GEOGRAPHIC VARIATION OF THE HUMAN GENE POOL: THE GLOBAL PATTERNS

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The studies of genetic variation in human populations started 100 years ago: in 1914, the pronounced differences in frequencies of blood groups were revealed for the first time. During the century-long history of intensive research the arsenal of population geneticists has changed six times. The immunological markers or blood groups (1) were only available genetic systems for decades until biochemical markers (2) were widely introduced in 1960s. Both types are now known as "classical markers"; the datasets on their variation in human variations worldwide are large and have been summarized by both Western (Cavalli-Sforza et al., 1994) and Russian (Gene pool and gene geography of USSR) scientific gene geographical schools. The classical markers are virtually out of experimental use in present days. But because their variation has been well described and analyzed, these generalized conclusions are widely used as a background for current research. Since 1990s, the mitochondrial DNA (3) and Y-chromosome (4) became the most popular genetic systems in population studies. Hundreds of papers were dedicated to their variation, and accumulated datasets include hundreds thousands of samples from thousands of populations worldwide. However, the number of papers on these markers decreases each year, because the genome-wide (5) and full genome (6) markers are becoming the new favorite tools in the arsenal of researchers, but data on these genetic systems are not abundant yet. Thus, the crucial task is to summarize the accumulated data on mitochondrial DNA and Y-chromosomal variation, to extract the generalized patterns and to make the overall conclusions of the global gene pool structure from these two kinds of genetic data. Then these systems will be for many decades the great story-tellers about general trends in human variation and value of these "new classics" will be high even when no living researcher will remember experimental methods for their analysis. The talk will present the largest databases on mitochondrial DNA and Y-chromosomal variation worldwide and the cartographic atlases summing up both particular and universal patterns of these systems. This study was supported by RFBR grants 13-04-0171, 13-06-00670, 14-06-00384, 13-04-90420, 14-06-31331 and Presidium RAS program "Molecular and cell biology".

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