A NOVEL GLUCOKINASE GEN MUTATION: MODY TYPE-2 CASE
Aslıhan Araslı Yılmaz¹, Selin Elmaoğulları¹, Fatma Demirel¹,² Meltem Tayfun¹, Seyit Ahmet Uçaktürk¹, Fatih Gürbüz¹, Ali Kemal Topaloğlu³

1 Ankara Children’s Hematology-Oncology Training Hospital, Department of Pediatric Endocrinology, Ankara, Turkey
2 Yıldırım Beyazıt University Faculty of Medicine, Ankara, Turkey
3 Çukurova University Faculty of Medicine, Department of Pediatric Endocrinology, Adana, Turkey

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.

BACKGROUND

- 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.

CASE

Table I - Laboratuary findings

| Glucose (Fasting): 114 mg/dL | Fasting Insulin: 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.

<table>
<thead>
<tr>
<th>Time</th>
<th>Glucose (mg/dl)</th>
<th>Insulin (µIU/ml)</th>
</tr>
</thead>
<tbody>
<tr>
<td>0’</td>
<td>103</td>
<td>4,23</td>
</tr>
<tr>
<td>120’</td>
<td>153</td>
<td>20,88</td>
</tr>
</tbody>
</table>

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.