A RARE CLINICAL CASE OF A COMBINATION OF MONOGENIC AND AUTOIMMUNE DIABETES MELLITUS.

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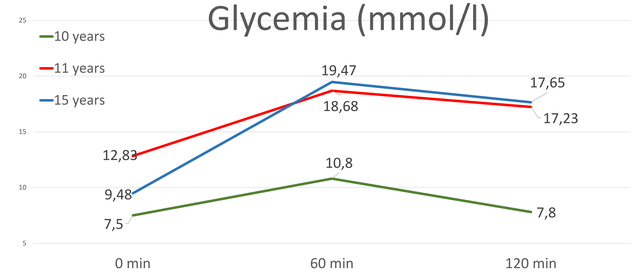
INTRODUCTION

Type 1 diabetes mellitus (T1D) is the most common form of carbohydrate metabolism disorder in childhood. There are also monogenic forms of diabetes.

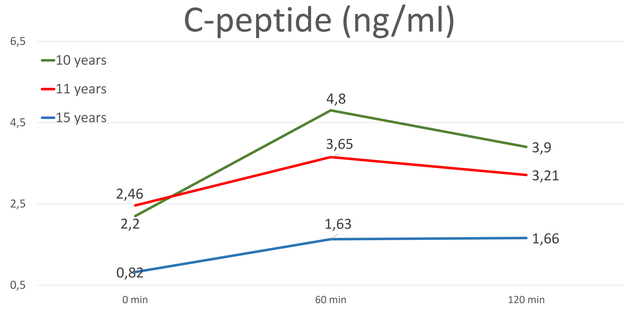
In the world literature, there are isolated mentions of the combination of monogenic and autoimmune forms of diabetes mellitus.

CLINICAL CASE

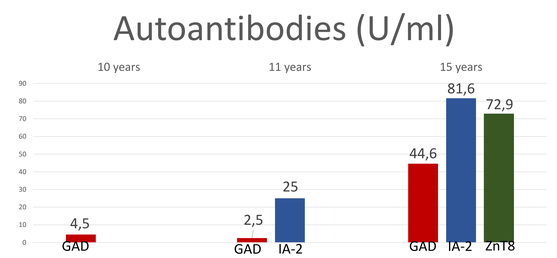
✓**A 10-year-old** obese patient presented with hyperglycemia. At hospitalization: height 168 cm, weight 87 kg, SDS BMI +3.0. Glycemia 14.4 mmol/l, HbA1c - 8.9%.
MMTT: fasting glucose 12.8 mmol/l, C-peptide 2.46 ng/ml; after 2 hours - glucose 17.2 mmol/l, C-peptide 3.21 ng/ml.
Autoantibodies - GAD 4.5 U/ml (N 0-1), IA-2 - 25 U/ml (N 0-1).
HLA typing: DRB1*01:07-DQA1*0101,0201-DQB1*0501:0201 (haplotypes: predisposing and neutral).
The patient was diagnosed with T1D and started on insulin therapy at a daily dose of 0.2 U/kg.



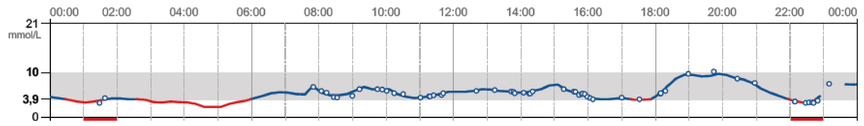
✓On examination **at the age of 11**: height 174 cm, weight 108 kg, SDS BMI +3.35, HbA1c 10.6%.
The daily dose of insulin was 0.95 U/kg/day. Due to the presence of obesity and intact insulin secretion, metformin was started at a dose of 1000-1350 mg/day.
The daily dose of insulin was reduced to 0.60-0.65 U/kg.



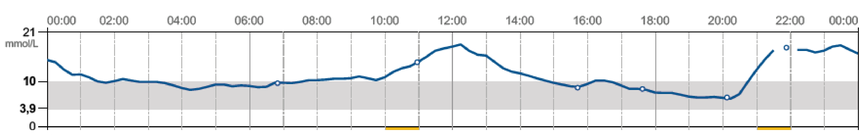
✓**At the age of 15**: height 168.5 cm, weight 90 kg, SDS BMI +3.4; HbA1c 10.7%;
MMTT: fasting glucose 9.5 mmol/l, C-peptide 0.83 ng/ml; after 2 hours - glucose 17.6 mmol/l, C-peptide 1.66 ng/ml.
Autoantibodies: ZnT8 72.9 U/ml, IA-2 81.6 U/ml, GAD 44.6 U/ml.



✓We believe the patient has high HbA1c levels due to low compliance, as glycemia were always stable (4-10 mmol/l) in the hospital when the patient maintained the diet and insulin therapy.

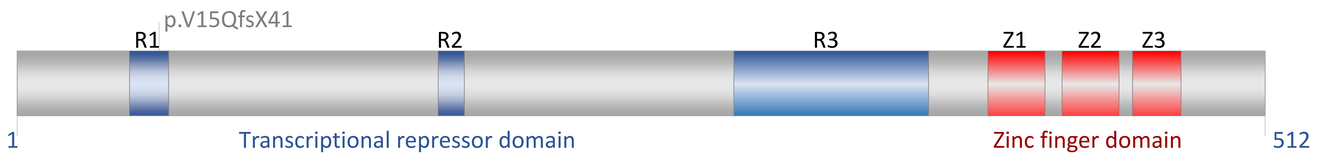


✓Glycemic variability in hospital



✓Glycemic variability in home

✓Whole exome sequencing revealed a likely pathogenic heterozygous variant c.40-41dupGC (p.Val15GlnfsTer41) in the KLF11 gene.



✓Mutations in the KLF11 gene may lead to MODY7. To date, fewer than 10 mutations in the KLF11 gene have been described, and therefore, data on the clinical picture are rather scarce. Basically, the disease proceeds similarly to type 2 diabetes. Although, there are references to insulin requirements.

CONCLUSIONS

This clinical case is a combination of monogenic and autoimmune diabetes mellitus. We suppose the patient presented with MODY7 at first with later onset of autoimmune diabetes mellitus due to mild disease course, relatively low titer of pancreatic autoantibodies, average T1D risk based on HLA typing, and presence of insulin secretion at the beginning.

CONTACT INFORMATION

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