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Idiopathic pulmonary fibrosis (Allen, 2019)

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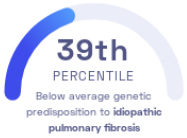
Lungs

STUDY SUMMARY

Identification of 5 novel variants associated with *idiopathic pulmonary fibrosis*.

YOUR RESULT



STUDY DESCRIPTION

Idiopathic pulmonary fibrosis (IPF) is a *progressive* lung disease that is characterized by scarring of lungs which makes it hard to breathe. Over 15,000 new cases of IPF are reported yearly in the United States, however, the cause of IPF is not known and there is also no cure. This study sought to identify genetic factors that contribute to the risk of developing IPF. To this end, the researchers conducted the largest genome-wide association study of IPF to date, utilizing data from over 11,000 individuals of European descent. They identified 5 novel variants associated with susceptibility to IPF, and confirmed 11 previously reported variants. Two of the novel variants are near genes that regulate *chromosome* separation during *cell division*, suggesting that risk for IPF is tied to fundamental cellular level processes.

DID YOU KNOW?

Although the cause of IPF is unknown, some possible risk factors include a history of smoking and working around dust or fumes. Age is also a risk factor – the disease develops gradually, meaning symptoms may not be apparent until they are severe.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to idiopathic pulmonary fibrosis we summed up the effects of genetic variants that were linked to idiopathic pulmonary fibrosis 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 idiopathic pulmonary fibrosis. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to idiopathic pulmonary fibrosis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to idiopathic pulmonary fibrosis. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for idiopathic pulmonary fibrosis to be 0.22**. 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 idiopathic pulmonary fibrosis is in the **39th percentile**. This means that it is higher than the polygenic scores 39% of people. We consider this to be a **below average genetic predisposition to idiopathic pulmonary fibrosis**. 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
rs35705950_T	G / G	1.58 (–)	15%	1.18×10^{-203}
rs2076295_G	T / G	0.38 (↑)	47%	2.79×10^{-30}
rs7725218_A	G / A	-0.33 (↓)	33%	1.54×10^{-20}
rs2077551_C	T / C	-0.34 (↓)	19%	2.83×10^{-16}
rs59424629_G	T / T	-0.26 (–)	46%	7.30×10^{-16}
rs2897075_T	C / T	0.26 (↑)	39%	3.10×10^{-14}
rs2013701_T	G / T	-0.25 (↓)	49%	3.30×10^{-13}
rs12696304_G	C / C	0.27 (–)	28%	7.09×10^{-13}
rs12699415_A NEW	A / A	0.25 (↑)	42%	7.15×10^{-13}
rs12610495_G	A / A	0.27 (–)	31%	2.92×10^{-12}
rs62023891_A	G / G	0.24 (–)	30%	1.27×10^{-10}
rs9577395_G	C / C	-0.26 (–)	21%	1.34×10^{-10}
rs78238620_A NEW	T / T	0.46 (–)	5%	5.12×10^{-10}
rs41308092_A NEW	NA	0.75 (–)	2%	7.65×10^{-10}
rs28513081_G NEW	A / A	-0.20 (–)	43%	1.20×10^{-9}
rs537322302_G NEW	NA	2.06 (–)	< 1%	3.43×10^{-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.