

Clinical Cases of Rare Types of 'Maturity Onset Diabetes of the Young' Diabetes

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Abstract : In Siberia endocrinologists increasingly noted young patients with the course of diabetes mellitus differing from 1 and 2 types. Therefore we did a molecular genetic study for this group of patients to verify the monogenic forms of diabetes mellitus in them and researched the characteristics of this pathology. When confirming the monogenic form of diabetes, we performed a correction therapy for many patients (transfer from insulin to tablets), prevented specific complications, examined relatives and diagnosed their diabetes at the preclinical stage, revealed phenotypic characteristics of the pathology which led to the high significance of this work. **Materials and Methods:** We observed 5 patients (4 families). We diagnosed MODY (Maturity Onset Diabetes of the Young) during the molecular genetic testing (direct automatic sequencing). All patients had a full clinical examination, blood samples for biochemical research, determination of C-peptide and TSH, antibodies to b-cells, microalbuminuria, abdominal ultrasound, heart and thyroid ultrasound, examination of ophthalmologist. **Results:** We diagnosed 3 rare types of MODY: two women had MODY8, one man - MODY6 and man and his mother - MODY12. Patients with types 8 and 12 had clinical features. Age of onset hyperglycemia ranged from 26 to 34 years. In a patient with MODY6 fasting hyperglycemia was detected during a routine examination. Clinical symptoms, complications were not diagnosed. The patient observes a diet. In the first patient MODY8 was detected during first pregnancy, she had itchy skin and mostly postprandial hyperglycemia. Upon examination we determined glycated hemoglobin 7.5%, retinopathy, non-proliferative stage, peripheral neuropathy. She uses a basic bolus insulin therapy. The second patient with MODY8 also had clinical manifestations of hyperglycemia (pruritus, thirst), postprandial hyperglycemia and diabetic nephropathy, a stage of microalbuminuria. The patient was diagnosed autoimmune thyroiditis. She used inhibitors of DPP-4. The patient with MODY12 had an aggressive course. In the detection of hyperglycemia he had complaints of visual impairment, intense headaches, leg cramps. The patient had a history of childhood convulsive seizures of non-epileptic genesis, without organic pathology, which themselves were stopped at the age of 12 years. When we diagnosed diabetes a patient was 28 years, he had hypertriglyceridemia, atherosclerotic plaque in the carotid artery, proliferative retinopathy (laserocoagulation). Diabetes and early myocardial infarction were observed in three cases in family. We prescribe therapy with sulfonylureas and SGLT-2 inhibitors with a positive effect. At the patient's mother diabetes began at a later age (30 years) and a less aggressive course was observed. She also has hypertriglyceridemia and uses oral hypoglycemic drugs. **Conclusions:** 1) When young patients with hyperglycemia have extrapancreatic pathologies and diabetic complications with a short duration of diabetes we can assume they have one of type of MODY diabetes. 2) In patients with monogenic forms of diabetes mellitus, the clinical manifestations of hyperglycemia in each succeeding generation are revealed at an earlier age. Research had increased our knowledge of the monogenic forms of diabetes. The reported study was supported by RSCF, research project No. 14-15-00496-P.

Keywords : diabetes mellitus, MODY diabetes, monogenic forms, young patients

Conference Title : ICDM 2018 : International Conference on Diabetes and Metabolism

Conference Location : Rome, Italy

Conference Dates : May 03-04, 2018