

BCHE (OP Sensitivity / Neurodegeneration)

Gene Report

REPORT CATEGORIES —



Sample Client

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Personal information

NAME

Sample Client

SEX AT BIRTH

Male

HEIGHT

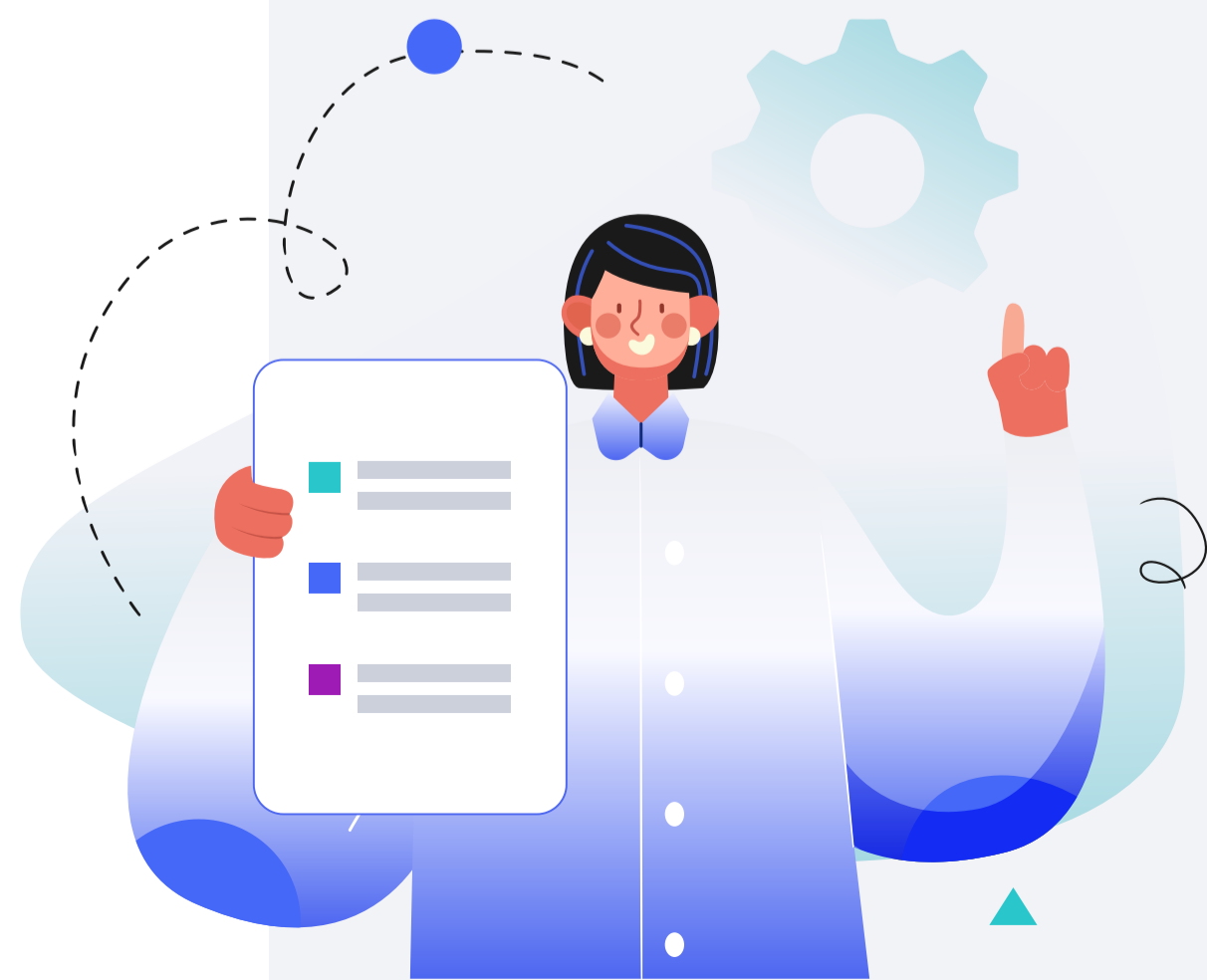
5ft 5" 165cm

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.



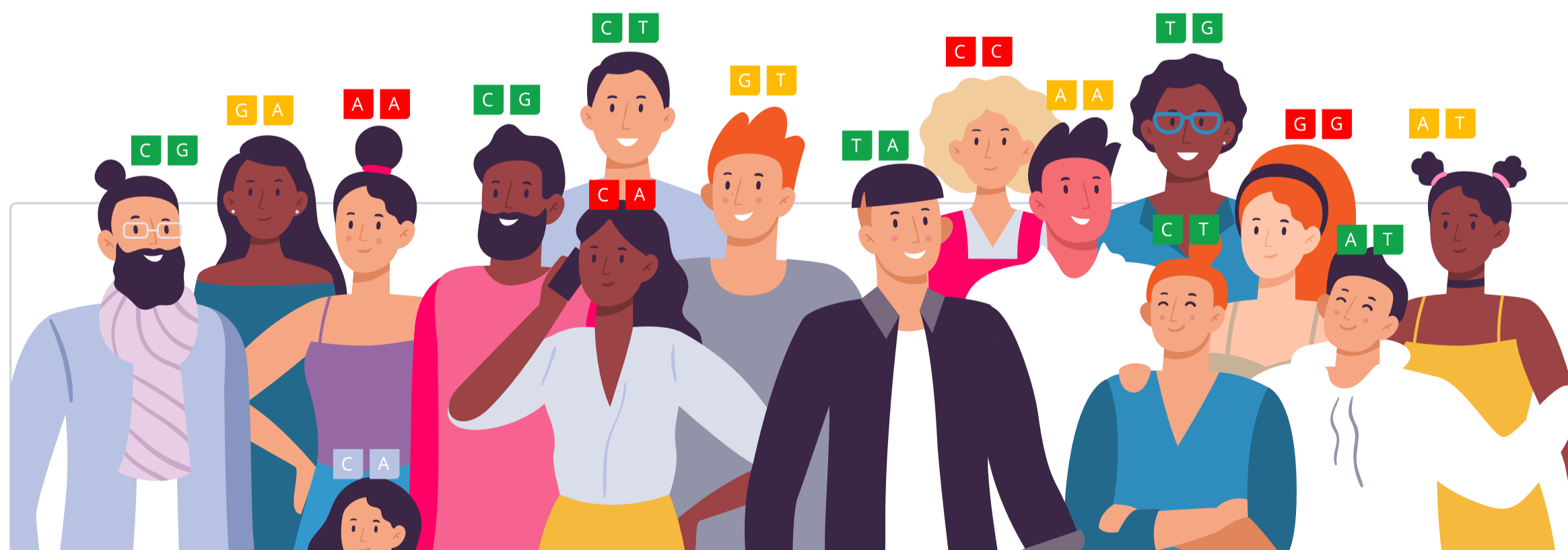
Similarly, our Trait Reports look at how your DNA influences your traits.



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 [BCHE](#) gene encodes the pseudocholinesterase enzyme, also known as butyrylcholinesterase. This enzyme contributes to the breakdown of the neurotransmitter acetylcholine in part directly, but mainly by inactivating substances that inhibit the main enzyme responsible for degrading it (acetylcholinesterase). Some of these substances include [\[R\]](#), [\[R\]](#), [\[R\]](#):

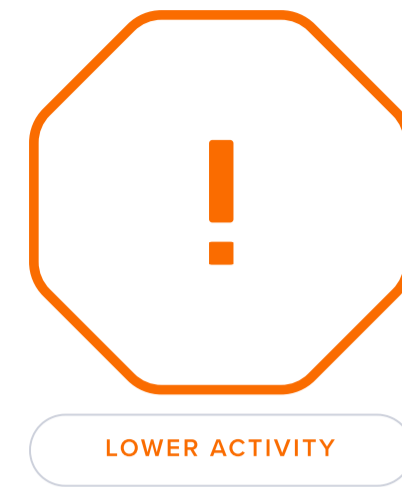
- Choline esters (muscle relaxant drugs)
- Organophosphate pesticides
- Nerve agents (e.g., sarin gas)
- Alkaloids from nightshades (e.g, solanine)

BCHE expression is highest in the liver, followed by belly fat, esophagus, colon, Fallopian tube, uterus, cervix, and lungs [\[R\]](#).

Mutations in this gene have been associated with pseudocholinesterase deficiency, a genetic condition with inefficient choline ester drugs, leading to abnormally prolonged effects of these drugs [\[R\]](#).

BCHE Genetics

The best-characterized *BCHE* polymorphism is [rs1803274](#) (Ala567Thr), commonly referred to as the ‘K-variant’ (for ‘Kalow’). Its minor ‘T’ allele reduces BCHE activity by 11-33% and has been associated with [\[R\]](#), [\[R\]](#), [\[R\]](#):



Predisposed to lower BCHE activity based on 8 genetic variants we looked at

Your top variants that most likely impact your genetic predisposition:

GENE	SNP	GENOTYPE
BCHE	rs1803274	TC
BCHE	rs3495	CT
BCHE	rs1126680	TC
BCHE	rs2668207	CT
BCHE	rs2048493	GC
BCHE	rs1799807	TT
BCHE	rs28933390	CC
BCHE	rs28933389	GG

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

- Increased risk of Parkinson’s disease in people exposed to pesticides [\[R\]](#)
- Higher triglyceride levels in people exposed to pesticides [\[R\]](#)
- Prolonged effects of the muscle relaxant succinylcholine [\[R\]](#)
- Higher levels of pro-inflammatory cytokines in workers exposed to toxic chemicals [\[R\]](#)
- Increased risk of Alzheimer’s disease, and impaired cognition and decreased response to rivastigmine in patients with this disease [\[R\]](#), [\[R\]](#), [\[R\]](#)
- Increased risk and worse prognosis of breast cancer [\[R\]](#)
- Slightly increased risk of hyperhidrosis (when combined with rs1126680) [\[R\]](#)
- Increased risk of major depressive disorder [\[R\]](#)
- Increased risk of obesity in adults but decreased risk in adolescents [\[R\]](#), [\[R\]](#)
- Increased risk of ischemic stroke in CKD patients [\[R\]](#)
- Increased risk of coronary in-stent restenosis after heart surgery [\[R\]](#)
- Lower triglyceride levels in obese women [\[R\]](#)
- Increased risk of crack cocaine use [\[R\]](#)

Another well-researched polymorphism is [rs3495](#). Its minor ‘A’ allele may decrease BCHE activity and has been associated with [\[R\]](#):

- Increased risk of organophosphate pesticide toxicity [\[R\]](#)
- Increased risk of obesity [\[R\]](#), [\[R\]](#)
- Higher triglyceride levels [\[R\]](#), [\[R\]](#)
- Slightly increased risk of substance abuse disorder [\[R\]](#)
- Decreased risk of major depressive disorder in women [\[R\]](#)

The rare ‘C’ allele of [rs1799807](#) (Asp70Gly), also known as the ‘A-variant’ (for ‘atypical’), may decrease BCHE activity even more than the K-variant. This variant has been associated with [\[R\]](#), [\[R\]](#):

- Prolonged effects of the muscle relaxant succinylcholine [\[R\]](#)
- Increased risk of leprosy [\[R\]](#)

Two other polymorphisms, commonly known as *F1* and *F2*, are [rs28933389](#) and [rs28933390](#). Their minor 'A' alleles may decrease BCHE activity and have been associated with prolonged effects of the muscular relaxant succinylcholine [\[R\]](#).

The T' allele of [rs1126680](#) lowers BCHE activity and has been associated with [\[R, R, R\]](#):

- Increased risk of hyperhidrosis (in combination with the K-variant) [\[R\]](#)
- Increased risk of Alzheimer's disease [\[R\]](#)
- Decreased risk of obesity in adolescents [\[R\]](#)
- Higher triglyceride levels in obese adolescents [\[R\]](#)
- Decreased risk of Parkinson's disease [\[R\]](#)

Finally, a study of 287 farmworkers exposed to organophosphate pesticides identified two variants that also reduce BCHE activity, but only by 4-9.5%: the 'C' allele of [rs2668207](#) and the 'G' allele of [rs2048493](#). These variants are usually inherited together, so you will most likely have both or neither of them [\[R\]](#).