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Mauriac Syndrome: A Rare Cause of Hepatomegaly

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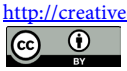

Abstract

Uncontrolled type 1 diabetes mellitus (T1DM) can lead to complications, including hepatic glycogenic overload, also known as Mauriac syndrome. This syndrome, initially described in children with T1DM, is characterized by excessive hepatic glycogen accumulation due to fluctuating hyperglycemia and high insulin doses. It is often underdiagnosed due to clinical overlap with metabolic dysfunction-associated steatotic liver disease (MASLD). We report the case of a 15-year-old boy with poorly controlled T1DM and recurrent diabetic ketoacidosis (DKA) who presented with abdominal distension, hepatomegaly, growth retardation, and delayed puberty. Laboratory tests showed elevated transaminases, hyperglycemia, and poor glycemic control, with a negative etiological workup for other liver diseases. Imaging confirmed hepatomegaly, and histological analysis via liver biopsy demonstrated glycogenic hepatopathy with intracytoplasmic glycogen deposits. Management was based on stringent glycemic control, which led to a marked improvement in liver tests and hepatomegaly. This case underscores the importance of considering Mauriac syndrome in T1DM patients presenting with hepatomegaly and abnormal liver enzymes.

Subject Areas

Gastroenterology & Hepatology

Keywords

Mauriac Syndrome, Glycogenic Hepatopathy, Diabetes, Hepatomegaly

1. Introduction

Mauriac syndrome, initially described by Pierre Mauriac in 1930, represents a rare

but significant complication of poorly controlled type 1 diabetes mellitus (T1DM) in pediatric and adolescent populations. It underscores the systemic impact of T1DM and is characterized by hepatomegaly, growth retardation, and delayed pubertal development. With the advent of modern insulin analogs and improved diabetes management protocols, the prevalence of this syndrome has significantly declined. However, reports of the syndrome persist, particularly in under-resourced settings or among patients with poor adherence to therapy. This case report elucidates the clinical manifestations, diagnostic challenges, and therapeutic approach in a patient diagnosed with Mauriac syndrome.

2. Case Presentation

We report the case of a 15-year-old male with a five-year history of T1DM, managed on a subcutaneous basal-bolus insulin regimen. The patient demonstrated poor adherence to treatment, resulting in recurrent episodes of diabetic ketoacidosis (DKA). His family history was unremarkable for hepatic disorders.

The patient was admitted with DKA, characterized by a five-day history of periumbilical abdominal pain, vomiting, polyuria, and polydipsia without associated fever. Initial management included intravenous fluids and insulin therapy with close metabolic monitoring. The patient exhibited no signs of jaundice or pruritus.

Upon physical examination, the patient appeared hemodynamically and respiratorily stable. Anthropometric measurements indicated a weight of 40 kg (below the 2nd percentile), height of 151 cm (below the 2nd percentile), and BMI of 17.5 kg/m^2. Abdominal examination revealed distension with hepatomegaly, as evidenced by a liver span of 19 cm and a smooth, palpable liver edge. There were no signs of splenomegaly, ascites, or stigmata of chronic liver disease. Genital examination demonstrated Tanner stage I pubertal development.

Laboratory tests revealed a complete blood count showing leukocyte levels of 5000/µL, hemoglobin level of 13 g/dL, and platelet count of 247,000/µL. Liver function tests indicated abnormalities with aspartate aminotransferase (AST) at 63 IU/L, alanine aminotransferase (ALT) at 115 IU/L, gamma-glutamyl transferase (GGT) at 180 IU/L, alkaline phosphatase (ALP) at 164 IU/L, and total bilirubin at 53 µmol/L, while albumin was within normal limits and prothrombin time was 97%. Renal function tests showed urea at 0.16 g/L and creatinine at 7.86 mg/L, both within acceptable ranges. Additional findings included triglycerides at 110 mg/dL, total cholesterol at 160 mg/dL, hyperglycemia at 1.89 g/L, and HbA1c at 12.2%, indicating poor glycemic control. Ferritin levels were 181 ng/mL, and bone age assessment corresponded to 10 years.

Hepatic etiological workup, including viral serologies, autoimmune diseases, and testing for Wilson's disease, yielded negative results. Abdominal ultrasonography identified hepatomegaly with homogeneous echotexture and normal hepatic vein dimensions, without evidence of cirrhosis or portal hypertension (**Figure 1**). Computed tomography (CT) confirmed an enlarged liver measuring 22 cm (**Figure 2**,

Figure 3). Histopathological evaluation of two liver biopsy specimens (1.5 cm each, encompassing approximately ten portal spaces) demonstrated glycogenic hepatopathy (Figure 4).

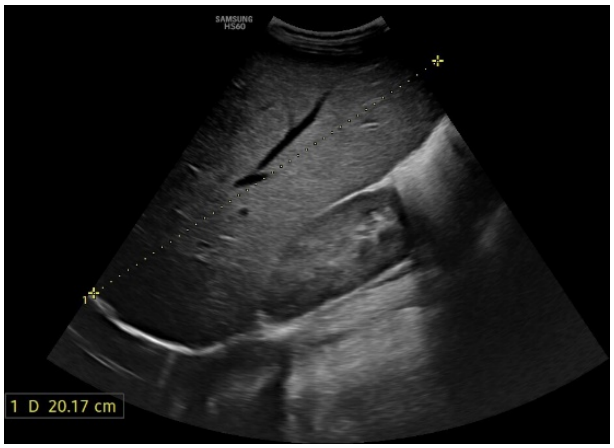

Figure 1. Longitudinal ultrasound section showing hepatomegaly with a liver span of 20 cm.

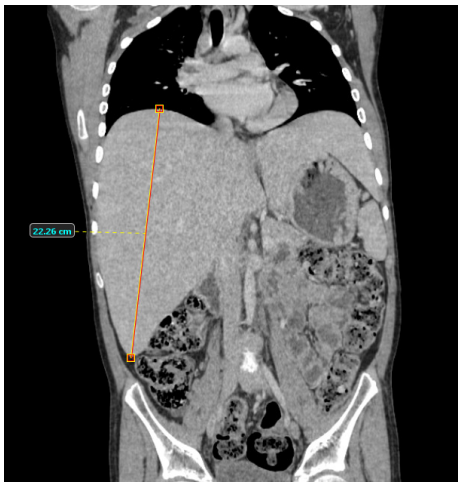

Figure 2. Coronal CT image of the abdomen showing a hepatomegaly.

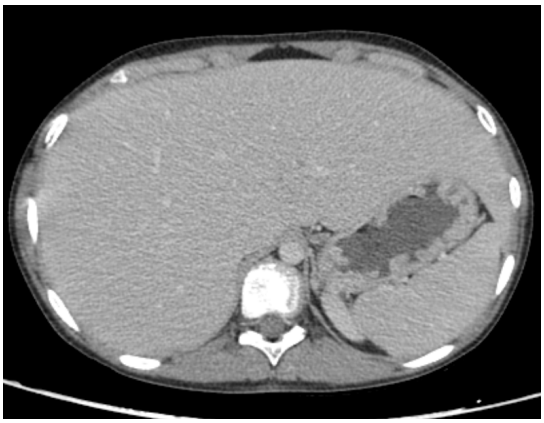

Figure 3. Transverse CT image of the abdomen showing an enlarged liver.

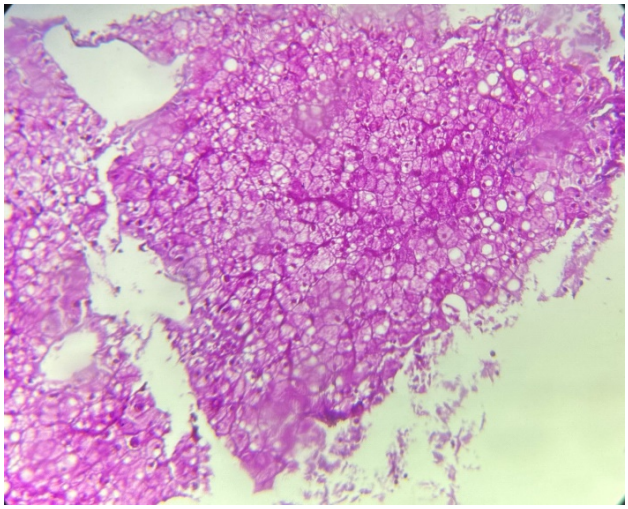

Figure 4. Hepatocytes with microvacuolar cytoplasm, strongly positive for PAS staining (HE × 40), showing intracytoplasmic glycogen deposits.

The patient's clinical status improved significantly with a multidisciplinary approach. This included structured diabetes education programs tailored to both the patient and his caregivers, optimized glycemic management with strict adherence to insulin therapy, proper nutritional supplementation, and consistent, close follow-up care. These combined efforts were essential in achieving glycemic control, and the patient's liver function tests quickly returned to normal.

3. Discussion

Mauriac syndrome, a glycogenic hepatopathy, was first described as excessive hepatic glycogen accumulation leading to hepatomegaly in association with unstable T1DM [1] [2]. Its other main features include growth retardation, delayed puberty, cushingoid signs, and elevated transaminases [1]-[4]. The pathophysiology remains only partially understood and involves marked fluctuations in plasma glucose levels, with alternating episodes of hyperglycemia and hyperinsulinism [2]. During hyperglycemia, glucose passively enters hepatocytes, promoting glycogen synthesis and leading to excessive hepatic glycogen accumulation. This process is further augmented by the administration of insulin at supraphysiologic doses, which stimulates glycogenesis and inhibits glycogenolysis by activating glucokinase and glycogen synthase while suppressing glucose-6-phosphatase [3].

Investigations for hepatomegaly and elevated aminotransferases involve screening for infectious, metabolic, obstructive, oncologic, and autoimmune causes to rule out all potential contributors to liver damage [5]. Diagnostic imaging, such as ultrasound and computed tomography, aids in identifying hepatomegaly, assessing liver tissue characteristics, and excluding other causes of liver enlargement [2].

Mauriac syndrome is often underdiagnosed due to its overlap with metabolic dysfunction-associated steatotic liver disease (MASLD) [3]. These two conditions cannot be reliably distinguished through history, physical examination, biological

markers, or radiological findings, making liver biopsy the gold standard for diagnosis [6] [7]. Mauriac syndrome typically occurs in individuals with T1DM, often in association with acute ketoacidosis or recurrent hypoglycemia, whereas MASLD is more commonly linked to type 2 diabetes [1]. The majority of children with MASLD have a body mass index in the overweight or obese range [8]. Both Mauriac syndrome and MASLD are associated with significant dyslipidemia, and radiological imaging reveals similar liver appearances [9]. Proper tissue preparation, such as using Carnoy's solution [10], and specialized staining methods like periodic acid-Schiff (PAS), are crucial for identifying glycogen accumulation. Histologically, hepatic glycogenosis is characterized by pale, swollen hepatocytes with marked glycogen deposition, minimal fatty change, negligible inflammation, and intact liver architecture with no significant fibrosis [11]

Management primarily involves stringent glycemic control to reverse hepatic glycogen accumulation. Improvement in glycemic control typically leads to normalization of liver function and resolution of hepatomegaly within weeks to months [12] [13]. While long-term evaluation of outcomes in patients with Mauriac syndrome is necessary, the condition is generally considered benign, with a low risk of progression to fibrosis, unlike MASLD.

This case underscores the critical need for multidisciplinary management involving endocrinologists, hepatologists, and dietitians to optimize glycemic control and ensure comprehensive patient care, ultimately improving outcomes.

4. Conclusion

Despite significant advancements in insulin therapy, consistent monitoring and proactive interventions remain pivotal, particularly in patients with suboptimal glycemic control. In the future, the advancement of noninvasive imaging methods may facilitate the differentiation with MASLD.

Conflicts of Interest

The authors declare no conflicts of interest.

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