**Hyperglycaemia in a boy of 13 years old: Not always Type 1 Diabetes Mellitus. A case report**

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**Topic:** Diabetes

**Disclosure:** No conflict of interest

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**Introduction**

Type 1 Diabetes (T1D), the most frequent type of diabetes in Paediatrics, can be easily misdiagnosed.

**Objectives**

We report a 13 year old boy with monogenic diabetes, initially diagnosed and treated as T1D.

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**Methods**

The patient presented at 7.5 years of age with a febrile illness and mild hyperglycaemia. An Oral Glucose Tolerance Test (OGTT) then was normal, HbA1c 6.3% (45 mmol/mol), (table 1). Slowly progressing T1D was diagnosed; he stayed under follow-up with routine BMIsix measuring at home (max blood glucose (BG) 153 mg/dl (8.5mmol/l)). A repeat OGTT at the age of 9y showed BG 127 mg/dl (7.1mmol/l) at 0’ and 258 mg/dl (14.3mmol/l) at 120’, HbA1c 6.7% (50mmol/mol) (table 2). He started on small doses of insulin. His glycaemic control was excellent; he remained on small doses of insulin (0.1U/Kg/d) for four years. The patient discontinued insulin without medical advice. Six months later, he had mild fasting hyperglycaemia, (BG 107-148mg/dl (6-8mmol/l)), HbA1c 6.2% (44 mmol/mol); Anti-GAD, ICA and IAA were negative (table 3). OGTTs were normal for father and younger sister aged 2 years. His mother, 37 year old, had gestational diabetes, her OGTT showed BG 147mg/dl (8.2 mmol/l) at 0’ and 121mg/dl (6.7mmol/l) at 120’, HbA1c 6.4% (46 mmol/l); negative anti-IAA antibodies. DNA analysis was carried out for the presence of mutations in HNF1A and GCK genes employing bidirectional sequencing of the coding regions of the two genes. MLPA was employed to search for deleterions in the genes GCK, HNF1A, HNF4A, HNF1B.

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**Results**

Point mutations were not detected in the genes GCK and HNF1A. The MLPA revealed that the patient and his mother harbor a heterozygote GCK gene deletion (exons1-10), confirming the diagnosis of maturity onset diabetes of the young type 2 (MODY 2).

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**Conclusions**

MODY is a heterogeneous group of monogenic diabetes that result in β-cell dysfunction (table 4, 5 &6). Diagnosis in paediatric patients may be challenging. It has an estimated prevalence of just 1%-2% of all diabetes in industrialized countries, however this prevalence is probably underestimated since large population screening studies have not been performed. MODY2 is characterized by mildly elevated fasting blood sugars and HbA1c ranging from 5.6–7.6% (38-60 mmol/mol). It is frequently unrecognized or misdiagnosed as T1D or T2D, resulting in unnecessary insulin treatment. The suggested treatment for MODY 2 is normally a lifestyle modification with regular physical activity and a well balanced diet. Molecular diagnosis is, therefore, very important for recognising the type of MODY, deciding the appropriate treatment for the patient and providing a reliable long term prognosis for individual patients and their relatives.

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**References:**


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**Table 1. Initial OGTT, age 7.5 y old**

<table>
<thead>
<tr>
<th>Glu (mg/dl)</th>
<th>0'</th>
<th>120'</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hba1c (6.3%)</td>
<td>115</td>
<td>91</td>
</tr>
</tbody>
</table>

**Table 2. OGTT, age 9 y old**

<table>
<thead>
<tr>
<th>Glu (mg/dl)</th>
<th>0'</th>
<th>120'</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hba1c (4.3-5.7%)</td>
<td>127</td>
<td>258</td>
</tr>
</tbody>
</table>

**Table 3. OGTT, 13y old**

<table>
<thead>
<tr>
<th>Glu (mg/dl)</th>
<th>0'</th>
<th>120'</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hba1c (4.3-5.7%)</td>
<td>6.7%</td>
<td>6.7%</td>
</tr>
</tbody>
</table>

**Table 4. MODY types**

<table>
<thead>
<tr>
<th>MODY</th>
<th>Frequency</th>
<th>Characteristics</th>
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<tbody>
<tr>
<td>MODY 2</td>
<td>5-10%</td>
<td>Mild but can be severe</td>
</tr>
</tbody>
</table>

**Table 5. MODY2 pathophysiology**

**Table 6. MODY2 inheritance**

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**References:**


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