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Alcohol-related liver cirrhosis (Schwantes-An, 2020)

Tae-Hwi Schwantes-An, et al.
Hepatology

Liver

STUDY SUMMARY

Discovery of a novel region of the genome associated with alcohol-related liver cirrhosis.

YOUR RESULT

STUDY DESCRIPTION

The liver is a large organ that sits on the right side of the abdomen. It filters blood to detoxify chemicals, including drugs and alcohol. After long periods of heavy alcohol use, healthy liver tissue is replaced by scar tissue. Over time, the build-up of scar tissue can impair the functioning of the liver, leading to a condition called cirrhosis. It is estimated that 10-20% of heavy drinkers will develop cirrhosis, which can eventually lead to liver failure. This genome-wide association examined about 1,750 individuals of European ancestry to identify genetic factors that may put heavy drinkers at an increased risk for developing liver cirrhosis. The study found one novel genomic region associated with liver cirrhosis. This region harbors the FAF2 gene that plays a role in fat metabolism, in particular how fats are organized within cells.

DID YOU KNOW?

The liver is the only internal organ that has the potential to regenerate. As little as 51% of the original liver can regenerate to a full-sized organ. Prolonged alcohol use, though, can affect how well the cells of the liver can regrow.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to alcohol-related liver cirrhosis we summed up the effects of genetic variants that were linked to alcohol-related liver cirrhosis 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 alcohol-related liver cirrhosis. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to alcohol-related liver cirrhosis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to alcohol-related liver cirrhosis. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for alcohol-related liver cirrhosis to be 0.24**. 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 alcohol-related liver cirrhosis is in the **60th percentile**. This means that it is higher than the polygenic scores 60% of people. We consider this to be an **average genetic predisposition to alcohol-related liver cirrhosis**. 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
rs2294915_T	C / T	0.73 (↑)	32%	1.28×10^{-53}
rs10401969_C	T / T	0.40 (–)	8%	2.40×10^{-9}
rs10433937_G	T / G	-0.25 (↓)	25%	2.85×10^{-9}
rs11134977_C NEW	T / C	-0.24 (↓)	46%	1.56×10^{-8}
rs28929474_T	NA	0.64 (–)	2%	1.99×10^{-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.