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## Familial short stature (Lin, 2020)

Ying-Ju Lin, et al.
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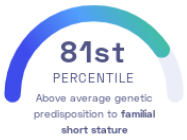
Appearance

### STUDY SUMMARY

Discovery of 10 genetic variants associated with familial short stature.

### YOUR RESULT




### STUDY DESCRIPTION

Height varies wildly across the globe! In the Netherlands, the average height for men and women is 6ft and 5ft 7in, respectively. Across the ocean, men and women in Peru are, on average, 5ft 5in and 4ft 11in tall. While environmental factors do contribute to height differences, studies of twins have estimated that nearly 86% of a person's height may be due to genetics. This study aimed to understand the genetics of familial short stature, which is defined as having a shorter stature than 97% of people of the same age and gender. The researchers examined the genomes of 1,163 individuals of Han Chinese ancestry who were diagnosed with familial short stature. The study discovered 10 novel genetic variants that have not been previously associated with general height. Multiple variants are in genes that play a role in production of *hormones* that control growth and development.

### DID YOU KNOW?

Some studies have found a potential link between shorter stature and a decreased risk of developing some cancers. For example, taller women are more likely to develop breast cancer, with a 1.2% increased risk for every 4 inches of height. For men, the risk of prostate cancer also increases with height, roughly 1% for every 4 inches of height.

### YOUR DETAILED RESULTS

To calculate your genetic predisposition to familial short stature we summed up the effects of genetic variants that were linked to familial short stature 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 familial short stature. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to familial short stature. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to familial short stature. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for familial short stature to be 11.14**. 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 familial short stature is in the **81st percentile**. This means that it is higher than the polygenic scores 81% of people. We consider this to be an **above average genetic predisposition to familial short stature**. 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 |
|---|---|---|---|---|
| rs7659854_C NEW | T / C | 1.73 (↑) | 13% | $1.44 \times 10^{-149}$ |
| rs2375843_C NEW | T / T | 1.39 (–) | 40% | $3.95 \times 10^{-90}$ |
| rs525537_G NEW | G / G | 1.72 (↑) | 41% | $5.40 \times 10^{-76}$ |
| rs116988614_G NEW | NA | 2.73 (–) | < 1% | $3.52 \times 10^{-70}$ |
| rs7033295_G NEW | G / T | 1.56 (↑) | 48% | $1.42 \times 10^{-53}$ |
| rs13183322_C NEW | T / T | 0.64 (–) | 12% | $7.76 \times 10^{-20}$ |
| rs202128628_T NEW | NA | 2.26 (–) | 1% | $1.80 \times 10^{-17}$ |
| rs117002249_T NEW | T / T | 2.21 (↑) | > 99% | $3.37 \times 10^{-16}$ |

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.