BITS :: Call for Abstracts 2022 - Oral communication

Type	Oral communication
Session	Molecular Evolution analysis
Title	Ancestral genomic contributions to complex traits in contemporary Europeans
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Motivation

The contemporary European genetic makeup formed in the last 8000 years when local Western Hunter-Gatherers mixed with incoming Anatolian Neolithic farmers and Pontic Steppe pastoralists. Previous studies inferred phenotypes in these source populations and investigated how they were influenced by natural selection. However, how ancient populations contributed to present-day phenotypic variation is poorly understood. Here we investigate how the unique tiling of genetic variants inherited from different ancestral components drives the complex traits landscape of contemporary Europeans, and quantify selection patterns associated with these components.

Methods

Using matching individual-level genotype and phenotype data for 27 traits in the Estonian biobank and genotype data directly from the ancient source populations, we quantify the contributions from each ancestry to present-day phenotypic variation in each complex trait. To do so we introduce covA, a measure of the relative similarity between any contemporary individual and the ancestries that contributed to its genetic makeup, which can be computed genome-wide or for regions of interest. We then apply it to candidate regions derived from GWAS catalog hits for each trait analyzed.

Results

We find substantial differences in ancestry for eye and hair colour, body mass index, waist/hip circumferences and their ratio, height, cholesterol levels, caffeine intake, heart rate and age at menarche. Furthermore, we find evidence for recent positive selection linked to four of these traits and, in addition, sleep patterns and blood pressure.
Our results show that these ancient components were differentiated enough to contribute ancestry-specific signatures to the complex trait variability displayed by contemporary Europeans.

Info

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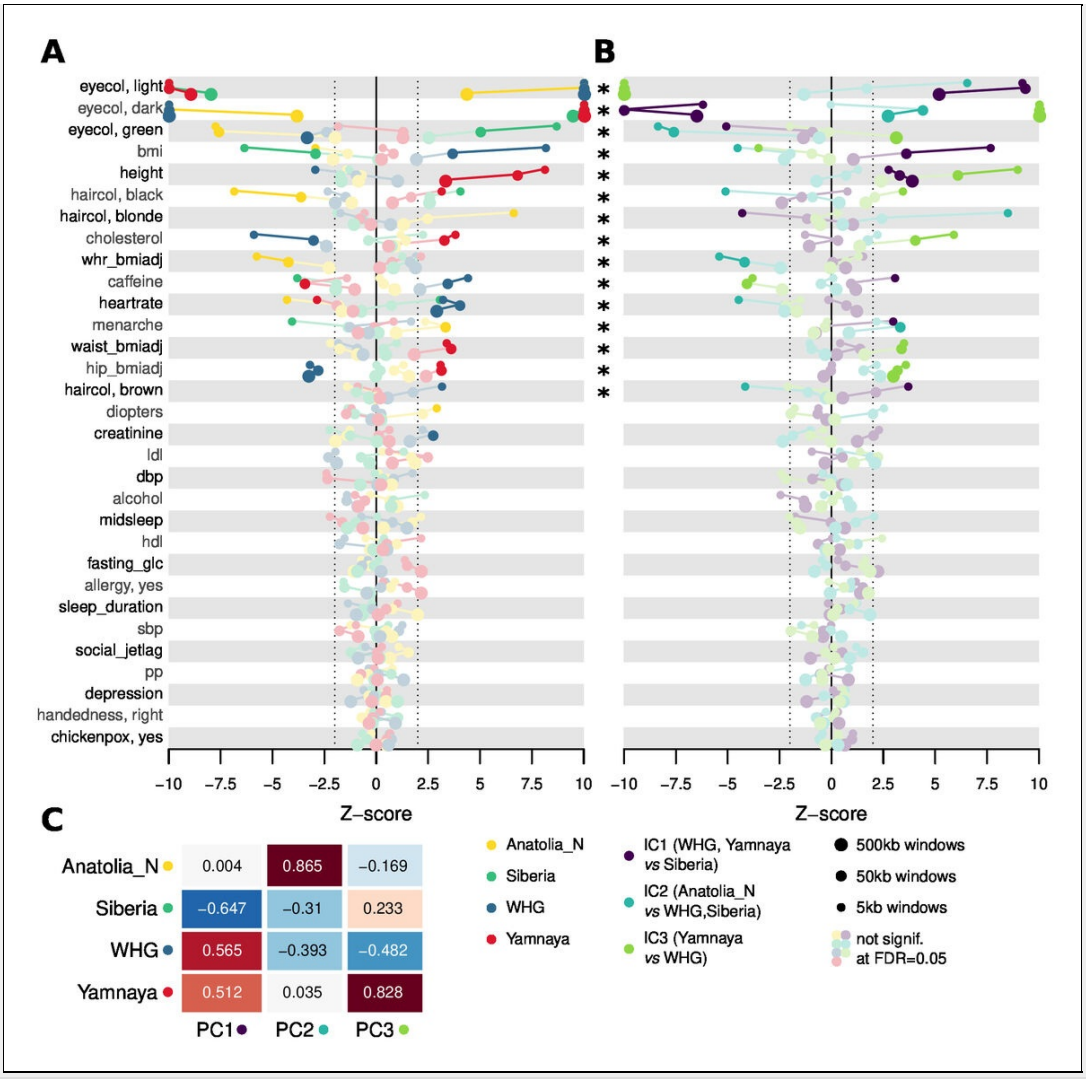
Image caption:
Ancestry-trait association on candidate regions. (A) Z-scores of covA coefficients, the color refers to the ancestry tested. (B) Z-scores of coefficients associated with covA independent components (IC) computed with whole genome-based covA PC loadings. Each color is associated with one of the three ICs. For each trait we show the Z-score of the standardized coefficient associated with candidate regions against a distribution of 50 random genomic regions of matching size. Candidate regions are determined around GWAS hits for appropriate traits as windows with three different widths: 5 (small dot), 50 (medium dot), and 500 (large dot) kilobases. Pastel dots are deemed not significant at Benjamini-Hochberg FDR = 0.05, p value from double-sided Z-test; asterisks mark traits to be considered significant according to (B); dotted lines correspond to absolute Z-scores =2. (C) Loading matrix for genome-wide covAs and their PCs, used to transform covAs into their ICs.

filename	candidatecova.jpg

Figure



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