

Searching genomic signals of natural selection and local adaptation in populations of Russia

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The identification of genomic loci that are subjected to natural selection provides both deeper insights into the existence of population genetic structures and understanding the genetic bases of population-level differences in the distribution of common diseases and traits. In our study we searched the signatures of positive selection in the genome-wide data of 10 populations from Russia that, until recently, remained a white spot on the genomic map of human local adaptations. To detect signals of selection, we used the population branch statistic (PBS), which was calculated for sliding windows of 20 SNPs with a step size of 5 SNPs. The chromosome regions containing windows with PBS values above the 99.9th percentile of the empirical distribution were candidate regions and were further annotated. Totally, 936 genes were annotated (72 to 169 genes per population). The functional evaluation of the genes showed that many of them were significantly overrepresented in particular biological processes (metabolic pathways). Some of these pathways were identified across multiple populations – groups of populations, united by their history of origin or geography of residence: 1) the processes of alcohol metabolism, retinol and retinoic acid associated with ADH1A, ADH1B and ADH1C in Veps, Komi Priluzski, Khanty, Mansi and Nenets, 2) the metabolism of unsaturated fatty acids associated with FADS1, FADS2 and FADS3 in Khanty, Mansi, Nenets and Yakuts, and 3) the metabolism of thyroid hormones associated with DUOX1, DUOX2, DUOXA1 and DUOXA2 in Nenets and Yakuts. Others were population-specific: glucuronization processes (Veps), retinoic acid metabolism by cytochromes (northern Russians), cornification (Komi Izhemski), etc. At the same time, the vast majority of the genes were not associated with any pathways and processes. This suggests the complexity of adaptation processes, which in each specific case could be going or supplemented by adjusting individual elements of a variety of metabolic pathways to the characteristics of the habitat.

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