Case Report: 11 year old overweight boy with hyperglycemia



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Past Medical History

- Three fractures of lower limbs since the age of 10 years
- After second fracture of the femur at the age of 11 years the healing was not appropriate.
 He had surgery, metal clips inserted. The surgical site got infected with pus formation
- Blood glucose was measured and FPG 135 mg/dl + obesity
- He was diagnosed as having DM type 2 and offered treatment with Metformin.

Past Medical History



- After starting treatment with Metformin, in few days he developed severe diarrhea
- Parents discontinued the use of medicine
- Diet + swimming
- FPG 128-115 mg/dl
- Bone health markers were not studied

Family History

- Mothers side Grandmother and aunt had DM
- Fathers side:

Father 48 years old – Overweight

Uncle –DM since the age of 40 years

Above mentioned uncles son – DM since the age of 17 years, now 22 (was not overweight when diagnosed)

Grandfather – DM

Giorgi, 13 years old

- Limp during walking, uses cane
- Weight 62 kg -90th percentile
- Height 152 cm- 25 th percentile
- $BMI 27 -> 90^{th}$ percentile
- No acanthosis or rush
- Mother 159 cm, Father 164 cm, MPH -168 cm,
 10-th percentile for population

Labs

- FBG 115 mg/dl
- C-peptide 1.27 ng/ml (N 0.81-3.85)
- HDL 39 (N 40-60)
- HbA1c 6.2 % (N 4.0-6.0)
- GAD-65 Ab < 1.0
- IA-2 Ab < 0.8
- Urinalysis

Specific gravity -1.015

Trace amount of ketones and epithelial cells

Patient has twin sister. She has normal weight, Tanner stage 4. We measured her FBG -106 mg/dl



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- What do you think is a cause of hyperglycemia in this patient?
- Metabolic Syndrome? DM type 2?
- Any other options?
- What other do you recommend?
- What management strategy would you recommend?

When to suspect a diagnosis of type 1 diabetes in children may not be correct?

- Diabetes presenting before 6 months of age as type 1 diabetes is extremely rare in this age group
- Family history of diabetes in one parent and other first degree relatives of that affected parent.
- Absence of islet auto antibodies, especially if measured at diagnosis.
- Preserved β-cell function, with low insulin requirements and detectable C-peptide (either in blood or urine) over an extended partial remission phase (5 yr after diagnosis)

When to suspect a diagnosis of type 2 diabetes in children may not be correct?

- Absence of severe obesity
- Lack of acanthosis nigricans and/or other markers of metabolic syndrome
- Ethnic background with a low prevalence of type 2 diabetes, e.g., European Caucasian
- Strong family history of diabetes without obesity

- Monogenic diabetes is uncommon, it accounts for 1–4% of pediatric diabetes cases
- In familial autosomal dominant symptomatic diabetes, mutations in the hepatocyte nuclear factor 1α (HNF1A) gene (HNF1A-MODY) should be considered as the first diagnostic possibility, while mutations in the GCK gene are the most common cause in the absence of symptoms or marked hyperglycemia
- Results of genetic testing should be reported and presented to families in a clear and unambiguous manner, as results may have a major effect on clinical management
- Some forms of MODY diabetes are sensitive to sulfonylureas
- Mild fasting hyperglycemia due to GCK-MODY is not progressive during childhood; patients do not develop complications and do not respond to low dose insulin or oral agents, so should not receive treatment

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