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# Not every obese child has type 2 Diabetes Mellitus


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## NOT EVERY OBESE CHILD HAS TYPE 2 DIABETES MELLITUS

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### CASE PRESENTATION

- 11.84 years female
- Gradually gaining weight after 8 years
- Presented at 11.03 years with obesity
- BMI: 31.5kg/m<sup>2</sup>; BMI z-score: 3.55
- No significant past health issues

**Clinical examination** Signs of metabolic syndrome

- Mild acanthosis nigricans
- Arterial pressure: 129(>99<sup>th</sup>le) /80 mmHg (>95<sup>th</sup>le)
- Arterial pressure: 111 (<90<sup>th</sup>le) /80 mmHg (>95<sup>th</sup>le)

**FOLLOW-UP**

- Following 3 months dietary/lifestyle intervention
- OGTT repeated
- 120' glucose: 225mg/dl
- T1DM antibodies negative, C-peptide normal
- Started on Metformin (850 mg BD)

OGTT 29/7/2020	0'	30'	60'	120'
Glucose (mg/dl)	98	229	225	225
Insulin (μU/ml)	31.2			
HbA1c (%)	5.5			
Glucose in urine	Negative			

>At 11.84 years BMI progressively increases (BMI z-score: 3.98 from )

>On metformin HbA1c unchanged but normal (5.5%)

>A new OGTT was performed to better assess response on metformin

OGTT 19/9/2021	0'	30'	60'	90'	120'	150'	180'
Glucose (mg/dl)	123	125	170	227	234	130	216
Insulin (mg/dl)	32.87	29.91	63.48	106.0	102.1	70.14	98.63

Unchanged → Question of adherence  
Increased metformin to 1000mg BD

WAS IT TYPE 2 DIABETES OR SOMETHING ELSE?

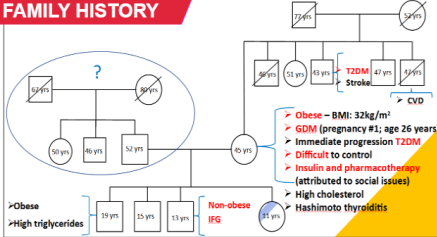
**Young people with T2DM**

- Present around puberty
- Majority are obese

**Patients with monogenic diabetes**

- May also be obese
- Can be difficult to distinguish from T2DM

**FAMILY HISTORY**



**REVIEW OF MANAGEMENT**

Features that suggest T2DM:

- Signs of metabolic syndrome
- Severe obesity
- Acanthosis nigricans (mild)

NGS panel for MODY

Mutation p.R200Q/N of HNF1A mother and patient

**HNF1A-MODY (MIM# 600496) or MODY3**

Features that suggest monogenic diabetes:

- Family history
- Early onset diabetes in the mother
- Obese, GDM and T2D at 26 years
- Patient's brother non-obese - IFG
- Mother's brother T2D and stroke
- Strong family history of CVD

**Molecular diagnosis**

- Choice of most appropriate treatment
- Optimize blood glucose control
- Reduce long-term complications
- Proper genetic counseling

**SUMMARY**

- Cases of MODY may be difficult to distinguish from T2DM
- Importance of Family History – Low threshold perform genetic testing
- Importance of Genetic testing
- Can drive the choice of treatment
- Assess the risk for complications
- Provide genetic counseling
- Treatment adherence requires regular follow-up – Clinical interview skills

**CONCLUSIONS**

- Cases of MODY may be difficult to distinguish from T2DM.
- Studies suggest that 5% of subjects diagnosed with diabetes before the age of 45 years have MODY, with 80% of them having been incorrectly diagnosed as having T1DM or T2DM.
- Importance of Family History – Low threshold perform genetic testing.
- Genetic testing allows confirmation of the correct diagnosis and leads to optimal treatment.

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**Background:** Maturity onset Diabetes of the young (MODY) is a rare form of diabetes with specific features that distinguish it from diabetes mellitus type 1 (DM1) or type 2 (Dm<sup>2</sup>). Research studies suggest that 5% of subjects diagnosed with diabetes before the age of 45 years have MODY, with 80% of them having been incorrectly diagnosed as having DM1 or Dm<sup>2</sup>. Genetic testing may enable correct diagnosis and treatment, optimize glycemic control, reduce the risk of hypoglycemic events and long-term complications, and enable proper genetic counseling.

**Case description:** We present the case of a 11.84 year old female, who presented with obesity and a history of gradual weight gain since the age of 8 years. Her mother had obesity (BMI: 32 kg/m<sup>2</sup>) and a history of gestational diabetes mellitus (GDM) that progressed to Dm<sup>2</sup>, which was difficult to control despite treatment with insulin and metformin. Her elder 13 year old brother had impaired fasting glucose despite normal BMI, while her uncle had also been diagnosed with Dm<sup>2</sup>. On clinical examination she had obesity (BMI: 31.5kg/m<sup>2</sup>, BMI z-score: 3.55), mild acanthosis nigricans and hypertension. Endocrinologic evaluation revealed elevated fasting plasma glucose (FG: 122 mg/dl), and impaired glucose tolerance on OGTT (glucose @ 120': 182mg/dl, insulin 0' = 43.38µUI/ml). Despite the implementation of a personalized intervention program of diet and exercise, her glycemic control deteriorated. At 11.14 years, her FG (118 mg/dl) and insulin concentrations (31.5 µUI/ml) remained elevated. A repeat OGTT confirmed the diagnosis of diabetes (glucose @ 120': 225mg/dl) and the patient was commenced on metformin. Her HbA1C (5.5%) and C-peptide (3.8 ng/ml) were normal, and the antibody screen for DM1 was negative. The diagnosis of MODY was suggested by the strong family history, and genetic testing was undertaken. A heterozygous mutation in HNF1A gene was detected, thereby confirming the diagnosis of MODY3. The management plan now includes institution of sulfonylurea treatment.

**Discussion:** Patients with MODY may present with obesity and may be difficult to distinguish from Dm<sup>2</sup>. Genetic testing allows confirmation of the correct diagnosis and leads to optimal treatment.

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