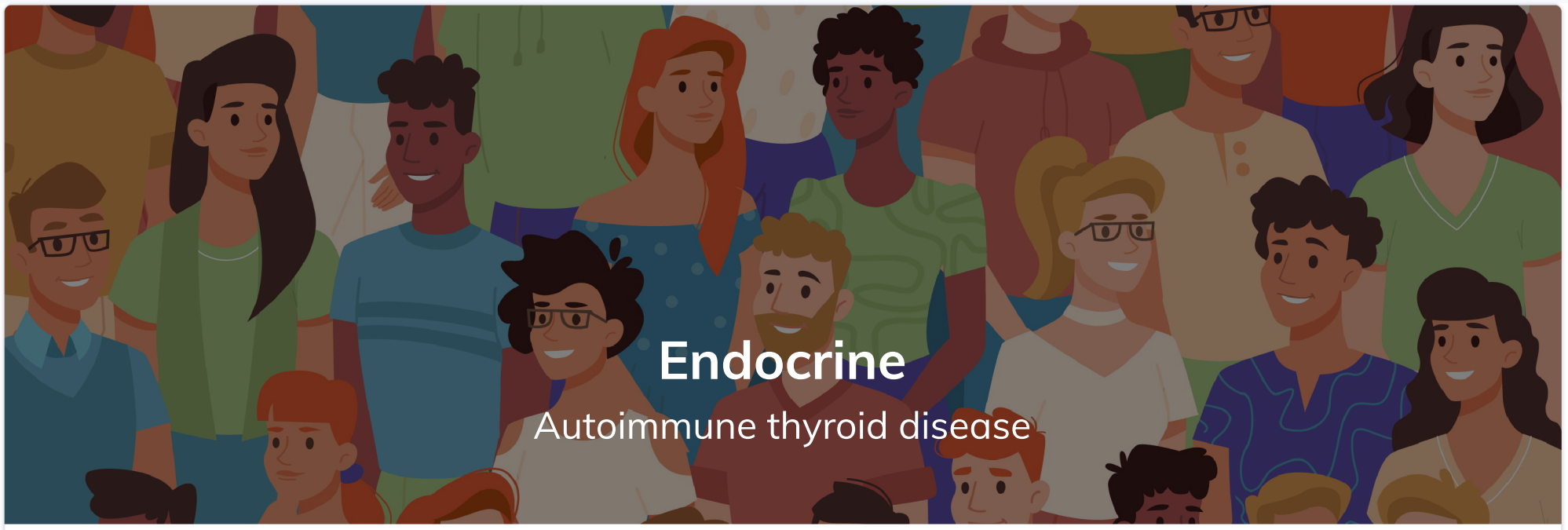
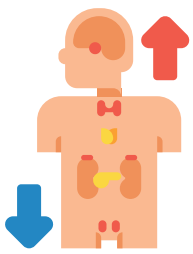


DEMO REPORT

Autoimmune thyroid disease



Autoimmune thyroid disease is the most common autoimmune disease and is highly heritable1. Here, by using a genome-wide association study of 30,234 cases and 725,172 controls from Iceland and the UK Biobank, we find 99 sequence variants at 93 loci, of which 84 variants are previously unreported2-7. A low-frequency (1.36%) intronic variant in FLT3 (rs76428106-C) has the largest effect on risk of autoimmune thyroid disease (odds ratio (OR) = 1.46, P = 2.37 × 10-24). rs76428106-C is also associated with systemic lupus erythematosus (OR = 1.90, P = 6.46 × 10-4), rheumatoid factor and/or anti-CCP-positive rheumatoid arthritis (OR = 1.41, P = 4.31 × 10-4) and coeliac disease (OR = 1.62, P = 1.20 × 10-4). FLT3 encodes fms-related tyrosine kinase 3, a receptor that regulates haematopoietic progenitor and dendritic cells. RNA sequencing revealed that rs76428106-C generates a cryptic splice site, which introduces a stop codon in 30% of transcripts that are predicted to encode a truncated protein, which lacks its tyrosine kinase domains. Each copy of rs76428106-C doubles the plasma levels of the FTL3 ligand. Activating somatic mutations in FLT3 are associated with acute myeloid leukaemia8 with a poor prognosis and rs76428106-C also predisposes individuals to acute myeloid leukaemia (OR = 1.90, P = 5.40 × 10-3). Thus, a predicted loss-of-function germline mutation in FLT3 causes a reduction in full-length FLT3, with a compensatory increase in the levels of its ligand and an increased disease risk, similar to that of a gain-of-function mutation.

Your genetic results

Based on your genetic results, your predisposition to this trait is

Higher genetic predisposition

Your genetic predisposition is higher than the 98% average person from your genetic population

Confidence Score

★★★★★ High

Disclaimer This report is intended as educational information. It is not intended to provide medical advice or be used solely by the customer in the diagnosis, cure, mitigation, treatment or prevention of disease. If you have any serious medical condition(s), including but not limited to, being over or under weight, or having diabetes or heart disease, you should not make any changes to your diet or exercise without consulting your doctor. Under no circumstances, should you make changes to your medication or other medical care without consulting your physician Find more information at this link

Scientific Details

We looked at a genetic marker (SNP) that influences whether you are more likely to have this trait. For each genetic marker, we calculated a score and compare it to the average score of your main ancestry. This approach allows us to predict how common is your genetic score compared to this population.

Genes

We have tested the following 73 genes that are associated with your genetic predisposition to this trait.

ACAP1

ArfGAP with coiled-coil, ankyrin repeat and PH domains 1

More information about this gene in GENECARDS

Location

This gene is located at the following position in your dna:

Chromosome 17

Position: (HG19) 7239848 - 7254796

SNP	Genotype
rs61759532	CT

ADCY7

adenylate cyclase 7

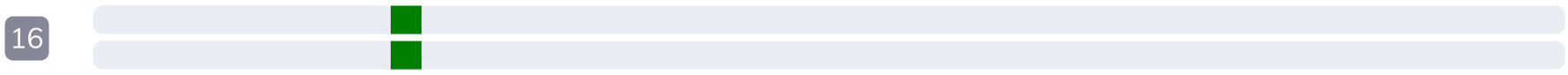
More information about this gene in GENECARDS

Location

This gene is located at the following position in your dna:

Chromosome 16

Position: (HG19) 50300462 - 50352046



SNP	Genotype
rs78534766	CC

View 71 more genes

Tested SNPs

These are the SNPs we have tested for this trait. The variants found in your genome are indicated in the column "Variant Found". The SNPs that has "--" genotype means they were not found in your DNA file. Sometimes the DNA files does not contains all the DNA markers we use for the analysis or the genotype couldn't be read by the laboratory.

Show 10 entries

Search:

SNP	Chromosome	Genotype	Variant allele	Variant allele frequency
rs12117927	1	**A A**	A	48.40%
rs926103	1	T **C**	C	35.40%
rs4320727	1	**A A**	A	62.00%
rs2476601	1	**G G**	G	9.70%
rs72922276	1	G G	A	89.10%
rs61776678	1	G G	A	59.60%
rs12756886	1	T T	C	11.80%
rs1214598	1	G G	A	37.40%
rs2234167	1	**A** G	A	13.30%
rs146750254	1	T T	C	3.90%

Showing 1 to 10 of 99 entries

Previous 1 2 3 4 5 ... 10 Next

DNA Genics always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other companies may report genotypes using the opposite strand, so the genotypes may not be the same (they have to be reverted).

References

1. FLT3 stop mutation increases FLT3 ligand level and risk of autoimmune thyroid disease (2020 Aug)
   Saevarsdottir S, Olafsdottir TA, Ivarsdottir EV, Halldorsson GH, Gunnarsdottir K, Sigurdsson A, Johannesson A, Sigurdsson JK, Juliusdottir T, Lund SH, Arnthorsson AO, Styrmisdottir EL, Gudmundsson J, Grondal GM, Steinsson K, Alfredsson L, Askling J, Benediktsson R, Bjarnason R, Geirsson AJ, Gudbjornsson B, Gudjonsson H, Hjaltason H, Hreidarsson AB, Klareskog L, Kockum I, Kristjansdottir H, Love TJ, Ludviksson BR, Olsson T, Onundarson PT, Orvar KB, Padyukov L, Sigurgeirsson B, Tragante V, Bjarnadottir K, Rafnar T, Masson G, Sulem P, Gudbjartsson DF, Melsted P, Thorleifsson G, Norddahl GL, Thorsteinsdottir U, Jonsdottir I, Stefansson K

   - **Journal**: Nature
   - **Sample size and ancestry**: 755406 individuals with european ancestry

   ★★★★ **Very strong scientific evidence**