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P-21 Maturity onset diabetes of the young 7: Case report

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Introduction: Maturity Onset Diabetes of the Young (MODY) is a rare form of diabetes which is different from both type 1 and type 2 diabetes, and runs strongly in families. MODY is autosomal dominant inherited type of diabetes, caused by a mutation (or change) in a single gene. If a parent has this gene mutation, any child they have, has a 50% chance of inheriting it from them. Recently, 14 gene mutations have been found to be associated with MODY. The Krüppel-like factor 11 (KLF11) gene mutation is the pathogenic gene of MODY7. **Clinical Case:** A 17-year-old female patient with a previous history of obesity was admitted at outpatient office for regular control of weight and body mass index (BMI). Two years ago the patient was diagnosed with obesity, hyperinsulinemia and insulin resistance, with body weight (BW) 90 kg, body height (BH) 167 cm and BMI 32,3. The patient is on therapy with Metformin 2×500 mg. Results of the moment of examination: BW 100 kg, BH 168 cm, BMI 35,4; HbA1c 5,96%; Fasting plasma glucose (FPG) 6.8 mmol/l; Insulinemia 13,17; C-peptide 1,00; oral glucose tolerant test (OGTT): 0' 6,5 mmol/l 120' 11,0 mmol/l; insulinemia during OGTT (μU/mL): 0' 14,05; 120' 98,75; C-peptide during OGTT (ng/mL): 0' 1,00; 120' 6.95; Fundus examination and Doppler ultrasonography of both lower limbs showed no abnormalities. Family history was positive for Diabetes mellitus type 2 (mother, grandmother). The Immunological tests for Diabetes mellitus type 1 were negative for antibodies: GAD65, IAA, ICA and IA-2; Because there was doubt for MODY, genetic tests were performed with targeted resequencing of 4800 clinically relevant genes. The presence in the KLF11 gene associated with MODY7 was determined, c.911>G, p.(Lys304Arg). The change is found in heterozygous form and is inherited from the mother. The patient continued treatment with metformin and lifestyle changes and this resulted in weight loss and good glycoregulation. Control one year later showed weight loss and good glycoregulation, FPG 5.1 mmol/l; HbA1c 5.5%; BW 89 kg, BH 168 cm, BMI 31.5; The level of blood glucose was controlled and stable. The patient's mother and grandmother were also treated with metformin. **Conclusion:** Young patients with signs of DM2 should be tested for genetic types of diabetes. The number of patients with MODY is small, especially regarding MODY7, which rarely occurs. MODY is the most common type of monogenic β-cell function defect. It accounts for 2% to 15% of young diabetic patients. Studies have found that oral hypoglycemic drugs and dietary interventions are beneficial for MODY7 patients, and the control of the intake of staple food carbohydrates helps to control blood glucose which was also shown in our patient (1).

1. Chen L, Hou X. Clinical and Functional Characteristics of a Novel KLF11 Cys354Phe Variant Involved in Maturity-Onset Diabetes of the Young. J Diabetes Res. 2021;2021:7136869