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Gallstones (Fairfield, 2021)

Cameron Fairfield, et al.
Hepatology

Stomach | Diet | Liver

Gallstones form from the accumulation of bile.

STUDY SUMMARY

This report is based on a study that discovered 46 novel genetic variants associated with gallstone development.

YOUR RESULT

5th
PERCENTILE
Very low genetic predisposition to **gallstones**

STUDY DESCRIPTION

The gallbladder is a small, pear-shaped pouch located under the liver. It is connected to the intestines and liver by small tubes called bile ducts. Bile ducts carry bile, a yellow-green fluid produced by the liver, which helps with digestion. Occasionally, bile can form hardened clumps known as gallstones, which can cause pain, and in severe cases require surgical removal. Some individuals appear to have a higher propensity for gallstone formation, though the reasons are not clear. This study attempted to identify genetic factors that influence an individual's predisposition to develop gallstones. Through an analysis of over 375,000 individuals of European ancestry, researchers identified 75 genetic variants associated with gallstone formation, 46 of which were newly discovered in this study. Among the many genes linked to gallstones, ANO1 and TMEM147 are both connected to the gut's ability to expand and contract during digestion.

DID YOU KNOW?

A diet that would be considered 'heart-healthy' is 'gallbladder-healthy' too. Fats found in foods such as nuts, avocados, seeds, olives, peanut butter, fatty fish are generally okay. However, foods that increase the cholesterol level also increase the risk of 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 1.71**. 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 **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 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	GENE	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs11887534_G	G / C	ABCG8	0.70 (↑)	7%	4.96×10^{-324}
rs4681515_A	A / A	TM4SF4	-0.12 (↓)	44%	2.84×10^{-39}
rs2290846_G	G / A	LRBA	0.12 (↑)	29%	5.89×10^{-38}
rs7802555_A	A / A	ABCB4	-0.16 (↓)	14%	1.37×10^{-34}
rs681343_C	T / T	FUT2	0.11 (–)	49%	2.76×10^{-32}
rs1800961_C	NA	HNF4A	0.25 (–)	3%	1.55×10^{-29}
rs1993453_A	A / G	CYP7A1/UBXN2B	-0.09 (↓)	34%	1.62×10^{-29}
rs3802548_T	T / T	MARCHF8	0.11 (↑)	24%	2.02×10^{-29}
rs2393775_G	G / A	HNF1A	0.09 (↑)	38%	1.32×10^{-23}
rs686030_C	C / A	TTC39B	0.12 (↑)	14%	1.96×10^{-21}
rs34255979_C	C / T	IRF2BP1/FOXA3	0.12 (↑)	12%	8.54×10^{-20}
rs1260326_T NEW	T / C	GCKR	0.08 (↑)	39%	6.80×10^{-18}
rs2393969_A	A / C	JMJD1C	0.07 (↑)	47%	1.16×10^{-17}
rs12532734_G NEW	G / G	Intergenic	-0.08 (↓)	23%	1.18×10^{-16}
rs56363382_C NEW	C / C	ANO1	0.12 (↑)	8%	6.51×10^{-16}
rs17240268_G	G / G	ANPEP	-0.12 (↓)	9%	3.03×10^{-13}
rs708686_C	C / C	FUT6	0.06 (↑)	27%	4.18×10^{-12}
rs17138478_C	C / A	HNF1B	0.09 (↑)	13%	2.05×10^{-11}
rs13280055_G	G / G	GATA4	0.08 (↑)	13%	2.19×10^{-11}
rs7599_A NEW	G / G	TMEM147	-0.05 (–)	37%	2.49×10^{-11}
rs3784924_A	A / G	LITAF	0.06 (↑)	31%	2.67×10^{-11}
rs438568_A NEW	G / G	SPTLC3	-0.06 (–)	39%	4.85×10^{-11}
rs9427114_T NEW	T / T	ADAR	0.05 (↑)	49%	1.47×10^{-10}
rs10828250_C	C / G	MLLT10/SKIDA1	0.06 (↑)	31%	2.30×10^{-10}
rs174574_A	C / C	FADS2	-0.06 (–)	35%	3.04×10^{-10}
rs2469991_A	A / A	MAL2/SNORA32	-0.06 (↓)	29%	5.10×10^{-10}
rs11089985_A NEW	A / A	TNRC6B	0.05 (↑)	35%	8.38×10^{-10}
rs56339318_A NEW	A / A	LIN28B	-0.08 (↓)	10%	1.38×10^{-9}
rs4493564_A NEW	G / G	SHROOM3	0.05 (–)	35%	5.21×10^{-9}
rs60360195_A NEW	A / A	SH3BP4	0.06 (↑)	20%	6.66×10^{-9}
rs10898881_A NEW	A / G	ATG16L2	-0.05 (↓)	45%	1.88×10^{-8}
rs115478735_A NEW	A / T	ABO	0.05 (↑)	19%	2.33×10^{-8}
rs6936023_G NEW	G / A	GMDS-DT	-0.06 (↓)	18%	4.71×10^{-8}
rs7786376_A NEW	A / A	MLXIPL	0.05 (↑)	28%	4.97×10^{-8}
rs8051062_T NEW	T / C	CYB5B/NFAT5	-0.05 (↓)	42%	5.09×10^{-8}
rs13135475_T NEW	T / T	UGDH	-0.05 (↓)	46%	7.12×10^{-8}
rs7314929_C NEW	C / T	POC1B	0.06 (↑)	25%	7.46×10^{-8}
rs28601761_C NEW	C / C	TRIB1	0.05 (↑)	42%	8.10×10^{-8}

rs7412_C	C / C	APOE	0.08 (↑)	8%	8.48×10^{-8}
rs59893339_C	C / C	LOC107986957	0.07 (↑)	14%	2.43×10^{-7}
rs1476698_A	A / A	FARP2	0.05 (↑)	37%	3.55×10^{-7}
rs17219671_A	A / A	PTTG1IP	0.05 (↑)	27%	5.13×10^{-7}
rs2292553_G	G / G	TMBIM1	0.04 (↑)	44%	1.22×10^{-6}
rs7012637_G	G / A	RP11-115J16.1	-0.04 (↓)	48%	1.42×10^{-6}
rs55932961_G	G / G	CPS1	-0.05 (↓)	19%	2.62×10^{-6}
rs735286_C	C / T	VEGFA	0.04 (↑)	31%	3.34×10^{-6}
rs62185132_G	G / G	AP1S3	0.04 (↑)	38%	4.93×10^{-6}
rs9323382_A	A / A	SYT16	-0.05 (↓)	19%	8.32×10^{-6}
rs2618566_G	G / T	AL035045.1	0.04 (↑)	34%	1.01×10^{-5}
rs11189138_T	C / C	FRAT2	-0.04 (–)	41%	1.33×10^{-5}
rs7626199_T	T / T	ARHGEF3	-0.04 (↓)	46%	1.46×10^{-5}
rs113638914_T	T / T	SYNJ2	0.04 (↑)	21%	2.49×10^{-5}

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.