CLeft ventricular end-systolic volume (Pirruccello, 2020)

James Pirruccello, et al. Nature Communications

Heart

# STUDY SUMMARY

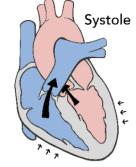
Identification of 28 genetic variants associated with the left ventricular end-systolic volume (LVESV).

#### 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-systolia volume (LVESV) is the volume of blood that remains in the left ventricle at the end of a pump. The average LVESV is 41mL for women and 58mL 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 LVESV. The study identified 28 genomic regions associated with LVESV adjusted for body-surface-area. The study also found that a high polygenic score for body-surface-area adjusted LVESV was



During the systole phase the heart muscle contracts, the heart chambers shrink and the blood is elected.

strongly correlated with the risk of dilated cardiomyopathy. This condition describes a heart muscle that is too stretched and thin and cannot pump efficiently.

## DID YOU KNOW?

The expression "laughter is the best medicine" has some truth to it. Laughter can be beneficial for your heart health, allowing arteries to relax and become more flexible, permitting easier blood flow.

### YOUR DETAILED RESULTS

To calculate your genetic predisposition to larger left ventricular end-systolic volume we summed up the effects of genetic variants that were linked to larger left ventricular end-systolic 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-systolic volume. The variants highlighted in blue have negative effects sizes and decrease your genetic predisposition to larger left ventricular end-systolic volume. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to larger left ventricular end-systolic volume to be 1.93. 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-systolic volume is in the 89th percentile. This means that it is higher than the polygenic scores 89% of people. We consider this to be an above average genetic predisposition to larger left ventricular end-systolic 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.

ra72840788_0   0 / 0   0.09 (1)   78%   170 x 10 x	variant <sup>©</sup>	YOUR GENOTYPE <sup>©</sup>	EFFECT SIZE®	VARIANT FREQUENCY <sup>©</sup>	SIGNIFICANCE <sup>®</sup>
ria2662245_T   T / T   0.09 (f)   80%   1.00 × 10 <sup>-52</sup> ria75028840_6   0 / 6   0.06 (f)   66%   6.20 × 10 <sup>-18</sup> ria57028840_6   0 / 6   0.06 (-)   20%   7.50 × 10 <sup>-18</sup> ria5776208_6   0 / 6   0.07 (f)   80%   1.20 × 10 <sup>-18</sup> ria5776328_6   0 / 6   0.07 (f)   80%   1.20 × 10 <sup>-18</sup> ria54673806_C   0 / 7   0.06 (f)   71%   1.50 × 10 <sup>-18</sup> ria5063213_T   T / 6   0.04 (f)   62%   7.60 × 10 <sup>-18</sup> ria5063213_T   T / 6   0.04 (f)   62%   7.60 × 10 <sup>-18</sup> ria7060230_C   0 / 7   0.04 (f)   62%   7.60 × 10 <sup>-18</sup> ria7060230_C   0 / 7   0.04 (f)   62%   6.00 × 10 <sup>-19</sup> ria106210_A_T   0 / 7   0.04 (f)   62%   6.00 × 10 <sup>-19</sup> ria106220_A_T   7 / 7   0.04 (f)   62%   6.00 × 10 <sup>-19</sup> ria10620_A_T   7 / 7   0.04 (f)   62%   2.10 × 10 <sup>-19</sup> ria10620_A_T   7 / 7   0.	rs72840788_G	G / G	0.09 (1)	79%	1.70 x 10 <sup>-28</sup>
re27028889_6	rs945425_T	T/C	-0.07 (↓)	32%	8.50 x 10 <sup>-23</sup>
ref50000t_6   A / A   -0.06 (-)   20%   7.50 x 10^-8     res176320_6   9 / 9   0.07 (t)   80%   180 x 10^-4     res10421891_A   A / A   -0.06 (4)   66%   120 x 10^-8     res12482387_T   7 / C   0.06 (t)   71%   180 x 10^-8     res263283_T   7 / C   0.07 (t)   84%   2.70 x 10^-8     res76602300_C   0 / C   0.04 (t)   62%   7.60 x 10^-11     res1063218_T   7 / C   0.05 (t)   80%   4.40 x 10^-10     res76602300_C   0 / C   0.05 (t)   80%   4.40 x 10^-10     res1063218_T   7 / C   0.04 (-)   45%   6.00 x 10^-10     res1063200_C   7 / C   0.04 (t)   52%   6.50 x 10^-10     res1063200_C   7 / C   0.04 (t)   99%   2.10 x 10^-10     res1063200_C   7 / C   0.04 (t)   50%   2.20 x 10^-10     res1063200_C   7 / T   0.04 (t)   50%   2.00 x 10^-10     res8063200_C   7 / T   0.04 (t) <th< td=""><td>rs2562845_T</td><td>T/T</td><td>0.09 (1)</td><td>80%</td><td>1.60 x 10<sup>-22</sup></td></th<>	rs2562845_T	T/T	0.09 (1)	80%	1.60 x 10 <sup>-22</sup>
180	rs73028849_G 💮	G / G	0.06 (1)	66%	6.20 x 10 <sup>-19</sup>
ra10421891_A   A/A   -0.05 (4)   66%   1.20 x 10 '8'     rs12452397_T   T/C   0.06 (f)   71%   1.80 x 10 '8'     rs34373305_C   0/C   0.07 (f)   84%   2.70 x 10 '8'     rs8063213_T   T/B   0.04 (f)   62%   7.80 x 10 '1'     rs79602300_C   C/C   0.05 (f)   80%   4.40 x 10 '0'     rs108210_T   C/C   -0.04 (-)   45%   6.40 x 10 '0'     rs1102305_A   A/A   0.04 (f)   62%   6.50 x 10 '0'     rs1102805_A   A/A   0.04 (f)   99%   2.10 x 10 '0'     rs188660884_C   C/C   0.21 (f)   99%   2.10 x 10 '0'     rs149883_T   T/T   0.04 (f)   69%   2.80 x 10 '9'     rs149883_T   T/C   0.04 (f)   88%   4.00 x 10 '9'     rs17785_G   G/A   0.07 (f)   88%   4.00 x 10 '9'     rs1087785_G   G/B   0.04 (f)   49%   6.00 x 10 '9'     rs10804997_G   G/B   0.03 (f)   99%   1.80 x 10 '	rs5760061_G	A / A	-0.06 (-)	20%	7.50 × 10 <sup>-15</sup>
re12452367_T   T / C   0.06 (†)   71%   180 x 10 - 13     re34373805_C   C / C   0.07 (†)   84%   2.70 x 10 - 13     re8063213_T   T / G   0.04 (†)   62%   7.60 x 10 - 11     re378602300_C   C / C   0.05 (†)   80%   4.40 x 10 - 10     re378602300_C   C / C   0.04 (-)   46%   6.40 x 10 - 10     re31002306_A   A / A   0.04 (†)   62%   6.60 x 10 - 10     re31002306_A   A / A   0.04 (†)   62%   6.60 x 10 - 10     re31002306_A   T / T   0.04 (-)   42%   2.10 x 10 - 8     re3802661_C   T / T   0.04 (†)   69%   2.80 x 10 - 9     re3409813_T   T / T   0.04 (†)   69%   2.80 x 10 - 9     re3409813_T   T / T   0.04 (†)   88%   4.80 x 10 - 9     re3409813_T   T / C   0.04 (†)   73%   6.90 x 10 - 9     re31087753_G   6 / G   0.04 (†)   98%   1.10 x 10 - 8     re3108047053_G   6 / G   0.04 (†)	rs3176326_G 💮	G / G	0.07 (1)	80%	1.80 × 10 <sup>-14</sup>
rs34373806_C   C / C   0.07 (†)   84%   2.70 × 10 · 5     rs8063213_T   T / 6   0.04 (†)   82%   7.60 × 10 · 1     rs79602300_C   C / C   0.05 (†)   80%   4.40 × 10 · 10     rs1962104_T   C / C   -0.04 (-)   45%   6.60 × 10 · 10     rs1023069_A   A / A   0.04 (†)   52%   6.50 × 10 · 10     rs19866984_C   C / C   0.21 (†)   99%   2.10 × 10 · 9     rs1498813_T   T / T   0.04 (†)   59%   2.80 × 10 · 9     rs2202456_G   G / A   0.07 (†)   88%   4.60 × 10 · 9     rs11748963_T   T / C   0.04 (†)   73%   5.90 × 10 · 9     rs11690497_G   G / G   0.04 (†)   49%   6.00 × 10 · 9     rs119003861_C   G / G   0.03 (†)   99%   1.10 × 10 · 8     rs19003861_C   G / G   0.03 (†)   99%   1.60 × 10 · 8     rs19003861_C   G / G   0.03 (†)   99%   1.60 × 10 · 8     rs19003861_C   G / G   0.04 (-)	rs10421891_A 💮	A / A	-0.05 (↓)	65%	1.20 x 10 <sup>-13</sup>
rs8065213_T   T/6   0.04 (†)   62%   7.60 x 10 <sup>-14</sup> rs79602300_C   C/C   0.05 (†)   80%   4.40 x 10 <sup>-10</sup> rs1962104_T   C/C   -0.04 (-)   45%   6.40 x 10 <sup>-10</sup> rs11023069_A   A/A   0.04 (†)   62%   6.60 x 10 <sup>-10</sup> rs189669984_C   C/C   0.21 (†)   99%   2.10 x 10 <sup>-9</sup> rs1499813_T   T/T   0.04 (-)   42%   2.10 x 10 <sup>-9</sup> rs2302456_G   G/A   0.07 (†)   88%   4.60 x 10 <sup>-9</sup> rs11748963_T   T/C   0.04 (†)   49%   6.00 x 10 <sup>-9</sup> rs116904997_G   G/G   0.04 (†)   49%   6.00 x 10 <sup>-8</sup> rs1190093681_C   C/C   0.37 (†)   98%   1.10 x 10 <sup>-8</sup> rs286037_G   G/A   -0.04 (-)   68%   1.90 x 10 <sup>-8</sup>	rs12452367_T 💮	T/C	0.06 (1)	71%	1.80 x 10 <sup>-13</sup>
rs79602300_C   C / C   0.06 (†)   80%   4.40 x 10 <sup>-10</sup> rs1962104_T   C / C   -0.04 (-)   45%   6.40 x 10 <sup>-10</sup> rs1962104_T   A / A   0.04 (†)   62%   6.60 x 10 <sup>-10</sup> rs1989669984_C   C / C   0.21 (†)   99%   2.10 x 10 <sup>-9</sup> rs9892661_C   T / T   0.04 (-)   42%   2.10 x 10 <sup>-9</sup> rs1499813_T   T / T   0.04 (†)   69%   2.80 x 10 <sup>-9</sup> rs4280436_G   G / A   0.07 (†)   88%   4.60 x 10 <sup>-9</sup> rs17749963_T   T / C   0.04 (†)   73%   6.90 x 10 <sup>-9</sup> rs10871763_G   G / G   0.04 (†)   49%   6.00 x 10 <sup>-9</sup> rs190093681_C   G / G   -0.13 (4)   98%   1.10 x 10 <sup>-8</sup> rs709208_A   • / G   -0.04 (-)   68%   1.90 x 10 <sup>-8</sup> rs288637_G   G / A   -0.04 (-)   49%   2.90 x 10 <sup>-8</sup>	rs34373805_C	C/C	0.07 (1)	84%	2.70 x 10 <sup>-13</sup>
res1982104_T   C / C   -0.04 (-)   45%   6.40 x 10 <sup>-10</sup> res1023069_A   A / A   0.04 (1)   62%   6.50 x 10 <sup>-10</sup> res198669984_C   C / C   0.21 (1)   99%   2.10 x 10 <sup>-9</sup> res9892661_C   T / T   0.04 (-)   42%   2.10 x 10 <sup>-9</sup> res1498913_T   T / T   0.04 (1)   69%   2.80 x 10 <sup>-9</sup> res2302465_G   G / A   0.07 (1)   88%   4.60 x 10 <sup>-9</sup> res17478963_T   T / C   0.04 (1)   73%   5.90 x 10 <sup>-9</sup> res10871763_G   G / G   0.04 (1)   49%   6.00 x 10 <sup>-9</sup> res109093681_C   G / G   0.03 (1)   98%   1.10 x 10 <sup>-8</sup> res109093681_C   C / C   0.37 (1)   > 99%   1.60 x 10 <sup>-8</sup> res286037_G   G / A   -0.04 (-)   68%   1.90 x 10 <sup>-8</sup>	rs8063213_T 🌼	T/G	0.04 (1)	62%	7.60 × 10 <sup>-11</sup>
res11023059_A A / A 0.04 (1) 62% 6.50 x 10 -10   res189669984_C C / C 0.21 (1) 99% 2.10 x 10 -8   res9892651_C T / T 0.04 (-) 42% 2.10 x 10 -9   res1499813_T T / T 0.04 (1) 59% 2.80 x 10 -9   res2302455_G G / A 0.07 (1) 88% 4.60 x 10 -9   res11748963_T T / C 0.04 (1) 73% 5.90 x 10 -9   res10871763_G G / G 0.04 (1) 49% 6.00 x 10 -8   res116904997_G G / G 0.03 (1) 98% 1.10 x 10 -8   res190093681_C C / C 0.37 (1) >99% 1.60 x 10 -8   res2880037_G 6 / A -0.04 (-) 68% 1.90 x 10 -8   res2880037_G G / A -0.04 (4) 49% 2.90 x 10 -8	rs79502300_C 🌼	C/C	0.05 (1)	80%	4.40 x 10 <sup>-10</sup>
rs189669984_C ©   C / C   0.21 (1)   99%   2.10 x 10 · 9     rs9892661_C ©   T / T   0.04 (-)   42%   2.10 x 10 · 9     rs1499813_T ©   T / T   0.04 (1)   59%   2.80 x 10 · 9     rs2302455_G ©   G / A   0.07 (1)   88%   4.60 x 10 · 9     rs11748963_T ©   T / C   0.04 (1)   73%   5.90 x 10 · 9     rs10871753_G ©   G / G   0.04 (1)   49%   6.00 x 10 · 9     rs116904997_G ©   G / G   -0.13 (4)   98%   1.10 x 10 · 8     rs190093681_C ©   C / C   0.37 (1)   > 99%   1.60 x 10 · 8     rs709208_A ©   • / G   -0.04 (-)   68%   1.90 x 10 · 8     rs2886037_G ©   G / A   -0.04 (4)   49%   2.90 x 10 · 8	rs1962104_T 🌼	C/C	-0.04 (-)	45%	6.40 x 10 <sup>-10</sup>
rs9892661_C T / T 0.04 (-) 42% 2.10 x 10 ·9   rs1499813_T T / T 0.04 (1) 59% 2.80 x 10 ·9   rs2302466_G 9 / A 0.07 (1) 88% 4.60 x 10 ·9   rs11748963_T T / C 0.04 (1) 73% 5.90 x 10 ·9   rs10871753_G 9 / 9 0.04 (1) 49% 6.00 x 10 ·9   rs116904997_G 9 / 9 -0.13 (1) 98% 1.10 x 10 ·8   rs190093681_C C / C 0.37 (1) > 99% 1.60 x 10 ·8   rs709208_A • / G -0.04 (-) 68% 1.90 x 10 ·8   rs2886037_G 6 / A -0.04 (1) 49% 2.90 x 10 ·8	rs11023059_A 🌼	A / A	0.04 (1)	52%	6.50 x 10 <sup>-10</sup>
rs1499813_T   T / T   0.04 (1)   59%   2.80 x 10·9     rs2302455_6   9 / A   0.07 (1)   88%   4.60 x 10·9     rs11748963_T   T / C   0.04 (1)   73%   5.90 x 10·9     rs10871753_G   9 / G   0.04 (1)   49%   6.00 x 10·9     rs116904997_G   6 / G   -0.13 (4)   98%   1.10 x 10·8     rs190093681_C   C / C   0.37 (1)   > 99%   1.60 x 10·8     rs709208_A   • / G   -0.04 (-)   68%   1.90 x 10·8     rs2886037_G   9 / A   -0.04 (4)   49%   2.90 x 10·8	rs189569984_C 🖐	C/C	0.21 (1)	99%	2.10 x 10 <sup>-9</sup>
rs2302455_6 6 / A 0.07 (1) 88% 4.60 x 10.9   rs11748963_T 7 / C 0.04 (1) 73% 5.90 x 10.9   rs10871753_G 6 / G 0.04 (1) 49% 6.00 x 10.9   rs116904997_G 6 / G -0.13 (4) 98% 1.10 x 10.8   rs190093681_C C / C 0.37 (1) > 99% 1.60 x 10.8   rs709208_A + / G -0.04 (-) 68% 1.90 x 10.8   rs2886037_G G / A -0.04 (4) 49% 2.90 x 10.8	rs9892651_C 🌼	T/T	0.04 (-)	42%	2.10 x 10 <sup>-9</sup>
rs11748963_T T / C 0.04 (1) 73% 5.90 x 10 · 9   rs10871763_G 9 / 6 0.04 (1) 49% 6.00 x 10 · 9   rs116904997_G 9 / 6 -0.13 (4) 98% 1.10 x 10 · 8   rs190093681_C C / C 0.37 (1) > 99% 1.60 x 10 · 8   rs709208_A • / G -0.04 (-) 68% 1.90 x 10 · 8   rs2886037_G 6 / A -0.04 (4) 49% 2.90 x 10 · 8	rs1499813_T 🌼	T/T	0.04 (1)	59%	2.80 x 10 <sup>-9</sup>
rs10871753_6   6 / 6   0.04 (1)   49%   6.00 x 10 · 9     rs116904997_6   6 / 6   -0.13 (1)   98%   1.10 x 10 · 8     rs190093681_0   C / C   0.37 (1)   > 99%   1.60 x 10 · 8     rs709208_A   + / G   -0.04 (-)   68%   1.90 x 10 · 8     rs2886037_6   G / A   -0.04 (1)   49%   2.90 x 10 · 8	rs2302455_G 🖐	G / A	0.07 (1)	88%	4.60 × 10 <sup>-9</sup>
rs116904997_6	rs11748963_T 💮	T/C	0.04 (1)	73%	5.90 x 10 <sup>-9</sup>
rs190093681_C   C / C   0.37 (↑)   > 99%   1.60 x 10 -8     rs709208_A   + / G   -0.04 (−)   68%   1.90 x 10 -8     rs2886037_G   G / A   -0.04 (↓)   49%   2.90 x 10 -8	rs10871753_G 👴	G / G	0.04 (1)	49%	6.00 × 10 <sup>-9</sup>
rs709208_A • / 6	rs116904997_G 🌼	G / G	-0.13 (↓)	98%	1.10 x 10 <sup>-8</sup>
rs2886037_G	rs190093681_C 🌼	C/C	0.37 (1)	> 99%	1.60 x 10 <sup>-8</sup>
	rs709208_A 🌼	*/G	-0.04 (-)	68%	1.90 × 10 <sup>-8</sup>
rs242562_G 💮 6/6 -0.04 (\dagger{4}) 62% 3.00 x 10 <sup>-8</sup>	rs2886037_G 🌼	G / A	-0.04 (\1)	49%	2.90 x 10 <sup>-8</sup>
	rs242562_G 🌼	G / G	-0.04 (↓)	62%	3.00 x 10 <sup>-8</sup>