Heterozygous mutation in G6PC2 (glucose 6 phosphatase catalytic subunit 2) gene in a female patient with atypical diabetes characteristics; does this mutation explain clinic features or is just a polymorphism

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Introduction

Some of the mutations that cause monogenic diabetes increase the susceptibility to Type 2 DM. Glycokinase is one of these. The activity of the GCK gene is regulated by G6PC2 (glucose 6 phosphatase catalytic subunit 2) in beta cells and by the GCK regulator protein in the liver. Variations in the GCK and G6PC2 genes may cause type 2 DM susceptibility by disrupting the relationship between them. A 10-year-old girl with a diabetes mellitus was presented for showing a rare mutation / variation in G6PC2 by genetic analysis.

Case

- She was first diagnosed with obesity at 9 years and 7 months in our clinic. Blood glucose was normal at that time. Her medical history was not remarkable. But there was a strong family history about diabetes.
- At the age of 10 years and 6 months, blood glucose was found 570 mg / dl coincidentaly.
- **In her physical examination** weight was 54 kg (99%p) height was 146,7 cm (99% p) body mass ındex was 25 (98% p) and pubertal stage was 3.
- **In her laboratuar examination**; fasting blood glucose was 363 mg/dl , no acidosis and ketone was detected and insuline, c peptide and HbA1C level were 3,76 mıu/ml, 1 ng/ml, 10% respectively. Fasting lipid, thyroid hormon and tyhroid autoantybody levels were in normal range.
- Anti GAD was found 3 ıu/ml(0-1)
- Patient were considered Type 1 diabetes, was began subcutaneous insulin therapy and on follow metformin was added because the patient had insulin resistance findings.
- Our patient had good blood glisemic control despite low dose of insulin requirement.
- At the end of 3 years because of insuline reserve, monogenic diabetes genes were studied for 23.
- A heterozygous potential pathological mutation in G6PC2 was detected.
- Although this mutation has been reported to be pathological, there is not enough data to explain clinical findings in ourpatient.

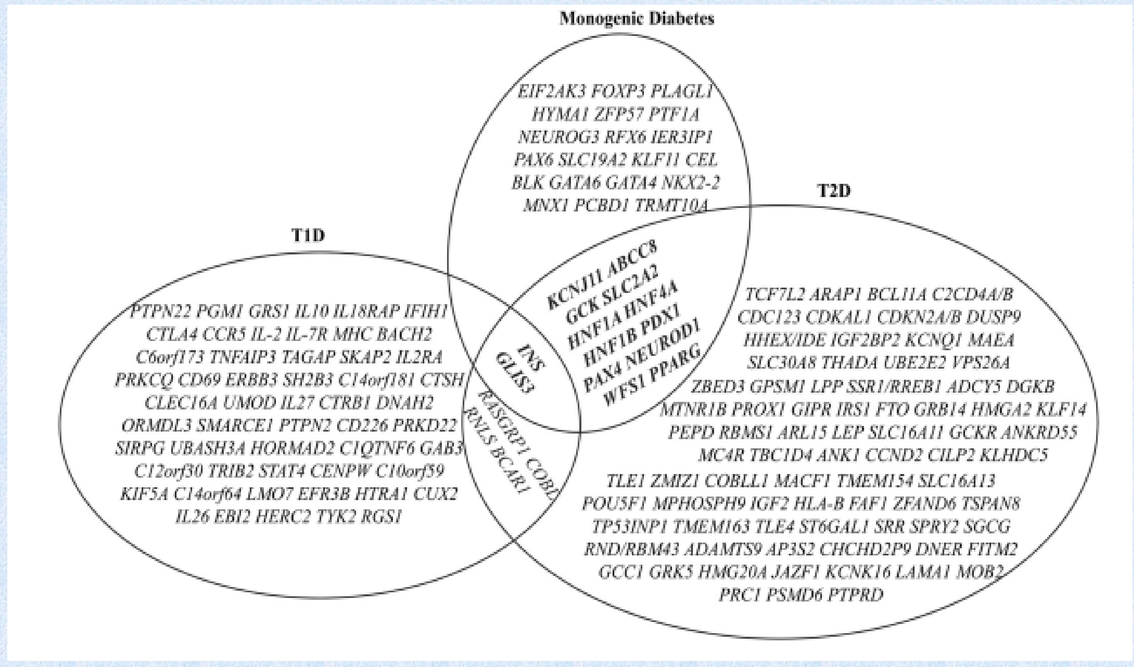

Figure 1: Genes to cause Diabetes

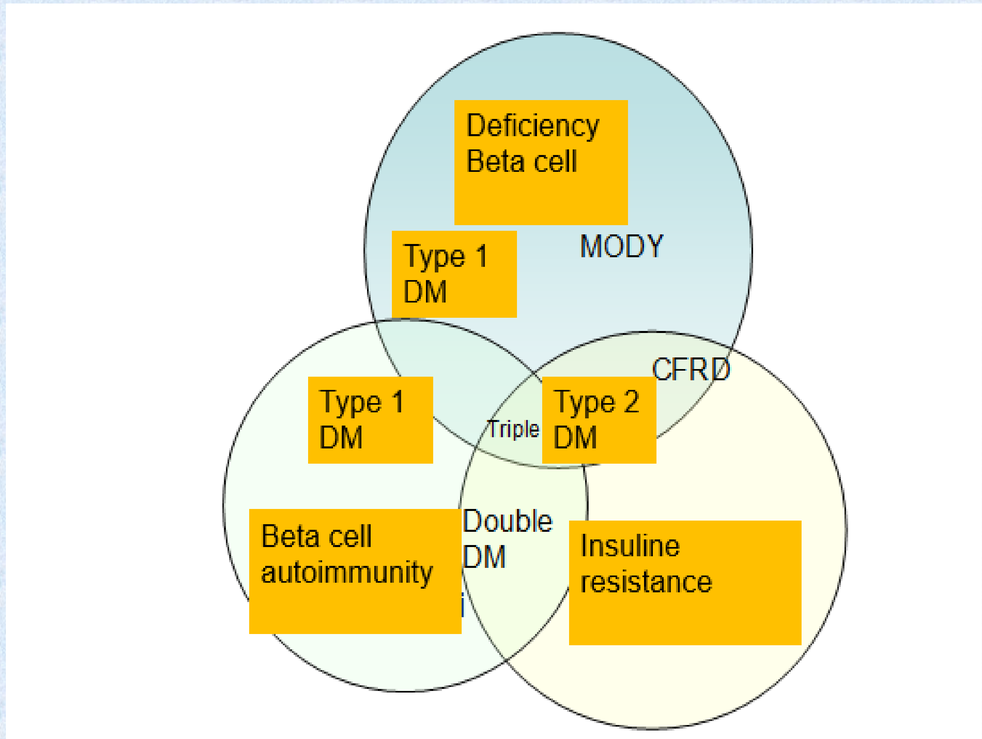

Figure 2 : Beta cell insufficiency and spectrum of Insulin resistance syndrome (Clasification of Diabetes Mellitus, Pediatric Clinics of North America volum 52 number -6)

Discussion

G6PC2 gene is known as regulating fasting blood glucose and its mutation is prone to type 2 diabetes. Although our patient was treated as Type 1 diabetes, some patients can show the characteristics of all types of diabetes. So diabetes classification can be made more clearly with advanced genetic tests.