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Case Report

Isolated Hepatomegaly Revealing a Mauriac Syndrome (Case Report)

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Abstract

Background: Poor insulin compliance exposes type 1 diabetic patients to several complications. Hepatic glycogenosis (HG) or Mauriac syndrome is one of them. HG results from a significant accumulation of glycogen in the hepatocytes. It's manifested by short stature, growth maturation delay, dyslipidemia, protuberant abdomen with hepatomegaly. Its management is based on the appropriate control of diabetes.

Case Presentation: We report a case of 15-years-old female, with type 1 diabetes, who was admitted for marked hepatomegaly. Several causes of hepatopathy were excluded with blood investigations and liver tests. The diagnosis of hepatic glycogenosis was retained histologically after a liver biopsy.

Conclusion: Although hepatic glycogenosis is a rare entity, it should be considered in cases of hepatomegaly in a context of unbalanced type 1 diabetes.

Keywords: *Type 1 Diabetes Mellitus; Hepatic Glycogenosis; Mauriac Syndrome; Liver Biopsy*

Introduction

Hepatic glycogenosis (GH), also known as Mauriac syndrome, is a complication of unbalanced type 1 diabetes (T1DM). It results from an important accumulation of glycogen in the hepatocytes [1,2]. It was described in 1930, by Mauriac, in children and adolescents, with type 1 diabetes and poor insulin compliance [3]. It associates classically an hepatomegaly with abnormal liver enzymes, growth failure, delayed puberty, dyslipidaemia and cushingoid appearance [2,4]. Before retaining the diagnosis of HG, it's necessary to eliminate the other causes of hepatopathy, in particular viral, metabolic and auto immune diseases [1]. The incidence of HG is unknown and mostly published as case reports [5].

We report here the case of a patient with T1DM, since the age of 3 years old, admitted for exploration of hepatomegaly.

Case Report

We present the clinical case of a 15 years old girl, affected by T1DM since she was 3 years old. She had poor glycemic control, due to poor therapeutic compliance, with several hospitalizations for recurrent episodes of diabetic ketoacidosis.

She was admitted for exploration of a progressive abdominal distension associated to a right hypochondrium and epigastric pain.

At the admission, she presented a body mass index (BMI) of 17.35 kg/m^2, height = 146 cm and weight = 37 kg (-2 DS of expected height and weight for age and sex). Menarche and secondary sexual characters were absents. The clinical examination revealed a patient in a good general condition with an homogeneous hepatomegaly (19 cm). We didn't note jaundice, splenomegaly or ascites.

Laboratory tests showed a very unbalanced diabetes (Hba1c = 9.2%), hypertriglyceridemia (2,35 g/L) and hypercholesterolemia (2,36 g/l). Liver tests showed normal levels of total bilirubin, alkaline phosphatase, gamma-GT and transaminases. A normal levels of albumin and prothrombin time was also noted. To exclude viral origin, serologies of hepatitis B, C, CMV (cytomegalovirus), HSV (Herpes simplex virus) and EBV (Epstein Barr virus) were performed and proved negative. To rule out autoimmune hepatitis, liver-specific autoantibodies were analyzed: antinuclear antibodies, anti-smooth muscle, anti-LKM1, anti-SLA and antimitochondrial antibodies, which all resulted negative.

Ultrasound and abdominal scanner showed an important homogeneous hepatomegaly with a hepatic arrow of 210 mm, without other associated anomaly (Figure 1).

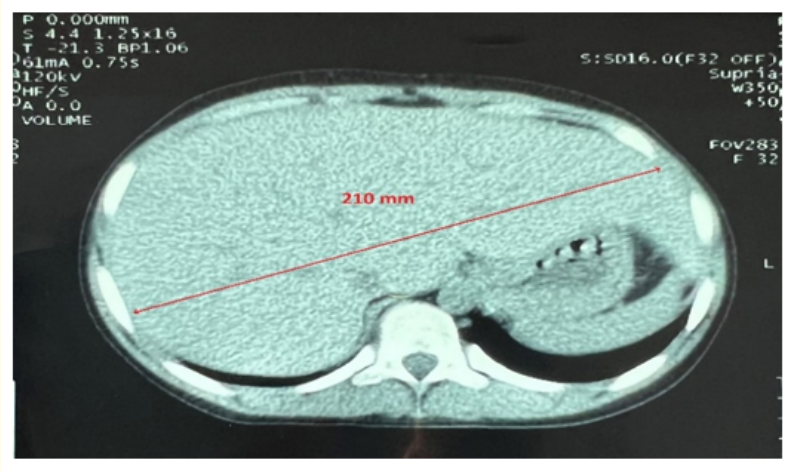

Figure 1: *Image of patient's abdominal CT showing a massive hepatomegaly (estimated at 210 mm), with regular contours.*

Liver biopsy showed clarified hepatocytes with a vegetative appearance and regular rounded nuclei. Periodic Schiff's acid (PAS), staining before and after diastasis, revealed the presence of intracytoplasmic glycogen, which was consistent with hepatic glycogenosis (Figure 2).

At this stage, the diagnosis of hepatic glycogenosis was established in our patient.

She was referred to an endocrinology unit to control her diabetes and for therapeutic education.

Discussion

The increasing prescription of intensive insulin therapy in patients with T1DM has led to a significant decrease in the incidence of hepatic glycogenosis (GH). Nevertheless, it still exists, and is probably under-diagnosed, as it poses problem of differential diagnosis with non-alcoholic fatty liver disease (NAFLD) [1].

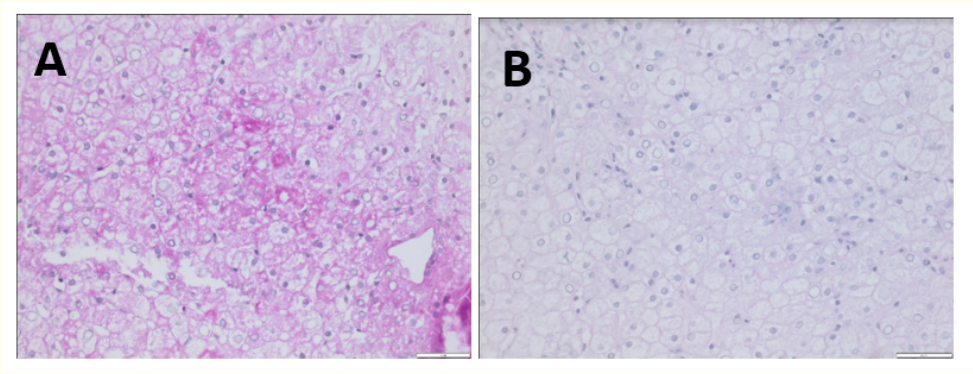

Figure 2: *Patient percutaneous liver biopsy: A-Positively stained hepatocyte with periodic acid-Schiff suggesting hepatic glycogenosis (×200). B-Staining glycogen disappeared after diastase treatment, confirming the present of glycogen (×200).*

The physiopathology of HG is probably related to the combination of hyperglycemic episodes and insulin excess. In fact, insulin excess induces activation of glucokinase and glycogen synthetase, and an inhibition of glucose-6-phosphatase, which leads to hyperstimulation of glycogenesis and inhibition of glycogenolysis, resulting in an excessive storage of circulating glucose in the form of intrahepatic glycogen [6].

Hepatomegaly results from the deposition of glycogen in the liver. In case of major hyperglycemia, glucose, via GLUT 2 (glucose transporter 2), enters the hepatocytes and is converted into 6P-glucose by glucokinase. High insulin administration stimulates glycogen synthetase which converts 6P-glucose into glycogen [7,8].

In case of repeated ketoacidosis, high doses of intravenous insulin are administered, and consequently this phenomenon is amplified [9], as in the case of our patient.

The lack of glucose in the organs, the absence of insulin as a growth factor and hypercorticism may explain the delayed growth and puberty encountered in this context [4,10].

Hepatic glycogenosis is a diagnosis of elimination. Before retaining it, it is imperative to rule out viral, metabolic, obstructive and autoimmune causes [9]. In the context of a metabolic syndrome (diabetes, obesity, dyslipidemia), the diagnosis of non-alcoholic fatty liver disease (NASH) should also be considered [11].

The only test that can formally confirm the diagnosis is a liver biopsy, which reveals a glycogen overload after staining with PAS (periodic-acid-Schiff) [12].

However, liver biopsy is not routinely recommended. An improvement of the symptomatology with a good glycemic control, for at least 4 weeks, allows to retain the diagnosis of HG and to avoid the recourse to an invasive examination. However, the liver biopsy must be performed, if glycemic control can't be achieved, as in our patient's case, or if there is a doubt about the diagnosis [1].

The evolution is usually favorable with good glycemic control, and liver damage usually disappears in two to four weeks [2].

Conclusion

Although hepatic glycogenosis or Mauriac syndrome is a rare entity, it should be considered in cases of hepatomegaly in T1DM with history of frequent diabetic ketoacidosis. The certainty diagnostic is histological, although rarely obtained, and treatment is based on the control of diabetes.

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