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Autoimmune thyroid disease (Saevarsdottir, 2020)

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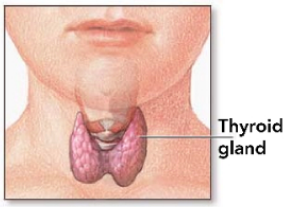
Autoimmunity | Thyroid



The thyroid gland produces hormones that regulate protein, fat, and carbohydrate metabolism.

STUDY SUMMARY

Identification of 99 genetic variants associated with autoimmune thyroid disease.

YOUR RESULT

8th
PERCENTILE
Low genetic predisposition to **autoimmune thyroid disease**

STUDY DESCRIPTION

The human immune system evolved to defend our bodies against disease-causing microorganisms. However, sometimes a person's immune system may attack the body's own cells, causing an autoimmune disease. When the immune system attacks and destroys the thyroid gland, a bowtie-shaped, hormone-producing organ in the neck, it can slow the heartbeat, cause weight gain, and lead to feeling cold, tired, and depressed. This study examined genetic data of over 30,000 patients with autoimmune thyroid disease and over 700,000 healthy controls (European ancestry) and discovered 99 genetic variants, 84 of which are novel, that are associated with autoimmune thyroid disease. One of the discovered variants reduces the production of the FLT3 *receptor*. In response, the body produces more of the signaling molecule that binds to the FLT3 *receptor* protein, which affects the function of the immune system and increases the risk of autoimmune thyroid disease.

DID YOU KNOW?

Hashimoto's disease and Grave's disease are both caused by the immune system attacking the thyroid gland. In Hashimoto's disease, the immune system kills thyroid cells, leading to reduced function of the thyroid gland. In Grave's disease, the immune system actually stimulates the thyroid cells which results in overproduction of thyroid hormones.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to autoimmune thyroid disease we summed up the effects of genetic variants that were linked to autoimmune thyroid 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 autoimmune thyroid disease. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to autoimmune thyroid disease. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to autoimmune thyroid disease. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for autoimmune thyroid disease to be -0.10**. 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 autoimmune thyroid disease is in the **8th percentile**. This means that it is higher than the polygenic scores 8% of people. We consider this to be a **low genetic predisposition to autoimmune thyroid 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
rs9272426_G	A / G	0.27 (↑)	45%	1.40×10^{-195}
rs2476601_A	G / G	0.36 (-)	10%	2.20×10^{-160}
rs3184504_T	T / C	0.21 (↑)	46%	9.40×10^{-115}
rs925489_C	T / T	-0.22 (-)	33%	8.50×10^{-110}
rs231775_G	A / G	0.17 (↑)	39%	1.60×10^{-74}
rs371058848_G	/	0.22 (-)	9%	3.80×10^{-62}
rs654537_G NEW	G / A	-0.13 (↓)	38%	5.10×10^{-42}
rs7568275_G NEW	C / C	0.14 (-)	23%	4.50×10^{-39}
rs13080163_T NEW	T / T	0.12 (↑)	44%	1.10×10^{-37}
rs2757041_G	G / G	-0.13 (↓)	35%	1.10×10^{-37}
rs4409785_C NEW	T / T	0.14 (-)	17%	2.20×10^{-31}
rs71508903_T NEW	C / T	0.13 (↑)	19%	2.30×10^{-31}
rs229527_A	C / A	0.10 (↑)	42%	2.90×10^{-30}
rs6535628_G	G / G	-0.13 (↓)	22%	5.40×10^{-30}
rs7090530_C NEW	C / A	-0.09 (↓)	40%	2.10×10^{-26}
rs3764022_G NEW	C / G	-0.09 (↓)	33%	2.20×10^{-24}
rs76428106_C NEW	NA	0.38 (-)	1%	2.40×10^{-24}
rs17364832_G NEW	T / G	0.10 (↑)	27%	2.10×10^{-23}
rs11052754_A NEW	T / T	-0.11 (-)	32%	3.50×10^{-22}
rs11675342_T	C / T	0.09 (↑)	42%	1.40×10^{-21}
rs2921071_C NEW	C / C	-0.08 (↓)	48%	2.50×10^{-21}
rs1534430_T NEW	T / T	-0.08 (↓)	39%	3.10×10^{-20}
rs4293777_C NEW	G / C	0.08 (↑)	47%	2.30×10^{-19}
rs397746317_T NEW	/	0.10 (-)	24%	7.40×10^{-19}
rs6914622_T	G / T	0.09 (↑)	32%	2.20×10^{-18}
rs7173665_C NEW	T / T	0.09 (-)	27%	9.40×10^{-18}
rs10742340_C NEW	T / C	0.08 (↑)	39%	1.50×10^{-17}
rs10686842_T NEW	TAAA / TAAA	-0.08 (-)	37%	4.40×10^{-17}
rs2445610_G NEW	A / G	-0.07 (↓)	37%	1.30×10^{-16}
rs61759532_T NEW	C / C	0.09 (-)	24%	1.30×10^{-16}
rs11079788_T NEW	C / T	0.09 (↑)	22%	2.10×10^{-16}
rs71198528_C NEW	C / C	-0.07 (↓)	46%	5.10×10^{-16}
rs12582330_G NEW	T / T	0.08 (-)	28%	7.10×10^{-16}
rs76226393_T NEW	G / G	0.09 (-)	74%	8.80×10^{-16}
rs78534766_A NEW	NA	0.38 (-)	1%	9.70×10^{-16}
rs7441808_G NEW	A / G	0.08 (↑)	29%	9.80×10^{-16}
rs9533118_G NEW	G / G	-0.07 (↓)	46%	9.90×10^{-16}

rs1990760_C	C / T	-0.07 (↓)	41%	1.10×10^{-14}
rs1050976_T	C / T	0.01 (↑)	52%	2.50×10^{-14}
rs2123340_G	A / A	0.07 (–)	36%	5.80×10^{-14}
rs244688_T	C / C	0.10 (–)	14%	1.10×10^{-13}
rs34205992_T	TA / TA	0.07 (–)	41%	1.20×10^{-13}
rs6457834_C	A / A	-0.07 (–)	32%	5.10×10^{-13}
rs34536443_C	NA	-0.16 (–)	4%	8.30×10^{-13}
rs35667974_C	NA	-0.24 (–)	2%	8.80×10^{-13}
rs4841567_G	G / G	-0.06 (↓)	48%	1.20×10^{-12}
rs61382_A	/	0.09 (–)	16%	1.20×10^{-12}
rs2069556_A	G / G	0.07 (–)	39%	1.40×10^{-12}
rs2745803_G	A / A	-0.08 (–)	21%	1.50×10^{-12}
rs146750254_C	NA	0.20 (–)	4%	2.00×10^{-12}
rs6867654_A	G / G	0.07 (–)	39%	2.90×10^{-12}
rs3775291_T	C / T	-0.07 (↓)	29%	5.90×10^{-12}
rs301802_T	A / A	0.06 (–)	42%	7.40×10^{-12}
rs13250295_T	C / C	-0.06 (–)	30%	7.51×10^{-12}
rs10748781_C	C / A	0.06 (↑)	43%	7.60×10^{-12}
rs911760_A	C / A	0.08 (↑)	21%	9.40×10^{-12}
rs9494389_C	T / T	0.07 (–)	31%	9.50×10^{-12}
rs9878908_C	T / T	0.08 (–)	20%	9.90×10^{-12}
rs926103_T	T / C	0.01 (↑)	35%	1.00×10^{-11}
rs202157095_INS	T / TGAAAAGG	0.10 (–)	10%	1.10×10^{-11}
rs6505765_G	C / G	0.07 (↑)	34%	1.90×10^{-11}
rs12117927_A	C / A	0.06 (↑)	48%	2.00×10^{-11}
rs2271194_A	T / T	0.06 (–)	43%	2.00×10^{-11}
rs2234167_A	G / G	0.09 (–)	13%	2.10×10^{-11}
rs114378220_T	C / C	0.11 (–)	7%	2.10×10^{-11}
rs11714843_A	T / A	0.08 (↑)	18%	2.50×10^{-11}
rs7831557_G	G / G	-0.06 (↓)	49%	2.80×10^{-11}
rs7251_G	G / G	-0.06 (↓)	33%	3.10×10^{-11}
rs12697352_A	G / A	-0.06 (↓)	33%	3.90×10^{-11}
rs3784099_A	G / G	-0.06 (–)	29%	3.90×10^{-11}
rs1991797_T	G / G	-0.06 (–)	32%	4.70×10^{-11}
rs763361_T	T / C	0.06 (↑)	48%	5.10×10^{-11}
rs2823272_A	A / A	-0.06 (↓)	32%	5.10×10^{-11}
rs1257920_A	A / A	-0.06 (↓)	52%	5.20×10^{-11}
rs11720041_T	C / T	0.08 (↑)	17%	9.70×10^{-11}
rs11898293_C	T / T	-0.06 (–)	49%	1.30×10^{-10}
rs2111567_G	A / A	-0.06 (–)	45%	1.43×10^{-10}
rs61776678_A	G / A	-0.06 (↓)	40%	1.50×10^{-10}
rs1214598_A	G / G	0.06 (–)	37%	2.50×10^{-10}
rs9683415_A	A / G	0.06 (↑)	33%	3.10×10^{-10}
rs4320727_G	G / A	-0.06 (↓)	38%	3.30×10^{-10}
rs111352680_A	G / A	-0.06 (↓)	26%	3.90×10^{-10}
rs7218886_G	T / T	-0.08 (–)	16%	4.40×10^{-10}
rs72922276_A	G / G	-0.09 (–)	11%	4.60×10^{-10}
rs12114596_T	C / C	0.06 (–)	39%	4.90×10^{-10}
rs7428218_C	T / C	0.08 (↑)	14%	5.30×10^{-10}
rs221796_C	G / G	-0.09 (–)	11%	7.40×10^{-10}
rs7758816_C	T / T	-0.05 (–)	49%	7.90×10^{-10}
rs1561924_A	G / G	-0.08 (–)	13%	8.02×10^{-10}
rs1045216_A	A / A	-0.05 (↓)	40%	8.30×10^{-10}
rs607404_T	A / T	-0.06 (↓)	28%	1.00×10^{-9}
rs12756886_C	T / T	0.08 (–)	12%	3.10×10^{-9}
rs1079418_G	G / G	-0.06 (↓)	31%	3.50×10^{-9}
rs7594497_C	T / T	0.10 (–)	6%	4.10×10^{-9}
rs12793348_G	A / A	-0.11 (–)	8%	5.30×10^{-9}
rs1058900_T	T / C	0.05 (↑)	42%	8.60×10^{-9}

rs35677470_A NEW	G / G	0.10 (–)	8%	8.90×10^{-9}
rs8053655_T NEW	T / T	0.12 (↑)	6%	2.30×10^{-8}
rs2681417_G NEW	G / A	0.10 (↑)	8%	3.30×10^{-8}

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.