A NOVEL GLUCOKINASE GEN MUTATION: MODY TYPE-2 CASE

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BACKGROUND

Maturity-Onset Diabetes of the Young (MODY) is a rare monogenic form of diabetes that result in β-cell dysfunction. MODY accounts for 2%–5% of all diabetes cases. MODY2 patients are characterized by glucose sensing defects, leading to have mild fasting hyperglycemia throughout life, and rarely require medication or develop microvascular complications. We presented here a family with MODY2 caused by a novel heterozygous p.L164I (c.490 C>A) mutation of the GCK gene.

CASE

- 15,5 years –old, girl
- Fasting hyperglycemia detected in routine control.
- No diabetes symptoms .
- Parents had no consanguinity.
- Her mother was 26 years old with a diagnosis of gestational diabetes in her second pregnancy, used metformin for eight years after having been diagnosed as diabetes.
- It was learned that her aunt and grandmother had diabetes and her cousin had gestational diabetes.

Table I- Laboratuary findings

Glucose (Fasting): 114 mg/dL	**Fasting İnsulin : 5,08 µIU/ml**
Hemoglobin A1c: %5,56	**C-Peptid: 1,39 ng/ml (0.9 - 7.1)**
AntiGAD: Negative	**ICA: Negative IAA: Negative**

Table II- Glucose and insulin concentrations during a standard oral glucose tolerance test with 75 g glucose equivalent.

Time	Glucose (mg/dl)	Insulin (µIU/ml)
0'	103	4,23
120'	153	20,88

Figure I-Pedigree of family

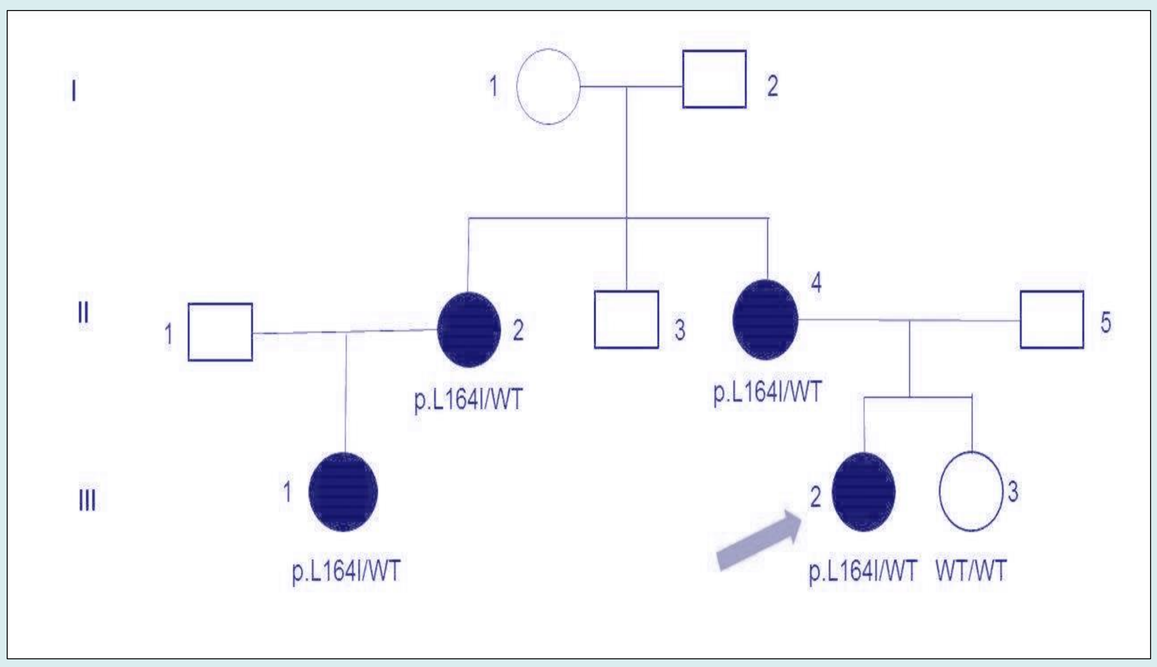
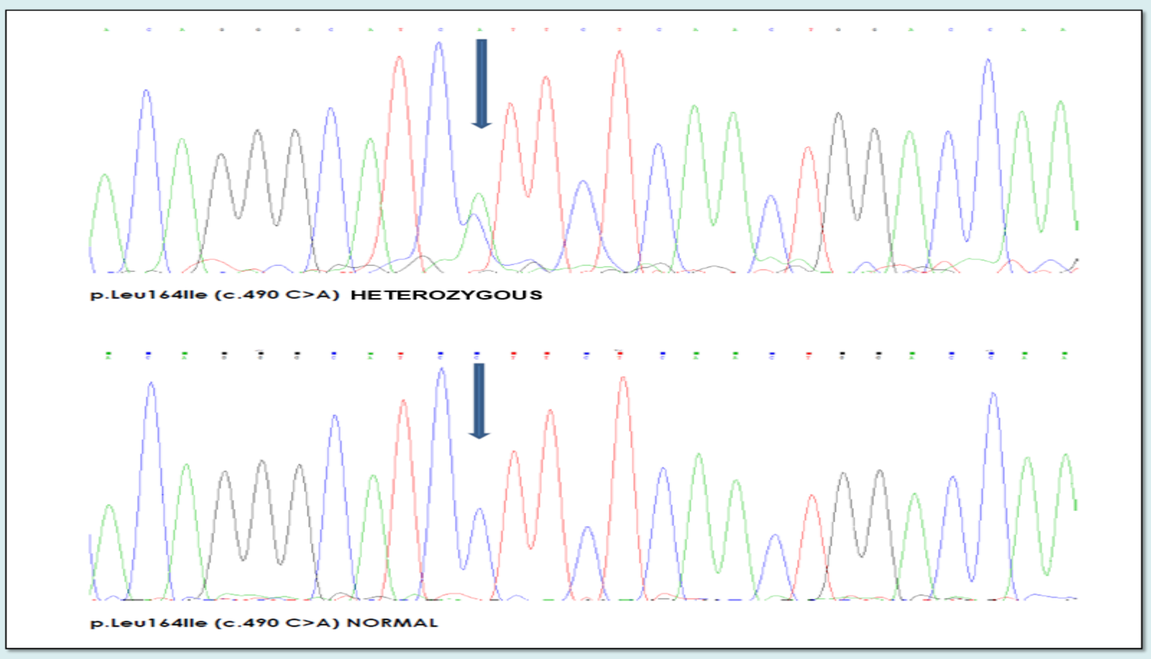


Figure II- GCK gene mutation analysis



CONCLUSION

MODY should be suspected in children who is found to have a random rise of blood sugar and has a family history of diabetes. Cases and individuals who have a family history of diabetes should be screened respectively for mutation. A precise molecular diagnosis is crucially essential because it leads to optimal treatment of the patients and allows early diagnosis for their asymptomatic family members.

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