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☆ Nonsyndromic cleft lip with/without cleft palate (Lu, 2019)

Yun Lu, et al.
Human Mutation

Mouth



Cleft Lip

Cleft Palate

STUDY SUMMARY

Nonsyndromic cleft lip with/without cleft palate is associated with a genetic variant in a long, *noncoding* region of the genome.

STUDY DESCRIPTION


Nonsyndromic cleft lip with/without cleft palate (NSCL/P) is one of the most common human birth defects. The term nonsyndromic indicates that these defects are not associated with any other abnormalities or symptoms besides those directly caused by a cleft lip or palate (like difficulties eating and speaking clearly). A cleft lip occurs when the tissue that forms the lip does not completely join while a cleft palate is when tissues that form the roof of the mouth do not join together. This study examined 2,573 individuals from a Chinese population to identify genetic variants associated with NSCL/P. It found one significant variant in a long *noncoding* region of the genome.

DID YOU KNOW?

Surgery can often successfully treat cleft lip and cleft palate if done within a year of birth. The risk of this birth defect can be decreased by taking folic acid and not drinking or smoking during pregnancy.

YOUR DETAILED RESULTS

The variants highlighted in green have **positive effect sizes** and increase your genetic predisposition to NSCL/P. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to NSCL/P. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to NSCL/P. 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 [ⓘ]
rs2262261_C 	C / G	-0.21 (↓)	65%	1.00 x 10 ⁻³