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Description of the clinical case of the patient *HNF1A* (MODY3) with familial segregation of the disease in five generations

Alla Ovsyannikova[a], **Oksana Rymar**[a], **Elena Shakhtshneider**[a] and **Mikhail Voevoda**[a]
[a]IIPM-ICG SB RAS, Russia

Aim: The purpose of the research was to determine family segregation in a proband family with a confirmed mutation in the *HNF1A* gene (MODY3 diabetes).

Materials & Methods: The diagnosis of HNF1A-MODY was verified by the proband and his relatives on the basis of direct automatic sequencing and sequencing by Sanger.

Results: Proband - a woman of 50 years, gestational diabetes mellitus (DM) is diagnosed at the age of 21 during pregnancy, insulin therapy was done in the basis-bolus regimen. After delivery insulin therapy was canceled, the patient had a strict diet. Decompensation of carbohydrate metabolism was at the age of 28 years on the background of stress, insulin therapy was prescribed in the basal-bolus regimen, which is still preserved. Non proliferative diabetic retinopathy, peripheral neuropathy, dyslipidemia were defined. Antibodies to B-cells and glutamate decarboxylase were negative and the C-peptide was slightly reduced. DM was at the great-grandfather, the grandmother from 65 years, mother of the proband from 45 years, at the son from 21 years. Mother and son of the proband had dyslipidemia with increased cholesterol of low density lipoproteins, in grandmothers and great grandfathers- macrovascular complications (acute cerebrovascular accident) at a late age. A previously unrecognized mutation in the 1 exon of the *HNF1A* gene in a proband, her mother and her son was revealed according to molecular genetic research. An identical mutation was detected in the granddaughter of a proband at the age of 2 months.

Conclusions: The pedigree of HNF1A-MODY demonstrates the phenomenon of genetic prediction that is the gradual decrease in the age of diagnosis in subsequent generations probably due to increased awareness leading to earlier testing of glucose levels.

The decrease in the age of development of dyslipidemia in patients with a mutation in the *HNF1A* gene was also determined which may be interrelated characteristics and require further study.

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Biography

Alla Ovsyannikova finished Novosibirsk Medical University, Russia in 2008 year with Honors. She had two certificates of specialists in Internal Medicine and Endocrinologist. She is a PhD since 2013 with dissertation work "Diabetes mellitus in young people: some clinical and molecular genetic aspects". Currently, she is working as an Endocrinologist and Scientist in IIPM-Branch of IC&G SB RAS. Her research work is about monogenic types of diabetes mellitus (especially MODY diabetes) in young patients. She investigates the characteristics of the clinical course, treatment and genetic features in Siberian and Russian population. She has published more than 20 abstracts in national refereed journals, 30 abstracts in conference with international participation. She has participated in international conferences.

aknikolaeva@bk.ru

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