09/2021

Eosinophilic esophagitis (Chang, 2021)

Xiao Chang, et al.
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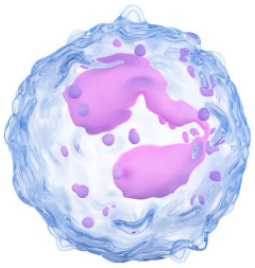
Allergy | Intestines | Inflammation

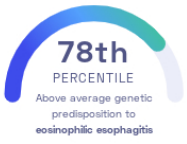


Eosinophils can protect against infections, but can also contribute to allergies.

STUDY SUMMARY

This report is based on a study that discovered 11 novel genetic variants associated with eosinophilic esophagitis.

YOUR RESULT



STUDY DESCRIPTION

Eosinophils are a type of white blood cell that help to fight off infections in the body by promoting inflammation. However, sometimes eosinophils can be damaging to the body. When eosinophils cause inflammation in the esophagus, the tube that connects the mouth to the stomach, a condition called eosinophilic esophagitis (EoE) can develop. EoE is often caused by food allergies or acid reflux and can result in pain and difficulty swallowing. To identify regions of the genome associated with an individual's predisposition to developing EoE, this study analyzed the genetic data of over 15,000 individuals of European ancestry. In total, 11 regions of the genome were associated with EoE development. Many of the genes located in these regions, including SMAD3 and RORA, have previously been associated with other allergy-related diseases.

DID YOU KNOW?

Males are approximately three times more likely to develop EoE than females, though the reasons for this are not yet understood.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to eosinophilic esophagitis we summed up the effects of genetic variants that were linked to eosinophilic esophagitis 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 eosinophilic esophagitis. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to eosinophilic esophagitis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to eosinophilic esophagitis. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for eosinophilic esophagitis to be -0.14**. 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 eosinophilic esophagitis is in the **78th percentile**. This means that it is higher than the polygenic scores 78% of people. We consider this to be an **above average genetic predisposition to eosinophilic esophagitis**. 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
rs1438673_C	T / T	TSLP/WDR36	-0.36 (–)	43%	6.12×10^{-22}
rs143457388_A	T / T	CAPN14	0.57 (–)	8%	2.69×10^{-16}
rs61894547_T	C / C	EMSY	0.58 (–)	7%	4.69×10^{-16}
rs35099084_T	C / C	CLEC16A	-0.33 (–)	17%	1.92×10^{-12}
rs2279293_G NEW	C / C	RORA	-0.37 (–)	11%	4.66×10^{-11}
rs56062135_T NEW	C / C	SMAD3	0.25 (–)	28%	3.79×10^{-10}
rs887992_C NEW	A / A	TMEM182	-0.29 (–)	30%	4.43×10^{-10}
rs1620996_T NEW	T / C	SOX4	-0.37 (↓)	8%	2.70×10^{-8}
rs2106984_A NEW	T / A	RAD50	0.23 (↑)	25%	4.11×10^{-8}