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Whole-exome sequencing for monogenic diabetes in Russian children reveals high frequency of genetic variants in MODY-related and unrelated genes

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Using whole-exome sequencing, we identified the frequency and the spectrum of genetic variants causative of monogenic diabetes in 60 Russian children with non-type 1 diabetes mellitus. Genetic variants were screened in a total of 35 genes: 13 genes causative of MODY (HNF4A(MODY1), GCK(MODY2), HNF1A(MODY3), PDX1(MODY4), HNF1B(MODY5), NEUROD1(MODY6), KLF11(MODY7), CEL(MODY8), PAX4(MODY9), INS(MODY10), BLK(MODY11), ABCC8(MODY12), KCNJ11(MODY13)), and 22 genes causative of transient or permanent neonatal diabetes, including the ones related to specific syndromes (EIF2AK3, RFX6, WFS1, ZFP57, FOXP3, AKT2, PPARG, APPL1, PTF1A, GATA4, GATA6, GLIS3, IER3IP1, LMNA, NEUROG3, PAX6, PLAGL1, SLC19A2, SLC2A2, SH2B1, SERPINB4, MADD). Overall, 33 out of 60 patients (55 %) had genetic variants in the target genes. Of all 33 positive patients, 27 (81.8 %) had genetic variants in MODY-related genes. The majority of these patients – 19 out of 27 – had genetic variants in GCK (MODY2). We analyzed the relationship of the detected genetic variants to the patients' diabetic phenotypes. Among 38 detected genetic variants, 23 have been reported previously to be linked to monogenic diabetes and 15 were novel ones. Thus, our results together with the results of other studies show that a higher mutation detection rate may be achieved by increasing number of genes tested. In this regard, one more advantage of WES should be mentioned: DNA sequencing data may be easily stored for further analysis of newly discovered candidate genes. Ethnic differences play an important role in determining epidemiology of monogenic diabetes, especially of MODY.

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