

PTPN22 (Autoimmunity)

Gene Report

REPORT CATEGORY —



INFLAMMATION &
AUTOIMMUNITY

Sample Client

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Personal information

NAME

Sample Client

SEX AT BIRTH

Male

HEIGHT

5ft 5" 165cm

WEIGHT

137lb 62kg

DISCLAIMER

This report does not diagnose this or any other health conditions. Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.



How this works

Our Wellness Reports analyze how your DNA influences your health.

We then use this analysis to give you personalized risk estimates and recommendations.



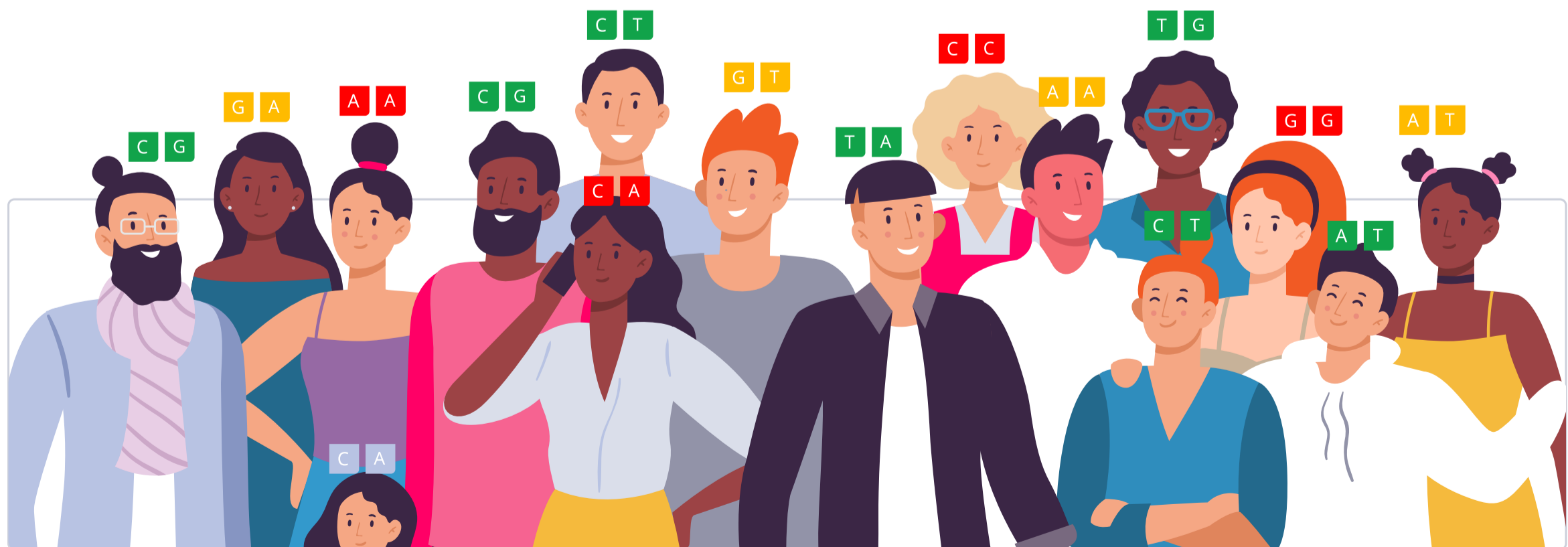
Similarly, our Trait Reports look at how your DNA influences your traits.



Your DNA is like an instruction manual — it contains a lot of information.

You can think of it as a blueprint for your body.

Genetic variants are parts of DNA that differ from person to person. Some can make you more vulnerable to certain health issues, while others may influence traits such as eye color.



We use artificial intelligence and machine learning to analyze all this information. We then summarize your results as a risk score or display it on a gauge.

In total, we analyze up to 83 million genetic variants.

When we give a risk score, the risk icon tells you if you are at a higher or lower risk compared to other people:



Genotype color info:

- AA** You don't have any risk alleles
- AA** You have 1 risk allele
- AA** You have 2 risk alleles

Your risk is also displayed as a percentile. This will tell you how your risks compare to our sample population. The lower your percentile number, the lower your risk. The "50th percentile" would be an average risk.

Similarly, the gauge tells you your relative risk score compared to our sample population, or it indicates a specific trait or haplotype you are more likely to have based on your genetic variants.

When applicable, we also list top evidence-based recommendations that may help lower your risk. The focus is on recommendations that may be of benefit to you, based on your genetics.

Our recommendations come in four categories: lifestyle, diet, supplements and drugs. The following icons tell you which category a recommendation falls into:



Our team of scientists also ranks each recommendation. We rank based on impact and the strength of evidence in the medical literature.

Impact shows how strongly a recommendation will affect your health in a certain area. Evidence is how much scientific support there is for the recommendation. Rankings are from 1 to 5 (low to high):



Impact

Impact scores range from 1-5. These scores reflect how much of an effect each recommendation can have. An impact score of 5 predicts the biggest effect.

When a recommendation affects something we can measure, we use those measurements to assign the impact score. For example, a recommendation that decreases cholesterol by 20% will have a higher impact score than one that decreases it by 5%.

Some recommendations affect things that we cannot directly measure, like stress or mood. For these, the impact score is based on how well they work relative to other recommendations and standard treatments. The best ones get the highest scores.

If there is a lot of research that shows a recommendation works especially well for your genotype, the impact score gets increased.

Recommendation Evidence

●●●●● 5 / 5

Recommendations that are considered effective and generally recommended by experts and medical bodies.

●●●●○ 4 / 5

Recommendations that are considered likely effective and that have multiple independent meta-analyses and a great many studies supporting them.

●●●○○ 3 / 5

Recommendations that are considered possibly effective and have many studies supporting them

●●○○○ 2 / 5

Recommendations that have insufficient evidence, with two or several clinical trials supporting them, or many studies but with ambiguous results.

●○○○○ 1 / 5

Recommendations that have insufficient evidence, with a single clinical trial, or with many studies most of which didn't find support for the recommendation.

○○○○○ 0 / 5

No evidence in humans.

Genotype-specific Evidence

●●●●● High-quality

Direct evidence that a recommendation helps more in people with your gene variant (many clinical trials, a few large clinical trials, or a meta-analysis).

●●●●○ Medium-quality

Direct evidence that a recommendation helps more in people with your gene variant (a few clinical trials or one large clinical trial).

●●●○○ Low-quality

Direct evidence that a recommendation helps more in people with your gene variant (a single clinical trial or more trials with inconsistent results).

●●○○○ Indirect

A recommendation may help more in people with your gene variant because it targets a specific gene or protein affected by your variant (e.g., MTHFR, dopamine).

●○○○○ In theory

A recommendation may help more in people with your gene variant because it targets a specific mechanism affected by your variant (e.g., inflammation, oxidative stress).

Some things to keep in mind:

- Genetics doesn't play a considerable role in a condition or a trait.
- There is not enough research available to estimate a genetic predisposition.
- There are technical limitations to estimating or presenting a genetic predisposition.
- The topic is sensitive, and a genetic predisposition should only be estimated and presented by a healthcare professional.

Introduction

The [PTPN22](#) (or *LYP*) gene encodes a protein from the PTP (protein tyrosine phosphatases) family. This protein regulates the development and function of T cells, B cells, and other immune system components [\[R\]](#).

More precisely, it controls the antiviral immune response and prevents excessive inflammation that can harm the healthy tissue and trigger autoimmunity [\[R\]](#).

Mutations in the *PTPN22* gene can cause defective protein synthesis and uncontrolled response, contributing to a range of [autoimmune disorders](#) [\[R\]](#).

PTPN22 Genetics

According to a study of almost 40,000 people, two *PTPN22* variants—[rs2476601](#) and [rs6679677](#)—are crucial genetic contributors to [hypothyroidism](#). Carriers of minor ‘A’ alleles on one of these variations have a 36% higher risk of hypothyroidism, compared with people who carry major alleles (G and C, respectively). These variants are almost always inherited together, so you will most likely carry either both or neither of them [\[R\]](#).

The rs2476601 variant also showed a significant connection with [Graves’ disease](#). In two British studies of 2,700 participants, the ‘A’ allele was associated with 43-88% higher rates. Two studies of over 900 Polish subjects confirmed this link and found that people with the ‘A’ allele get diagnosed younger. The same allele correlated with 85% higher rates of Graves’ disease in a Chinese meta-analysis [\[R, R, R, R, R\]](#).

The ‘A’ variants of these polymorphisms have also been associated with an increased risk of:

- [Rheumatoid arthritis](#) [\[R, R, R, R, R, R, R, R\]](#)
- [Lupus](#) [\[R, R, R, R, R, R, R\]](#)

Other conditions associated with the ‘A’ allele include [\[R, R, R, R, R, R\]](#):

- Juvenile idiopathic arthritis
- Type 1 diabetes
- Vitiligo
- Myasthenia gravis
- Alopecia areata
- Drug-induced liver injury
- Addison's disease
- Idiopathic inflammatory myopathies
- Immune thrombocytopenia
- Anti-neutrophil cytoplasmic antibody-associated vasculitis

However, this allele showed a protective effect (around 16% lower odds) in the case of [Crohn’s disease](#) [\[R\]](#).

Other *PTPN22* variants such as [rs2488457](#) and [rs33996649](#) are also associated with autoimmune



TYPICAL

Likely typical *PTPN22* genetics based on the genetic variants we looked at

81%

OF USERS SHARE THE SAME SCORE



You have the same genetic predisposition as 81% of our users.

Your top variants that most likely impact your genetic predisposition:

GENE	SNP	GENOTYPE
PTPN22	rs6679677	CC
PTPN22	rs2476601	GG

The number of "risk" variants in this table doesn't necessarily reflect your overall result.

diseases, especially rheumatoid arthritis, type 1 diabetes, and ulcerative colitis.

Scientists haven't figured out the exact mechanism by which these variations cause harm. The key may lie in defective PTP synthesis, which disables the protein and prevents the removal of self-attacking [lymphocytes](#) (T and B cells) [[R](#), [R](#)].

Risk variants may contribute to autoimmunity by:

- Being less capable of fighting infections
- Increasing T cell, Th1 and Th17 activity
- Suppressing regulatory T cells (Tregs)
- Activating B cells and self-attacking B cells

Your Recommendations

Your recommendations are prioritized according to the likelihood of it having an impact for you based on your genetics, along with the amount of scientific evidence supporting the recommendation.

You'll likely find common healthy recommendations at the top of the list because they are often the most impactful and most researched.

	DOSAGE		DOSAGE		
1	Exercise At Least One Hour a Day	1 hour	2	Curcumin	500 mg
3	Maintain Optimal Vitamin D Levels	1000 iu	4	Myo-inositol	4 g
5	CBD	10 mg	6	Garlic	
7	Avoid Exposure to Heavy Metals		8	Zinc	15 mg
9	Low-Level Laser Therapy (LLLT)	30 seconds			

1



Exercise At Least One Hour a Day

IMPACT

3 / 5

EVIDENCE

3 / 5

How to implement

Dedicate a minimum of 60 minutes to moderate-intensity activities such as brisk walking, swimming, or cycling. Do this most days of the week, aiming for at least 5 days to optimize benefits.

TYPICAL STARTING DOSE

1 hour

Description

Exercise can do wonders for your health. It can help you lose weight, improve your heart health, boost your mood, and more [\[R\]](#).

There are many ways you can be active. You can walk, run, swim, dance, or play team sports. **Everything counts, and it's never too late to start!**


Try to get at least **60 minutes of moderate physical activity on most days**, including walking. **Getting a mix of cardio (min 150 min/day) and strength training (min 2 times a week) may be optimal.**

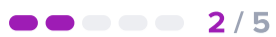
How it helps

Multiple animal- and human-based studies have reported that exercise may increase IL-2 production, boost Tregs, and decrease Th17 cell-mediated inflammatory cytokine release [\[R, R, R, R\]](#).

Exercise, especially cardio, may also improve physical fitness, metabolism, fatigue, inflammation, and complications in people with the following autoimmune conditions:

- Multiple sclerosis [\[R, R, R, R, R, R, R\]](#)
- IBD [\[R, R, R, R\]](#)
- Psoriasis [\[R\]](#)
- Lupus [\[R\]](#)
- Hashimoto's disease [\[R, R, R, R\]](#)
- Graves' disease [\[R\]](#)

2  **Curcumin**

IMPACT  **EVIDENCE** 

How to implement

Take a 500 mg curcumin supplement daily with food. To enhance absorption, take it with a meal that contains fats or oils since curcumin is fat-soluble.

TYPICAL STARTING DOSE

500 mg

Description

Curcumin is a compound found in turmeric known for its anti-inflammatory and antioxidant properties. It has been studied for its potential to reduce inflammation, support joint health, and contribute to overall well-being.

Turmeric is a yellow spice from India. It may reduce inflammation and [oxidative stress](#) [R].

The most important active compound in turmeric is **curcumin**. People use curcumin for [\[R, R, R, R, R, R\]](#):

- Joint pain
- Hay fever
- Mood
- High blood sugar
- Gut health
- Liver health

How it helps


Curcumin may suppress PTPN22 by boosting Tregs and anti-inflammatory IL-10, suppressing Th1 and Th17 activity, and inhibiting the activity of dendritic cells [\[R, R, R, R, R\]](#).

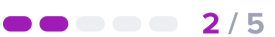
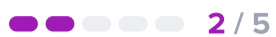
Curcumin may help relieve the symptoms in people with the following autoimmune conditions:

- IBD [\[R, R, R, R\]](#)
- Rheumatoid arthritis [\[R, R, R, R\]](#)
- Multiple sclerosis [\[R\]](#)

Note that curcumin is hard to absorb. Look for supplements with *bioavailable* curcumin, which is easier to absorb. Combining curcumin with [piperine](#) (a compound in black pepper) may also help [\[R, R\]](#).

Please note: curcumin may interfere with iron absorption due to its iron-chelating properties, potentially exacerbating anemia or making it harder to manage. If you have anemia, consult your healthcare provider before using curcumin or turmeric supplements [\[R, R, R\]](#).

3  **Maintain Optimal Vitamin D Levels**

IMPACT  **EVIDENCE** 

How to implement

Check your vitamin D levels, they should ideally be in the 30-66 ng/mL range. If your levels are lower than that, take a vitamin D supplement, 1000-4000 IU daily, to reach an optimal range.

TYPICAL STARTING DOSE

1000 iu

Description

Vitamin D, often referred to as the "sunshine vitamin," can be synthesized by the skin when exposed to sunlight, as well as being found in fish, eggs, and fortified milk. It helps regulate calcium absorption, promoting strong bones and a healthy immune system. Vitamin D deficiency can lead to conditions like rickets in children and osteoporosis in adults.

Your body needs [vitamin D](#) for strong bones. Vitamin D also plays a role in [\[R\]](#):

- Mood
- Immunity
- Heart health
- Blood sugar control

[Sunlight](#) is our main source of vitamin D. Experts recommend getting at least **5-15 minutes of midday sun, 2-3 times per week**. People with darker skin and those living at high latitudes may need longer periods of sun exposure [\[R, R\]](#).

Foods like fish, eggs, and fortified milk provide small amounts of vitamin D. **People lacking vitamin D should consider taking a supplement** [\[R\]](#).

How it helps


[Vitamin D](#) prevents autoimmunity by targeting the mechanisms behind your PTPN22 risk variant; it increases the activity of Tregs while suppressing inflammatory Th1 and Th17 cells [\[R, R, R\]](#).


At the same time, vitamin D stimulates Th2 activity and the production of anti-inflammatory cytokines such as IL-10 [\[R\]](#).


Vitamin D deficiency has been associated with worse symptom severity in people with the following autoimmune conditions:

- IBD [\[R, R, R\]](#)
- Psoriasis [\[R, R, R, R\]](#)
- Multiple sclerosis [\[R, R, R\]](#)
- Hashimoto's disease [\[R, R, R, R\]](#)
- Vitiligo [\[R, R, R\]](#)

Supplementation with vitamin D may help in some cases.

4  **Myo-inositol**

IMPACT  1 / 5

EVIDENCE  1 / 5

How to implement

Take 2 grams of myo-inositol supplement orally twice a day, preferably with meals to enhance absorption. This regimen should be followed daily for at least 6 months to observe significant benefits.

TYPICAL STARTING DOSE

4 g

Description

Myo-inositol is a naturally occurring compound that plays a role in various cellular functions. It is used in dietary supplements and is believed to have potential benefits for mental health, hormone regulation, and insulin sensitivity.

Inositol is a type of sugar that our bodies need to function. It was considered a vitamin (vitamin B8) until it was discovered that our bodies can make it from another common sugar, glucose [\[R\]](#).

[Myo-inositol](#) is the most common form of inositol. It plays a role in [\[R\]](#), [\[R\]](#), [\[R\]](#), [\[R\]](#):

- Heart health
- Normal blood sugar levels
- Mental health
- Cognitive function
- Reproductive health

Myo-inositol can convert to [D-chiro-inositol](#) in the body. These two forms of inositol are often combined in supplements. People use them to improve blood sugar control and PCOS [\[R\]](#), [\[R\]](#), [\[R\]](#).


How it helps


Pinitol from [carob](#) can directly suppress PTPN22, and reduce the ability of risk variants to spike [TNF-alpha](#) and other inflammatory markers. As a result, it may alleviate autoimmune conditions associated with this gene [\[R\]](#).


Carob also contains different types of inositol, which suppresses inflammation and supports the function of regulatory T cells [\[R\]](#), [\[R\]](#), [\[R\]](#).

However, tiny amounts of pinitol and inositol in carob probably wouldn't make a meaningful difference and suppress PTPN22. For more pronounced effects, you may want to take [myo-inositol](#) supplements.

Preliminary studies suggest that myo-inositol, especially when combined with selenium, may help restore normal thyroid function and prevent progression to hypothyroidism in patients with Hashimoto's disease [\[R\]](#), [\[R\]](#), [\[R\]](#).

5  **CBD**

IMPACT  1/5

EVIDENCE  1/5

How to implement

Take a CBD supplement in a dosage ranging from 10 to 40 milligrams per day. Start with the lowest dose and gradually increase it based on your body's response. This can be taken orally in the form of capsules, edibles, or oil drops under the tongue. For ongoing issues, such as anxiety or chronic pain, it may be used daily, while for acute issues, it might be used as needed.

TYPICAL STARTING DOSE

10 mg

Description

CBD is a non-psychoactive compound found in cannabis plants and is used in various forms, including oils and creams. It may have potential benefits for pain relief, anxiety reduction, and overall relaxation, although more research is needed to fully understand its effects.

Cannabidiol (CBD) is a compound found in cannabis. Unlike its cousin THC, **CBD is not psychoactive and can't get you "high"** [R].

People use CBD to:

- Reduce anxiety [R, R]
- Relieve pain [R, R, R]
- Prevent seizures [R]
- Improve sleep [R]

How it helps


Cannabidiol or CBD is a potent activator of regulatory T cells. By suppressing the Th1 immune response, it prevents excessive production of inflammatory cytokines such as **IL-17** while boosting anti-inflammatory IL-10 [R, R, R, R, R].


In a study of 58 people with rheumatoid arthritis, a cannabis extract containing both CBD and THC reduced pain and increased the quality of sleep. Some research suggests that the combination of CBD and THC is more effective than CBD alone [R].


A meta-analysis of 7 studies and 298 patients with multiple sclerosis concluded that CBD improves neuropathic pain in this condition [R].

Three meta-analyses (the most recent one with 9 trials and 1128 patients) found that an oral spray combining CBD and THC improves neuropathic pain and spasticity in patients with multiple sclerosis [R, R, R].

Please note: *CBD can interact with medications and raise the levels of liver enzymes. It may not be safe for pregnant women. Never use CBD without consulting your doctor. Also, make sure to verify the legality of CBD in your country or state* [R, R, R, R, R].

6  **Garlic**

IMPACT  1 / 5

EVIDENCE  1 / 5

How to implement

Incorporate 1-2 cloves of raw garlic into your meals daily. This can be achieved by finely chopping or crushing the garlic and letting it sit for a few minutes before adding it to your dishes to maximize its health benefits.

Description

Garlic is a popular herb known for its potential cardiovascular benefits, including lowering blood pressure and cholesterol levels. It also possesses antimicrobial properties and may support immune function.

[Garlic](#) is a vegetable often used to flavor food. It's been a part of traditional medicine for thousands of years [\[R\]](#).

Today, people take garlic to help control their blood pressure and cholesterol [\[R\]](#).

How it helps

A number of studies have demonstrated immune-modifying benefits of [garlic](#) or its extracts. For example, garlic (or its extracts) may increase the production of IL-2, elevate the number of Tregs, and reduce the number of Th17 cells [\[R, R, R\]](#).

In a study of 70 women with rheumatoid arthritis, those who consumed supplementary garlic (500 mg/day) had improved antioxidant status, pain scores, and quality of life [\[R\]](#).

Please note: *Garlic can interact with blood thinners (such as aspirin, Plavix, and Coumadin). In addition, garlic can irritate the stomach in some people. Talk to your doctor before taking garlic [\[R, R\]](#).*

7



Avoid Exposure to Heavy Metals

IMPACT

1 / 5

EVIDENCE

1 / 5

How to implement

To avoid exposure to heavy metals, ensure you're not using cosmetic products with heavy metals, opt for organic foods to minimize pesticide exposure, and use filters for drinking water to remove possible contaminants. Check for lead-based paints in older homes and avoid cooking or storing food in uncoated metal containers. When possible, choose glass or BPA-free plastics instead.

Description

Heavy metals are elements naturally found in the environment. They are also used for agricultural, industrial, and medicinal purposes. Some can even be found in small amounts in your body [\[R, R\]](#).

Long-term exposure to high amounts of heavy metals can be harmful to your health [\[R, R\]](#).

Heavy metals that are most often linked to health problems include [\[R, R\]](#):

- Lead
- Cadmium
- Arsenic
- Chromium
- Mercury


How it helps



A 2013 study named Th17 cells “key players in heavy-metal-elicited autoimmunity”. Mercury ions over-activate Th17 and spike inflammatory IL-17 in exposed humans and animals [\[R\]](#).

Heavy metals suppress Tregs and disrupt Th1/Th2 balance, which may trigger autoimmunity in susceptible individuals [\[R, R\]](#).

In line with this, exposure to heavy metals has been associated with an increased risk of the following autoimmune conditions:

- Psoriasis [\[R\]](#)
- Rheumatoid arthritis [\[R\]](#)
- Hashimoto's disease [\[R, R\]](#)

8  **Zinc**

IMPACT  **EVIDENCE** 

How to implement

Take a 15 mg zinc supplement daily, ideally with a meal to enhance absorption.

TYPICAL STARTING DOSE

15 mg

Description

Zinc is an essential mineral found in various foods, including meat, dairy, and nuts. It is crucial for immune function, wound healing, DNA synthesis, and maintaining healthy skin and nails. Zinc supplements are sometimes used to support immune health and manage zinc deficiencies.

[Zinc](#) is an essential mineral. Your body needs it to [\[R, R\]](#):

- Defend against disease
- Protect DNA from damage
- Heal wounds
- Control blood sugar

Some of the best sources of zinc include **shellfish, pork, beef, and beans**. It is also available as a supplement [\[R\]](#).

Adults should get **8-11 mg of zinc** per day [\[R\]](#).

How it helps

[Zinc](#) is well known to stimulate the production of IL-2. In addition, studies have suggested that its deficiency may promote Th17 activity while lowering the activity of Tregs [\[R, R, R\]](#).

Low zinc levels have been associated with the following autoimmune conditions:

- Rheumatoid arthritis [\[R, R, R\]](#)
- Lupus [\[R, R, R\]](#)

9  **Low-Level Laser Therapy (LLLT)**

IMPACT 1 / 5 **EVIDENCE** 1 / 5

How to implement

Use a low-level laser therapy device, as directed by its manual or a healthcare professional, on the affected area. Generally, treatment involves applying the laser for a specified duration, often between 30 seconds to several minutes, per treatment area. Sessions can be conducted 2-3 times per week for a period of 4-12 weeks, depending on the condition being treated and the device used.

TYPICAL STARTING DOSE**30 seconds**

Description

Low-level laser therapy (LLLT), also known as photobiomodulation, is a type of light therapy that uses low-intensity lasers to promote healing. It has been shown to be effective in treating a variety of conditions, including pain, inflammation, and wound healing.

How it helps

Low-level laser therapy (LLLT) also combats autoimmunity via PTPN22-related mechanisms. It can boost Tregs, fix the Th1/Th2 imbalance, and reduce inflammatory cytokines such as IL-1beta [[R](#), [R](#), [R](#)].

Studies suggest that low-level laser therapy is effective in improving thyroid function, reducing autoimmunity, and enhancing thyroid ultrasound features in patients with autoimmune thyroid disorders, particularly chronic autoimmune thyroiditis [[R](#), [R](#), [R](#), [R](#)].

Next Steps

Remember, your genes only tell one important part of your health story!

Now that you've seen your DNA-based results for this health topic, let's take a look at other contributing factors.

Your lab results

Your lab results are impacted by the combined effect of your genes, environment and lifestyle.

Lab tests will give you the best picture of your current health status, while your genes provide insight into your health predispositions and which recommendations are best for you.

