemia, dietary risk, bleeding history, or unexplained changes in red cell indices.
CMP with eGFR	Kidney function affects homocysteine and MMA interpretation, and famotidine is cleared renally.
TSH	Thyroid dysfunction is a common and treatable cause of fatigue.
Creatine kinase	Most useful when muscle symptoms raise concern about a statin-associated muscle problem, rather than as routine screening.

To discuss with your physician

- Confirm B12 before starting any substantial folate supplement. Folate can normalize the blood picture of B12 deficiency while nerve damage continues underneath. This sequence is the most important item on the page.
- Ask about periodic B12 assessment given long-term famotidine, rather than treating it as a one-off check.
- Review anything you want to add alongside atorvastatin, including red yeast rice, quercetin, high-dose niacin, and grapefruit.
- Confirm your monitoring plan for testosterone with the clinician who prescribes it.
- Raise the caffeine question with whoever manages your POTS rather than changing your intake on your own.
- Ask for a physical therapy referral for a hypermobility-specific strength program if you do not already have one.
- Discuss ongoing fatigue separately. This panel does not account for it. Sleep quality, thyroid function, iron status, medication effects, POTS control, and mood are higher-yield places to look.

PLEASE READ. This summary is educational. It is not a diagnosis, a prescription, or a treatment plan, and it does not replace the care of your physician or other treating clinicians. Please review it with the clinician who manages your health before making any change. Do not start, stop, or change the dose of any prescribed medication or supplement without speaking to the prescriber. Where this summary indicates that a product is not required, raise it with your clinician rather than acting independently. Dr. Lauren Cherrier, PharmD, is a licensed pharmacist and holds active pharmacist licensure in Kentucky, Arizona, Indiana, and Florida. She is not the patient's treating physician. MyTraitRx provides pharmacist-led health education and consulting and does not diagnose or prescribe. Findings reflect the panel supplied and the evidence available at the date of review. Variant identifiers, genotypes, methodology, and analytic limitations are held in the source laboratory report and the clinician file for this panel.

Selected references

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Dr. Lauren Cherrier, PharmD, Consulting Pharmacist | MyTraitRx | mytraitrx.com | info@mytraitrx.com