Pathogenetic heterogeneity of diabetes mellitus in children of Saint-Petersburg city.

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Aim of our scientific work:

The goal is to determine the frequency of occurrence and molecular-genetic characteristics of MODY in patients aged between 1 and 18 years old - residents of St. Petersburg.

Methods:

We examined 54 patients with suspected hereditary variants of diabetes with chronic hyperglycemia, detectable c-peptide for 2 years after the diagnostic of the disease, the absence of diabetic autoantibodies and the absence of signs of a metabolic syndrome.

Results:

In our study of DNA of patients with suspicion of MODY was performed by next generation sequencing. NGS-diagnostic panels were used to study the coding regions of genes, including the following: *HNF1A, GCK, HNF4A, HNF1B, PDX1, NEUROD1, KLF11, CEL, PAX4, INS, BLK, EIF2AK3, RFX6, WFS1, ZFP57, FOXP3, KCNJ11, ABCC8, GLUD1, HADH (SCHAD), SLC16A1, UCP2, INSR, AKT2, GCG, GCGR, PPARG, PTF1A.*

Clinical diagnosis was confirmed by molecular-genetic analysis in 32 children, which was 59% of all examined. The most common mutations in the *GCK* gene were 81.25% (n=26), *HNF1A* 12.5% (n=4), *WFS1* 3.12% (n=1), *PAX4* = 3.12% (n=1). The prevalence of MODY among all cases of DM in pediatric patients, respectively, was 2%.

Conclusions:

When using the basic differential diagnostic criteria to establish MODY, the molecular genetic confirmation of the diagnosis among patients suspected for MODY amounts to 59%.