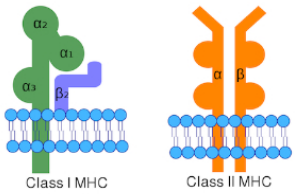
09/2011

Primary biliary cirrhosis (Mells, 2011)

George F. Mells, et al.
Nature Genetics

Autoimmunity | Liver



Major Histocompatibility Complex Proteins

STUDY SUMMARY

Genetic variants linked to pathways that promote inflammation may play a role in the development of primary biliary cirrhosis.

YOUR RESULT

STUDY DESCRIPTION

Primary biliary cirrhosis is a chronic, *autoimmune* liver disease. *Autoimmune* diseases are characterized by the immune system mistakenly attacking the body's own cells. For primary biliary cirrhosis, this eventually results in the destruction of your bile ducts, where bile is produced. Bile helps in processes like digestion and the elimination of toxins from the body. This study linked genetic variants in the HLA genes to primary biliary cirrhosis in 7,003 individuals of British or Irish ancestry. HLA genes encode for major histocompatibility complex (MHC) proteins that help recognize foreign molecules and elicit an immune response. Other genetic variants identified in this study provide additional evidence for the role of other immune system pathways in primary biliary cirrhosis.

DID YOU KNOW?

Reducing or eliminating alcohol, eating a healthy diet, and getting regular exercise are all ways to try and prevent liver damage and development of primary biliary cirrhosis.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to primary biliary cirrhosis we summed up the effects of genetic variants that were linked to primary biliary 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 primary biliary cirrhosis. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to primary biliary cirrhosis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to primary biliary cirrhosis. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for primary biliary cirrhosis to be 5.97**. 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 primary biliary cirrhosis is in the **100th percentile**. This means that it is higher than the polygenic scores 100% of people. We consider this to be a **very high genetic predisposition to primary biliary 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
rs7774434_C	C / C	0.47 (↑)	45%	3.86×10^{-34}
rs17129789_C	T / T	0.42 (-)	21%	9.48×10^{-20}
rs10931468_A NEW	C / A	0.41 (↑)	15%	2.35×10^{-19}
rs12531711_G	A / A	0.46 (-)	12%	8.90×10^{-17}
rs485499_T	C / C	0.32 (-)	61%	2.29×10^{-16}
rs12134279_T NEW	C / C	0.29 (-)	23%	2.06×10^{-14}
rs3745516_A	A / A	0.32 (↑)	25%	1.63×10^{-13}
rs6421571_C NEW	C / C	0.31 (↑)	82%	2.69×10^{-12}
rs12924729_G NEW	G / G	0.25 (↑)	70%	2.95×10^{-12}
rs7665090_G NEW	A / G	0.23 (↑)	55%	4.06×10^{-12}
rs860413_A NEW	A / C	0.26 (↑)	74%	1.02×10^{-11}
rs911263_T NEW	T / T	0.25 (↑)	74%	1.76×10^{-11}
rs2293370_G NEW	G / G	0.30 (↑)	81%	2.53×10^{-11}
rs11117432_G NEW	G / G	0.27 (↑)	80%	4.66×10^{-11}
rs968451_T NEW	T / T	0.24 (↑)	21%	1.08×10^{-9}
rs1800693_C NEW	T / T	0.20 (-)	42%	1.80×10^{-9}
rs6974491_A NEW	G / A	0.22 (↑)	19%	4.44×10^{-8}