

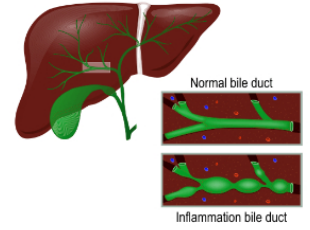
★ Primary biliary cholangitis (Cordell, 2021)

Heather Cordell, et al.
Journal of Hepatology

Liver Autoimmunity

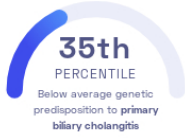
STUDY SUMMARY

This report is based on a study that discovered 57 genetic variants associated with primary biliary cholangitis, a chronic disease of the liver.



Primary biliary cholangitis causes inflammation and obstruction of bile ducts in the liver.

YOUR RESULT



STUDY DESCRIPTION

The liver is a large, football-shaped organ that sits on the right side of the abdomen. It performs many functions for the body, including removing toxins from the blood, assisting in digestion, and processing blood sugar. Primary biliary cholangitis (PBC) is an autoimmune disease that causes progressive destruction of the bile ducts of the liver. This leads to the accumulation of bile in the liver which results in further liver damage. Symptoms include abdominal pain, fatigue, and skin yellowing. This study sought to identify genetic variants associated with the risk of PBC by examining genetic data of over 30,000 individuals of European and East Asian ancestry. The researchers identified 57 genetic variants, 21 of which were newly associated with PBC in this study. Many variants were linked to genes that play a role in the functioning of the immune system. These included FCRL3, ID2, and RARB genes, all of which help T cells identify and eliminate foreign particles.



DID YOU KNOW?

The liver holds over 10% of the blood in your body at any given time, and nearly 1.5 liters of blood pass through the liver every minute.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to primary biliary cholangitis we summed up the effects of genetic variants that were linked to primary biliary cholangitis 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 cholangitis. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to primary biliary cholangitis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to primary biliary cholangitis. By adding up the effect sizes of the highlighted variants we calculated your polygenic score for primary biliary cholangitis to be **0.88**. To determine whether your score is high or low, we compared it to the scores of 5,000 other Nebula Average users. We found that your polygenic score for primary biliary cholangitis is in the **35th percentile**. This means that it is higher than the polygenic scores 35% of people. We consider this to be a **below average genetic predisposition to primary biliary cholangitis**. 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
rs7774434_C	C / C	HLA	0.46 (↑)	39%	2.72 x 10 ⁻¹¹⁶
rs6679356_C	T / T	IL12RB2	0.44 (-)	22%	4.85 x 10 ⁻⁶⁵
rs589446_T	T / T	IL12A	-0.33 (↓)	31%	2.23 x 10 ⁻⁵²
rs7119044_G	C / C	CXCR5	-0.36 (-)	21%	6.85 x 10 ⁻³⁸
rs6533022_C	T / C	NFKB1	0.23 (↑)	54%	2.21 x 10 ⁻³²
rs2293370_A	G / G	CD80	-0.29 (-)	19%	2.05 x 10 ⁻³¹
rs11889341_T	C / C	STAT4	0.25 (-)	21%	3.41 x 10 ⁻³¹
rs12919083_C	A / A	CLEC16A	-0.23 (-)	35%	3.63 x 10 ⁻²⁶
rs1322057_G	A / A	TNFSF15	0.44 (-)	9%	5.48 x 10 ⁻²⁶
rs11742270_A	G / A	IL7R	-0.24 (↓)	21%	4.44 x 10 ⁻²⁴
rs10802191_T	T / T	CD58	0.23 (↑)	86%	3.68 x 10 ⁻¹⁷
rs17641524_T	C / C	DENND1B	0.20 (-)	17%	2.00 x 10 ⁻¹⁶
rs859767_G	A / A	TMEM163	-0.16 (-)	34%	8.94 x 10 ⁻¹⁶
rs6550965_A	C / C	RARB	0.15 (-)	42%	1.50 x 10 ⁻¹⁴
rs3784099_A	G / G	RAD51B	-0.18 (-)	41%	1.69 x 10 ⁻¹⁴
rs1119132_A	G / G	IL21R	-0.21 (-)	11%	4.09 x 10 ⁻¹⁴
rs12419634_C	G / G	POU2AF1	0.15 (-)	53%	1.22 x 10 ⁻¹³
rs10581773_ATAT	A / A	CD28	0.21 (-)	14%	3.82 x 10 ⁻¹³
rs79344245_C	T / T	TNFSF11	0.14 (-)	39%	3.39 x 10 ⁻¹²
rs2546890_A	A / A	IL12B	0.13 (↑)	49%	4.61 x 10 ⁻¹²
rs1156336_A	A / T	PLCL2	-0.14 (↓)	63%	7.13 x 10 ⁻¹²
rs1808094_T	T / C	CD226	0.12 (↑)	54%	1.66 x 10 ⁻¹⁰
rs10230459_G	C / G	ELM01	0.21 (↑)	13%	2.59 x 10 ⁻¹⁰
rs968334_T	C / C	CCR6	0.12 (-)	40%	3.98 x 10 ⁻¹⁰
rs2007403_T	C / T	TET2	0.13 (↑)	49%	6.19 x 10 ⁻¹⁰
rs739496_A	A / G	SH2B3	0.16 (↑)	72%	6.29 x 10 ⁻¹⁰
rs799469_G	A / A	FAM177A1	0.15 (-)	24%	1.73 x 10 ⁻⁹
rs112500293_T	C / C	TRIM14	-0.16 (-)	33%	7.63 x 10 ⁻⁹
rs10893872_T	T / C	ETS1	0.11 (↑)	55%	9.77 x 10 ⁻⁹
rs11264790_T	C / T	FCRL3	-0.11 (↓)	54%	2.25 x 10 ⁻⁸
rs4904964_C	A / C	RIN3	-0.12 (↓)	36%	2.45 x 10 ⁻⁸
rs13416555_G	C / G	ID2	-0.12 (↓)	24%	2.95 x 10 ⁻⁸
rs742108_A	G / G	PRDM1	0.13 (-)	15%	3.16 x 10 ⁻⁸
rs6711622_A	G / A	DNMT3A	0.11 (↑)	34%	3.89 x 10 ⁻⁸
rs28568531_A	T / T	TNFSF14	0.11 (-)	41%	4.06 x 10 ⁻⁸
rs6874308_C	C / T	NDIFP1	-0.11 (↓)	33%	4.67 x 10 ⁻⁸
rs4733851_G	A / G	PVT1	-0.11 (↓)	55%	4.98 x 10 ⁻⁸
rs7922169_T	T / G	WDFY4	0.11 (↑)	48%	5.47 x 10 ⁻⁸

rs199565066_C	CA / C	DLEU1	-0.22 (↓)	8%	7.09×10^{-8}
rs6969930_T	T / C	IRF5	-0.10 (↓)	40%	1.61×10^{-7}
rs12122721_A 	G / G	INAVA	-0.11 (-)	23%	6.95×10^{-7}
rs12444221_T	C / T	IRF8	-0.12 (↓)	20%	3.26×10^{-6}
rs3760650_T	T / C	TYK2	0.09 (↑)	69%	9.50×10^{-6}
rs7786537_C 	G / G	ITGB8	-0.11 (-)	20%	1.12×10^{-5}