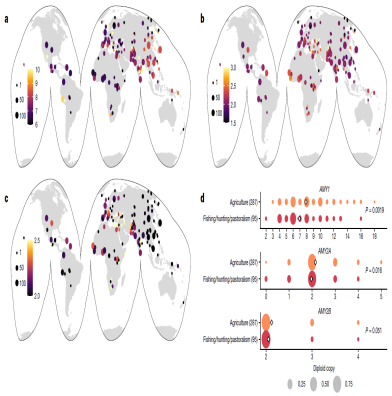
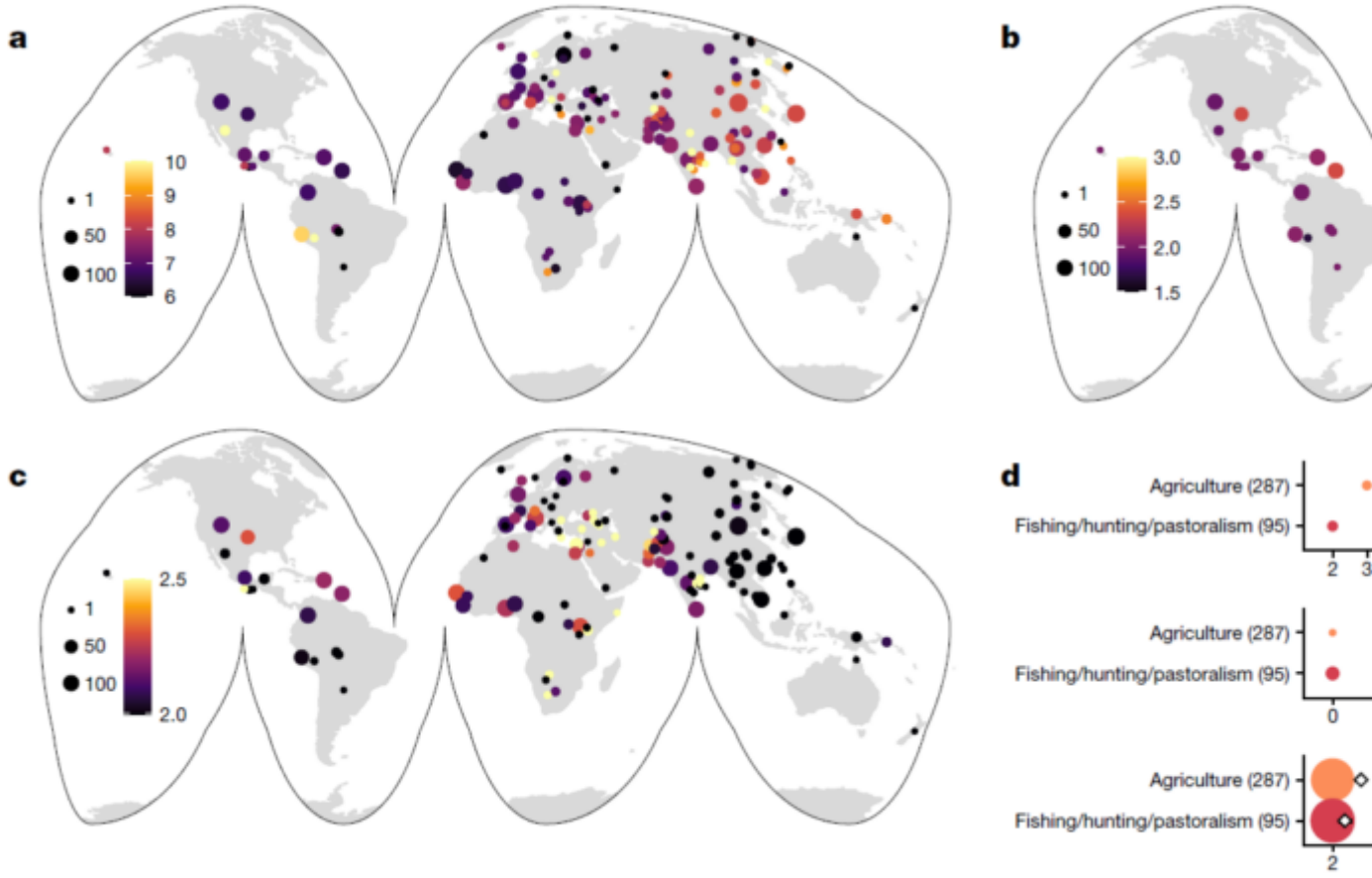


Recurrent evolution and selection shape structural diversity at the amylase locus

Description

The introduction of agriculture triggered a rapid shift toward starch-rich diets in the human population. Amylase genes facilitate starch digestion, and increased amylase copy number has been observed in some modern human populations with high starch intake, although evidence for recent selection is lacking. The authors used 94 long-read haplotype-resolved assemblies and short-read data from approximately 5,600 contemporary and ancient humans. They resolved the diversity and evolutionary history of structural variation at the amylase locus. They found that amylase genes have higher copy numbers in agricultural populations than in fishing, hunting, and herding populations.



Worldwide amylase copy number diversity. aâ??c, World maps indicating average AMY1 (a), AMY2A (b) and AMY2B (c) copy number in 147 different human populations. The point size indicates population sample sizes (ranging from 1 to 134), and the colour indicates the mean copy number. Copy number distributions across individual populations and continental groups are displayed in Extended Data Fig. 1. d, Copy number distributions of AMY1 (top), AMY2A (middle) and AMY2B (bottom) in 33 modern human populations with traditionally agricultural subsistence compared with fishing-based, hunting-based and pastoralism-based diets. Numbers in parentheses indicate sample size. Two-sided P values of a Studentâ??s t-test are shown without adjustment for multiple comparisons.

The authors have identified 28 different amylase structural architectures and show that nearly identical structures have repeatedly emerged on different haplotype backgrounds throughout recent human history. The AMY1 and AMY2A genes have each undergone multiple duplication/deletion events with mutation rates up to more than 10,000 times the single nucleotide polymorphism mutation rate. In contrast, the AMY2B gene duplications share a single origin. Using a pangenome-based approach, the authors infer in thousands of humans, identifying extensively duplicated haplotypes at higher haplotypes at higher frequencies in modern agricultural populations. Leveraging 533 ancient human genomes, they found that duplication-containing haplotypes (with more gene copies than the ancestral haplotype) have rapidly increased in frequency over the last 12,000 years in Western Eurasians, suggesting positive selection. The study highlights the potential impact of the agricultural revolution on human genomes and the importance of structural variation in human adaptation.

Category

1. News