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MODY3 early identification and diagnosis

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Background

MODY is monogenic. About 1% of diabetes has a monogenic cause but is frequently misdiagnosed as DM1 or DM2.

Objective

It is important to study family history of patients with atypical diabetes forms for verification of diagnosis and prognosis.

Subjects and Methods:

Genetic, biochemical and hormonal testing, 2 patients were examined comparisons.

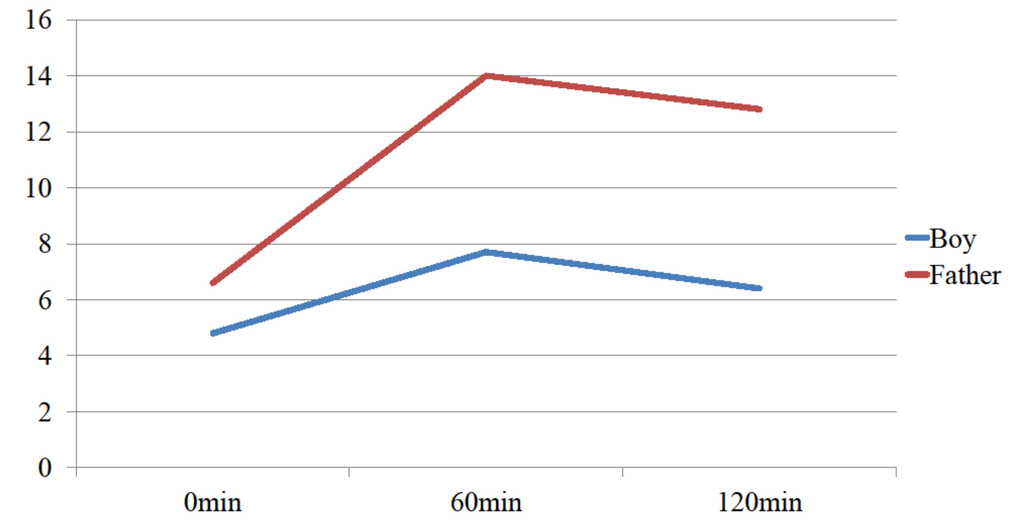
Results:

At pre-school medical examination a general practitioner noticed positive family history of diabetes of an 8 year old boy and sent him to an endocrinology center. It was noted that the proband's father had fasting hyperglycemia without symptoms of diabetes and obesity at the age of 18. He was not examined for 16 years as he did not have any diabetes symptoms. When he was 34, occasionally FPG 7.4 mmol/l and after meal 12.3 mmol/l without ketosis was detected. He was diagnosed DM1 and insulin was prescribed. At the moment of examination he had diabetes for 6 years and received insulin in dose 0.1U/kg/day. HbA1c was 6.6%. FPG was 6.6 mmol/l, stimulated glucose (after standard carbohydrate breakfast without insulin injection) was 14 mmol/l (1h), 12.8 mmol/l (2h). Basal insulin serum was 2.4U/l, stimulated insulin serum was 20.9 U/l (1h), 23.7 U/l (2h) (Pic. 1, 2). The boy's paternal grandmother had GDM at the age of 23 years. At the moment of examination she had diabetes for 40 years without complications and received insulin in dose 32U/d (0.5U/kg/day). Her two sisters had DM (Pic.3).

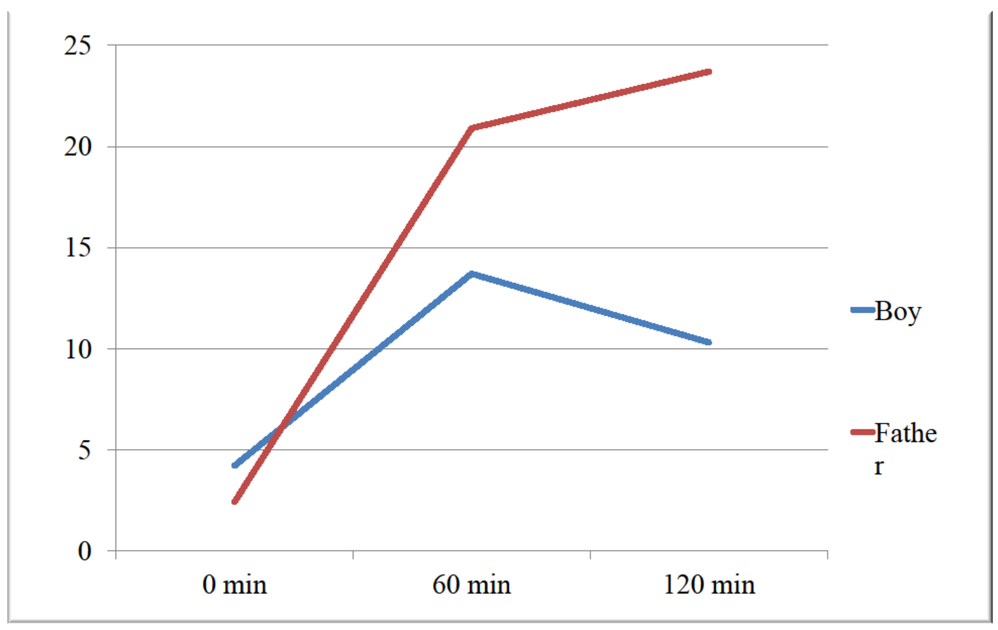
At the moment of examination the proband did not have hyperglycemia and diabetes symptoms. FPG was 4.2 mmol/l, stimulated glucose (OGTT) was 7.7 mmol/l (1h), 6.4mmol/l (2h). Basal insulin serum was 4.2U/l, stimulated insulin serum was 13.7 U/l (1h), 10.3 U/l (2h). (Pic. 1, 2).

Genetic testing revealed that the proband and his father had mutation **S355X in the *HNF1A* gene**. The proband's father was prescribed sulphonylurea tablets instead of insulin and this resulted in glycemia stabilization.

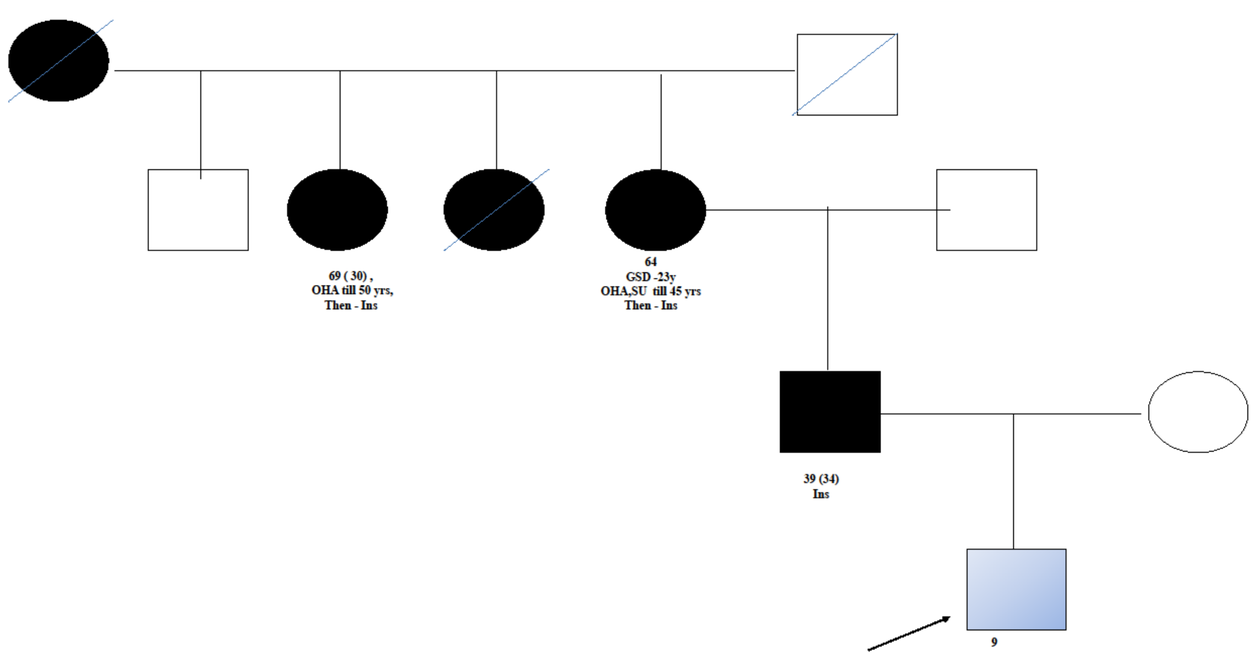
Pic.1 Fasting and postprandial glucose levels (mmol/l)



Pic.2 Fasting and postprandial insulin levels (mIU/ml)



Pic.3 Pedigree of the family



INS- insulin; OHA - oral hypoglycaemic agents; SU-sulfonylurea

Conclusion:

Correct genetic diagnosis is important for proper patients' treatment and prognosis and it enables predictive genetic testing for their asymptomatic relatives.

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