

★ Left ventricular end-diastolic volume (Pirruccello, 2020)

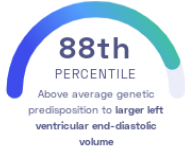
James Pirruccello, et al.  
Nature Communications

Heart

STUDY SUMMARY

Identification of 14 genetic variants associated with the left ventricular end-*diastolic* volume (LVEDV).

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. Left ventricular end-*diastolic* volume (LVEDV) is the volume of blood in a relaxed left ventricle right before it contracts and pumps blood into the body. The average LVEDV is 120mL for women and 150mL for men and can be used as a measure of heart health. This genome-wide association study examined the genomes of 36,000 individuals of European ancestry to better understand the genetic basis of LVEDV. The study identified 14 genomic regions associated with LVEDV adjusted for body-surface-area. The study also found a strong inverse relationship between the polygenic score for larger LVEDV and *hypothyroidism*.

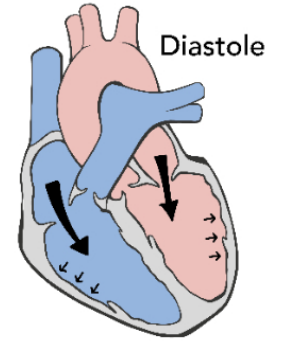
score for larger LVEDV and *hypothyroidism*.

DID YOU KNOW?

On average, the human heart pumps ~2,000 gallons (7,600 liters) of blood throughout the body every day.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to larger left ventricular end-diastolic volume we summed up the effects of genetic variants that were linked to larger left ventricular end-diastolic volume 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 larger left ventricular end-diastolic volume. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to larger left ventricular end-diastolic volume. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to larger left ventricular end-diastolic volume. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for larger left ventricular end-diastolic volume to be 0.29**. 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 larger left ventricular end-diastolic volume is in the **88th percentile**. This means that it is higher than the polygenic scores 88% of people. We consider this to be an **above average genetic predisposition to larger left ventricular end-diastolic volume**. 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.



During the diastole phase the heart muscle relaxes, the heart chambers expand and blood flows into the heart.

VARIANT <sup>Ⓞ</sup>	YOUR GENOTYPE <sup>Ⓞ</sup>	EFFECT SIZE <sup>Ⓞ</sup>	VARIANT FREQUENCY <sup>Ⓞ</sup>	SIGNIFICANCE <sup>Ⓞ</sup>
rs1873164_G	A / A	-0.07 (-)	20%	1.80 x 10 <sup>-16</sup>
rs9797817_C <span style="color: orange;">NEW</span>	C / C	-0.05 (↓)	65%	4.60 x 10 <sup>-12</sup>
rs72967533_T	T / C	-0.05 (↓)	52%	7.10 x 10 <sup>-12</sup>
rs72840788_G	G / G	0.06 (↑)	79%	1.00 x 10 <sup>-11</sup>
rs73028849_G <span style="color: orange;">NEW</span>	G / G	0.04 (↑)	66%	4.10 x 10 <sup>-9</sup>
rs1976402_G	G / G	0.04 (↑)	71%	1.10 x 10 <sup>-8</sup>
rs6458349_G <span style="color: orange;">NEW</span>	G / G	0.04 (↑)	27%	1.50 x 10 <sup>-8</sup>
rs9480737_A <span style="color: orange;">NEW</span>	A / A	-0.04 (↓)	68%	2.30 x 10 <sup>-8</sup>
rs7502466_G <span style="color: orange;">NEW</span>	G / A	0.06 (↑)	89%	2.30 x 10 <sup>-8</sup>
rs12499670_T <span style="color: orange;">NEW</span>	T / C	0.04 (↑)	40%	3.60 x 10 <sup>-8</sup>
rs10850034_T <span style="color: orange;">NEW</span>	T / A	0.04 (↑)	65%	3.90 x 10 <sup>-8</sup>