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CASE REPORT

Mauriac syndrome: A rare complication of uncontrolled type 1 diabetes mellitus in childhood - A case report

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Clinical Pearls

- **Unique Features of the Case:** Mauriac syndrome occurred in a child with poorly controlled T1DM, presenting with hepatomegaly, growth failure, delayed puberty, and elevated liver enzymes.
- **Primary Lesson for Clinicians:** Consider Mauriac syndrome in children with poorly controlled T1DM and hepatomegaly. Early recognition, sustained glycemic control, diabetes education, regular follow-up, psychosocial support, and treatment adherence may prevent recurrence and long-term complications.

ABSTRACT

Mauriac syndrome is a rare but reversible complication of long-standing, poorly controlled type 1 diabetes mellitus (T1DM) in children. It is characterized by growth failure, delayed puberty, hepatomegaly due to hepatic glycogen accumulation, and metabolic abnormalities. An 11-year-old girl with T1DM for 6 years, on irregular insulin therapy, presented with progressive abdominal distension for 1 year, poor linear growth, and visual disturbances. Examination revealed short stature, delayed pubertal development, hypertension, and marked hepatomegaly. Investigations showed poor glycemic control, elevated transaminases, hypertriglyceridemia, delayed bone age, and bilateral diabetic cataracts. Viral, autoimmune, metabolic, and endocrine causes of liver disease were excluded. Imaging revealed hepatomegaly without fibrosis. Liver biopsy demonstrated glycogen-laden hepatocytes, confirming glycogenic hepatopathy. The child was initiated on an intensive basal–bolus insulin regimen with comprehensive diabetes education. On follow-up, there was a significant reduction in hepatomegaly with better glycemic control. Mauriac syndrome should be considered in children with poorly controlled T1DM presenting with hepatomegaly and growth failure.

Keywords: Diabetes, growth failure, hepatomegaly.

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Introduction

Mauriac syndrome is a rare complication of long-standing, poorly controlled type 1 diabetes mellitus (T1DM) in children and adolescents. It is classically characterized by growth retardation, delayed pubertal development, and hepatomegaly secondary to excessive hepatic glycogen accumulation.[1,2] Although the incidence of Mauriac syndrome has markedly declined with the advent of modern insulin regimens and improved glycemic monitoring, sporadic cases continue to be reported, particularly among patients with poor metabolic control or erratic insulin adherence.[3]

There is hepatic glycogenesis, which manifests clinically as hepatomegaly and biochemically as mild-to-moderate elevations in serum transaminases.[4,5] In addition, disruption of the growth hormone–insulin-like growth factor-1 (GH–IGF-1) axis and relative hypercortisolism are thought to contribute to growth failure and delayed pubertal development observed in affected children.[6]

Despite its rarity in the current era, recognition of Mauriac syndrome remains clinically important because of its reversible nature. Optimization of insulin therapy and sustained glycemic control have been shown to result in regression of hepatomegaly, normalization of liver enzymes, and improvement in growth parameters.[7] This report aims to highlight the clinical features, diagnostic challenges, and therapeutic implications of Mauriac syndrome in contemporary pediatric diabetes care.

Case Report

An 11-year-old female child with a normal birth and development history was diagnosed with T1DM since 5 years of age and is on insulin therapy, but non-adherent to treatment. She presented with complaints of abdominal distension for 1 year, poor height gain for the last 5 years and blurring of vision for 2 months. Abdominal distension was not associated with jaundice, abdominal pain, bleeding manifestations, chronic diarrhea or constipation. She was started on regular insulin and long-acting insulin (glargine) at 1 unit/kg/day at diagnosis; however, she was not compliant due to financial constraints. She had poor glycemic control for the last 5 years, requiring four admissions to hospitals with complaints of vomiting and respiratory difficulty. The mother was being treated for hypothyroidism. On examination, she had pallor but no icterus, cyanosis, clubbing, edema, or lymphadenopathy. Anthropometry revealed short stature with a height-for-age Z score of -3.17. Midparental height could not be calculated as the father had died 5 years earlier due to alcoholic liver disease. Lipohypertrophy was noted at insulin injection sites. She had sexual maturity rating, Tanner stage 1. Blood pressure recordings at regular intervals with a digital sphygmomanometer showed hypertension (average systolic pressure above 95th percentile for her age, sex and height as per the Indian Academy of Pediatrics blood pressure charts).

Abdominal examination revealed a soft, distended abdomen with dilated superficial veins. The liver was palpable 8–12 cm below the right costal margin, firm, non-tender, with a smooth surface. The spleen was not palpable. The rest of the systemic examinations were normal. Investigations revealed poor glycemic control with hemoglobin A1c (HbA1c) of 11%. IGF-1 levels were not done due to financial constraints. Liver transaminases were elevated (aspartate aminotransferase 216 IU/L, alanine aminotransferase 462 IU/L) with normal serum bilirubin, serum albumin and coagulation profile. Her kidney function tests were normal. Triglyceride levels were elevated (485 mg/dL). Viral hepatitis markers and autoimmune hepatitis panel (antinuclear antibodies, anti-smooth muscle antibodies, and anti-liver kidney microsomal antibodies) were negative. Ophthalmological evaluation showed no Kayser–Fleischer ring with normal serum ceruloplasmin levels. Thyroid function tests and celiac serology were normal. Bone age was delayed (8–9 years). Other laboratory values are depicted in Table 1. Ultrasonography of the abdomen showed hepatomegaly with normal portal venous flow. FibroScan with M probe revealed steatosis with no evidence of hepatic fibrosis. Her 2D-Echocardiogram and renal artery Doppler were normal. Ophthalmological evaluation showed bilateral diabetic cataracts without evidence of diabetic retinopathy. A liver biopsy was done, and histopathology showed enlarged hepatocytes with pale cytoplasm and glycogen in hepatocytes with periodic acid–Schiff (PAS) staining. (Figs. 1, 2). Based on the clinical profile of growth failure, hepatomegaly, poor glycemic control, and supportive investigations of liver biopsy and FibroScan, a diagnosis of Mauriac syndrome (glycogenic hepatopathy) was made. Considering the clinical presentation, Cushing syndrome was considered a differential diagnosis and evaluated with morning 8 am cortisol of 40.20 (4.82–19.5). However, the histopathology of the liver showed glycogen deposition with no signs of fibrosis; raised cortisol was attributed to poorly controlled hyperglycemia, altering the circadian rhythm of cortisol. The child was initiated on a structured basal–bolus insulin regimen using insulin glargine and insulin aspart, with a total daily dose of approximately 1.25 U/kg/day. Intensive diabetes education, carbohydrate counting, hypoglycemia management, and injection site rotation were reinforced. Vitamin D and calcium supplementation were initiated, and antihypertensive therapy was started with close monitoring. At the 15-day follow-up, the child showed a reduction in abdominal distension, a decrease in liver size with liver palpable 2 cm below the right costal margin, and improved adherence to insulin therapy with better glycemic monitoring. We had planned to monitor liver function tests, HbA1c levels, liver ultrasound, growth monitoring and ophthalmic re-examination after at least 1 month of initiation of treatment; however, the child has not followed up after the first follow-up visit.

Discussion

Mauriac syndrome results from chronic insulin deficiency with intermittent hyperglycemia leading to excessive hepatic glycogen accumulation. Hepatomegaly, growth retardation, delayed bone age, and dyslipidemia are hallmark features.[4] Glycogenic hepatopathy must be differentiated from non-alcoholic fatty liver disease (NAFLD), as the former is potentially reversible with strict glycemic control. Early diagnosis and sustained metabolic control can lead

Table 1. Laboratory findings

	16/9/25	19/9/25	22/9/25	24/9/25	5/10/25
Hemoglobin (g/dL)	13.2			12.8	11.9
Total leukocyte count (×1000/µL)	6.5			10.1	7.52
Platelet count (×1000/µL)	488			437	520
Total bilirubin (mg/dL)	0.5			0.6	
Direct bilirubin (mg/dL)	0.2			0.3	
AST (U/L)	216			282	
ALT (U/L)	462			392	
Serum creatinine (mg/dL)	0.3			0.4	
Pre-feed blood glucose (mg/dL)	344	302	286	210	126
Post-feed blood glucose (mg/dL)	565	556	465	387	202
HbA1c (%)	11				
Serum cholesterol (mg/dL)	382				
LDL (mg/dL)	310				
HDL (mg/dL)	49.9				
Triglycerides (mg/dL)	485				
Cortisol (µ/dL)	40.20				

AST: Aspartate aminotransferase; ALT: Alanine aminotransferase; HbA1c: Hemoglobin A1c; LDL: Low-density lipoprotein; HDL: High-density lipoprotein.

to significant clinical improvement and prevent progression to liver fibrosis. The present case highlights the classical clinical spectrum of Mauriac syndrome. The child had long-standing poor glycemic control, evidenced by persistently elevated HbA1c levels, growth retardation with delayed bone age, delayed pubertal development, dyslipidemia, and marked hepatomegaly. The pathophysiology of Mauriac syndrome involves chronic insulin deficiency interspersed with episodes of relative insulin excess, which promotes excessive hepatic glucose uptake and glycogen deposition.[4] This abnormal glycogen accumulation leads to hepatic glycogenosis, manifesting as hepatomegaly and elevation of liver transaminases, as observed in this patient.[5] Growth failure in Mauriac syndrome is multifactorial. Poor metabolic control leads to disruption of the GH–IGF-1 axis, reduced hepatic IGF-1 production, and relative hypercortisolism, all

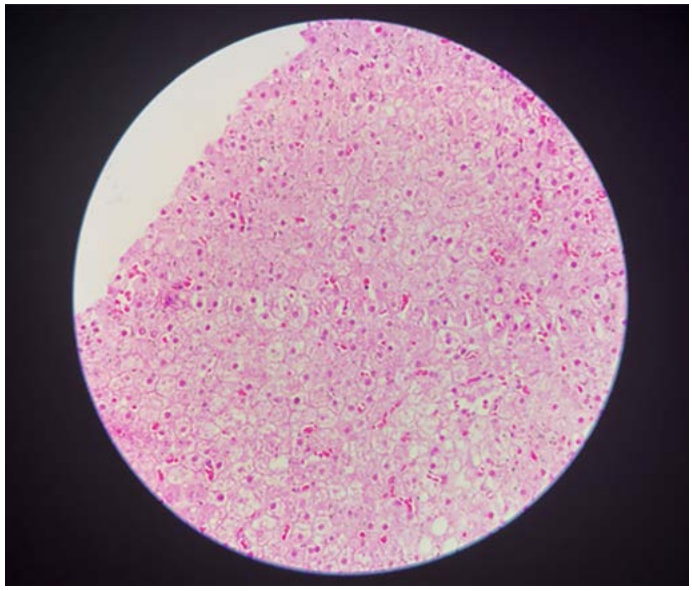


Figure 1. Liver histopathology with PAS staining showing glycogen accumulation in cytoplasm of hepatocytes.

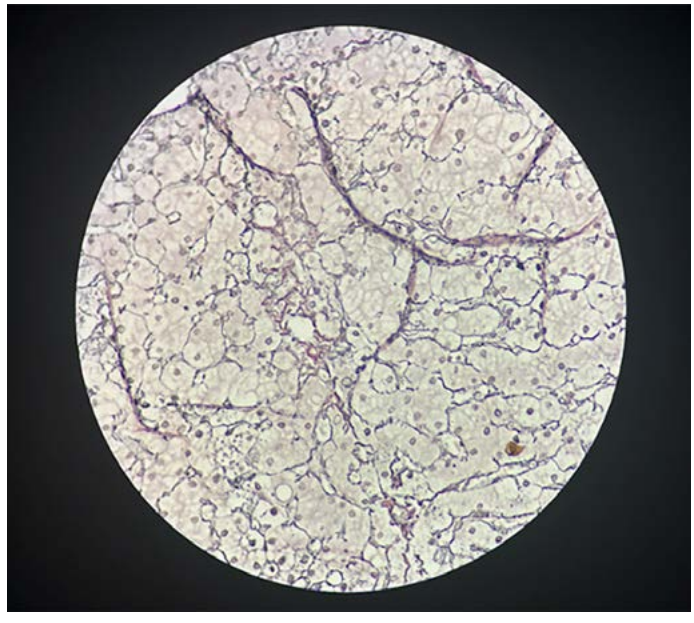


Figure 2. Liver histopathology showing pale swelling of hepatocytes.

of which contribute to impaired linear growth and delayed skeletal maturation.[6] In our case, the child had a significantly low height-for-age Z score and delayed bone age, supporting these mechanisms. In addition, delayed sexual maturation (Tanner stage I) further underscores the impact of prolonged metabolic derangement on pubertal development. Differentiating glycogenic hepatopathy from NAFLD is clinically important, as both conditions may present with hepatomegaly and elevated transaminases in children with T1DM. However, NAFLD is associated with insulin resistance and carries a risk of progression to fibrosis, whereas glycogenic hepatopathy is largely benign and reversible.[7] In this case, the absence of fibrosis on FibroScan, normal synthetic liver function, and liver biopsy findings of PAS-positive glycogen accumulation without significant inflammation or fibrosis confirmed the diagnosis of glycogenic hepatopathy. The presence of diabetic cataract in this child further reflects the chronicity and severity of hyperglycemia. Ocular involvement, though less frequently reported in Mauriac syndrome, emphasizes the systemic impact of prolonged poor glycemic control. Importantly, with initiation of a structured basal–bolus insulin regimen and intensive diabetes education, the child demonstrated clinical improvement, including reduction in hepatomegaly and abdominal distension. This favorable response reinforces existing evidence that strict glycemic control can lead to rapid and sustained reversal of hepatic and growth abnormalities in Mauriac syndrome.[7]

Conclusion

Mauriac syndrome should be considered in children with T1DM presenting with hepatomegaly and growth failure. Early recognition, continuous diabetes education and social support are essential to prevent recurrence. Mauriac syndrome, though rare in the modern era of diabetes management, remains a relevant clinical entity in children with poorly controlled T1DM. This case underscores the importance of considering Mauriac syndrome in children presenting with hepatomegaly, growth failure, delayed puberty, and elevated liver enzymes. Comprehensive diabetes education, regular follow-up, psychosocial support, and reinforcement of treatment adherence are crucial to prevent recurrence and long-term complications. Awareness of this condition among clinicians can facilitate timely intervention and significantly improve clinical outcomes.

Ethics Committee Approval: This is a single case report, and therefore ethics committee approval was not required in accordance with institutional policies.

Informed Consent: Written informed consent was obtained from the patient's parent/legal guardian for the publication of this case and accompanying images.

Conflict of Interest: All authors declare no conflicts of interest.

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