A CASE OF MATURITY ONSET DIABETES OF THE YOUNG

Covanțev Serghei[1], Chiriac Andrian[2], Perciuleac Ludmila[3]

[1] - Medical student SMPhU "Nicolae Testemițanu"
[2] - Department of Pediatric Endocrinology, Republican Clinical Hospital for Children "E. Coțaga"
[3] - Department of Molecular Biology and Genetics, SMPhU "Nicolae Testemițanu"

Summary

Diabetes is a worldwide problem with polygenic etiology. Nevertheless 10% of patients with type 1 and 5% of patients with type 2 diabetes have a monogenic form of this disease which has different treatment options and usually has fewer complications. We present a case analysis of a patient that has an unusual type of diabetes, presumably MODY, successfully treated with metformin.

Key words: MODY, monogenic diabetes.

Abstract

Un caz de diabet zaharat al tinerilor cu debut la maturitate

Diabetul este o problemă mondială cu etiologie poligenică. Cu toate acestea 10% dintre pacienții cu tipul 1 și 5% din pacienții cu diabet zaharat de tip 2 au o formă monogenică a acestei boli, care are diferite opțiuni de tratament și, de obicei, are mai puține complicații. Prezentăm o analiză de caz al unui pacient care are un tip neobișnuit de diabet, probabil MODY, tratat cu succes cu metformin.

Cuvinte cheie: MODY, diabet zaharat monogenic.

Introduction

An analysis of 199 counties estimated that the number of adults with diabetes has doubled from 153 million in 1980 to 347 million in 2008. By 2030 the number of individuals with diabetes worldwide may rise to 472 million. 80% of them will be in low and middle income countries. In some of these countries diabetic drugs and insulin are inaccessible or rather expensive which eventually affect the whole healthcare system [5].

Not all of these patients have the classical types of diabetes. Some of them may have monogenic forms with sometimes different presentations and better treatment options if they are diagnosed properly and in time.To date, several transcriptional factors and an enzyme are associated with maturity onset diabetes of the young (MODY). Current data describes 11 types of MODY [3]. These mutations are characterized by high level of penetrance. 95% of individuals that have MODY will become diabetic or will have an altered glucose metabolism before the age of 55. Misdiagnosis of MODY is a widespread problem. It is estimated that about 5% of patients with T2D and about 10% of type 1 diabetes (T1D) are misdiagnosed having actually MODY [1].

A study performed by G. Thanabalasingham et al., 2012 showed that 20 of 247 patients clinically labeled as T1D actually had HNF-1a or HNF-4a mutations. From 322 patients clinically labeled as T2D 80 had HNF-1a or HNF-4a mutations and 40 had GCK mutation [4]. E.-J. Schiopu, 2011 reports that MODY usually develops in 22% of cases due to GCK mutations, in 66% of cases due to mutations in transcription factors, less than 1% in case of insulin gene (INS) mutations, less than 1% due to carboxyl ester lipase mutation and about 11% due to other mutations sometimes of unknown etiology [2].

Material and Methods

In the case study we describe a patient with an unusual presentation of diabetes mellitus. The diagnosis was established using glucose tolerance test, glycated hemoglobin, blood biochemical analysis. Additional investigations included ultrasonography.

Results

A 15 year old female was diagnoses with diabetes (HbA1c 9.1%; blood glucose 7,8 mmol/l; 16,2 mmol/l - 1 hour after meal; 13,1 mmol/l – 2 hours after meal) which is clinically similar to type I diabetes. The mother and sister had a history of diabetes treated with insulin. Besides the altered glucose tolerance she had II grade obesity (with Body Mass Index 29,8 kg/m2), hyperlipidemia (with elevated cholesterol, lipoproteins), nontoxic (eutiroid) diffuse goiter. In a one year period she additionally developed diabetic nephropathy II gr. and liver steatosis. Metformin was used as a treatment option in this case with good results though there were still alterations in glucose metabolism.

In this case genetical testing could reveal the exact mutation of the gene and give us a better understanding of the disease and the possibility for better treatment options.

Discussions and conclusions

In Republic of Moldova there are approximately 60 thousand diabetic patients, which mean that represents somewhere 50% of the endocrine diseases. It means that there is a great possibility that not all diabetic patients receive the proper treatment. Some of them may need oral diabetic medication, others can live a proper life without any medication at all. Patients that receive insulin as drug of choice may have with time compromised β-cell function with a wide range of severe complications. Further investigation in this area can improve the possibilities for diagnosis and treatment of genetic diabetes, its complications and improve the live quality and expectancy in the country.

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Covanțev Serghei Catedra Biologie Moleculară și Genetică Umană USMF „Nicolae Testemițanu"
Chișinău, Bd. Ștefan cel Mare și Sfânt, 165 Tel.: 022548005 mob. 069948085 E-mail:kovantsev-serghei@gmail.com