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Clinical and genetic features of diabetes mellitus in young patients in Siberia

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The prevalence of diabetes mellitus (DM) is increasing worldwide in epidemic proportions, as well an increasing number of young patients with this nosology. First in Siberia we did molecular genetic studies of glucokinase gene (MODY 2) and researched the characteristics " of diabetes is not type 1" in young patients. First, we have developed an algorithm for the differential diagnosis of the type of diabetes in young patients in the presence of family history. Also for the first time, we had formed the indications for genetic research glucokinase gene under the Siberian region.

The aim of the research work was to investigate the clinical and laboratory characteristics and some molecular genetic determinants of diabetes in patients with onset before 25 years and with the presence of family history of diabetes in Siberia (on the example of the city of Novosibirsk).

Materials and Methods: We examined 78 patients with a debut of DM before 25 years old with a family history of diabetes: 33 patients with type 1 diabetes mellitus (DM 1), 33 - with type 2 diabetes mellitus (DM 2) and 12 - with type of diabetes is not verified. We did a full clinical examination, blood sampling for biochemical analysis, determination of C-peptide, glycosylated hemoglobin, antibodies to b-cells in all patients. We selected group of patients for molecular genetic studies of glucokinase gene (the most common subtype of MODY diabetes in Russian) after receiving results of laboratory data. It was 22 patients:

- 2 patients had atypical characteristics for type 1: absence of autoantibodies, the need for a small amount of insulin: 10 and 8 units per day for the duration of the disease 4 and 6 years, normal level of C- peptide;
- 8 patients had atypical characteristics for type 2 diabetes: the absence of obesity, diabetes asymptomatic;
- 12 patients had type of diabetes unverified.

Results : MODY 2 diabetes was confirmed by molecular genetic studies in 5 of 22 patients (23 % and 11 % for 'DM not type 1"): in 3 patients in this group was not verified by the type of diabetes early , one is a relative of a proband with confirmed MODY 2 and on one patient was previously verified DM 2. We diagnosed four missense mutations: two mutations in exon 7, two identical mutations in exon 4 (proband and the relative first degree of consanguinity), and mutation in exon 1, previously undescribed. We formed 3 groups of patients to interpret their results after conducting this study: 33 patients with diabetes type 1, 32 people with diabetes 2, 5 persons with MODY 2. Patients with diabetes type not verified will require further observation and their test results were not included.

We researched clinical and laboratory characteristics of their diabetes patients who develop up to 25 years based on the results. We found the "classical" symptoms of DM 1 in 94 % of patients. All patients had the C- peptide level below the reference value , 76% of patients achieved the target values HbA1c, antibodies to b- cells were positive for 3 patients (9 %). All patients used insulin.

59 % of patients with type 2 diabetes had not clinical manifestations of disorders of carbohydrate metabolism and the diagnosis was on the basis of laboratory data in routine surveys. Overweight and obesity had been diagnosed in only 16% of patients. Level of C- peptide in patients with type 2 diabetes was within reference range with a tendency to decrease, 47 % of patients used a balanced diet.

Age at diagnosis of MODY 2 ranged from 6 months to 25 years. In 3 of 5 patients had not clinical appearances of carbohydrate metabolism disorders. Overweight and obesity had not been detected in any patient. All patients diagnosed with MODY 2 diabetes were achieved targets HbA1c, C-peptide level was below the reference range. Three of the five patients used a balanced diet to achieve normoglycemia.

We revealed that the main differences from MODY 2 to DM 1 were: lack of antibodies to b- cells, lack of ketoacidosis at the time of diagnosis, no absolute need for exogenous insulin, a lower level of HbA1c, stored or slightly reduced secretory activity of b-cells. The main differences between MODY 2 of 2 DM were: lower age of patients at the time of diagnosis, a lower level of HbA1c, the absence of obesity.

Conclusions:

1. Ketoacidosis, weight loss, polydipsia and polyuria are a major clinical manifestation of symptoms in type 1 diabetes with onset before 25 years of age and family history of diabetes. We found preferential absence of characteristic clinical symptoms of carbohydrate metabolism disorders (polydipsia, polyuria), low rates of C-peptide in the blood serum and the absence of overweight and obesity in 84 % for patients with "diabetes is not type-1".

2. MODY 2 was detected in 11 % among the examined patients with "diabetes is not type-1". In patients with clinical manifestations of MODY 2 diabetes were found one new missense mutation and three known mutations in glucokinase. Identified mutations are located at 7, 4 and 1 glucokinase gene exons.

3. A family history of diabetes, mild or asymptomatic manifestation of the disease, lack of obesity, specific autoantibodies, normal values of C-peptide, no absolute need for exogenous insulin, achieving targets of carbohydrate metabolism - indications for molecular genetic studies definition of MODY 2 diabetes under the Siberian region.

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