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Gallstones (Ferkingstad, 2018)

Egil Ferkingstad, et al.
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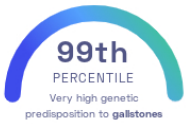
Inflammation | Gallbladder

STUDY SUMMARY

Discovery of 21 genomic regions associated with gallstone formation.

YOUR RESULT



STUDY DESCRIPTION

The gallbladder stores a fluid called bile. It releases bile into the intestines where it helps with digestion. Bile contains bile acids which are produced in the body from *cholesterol*. Gallstones are stones that can form in the gallbladder when there is too much *cholesterol* and not enough bile acids. Gallstones can cause pain in the upper right abdomen and may require surgery. To identify genetic variants associated with gallstone formation, this genome-wide association study examined the genomes of over 750,000 individuals of European descent. The study identified 28 genomic regions associated with gallstone disease, 21 of which are novel. Some of these genetic variants have previously been linked to diabetes and fat metabolism.

DID YOU KNOW?

According to the Guinness World Records, the most gallstones removed from a patient was ~24,000. In contrast, most patients who undergo surgery for gallstone disease have, on average, 2 to 20 gallstones.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to gallstones we summed up the effects of genetic variants that were linked to gallstones 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 gallstones. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to gallstones. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to gallstones. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for gallstones to be 1.09**. 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 gallstones is in the **99th percentile**. This means that it is higher than the polygenic scores 99% of people. We consider this to be a **very high genetic predisposition to gallstones**. 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
rs11887534_C	G / C	0.66 (↑)	5%	1.00×10^{-363}
rs212100_T	C / C	-0.15 (-)	16%	2.00×10^{-31}
rs12633863_G	G / G	0.10 (↑)	45%	3.60×10^{-30}
rs4148808_C	T / T	-0.15 (-)	16%	8.60×10^{-28}
rs2291428_C NEW	G / G	0.11 (-)	22%	2.40×10^{-27}
rs2290846_A NEW	G / A	0.11 (↑)	28%	4.70×10^{-27}
rs1800961_T NEW	NA	0.25 (-)	5%	5.70×10^{-26}
rs6471717_G	G / A	0.10 (↑)	31%	9.90×10^{-26}
rs601338_G NEW	A / A	-0.09 (-)	39%	9.40×10^{-22}
rs686030_C	C / A	-0.13 (↓)	13%	1.80×10^{-20}
rs756082276_CCT	NA	1.42 (-)	< 1%	1.20×10^{-17}
rs28929474_T NEW	NA	0.29 (-)	1%	1.80×10^{-17}
rs1260326_T	T / C	-0.08 (↓)	34%	2.00×10^{-16}
rs34861490_G NEW	A / G	0.11 (↑)	9%	4.20×10^{-15}
rs1169288_C NEW	A / C	-0.08 (↓)	31%	1.60×10^{-14}
rs756935975_T	NA	1.13 (-)	< 1%	1.80×10^{-14}
rs13280055_A NEW	G / G	0.10 (-)	13%	6.90×10^{-14}
rs56398830_A NEW	NA	0.31 (-)	1%	2.10×10^{-12}
rs174567_G NEW	A / A	0.07 (-)	39%	2.30×10^{-12}
rs708686_T NEW	C / C	0.08 (-)	23%	2.30×10^{-12}
rs11012737_A NEW	G / A	0.07 (↑)	27%	3.70×10^{-12}
rs1935_C NEW	C / G	-0.06 (↓)	48%	9.20×10^{-12}
rs2469991_T NEW	A / A	-0.07 (-)	32%	9.20×10^{-12}
rs17240268_A NEW	G / G	-0.11 (-)	12%	6.00×10^{-11}
rs2070959_G	A / G	0.07 (↑)	32%	8.50×10^{-11}
rs12004_G NEW	T / G	0.07 (↑)	31%	1.20×10^{-10}
rs55971546_T NEW	NA	0.14 (-)	4%	1.80×10^{-10}
rs11641445_T NEW	C / T	0.06 (↑)	34%	4.20×10^{-10}
rs17138478_A NEW	C / A	0.09 (↑)	13%	5.10×10^{-10}
rs45575636_T	NA	0.29 (-)	< 1%	7.90×10^{-9}
rs2292553_G NEW	G / G	-0.05 (↓)	46%	1.10×10^{-8}
rs12968116_T NEW	C / C	-0.08 (-)	16%	1.20×10^{-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.