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Lessons from the Whole Exome Sequencing Effort in Populations of Russia and Tajikistan

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Abstract

© 2016, Springer Science+Business Media New York. In contrast with the traditional methods applied to assessment of population diversity, high-throughput sequencing technologies have a wider application in clinical practice with greater potential to find novel disease-causing variants for multifactorial disorders. Widely used test panels may not meet their goal to diagnose the patient's condition with a full reliability since this method often does not take into account the population frequencies of analyzed genetic markers. Here, we analyzed 57 male individuals of five ethnic groups from Russia and Tajikistan using the whole exome sequencing technique (Ion AmpliSeq Exome), which resulted in detecting more than 299,000 single nucleotide polymorphisms. Samples formed clusters on the PCA plot according to the geographical location of the corresponding populations. Thereby, the methodology of whole-exome sequencing, in general, and the Ion AmpliSeq Exome panel, in particular, could be positively applied for the purposes of population genetics and for detection of the novel clinically relevant variants.

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Keywords

Diagnostic panel, Ion AmpliSeq Exome, North Eurasian populations, SNP, Whole exome sequencing