07/2019

Emphysema (Parker, 2019)

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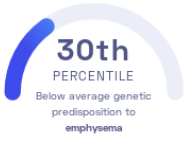
eLIFE

Lungs

STUDY SUMMARY

Genetic variants near the TGFβ2 gene may increase the risk of emphysema by elevating TGFβ2 expression in lung *fibroblasts*.

YOUR RESULT



STUDY DESCRIPTION

Emphysema is one of the main types of chronic obstructive pulmonary disease (COPD) that occurs when the air sacs in the lungs are damaged. This makes it difficult to breath and most individuals with emphysema develop a chronic cough. Although smoking is the most common cause of emphysema, air pollution, certain chemical fumes, and genetics are also risk factors. Therefore, this study looked at 10,192 non-Hispanic white and African American subjects to find how genetic variants near the previously-identified TGFβ2 gene influence the risk of emphysema. The study found that variants near the TGFβ2 gene, which are associated with increased emphysema risk, increase the expression of TGFβ in *fibroblasts*. The TGFβ2 protein helps regulate the expression of other genes and is involved in many cellular processes, such as cell growth and apoptosis (programmed cell death).

DID YOU KNOW?

To prevent emphysema, avoid cigarette smoke and air pollutants when possible. Additionally, wearing a mask if you work with chemical fumes can reduce your risk for this disease.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to emphysema we summed up the effects of genetic variants that were linked to emphysema 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 emphysema. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to emphysema. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to emphysema. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for emphysema to be 0.05**. 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 emphysema is in the **30th percentile**. This means that it is higher than the polygenic scores 30% of people. We consider this to be a **below average genetic predisposition to emphysema**. 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	COMMENTS	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs56077333_A	C / C	Moderate Centrilobular	0.02 (–)	67%	2.20×10^{-13}
rs17368582_T	T / T	Moderate Centrilobular	0.02 (↑)	14%	8.10×10^{-12}
rs56113850_T	C / C	Moderate Centrilobular	-0.02 (–)	59%	1.50×10^{-9}
rs796395_A	G / A	Moderate Centrilobular	0.01 (↑)	52%	6.10×10^{-9}
rs138641402_A	A / A	Moderate Centrilobular	0.01 (↑)	38%	3.30×10^{-8}
rs76756075_T NEW	NA	Panlobular	-0.01 (–)	2%	2.00×10^{-8}
rs78070126_T NEW	C / C	Panlonular	0.01 (–)	97%	2.80×10^{-8}
rs145770770_A	A / A	Panlonular	-0.01 (↓)	99%	3.80×10^{-8}
rs379123_T	T / T	Severe Centrilobular	-0.00 (↓)	40%	3.70×10^{-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.