MAURIAC SYNDROME: A COMPLICATION THAT STILL EXISTS IN CHILDREN WITH TYPE 1 DIABETES. REPORT OF A CASE

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INTRODUCTION

Mauriac Syndrome (MS) is currently an extremely rare complication in type 1 diabetes mellitus (DM1). It is characterized by the triad: poor metabolic control, dwarfism and hepatomegaly. Other findings are elevated transaminases, dyslipidemia, cushingoid features and delayed puberty

CASE

Male patient with DM1 since 5 years age. Coinciding with a family breakdown, from the age of 7 his metabolic control deteriorated significantly (HBA1c ~12% and recurrent ketoacidosis).

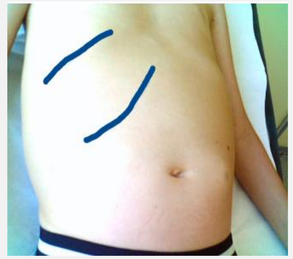
At 12 years of age he persisted with poor metabolic leading frequent hospitalizations for ketoacidosis and a progressive increase in insulin doses up to 2.7 U/kg/day with MDI therapy. Despite this, poor metabolic control and multiple hospitalizations persist. From the age of 8 years there is a lack of growth and weight together with hepatomegaly with increased transaminases.

At 15 years 2 months, his height was 130.3 cm (- 4.9 SD), BMI 16.2 kg/m2 (-1.8 SD); he had protruding abdomen with a smooth, firm consistency and not painful hepatomegaly of 5-6 cm under the costal margin, testes 2 cc bilateral, no pubic hair, thin limbs and hypotrophy of muscle masses

Laboratory: total cholesterol: 253 mg/dl, HDL: 92mg/dl, LDL: 134 mg/dl, triglycerides: 134mg/dl, AST: 369U/L, ALT :369 U/L with normal liver function. Normal blood count, celiac study, thyroid function and creatinine. Hepatitis A, B and C virus infection was ruled out. Basal GH: 2.89 ng/ml (reference value (VR) <1), and GH post cloninide test was 10,7 ng/ml, IGF-1: 346 ng/ml (VR 358 – 870), IGFBP-3: 5246 ng/ml (VR 2590 – 7280). Bone age with 2-year delay. Treatment with an insulin pump is started. Improved metabolic control and normalization of liver tests were obtained. Unfortunately, patient abandons controls. A follow-up carried out at 21 years of age he remains using an insulin pump with poor adherence, HBA1c ~11% and new hospitalizations for ketoacidosis.

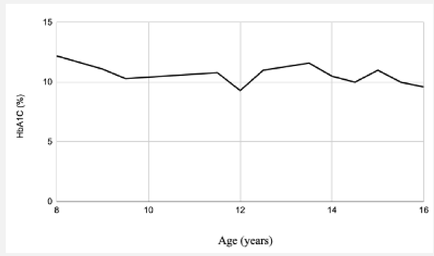
He reached a final height of 156.9 cm (-2.9 SD; Target height -0.9 SD)

Physical exam

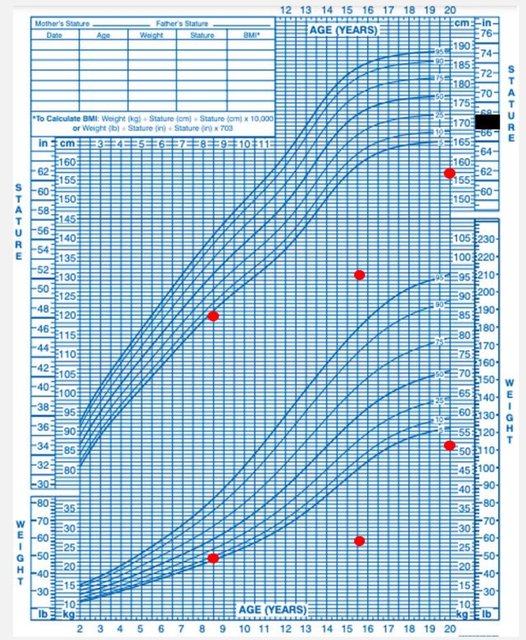


Hepatomegaly of 5-6 cm under the costal margin

HbA1c curve



Growth chart



CONCLUSIONS

MS is the consequence of serious metabolic compromise in very poorly controlled DM1 patients. Despite the advances in DM1 therapies, MS can still appear. SM can be presented in its full spectrum or just some of its characteristics. Differential diagnosis with viral hepatitis or glycogenosis should be considered. Growth impairment due to resistance to the action of GH, and a global and hepatic pro-inflammatory environment could be reversed with appropriate insulin therapy, but it can be definitive without it. Despite the large percentage of adolescents with poor metabolic control MS is rare. This leads to suppose the existence of certain genes that predispose to this pathology.

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