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International Collaboration and New Technology Reveal Rare Genes that Determine Height

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INTRODUCTION:
Height is a phenotype, an observable trait of the genes that we inherit from our parents. It’s a polygenic trait, meaning it is controlled by 2 or more genes. Because it is so easy to measure, it is an ideal characteristic to help us understand other polygenic traits. So far, genome-wide association studies (GWAS) have identified 700 common variants that determine a person's height. New discoveries have been made by the international group called Genetic Investigation of Anthropometric Traits (GIANT), revealing 83 genetic mutations that have additional effects on a person’s height.

METHODS:
Researchers used new technology called the Exomechip; this is a human genotyping microarray that can process thousands of DNA samples to identify mutations and structural variants. Common variants have a minor allele frequency (MAF) of > 5%, and are located outside of DNA coding regions. The Exomechip can capture low frequency (MAF=1–5%) and rare (MAF < 0.5%) variants. Because of the identification of these mutations, geneticists can now account for 27% of the genetics determining height, which is an increase from 20% based on earlier studies.

CONCLUSION:
This gives us a better understanding of human growth and how these variations influence the structure of a complex polygenic trait. By identifying rare genetic mutations, scientists can investigate uncommon DNA changes involved in other aspects of human health, such as diabetes and heart disease, giving them a better understanding of those diseases and may then lead to better treatments.

REFERENCES:
(3) https://www.illumina.com/techniques/popular-applications/genotyping/whole-genome-genotyping.html

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