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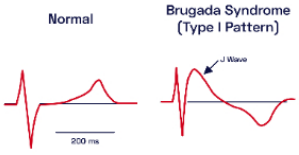
Brugada syndrome (Bezzina, 2013)

Connie Bezzina, et al.
Nature Genetics

Heart

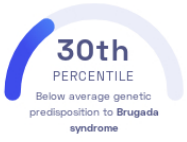


The characteristic electrocardiogram pattern of the Brugada syndrome.

STUDY SUMMARY

Identification of 3 genetic variants associated with Brugada syndrome, a rare heart disorder.

YOUR RESULT



STUDY DESCRIPTION

A healthy heart pumps blood with a regular rhythm that forms our "heartbeat". This rhythm allows blood to flow into the heart before being successfully pumped back out into the body. When the heart has an abnormal rhythm, known as arrhythmia, the heart does not pump blood as effectively which can impair the blood and oxygen supply of the body. Brugada syndrome, is a type of arrhythmia that can lead to fainting, difficulty breathing, and sudden death. To identify genetic risk factors for Brugada syndrome, this study analyzed nearly 1,500 individuals of European ancestry and found 3 variants that appear to be associated with Brugada syndrome. Two of these variants occur in genes (SCN5A and SCN10A) responsible for moving sodium in and out of cells, a process that is important for the heart's pumping action. The third identified variant is located in the HEY2 genes which is believed to play an important for the development of the heart.

DID YOU KNOW?

Healthy lifestyle choices can help prevent the development of arrhythmias. These can include adopting a low-fat diet rich in fruits and vegetables, limiting the consumption of caffeine, and avoiding stressful situations.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to Brugada syndrome we summed up the effects of genetic variants that were linked to Brugada syndrome 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 Brugada syndrome. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to Brugada syndrome. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to Brugada syndrome. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for Brugada syndrome to be 0.94**. 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 Brugada syndrome is in the **30th percentile**. This means that it is higher than the polygenic scores 30% of people. We consider this to be a **below average genetic predisposition to Brugada syndrome**. 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	COMMENTS	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs10428132_T NEW	T / G	Near SCN10A gene	0.94 (↑)	41%	1.01×10^{-68}
rs9388451_C NEW	T / T	Near HEY2 gene	0.46 (-)	50%	5.14×10^{-17}
rs11708996_C NEW	G / G	Near SCN5A gene	0.55 (-)	15%	1.02×10^{-14}