Abstract:

Detecting human genetic adaptation in recent human history is a major challenge of population genetics. The fundamental problem is to infer historical changes in the frequency of genetic variants (alleles), from data of contemporary genomes. With this we can identify unusual changes that are unlikely to have occurred in the absence of selective pressures. However, a generally applicable method to infer recent allele frequency changes is lacking. Instead, present methods can only detect frequency changes under very restrictive assumptions on the model of selection. Moreover, their time resolution is generally limited to prehistoric scales, on the order of the past 25-75 thousand years. To address these gaps we developed a novel statistical method, Singleton Density Score (SDS), that infers the recent changes in allele frequencies from local variation in contemporary genome sequences with specificity to historical timescales. Applied to data of ~3000 genomes from the UK10K project, SDS reveals that human genetic adaptation continued well into historical times. Over the past ~2000-3000 years, ancestors of modern Britons genetically evolved over a range of phenotypes related to diet, immunity, and physical appearance. Notably, we found that polygenic adaptation, whereby selection acting on many small-effect variants across the genome that together determine a single trait, has played a pervasive, previously undetected role in shaping present genotypic and phenotypic variation.

Reference: