

☆ Addison's disease (Eriksson, 2021)

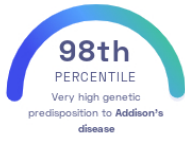
Daniel Eriksson, et al.
Nature Communications

Autoimmunity Hormones

STUDY SUMMARY

Discovery of 9 genetic variants associated with Addison's disease, a condition where the adrenal glands don't produce enough hormones.

YOUR RESULT



STUDY DESCRIPTION

The adrenal glands are acorn-sized glands located on the top of the kidneys. They produce multiple hormones including cortisol, which helps regulate the body's metabolism and also plays a role in the response to stress. If the adrenal glands become damaged, they may not be able to produce sufficient amounts of hormones. This can cause Addison's disease, which has symptoms including fatigue, weakness, and low blood sugar. If left untreated, it can become life-threatening. This genome-wide association study looked at over 5,300 individuals of European ancestry and found 9 regions of the genome associated with Addison's disease. Many of the variants are near genes that play a role in the immune system. For example, the CTLA4 gene helps to keep the immune system under control, and the AIRE gene normally works to prevent the immune system from attacking the body's own organs and

tissue.

DID YOU KNOW?

Addison's disease can be effectively treated by supplementation with hormones. However, the dose of these hormones needs to be adjusted regularly and in response to stress levels or various medical conditions.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to Addison's disease we summed up the effects of genetic variants that were linked to Addison's disease in the [study that this report is based on](#). These variants can be found in the table below. The variants highlighted in green have **positive effect sizes** and increase your genetic predisposition to Addison's disease. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to Addison's disease. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to Addison's disease. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for Addison's disease to be 5.43**. To determine whether your score is high or low, we compared it to the scores of 5,000 other Nebula Genomics users. We found that your polygenic score for Addison's disease is in the **98th percentile**. This means that it is higher than the polygenic scores 98% of people. We consider this to be a **very high genetic predisposition to Addison's disease**. However, please note that genetic predispositions do not account for important non-genetic factors like lifestyle. Furthermore, the genetics of most traits has not been fully understood yet and many associations between traits and genetic variants remain unknown. For additional explanations, click on the column titles in the table below and visit our [Nebula Library tutorial](#).

VARIANT [Ⓞ]	YOUR GENOTYPE [Ⓞ]	EFFECT SIZE [Ⓞ]	VARIANT FREQUENCY [Ⓞ]	SIGNIFICANCE [Ⓞ]
rs2476601_A	A / A	0.55 (↑)	11%	6.30×10^{-17}
rs11571303_G	G / G	0.33 (↑)	61%	5.00×10^{-11}
rs1464510_A	C / C	0.31 (-)	42%	7.30×10^{-12}
rs3998178_T	C / C	1.79 (-)	19%	3.50×10^{-179}
rs10806425_A	C / C	0.52 (-)	37%	2.80×10^{-27}
rs7137828_C	C / C	0.26 (↑)	46%	4.90×10^{-8}
rs8112143_A	NA	0.63 (-)	5%	1.40×10^{-8}
rs11203203_A	G / G	0.30 (-)	35%	8.60×10^{-10}
rs74203920_T	NA	1.23 (-)	2%	9.00×10^{-26}
rs2075876_G	G / G	0.77 (↑)	90%	7.80×10^{-14}

N/A indicates variants that could not be imputed using the 1000 genomes project datasets and variants that have a frequency of < 5%. Your genome was sequenced at 30x/100x coverage and is not imputed. However, to calculate percentiles, we need to compare your data with other users imputed data. To make the data comparable, we need to exclude some of the variants from your data.



Fatigue and Lethargy



Low Mood or Irritability



Weight loss



Muscle Weakness

Common symptoms of Addison's disease.