6/2016

# Squamous cell lung carcinoma (McKay, 2017)

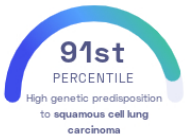
James McKay, et al.
Nature Genetics

Lung Cancer

## STUDY SUMMARY

Identification of 3 genomic regions associated with squamous cell lung carcinoma.

## YOUR RESULT




## STUDY DESCRIPTION

Lung cancer is a condition in which cells in the lungs divide uncontrollably. Squamous cells are flat-shaped cells that line many organs in the body. Squamous cell lung carcinoma is a type of lung cancer that is caused by mutations in squamous cells that line the airways of the lungs. This lung cancer subtype accounts for 30% of all lung cancer cases and is often linked to a history of smoking. In order to understand the genetic basis of squamous cell carcinoma, researchers examined the genomes of ~63,000 individuals. They identified 3 genomic regions that are associated with squamous cell carcinoma. These genetic variants explain approximately 6.2% of the heritability of squamous cell carcinoma.

## DID YOU KNOW?

The most common type of lung cancer in men is squamous cell carcinoma. This subtype of lung cancer is often easier to detect as it produces more symptoms than other subtypes of lung cancer.

## YOUR DETAILED RESULTS

To calculate your genetic predisposition to squamous cell lung carcinoma we summed up the effects of genetic variants that were linked to squamous cell lung carcinoma 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 squamous cell lung carcinoma. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to squamous cell lung carcinoma. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to squamous cell lung carcinoma. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for squamous cell lung carcinoma to be 0.00**. 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 squamous cell lung carcinoma is in the **91st percentile**. This means that it is higher than the polygenic scores 91% of people. We consider this to be a **high genetic predisposition to squamous cell lung carcinoma**. 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 |
|---|---|---|---|---|
| rs116822326_G | / | 0.22 (-) | 16% | $3.80 \times 10^{-19}$ |
| rs17879961_G | NA | -0.89 (-) | 1% | $5.70 \times 10^{-13}$ |
| rs7953330_C | G / G | -0.15 (-) | 32% | $7.30 \times 10^{-13}$ |

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.