

☆ Left ventricular ejection fraction (Pirruccello, 2020)

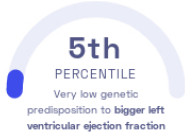
James Pirruccello, et al.
Nature Communications

Heart

STUDY SUMMARY

Identification of 22 genetic variants associated with the heart's left ventricular ejection fraction (LVEF).

YOUR RESULT



STUDY DESCRIPTION

The human heart is a muscle that pumps blood throughout the body. It consists of 4 chambers: 2 atria (left and right) and 2 ventricles (left and right). Blood that has been enriched with oxygen in the lungs enters the left atrium and then flows into the left ventricle from where it's pumped to all other parts of the body. When the left ventricle contracts, not all blood is ejected and some remains inside of the ventricle. Left ventricular ejection fraction (LVEF) is calculated by dividing the blood volume that leaves the left ventricle when the heart muscles contract by the blood volume that remains inside the left ventricle. A normal left ventricular ejection fraction (LVEF) ranges from 55% to 70%. To identify genetic variants associated with LVEF, this genome-wide association study examined over 36,000 individuals of European ancestry. The study identified 22 genetic variants including variants near genes that are responsible for the flexibility of the heart's muscle (TTN) and genes associated with the immune system. Together these variants explain ~ 31% of LVEF heritability. The study also found that a high polygenic score for LVEF was strongly correlated with the risk of dilated cardiomyopathy. This condition describes a heart muscle that is too stretched thin and cannot pump efficiently.

DID YOU KNOW?

Stress increases heart rate and blood pressure, both of which can negatively impact the health's health. Taking steps to control stress through activities such as meditation, exercise, and therapy may be effective in decreasing the risk of various heart conditions.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to bigger left ventricular ejection fraction we summed up the effects of genetic variants that were linked to bigger left ventricular ejection fraction 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 bigger left ventricular ejection fraction. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to bigger left ventricular ejection fraction. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to bigger left ventricular ejection fraction. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for bigger left ventricular ejection fraction to be -1.36**. 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 bigger left ventricular ejection fraction is in the **5th percentile**. This means that it is higher than the polygenic scores 5% of people. We consider this to be a **very low genetic predisposition to bigger left ventricular ejection fraction**. 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_G	G / G	-0.10 (↓)	79%	4.00 x 10 ⁻³²
rs1739837_C	C / T	0.07 (↑)	41%	2.90 x 10 ⁻²⁶
rs2662845_T	T / T	-0.08 (↓)	80%	5.70 x 10 ⁻²⁰
rs11710541_T ^{NEW}	T / T	-0.07 (↓)	66%	9.10 x 10 ⁻²⁰
rs3176326_G	G / G	-0.08 (↓)	80%	1.80 x 10 ⁻¹⁹
rs3807309_G	G / G	-0.08 (↓)	89%	4.10 x 10 ⁻¹⁶
rs12452367_T ^{NEW}	T / C	-0.06 (↓)	71%	1.30 x 10 ⁻¹³
rs2070458_A	T / T	0.06 (-)	20%	2.50 x 10 ⁻¹³
rs56099248_C ^{NEW}	C / C	-0.06 (↓)	81%	5.10 x 10 ⁻¹²
rs8023658_G	G / G	-0.05 (↓)	51%	4.60 x 10 ⁻¹¹
rs35999985_A ^{NEW}	A / G	0.05 (↑)	30%	3.80 x 10 ⁻¹⁰
rs5029142_T ^{NEW}	T / A	-0.04 (↓)	62%	3.90 x 10 ⁻¹⁰
rs10871753_G ^{NEW}	G / G	-0.04 (↓)	49%	6.10 x 10 ⁻¹⁰
rs10925197_C ^{NEW}	C / C	0.04 (↑)	46%	6.90 x 10 ⁻¹⁰
rs4073554_T ^{NEW}	C / C	0.04 (-)	48%	1.10 x 10 ⁻⁹
rs38029352_C ^{NEW}	T / T	-0.04 (-)	45%	1.30 x 10 ⁻⁸
rs113819537_C ^{NEW}	C / G	0.04 (↑)	75%	1.70 x 10 ⁻⁸
rs9274626_T ^{NEW}	C / C	-0.04 (-)	32%	2.70 x 10 ⁻⁸
rs2047273_T	T / T	-0.04 (↓)	68%	3.10 x 10 ⁻⁸
rs189569984_C ^{NEW}	C / C	-0.20 (↓)	99%	3.40 x 10 ⁻⁸
rs721067_T ^{NEW}	T / T	0.07 (↑)	92%	3.60 x 10 ⁻⁸
rs2503715_A ^{NEW}	A / G	-0.06 (↓)	13%	4.00 x 10 ⁻⁸