Converging evidence of positive selection at height-associated loci in Europe

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Background:
Human height exhibits pronounced geographic variation across Europe, with individuals from northern populations generally taller than those in the south. While previous studies have claimed natural selection as a driver of this pattern, concerns over residual population stratification in genome-wide association study (GWAS) summary statistics used to draw that inference have casted doubt on this claim. Here, we reassess the contribution of natural selection to height variation by integrating complementary methods and apply them to European ancestry populations.

Methods and results:
Leveraging high-powered multi-ancestry GWAS datasets, including within-family designs robust to population stratification, and allele frequency data from 13 populations, we detect signals of recent selection at height-associated SNPs using trait-specific Singleton Density Score (tSDS) analyses and provide new evidence of local adaptation across 13 European populations using the Q_X statistic. Importantly, these signals persist after accounting for ancient admixture events.

Conclusion:
Our findings provide converging lines of evidence supporting positive selection of height-increasing alleles in European populations and highlight the complexity of polygenic adaptation in humans.