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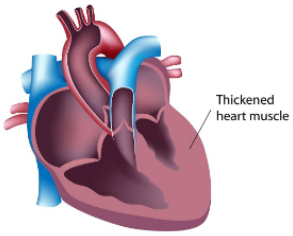
Hypertrophic cardiomyopathy (Harper, 2021)

Andrew Harper, et al.
Nature Genetics

Heart

STUDY SUMMARY

Discovery of 12 regions of the genome associated with hypertrophic cardiomyopathy, a disease that causes the heart's walls to become thicker than normal.



Hypertrophic cardiomyopathy is caused by a thickening of the heart muscle.

YOUR RESULT

Above average genetic predisposition to **hypertrophic cardiomyopathy**

STUDY DESCRIPTION

The heart pumps blood throughout the body, supplying oxygen and nutrients that are critical to sustaining life. Hypertrophic cardiomyopathy (HCM) is a condition that develops when the walls of the heart become thicker, which in turn reduces the amount of blood that can be pumped with each heartbeat. HCM can cause chest pain and abnormal heart rhythms, and can eventually contribute to strokes and heart failure. It is estimated that 1 in 500 people have HCM, though many are undiagnosed. To identify genetic factors associated with HCM, this study examined the genomes of over 50,000 individuals of different ethnicities. The researchers discovered 12 genetic variants, 11 of which have not been previously connected to HCM risk. Among the genes found linked to HCM was FHOD3. It encodes a protein that supports the formation of contracting fibers inside heart muscle cells.

DID YOU KNOW?

In the course of a day, your heart pumps around 2,000 gallons of blood.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to hypertrophic cardiomyopathy we summed up the effects of genetic variants that were linked to hypertrophic cardiomyopathy 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 hypertrophic cardiomyopathy. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to hypertrophic cardiomyopathy. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to hypertrophic cardiomyopathy. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for hypertrophic cardiomyopathy to be 2.11**. 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 hypertrophic cardiomyopathy is in the **65th percentile**. This means that it is higher than the polygenic scores 65% of people. We consider this to be an **above average genetic predisposition to hypertrophic cardiomyopathy**. 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
rs72840788_A NEW	G / G	0.42 (-)	21%	5.06×10^{-29}
rs4799426_G	A / A	0.32 (-)	35%	4.00×10^{-23}
rs1048302_T NEW	T / G	0.28 (↑)	33%	2.54×10^{-17}
rs2070458_A NEW	T / T	0.29 (-)	22%	7.12×10^{-16}
rs28768976_G NEW	G / G	0.25 (↑)	23%	4.12×10^{-12}
rs41306688_C NEW	NA	0.60 (-)	3%	1.08×10^{-11}
rs3176326_A NEW	G / G	0.25 (-)	21%	2.22×10^{-11}
rs12212795_C NEW	G / G	0.39 (-)	5%	2.51×10^{-10}
rs7210446_A NEW	A / A	0.22 (↑)	58%	6.82×10^{-10}
rs8033459_T NEW	C / C	0.19 (-)	47%	3.41×10^{-9}
rs13061705_C NEW	C / C	0.22 (↑)	69%	9.18×10^{-9}
rs7301677_C NEW	C / C	0.22 (↑)	73%	1.26×10^{-8}
rs118060942_T	NA	0.58 (-)	1%	2.35×10^{-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.