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Orofacial clefts (Mukhopadhyay, 2021)

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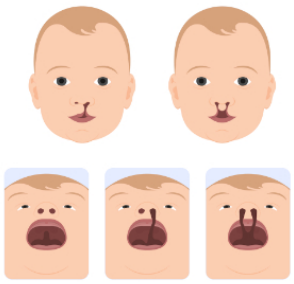
Mouth | Development



Types of orofacial clefts.

STUDY SUMMARY

This report is based on a study that discovered 18 genetic variants associated with orofacial clefts.

YOUR RESULT

STUDY DESCRIPTION

Cleft lips and cleft palates are birth defects that occur during the development of a baby's lips and mouth, respectively. Babies with these conditions are born with openings in their lips or roof of their mouth, which can affect activities such as speaking and eating. Together, these 2 birth defects are known as orofacial clefts. Orofacial clefts are a common condition, occurring in nearly 1 in 1000 births, but their causes are not fully understood. To study how genetic differences may account for an increased risk of orofacial clefts, this study examined genetic information of nearly 12,000 individuals of European, Asian, Native American, and African ancestry. The researchers identified 18 regions of the genome associated with orofacial clefts, 3 of which were newly discovered. Many genes that play a role in embryonic development, such as WNT3, WNT9B, and NTN1, were linked to the risk of orofacial clefts.

DID YOU KNOW?

Children born with orofacial clefts are more likely to suffer ear infections, develop hearing loss, and have problems with their teeth.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to orofacial clefts we summed up the effects of genetic variants that were linked to orofacial clefts 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 orofacial clefts. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to orofacial clefts. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to orofacial clefts. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for orofacial clefts to be 1.73**. 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 orofacial clefts is in the **92nd percentile**. This means that it is higher than the polygenic scores 92% of people. We consider this to be a **high genetic predisposition to orofacial clefts**. 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
rs72728755_A	T / T	NA	0.48 (–)	13%	2.80×10^{-29}
rs12543318_C	C / C	NA	0.24 (↑)	31%	7.45×10^{-11}
rs6745357_G	C / C	NA	0.25 (–)	43%	3.62×10^{-10}
rs16957821_G	C / G	NTN1	0.25 (↑)	22%	9.12×10^{-10}
rs17015217_A	NA	IRF6	-0.19 (–)	4%	1.46×10^{-9}
rs1588366_G	A / A	TANC2	-0.24 (–)	24%	1.09×10^{-8}
rs1975866_C	C / T	RBFOX3	0.39 (↑)	36%	3.13×10^{-8}
rs9439714_C	C / C	PAX7	0.15 (↑)	29%	3.27×10^{-8}
rs17075892_T	C / C	LOC105377732	-0.25 (–)	23%	6.95×10^{-8}
rs2003950_A	G / G	RHPN2	0.21 (–)	34%	1.21×10^{-7}
rs7216951_T	G / T	LRRC37A2	-0.20 (↓)	19%	1.42×10^{-7}
rs185266751_G	NA	STK3	0.50 (–)	1%	2.49×10^{-7}
rs9408874_T	C / T	NA	0.26 (↑)	18%	2.59×10^{-7}
rs181764204_T	NA	NA	0.54 (–)	< 1%	2.63×10^{-7}
rs150952246_C NEW	NA	ZNF503	0.37 (–)	< 1%	3.14×10^{-7}
rs62164740_A NEW	G / A	LOC101927967, LOC105374817	0.25 (↑)	10%	6.27×10^{-7}
rs118107597_A NEW	NA	SUFU	0.58 (–)	< 1%	8.21×10^{-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.