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Case Report

The Return of an Old Syndrome: Mauriac Syndrome

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Abstract

Introduction: Mauriac syndrome or hepatic glycogenosis was first described in 1930. The cardinal features are hepatomegaly, growth impairment, and cushingoid features in poorly controlled type 1 diabetes. Mauriac syndrome is now uncommon, especially with the advent of new insulin analogues and intensive insulin regimens. We report a case of Mauriac syndrome hospitalized at the pediatrics department of the CHU Mohammed VI Marrakesh.

Objective: To discuss the etiologies of hepatomegaly and the management of this pathology.

Clinical Case: 10-year-old child known as type 1 diabetic for 5 years. Who presented with unbalanced diabetes, staturo-ponderal delay and recently onset abdominal distension. The clinical examination had objectified a staturo-weight delay (weight: -2 SD and height -4 SD), a soft hepatomegaly at 17 cm without splenomegaly nor collateral venous circulation with Hb1Ac at 12%. Mauriac syndrome was retained after elimination of other etiologies, notably autoimmune hepatitis and celiac disease, and confirmed by biopsy liver puncture, which confirmed glycogen overload of hepatocytes. Optimization of the treatment of diabetes by insulin therapy has been instituted. The evolution is favorable.

Conclusion: The discovery of hepatomegaly with staturo-weight retardation in an unbalanced type 1 diabetic should evoke glycogenosis.

Keywords: *Mauriac Syndrome (MS); Diabetes Mellitus Type 1 (DM1); Hepatomegaly; Glycogenosis*

Introduction

Mauriac syndrome (MS) is a rare complication of poorly controlled diabetes mellitus type 1 (DM1) characterized by hepatomegaly, growth failure, delayed puberty, cushingoid features dyslipidemia and transaminase elevation [1,2]. MS has been described in children and adolescents with poor insulin compliance and brittle glycaemic control along with inadequate diet [3,4]. And can be present without the full spectrum of features described for MS [4]. High daily insulin dosage and recurrent ketoacidosis episodes significantly increase the risk for HG [4,5]. There may be different forms and etiologies involved in Mauriac syndrome. However, there are common features noted in these patients with adequate insulin treatment there is reversal of growth failure and hepatomegaly if present [2,6].

Case Presentation

We present the clinical case of an ten-year old boy, born from non-consanguineous parents who belonged to low socioeconomic strata, affected by T1D since he was 5 years old. Poorly controlled due to non-compliance with insulin-therapy and he had several hospitalizations for diabetic ketoacidosis. he was admitted in our department for unbalanced diabetes and abdominal distension. Physical examination revealed a staturo-weight delay (weight: -2 SD and height -4 SD), a soft hepatomegaly at 17 cm was noted at the abdominal palpation without others physical abnormalities. Laboratory analysis showed a marked aspartate aminotransferase (ASAT) 20 UI/L, aminotransferase (ALAT) 22 UI/L with normal gamma-glutamyltranspeptidase (GGT), bilirubin, alkaline phosphatase and coagulation tests. Patient's serum glucose was 17 mmol/l and haemoglobin A1 C was 12% with no ketonuria or acidosis.

Abdominal ultrasound confirmed hepatomegaly 17 cm showing an enlarged liver with normal echogenicity and vasculature. Our patient, there was no history of hepatotoxic agent use. Serologic tests for viral infections (Anti VHA Ig M, Ag HBs, Anti VHC, Epstein–Barr virus Ig M and Anti Cytomegalovirus Ig M) and auto-immune hepatitis were negative. An absence of specific anti-bodies and of duodenal mucosal atrophy had eliminated coeliac disease. Thyroid assessment was likewise normal. Given the remain uncertain diagnosis, a percutaneous liver biopsy was performed. Histology showed pale and swollen hepatocytes with prominent plasma membranes and numerous glycogenated nuclei. Cytoplasmic glycogen deposit was confirmed on a periodic acid Schiff stain (PAS). All the clinical and histological facts described above were consistent with the diagnosis of MS. An intensive insulin regime of 1 IU/kg/day divided into 4 doses per day (basal bolus) was implemented to control hyperglycemia and avoiding large variations of the glycemia. A better control was achieved on 7 months, with a reduction of HbA1c to 7,5%, a disappearance of hepatomegaly and improvement regarding growth.

Discussion

Mauriac syndrome became a rare disorder after the introduction of long-acting insulins for the treatment of DM1 and after the initiation of HbA1c as a marker of long-term glycemic control. Isolated patients with this syndrome are still reported [1,3,6]. The pathophysiology of MS has not been fully understood, but the main mechanism identified is the large fluctuation in glucose and insulin levels [6,8,9]. High glucose levels cause an influx of glucose into hepatocytes by passive diffusion. Glucose is then irreversibly converted to glucose-6-phosphate. Subsequent treatment of hyperglycemia with a high dose of insulin causes further polymerization of the glucose trapped in glycogen. In fact, the vicious cycle of high doses of short-acting insulin and administration of glucose to counteract the resulting hypoglycemia is described by several authors as the main mechanism of excessive glycogen accumulation in hepatocytes [6,9]. The pathogenesis of growth retardation and puberty is not clear but rather seems to be multifactorial: insufficient tissue glucose, lack of insulin as a growth factor and hypercorticism may contribute to it. The cushingoid signs present during glycogenosis are classically described in children [10,11]. Indeed, during the developmental period, the large-scale documented fluctuations between hyperglycemia and hypoglycemia (suggesting a pattern of over- and under-insulinization) are accompanied by activation of counter-regulatory hormones with hypercorticism reactive to excess insulin [10,12]. Hypercorticism seems to be responsible for the development of the secondary of Cushingoid obesity [11]. The clinical presentation of MS may range from an asymptomatic form with elevated liver enzymes and hepatomegaly to a symptomatic form with clinical signs of diabetic ketoacidosis such as abdominal pain and vomiting. In a few reports, patients had signs of acute hepatitis, jaundice, pruritus and elevated plasma lactate levels with or without diabetic ketoacidosis [7-9]. Our patient had hepatomegaly with growth failure which is a common clinical presentation in paediatric patients [7,8]. In Mauriac syndrome, hepatomegaly may [3,13] or may not [3,14] be associated with liver enzyme derangement. Transaminases are more frequently deranged than are alkaline phosphatase and bilirubin. Synthetic function, which is assessed by albumin and prothrombin time, is usually maintained [3]. The laboratory tests of our patient were normal. Abdominal imaging confirm hepatomegaly, Uniform liver's echogenicity is often described, indicative of glycogen storage but also suggesting non-alcoholic steatosis of the liver (NASH) which is associated with diabetes. Some reports

suggest that gradient dual echomagnetic resonance imaging (MRI) may be helpful to distinguish fat deposition in NASH from MS [7,8]. Our patient's abdominal ultrasound showed enlarged liver with no change in echogenicity.

The diagnosis of hepatic glycogenosis is a diagnosis of elimination. It is imperative to eliminate viral, metabolic, obstructive, autoimmune and non-alcoholic steatosis of the liver (NASH) causes before it is retained [11]. Several laboratory tests are usually perform to rule out there causes [7]. The only test that can formally confirm the diagnosis is liver biopsy, which reveals an aspect of glycogenic overload visible after staining with PAS (periodic-acid-Schiff) [9,11]. Nevertheless, some authors do not recommend biopsy if liver function tests are normalized with good glycemic control [5,11].

Glycemic control is usually achieved by increasing daily doses of insulin. A new therapeutic approach by using continuous insulin delivery was reported in few cases [1,7]. MS has a good prognosis with optimal glycemic control; resolution of hepatomegaly may take 2 weeks to 2 years (median, 5 to 6 months) [3]. Although hepatomegaly is reversible, close and continuous monitoring is necessary because this disorder may recur if glycemic control deteriorates [3].

Conclusion

The discovery of a hepatomegaly with staturoweight retardation in an unbalanced type 1 diabetic should evoke diabetic glycogenosis, in order to achieve an earlier diagnosis of mild forms and timely insulin therapy adjustment.

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