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Aerodigestive squamous cell cancer (Lesseur, 2021)

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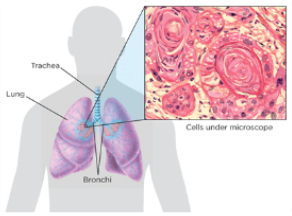
Cancer | Lungs | Mouth | Throat

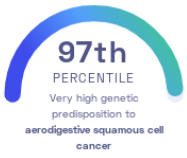


Squamous cell cancer of the lungs is often caused by smoking.

STUDY SUMMARY

This report is based on a study that discovered 9 genetic variants associated with squamous cell carcinomas in the aerodigestive tract.

YOUR RESULT



Very high genetic predisposition to **aerodigestive squamous cell cancer**

STUDY DESCRIPTION

Squamous cells can be found throughout the body where they form outer layers of the skin, the digestive system and the respiratory tract. Squamous cell carcinomas (SCCs) are a form of cancer that can occur when squamous cells begin to multiply uncontrollably. Often, SCCs form when squamous cells are damaged by exposure to UV light, smoke, or other environmental hazards. This study aimed to identify genetic variants associated with SCC of the aerodigestive tract, which includes the lungs, mouth, throat, and the food pipe. Following the analysis of genetic data from more than 75,000 individuals of European ancestry, 9 regions of the genome associated with squamous cell carcinomas were identified. One of the genes linked to SCCs of the aerodigestive tract was MDM4. This gene plays an important role in stopping cells with damaged DNA from dividing uncontrollably. Mutations in the MDM4 gene that disrupt its function may allow cells to become cancerous.

DID YOU KNOW?

Smoking is strongly associated with squamous cell carcinomas in the aerodigestive tract. For example, it is estimated to account for more than 70% of SCCs in the larynx (voice box).

YOUR DETAILED RESULTS

To calculate your genetic predisposition to aerodigestive squamous cell cancer we summed up the effects of genetic variants that were linked to aerodigestive squamous cell cancer 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 aerodigestive squamous cell cancer. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to aerodigestive squamous cell cancer. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to aerodigestive squamous cell cancer. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for aerodigestive squamous cell cancer to be 0.60**. 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 aerodigestive squamous cell cancer is in the **97th percentile**. This means that it is higher than the polygenic scores 97% of people. We consider this to be a **very high genetic predisposition to aerodigestive squamous cell cancer**. 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
rs11571815_A	NA	BRCA2	0.69 (–)	1%	2.30×10^{-21}
rs389884_G	A / A	STK19	0.23 (–)	9%	2.42×10^{-19}
rs9271611_G	A / G	HLA-DQA1	0.15 (↑)	41%	4.82×10^{-19}
rs7857345_T	T / T	CDKN2B-AS1	0.10 (↑)	30%	5.55×10^{-10}
rs1229984_T	C / C	ADH1B	-0.22 (–)	5%	1.89×10^{-9}
rs56321285_A NEW	G / G	TMEM237	-0.10 (–)	31%	6.99×10^{-9}
rs61494113_A NEW	G / G	ABHD8	0.09 (–)	30%	9.86×10^{-8}
rs13181561_G NEW	G / A	TMEM173	0.09 (↑)	28%	1.74×10^{-7}
rs12133735_G NEW	G / G	MDM4	0.08 (↑)	35%	2.16×10^{-7}

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.