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Case Report

Case report on Mauriac syndrome-revisiting a rare complication of a common condition

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ABSTRACT

Mauriac syndrome is a rare complication of poorly controlled type 1 diabetes mellitus, characterized by growth retardation, hepatomegaly, delayed puberty, and cushingoid features. We describe a 9-year-old girl with long-standing diabetes and poor glycemic control who presented with short stature, abdominal distension and dyslipidemia, with imaging findings suggestive of glycogen accumulation. These features established the diagnosis of Mauriac syndrome; a condition infrequently encountered today due to advances in insulin therapy. However, the case underscores that this syndrome still occurs in settings with limited resources and suboptimal treatment adherence. Early recognition is crucial, as improved glycemic control can reverse most clinical and biochemical abnormalities. This case report emphasizes the need for vigilance in diabetic children with growth failure or delayed puberty, and highlights the importance of education, monitoring, and multidisciplinary care in prevention of complications.

Keywords: Mauriac syndrome, Type 1 diabetes mellitus, Hepatomegaly

INTRODUCTION

Mauriac syndrome is a rare complication of poorly controlled T1DM in children, first identified by Pierre Mauriac in a 10-year-old girl in 1930.[1] It is distinguished by a unique combination of growth retardation, delayed puberty, cushingoid facies, truncal obesity, and hepatomegaly due to hepatic glycogen overload. The pathogenesis involves chronic insulin deficiency or inappropriate insulin therapy, leading to sustained hyperglycemia and disturbances in growth hormone dynamics, with consequent liver dysfunction. Though more prevalent in the pre-insulin era, this condition persists in patients with improperly managed diabetes poor adherence to treatment, particularly in regions with limited healthcare access. Early recognition of the characteristic clinical profile is crucial, as optimizing glycemic control can markedly improve growth, restore pubertal development, and resolve hepatomegaly in affected children.

CASE REPORT

A 9-year-old girl (Figure 1), second issue of a healthy non-consanguineous marriage, presented to our hospital with complaints of gradually progressive abdominal distension. The girl was a known case of T1DM, first diagnosed at 2 years age. She was put on basal-bolus insulin regimen (1.2 u/kg/d) seven years back but was poorly compliant and had a history of five hospital admissions for diabetic ketoacidosis. Antenatal and natal history were not contributory. Family history, including that of her 14-year-old brother, was seemingly normal. She had attained her childhood milestones in time and had average scholastic performance.

On examination, she had a pulse rate of 110/min, respiratory rate of 24/min, blood pressure of 104/70 mm Hg and SpO_2 97% in room air. She had pallor, pitting bipedal edema and abdominal distension. She also had cushingoid features in the form of moon facies, thin skin, easy bruisability and pendulous abdomen; acanthosis

nigricans; proportionate short stature, as evident from the anthropometry (Table 1); random blood sugar 180 mg/dl; Tanner stage 1 and height age of 2 years 8 months. The other significant finding on physical examination was non-tender hepatomegaly with a size of 5 cm below the right costal margin, smooth surface and rounded margin. There was no ascites or splenomegaly. Laboratory investigations are listed in Table 2. USG abdomen revealed moderately-enlarged fatty liver with a span of 16 cm. Owing to financial constraints, detailed workup, including hormonal analysis and liver biopsy, could not be performed.

A multimodal approach was devised with the involvement of a pediatric endocrinologist and a dietitian. Insulin therapy, exercise and nutrition formed the cornerstone of management plan. Medical nutrition therapy comprised minimising the JUNCS, i.e. junk foods, ultra-processed foods, nutritionally inappropriate foods, caffeinated/carbonated/coloured beverages and sugar-sweetened beverages. Various aerobic and anaerobic exercises were taught and the parents were asked to ensure their child indulged in 45-60 minutes of such exercise daily. Insulin therapy was optimized based on blood sugar levels and parents were counselled to maintain compliance. Sick day guidelines were also explained. As part of protocol, she was screened for other micro- and macrovascular complications prior to discharge.

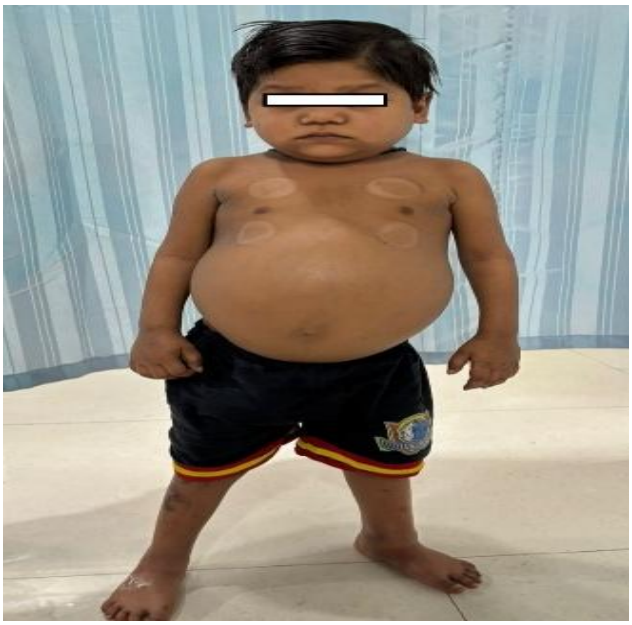

Figure 1: The patient.

Table 1: Anthropometry.

Parameters	Observed	Expected	Inference
Weight for age	14.5 kg	27 kg	<3rd centile
Height for age	92 cm	132 cm	<3rd centile
US:LS	1.13		
BMI (kg/m²)	17.13	15.9	>50th centile

Table 2: Laboratory investigations.

Parameters	Result	Reference range
Hgb	11.1	11.5-13.5 gm/dl
TLC	6.6	4-11×10³/µl
Platelet count	369	150-450×10³/µl
HbA1c	11.1	4-6%
Total protein	6.1	6-8.3 gm/dl
Serum albumin	3.2	3.5-5.2 gm/dl
Serum globulin	2.9	2.3-3.6 gm/gl
ALT	94	0-35 IU/l
AST	161	0-40 IU/l
ALP	311	90-320 IU/l
Urea/ creatinine	15 / 0.3	15-40/0.2-1.2 mg/dl
Serum Na^+	129	135-145 mEq/l
Serum K^+	4.2	3-6 mEq/l
TSH	5.462	0.5-5.5 µIU/ml
Total T_4	6.3	5.5-12.8 µg/dl
Total T_3	1.23	0.9-2.0 ng/ml
Urinalysis	Glucose 2000 mg/dl; absent ketones No significant bacteriuria	Nil glucose and ketones
Cholesterol	193	150-200 mg/dl
Triglycerides	233	100-150 mg/dl
HDL	35	40-70 mg/dl
LDL	149	60-150 mg/dl

DISCUSSION

Mauriac syndrome, also known as hepatic glycogenosis, is a relatively uncommon complication seen in patients with poorly controlled juvenile T1DM. Though more common in the past, it is rarely encountered these days due to better glycemic control with the help of improved varieties of insulin. It is said to be caused by a mutation in PHKG2 which is the catalytic subunit of the enzyme glycogen phosphorylase kinase (GPhK). GPhK is an enzyme complex responsible for the activation of glycogen phosphorylase.[2,3]

The pathophysiology of Mauriac syndrome revolves around poor glycemic control resulting in alternating hyperinsulinemia and hyperglycemia, thereby promoting glycogen accumulation in the liver and elevated corticosteroid levels in the blood, thus the cushingoid appearance. Despite recent studies, the exact mechanism is still not clear.[4,5]

The etiology of delayed growth in Mauriac syndrome is multi-factorial. These factors include inadequate tissue glucose, decreased growth hormone levels and IGF-1, impaired or resistant hormone receptor action and raised

cortisol. Hepatomegaly and impaired liver function are thought to be due to glycogen deposition in hepatocytes.[6]

This entity can be classified into two different forms according to the presence or absence of obesity. The first form includes patients with obesity and cushingoid features while the second form includes patients with persistent hyperglycemia but no obesity.[7]

The goal of treatment is improving the metabolic status of the patient which automatically leads to improvements in cushingoid features, growth and pubertal delay.[7]

Besides exploring the clinical and laboratory features of Mauriac syndrome, this case report also serves to highlight the ongoing challenges of pediatric diabetes care, emphasizing the need for vigilant monitoring and individualized therapy. It aims to enhance awareness and promote timely intervention among pediatricians managing kids with diabetes.

CONCLUSION

In conclusion, this case report on Mauriac syndrome in a poorly controlled pediatric T1DM patient underscores the critical interplay between chronic hyperglycemia, hepatic glycogenosis and growth impairment, highlighting an uncommon yet reversible manifestation which often goes undiagnosed in resource-limited settings. By detailing the diagnostic challenges, this case helps advance clinical understanding by demonstrating early recognition and multimodal management as key to preventing long-term complications like delayed puberty and metabolic sequelae.

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REFERENCES

1. Murata F, Horie I, Ando T, Isomoto E, Hayashi H, Akazawa S, et al. Gros ventre, hepatomegalie, troubles de las croissance chez les enfants diabetiques traits depuis plusieurs annes par l'insuline. Gax Hebd Med Bordeaux. 1930;26(8), 402-10.
2. Lombardo F, Passanisi S, Gasbarro A, Tuccari G, Ieni A, Salzano G. Hepatomegaly and type 1 diabetes: a clinical case of Mauriac's syndrome. Italian J Pediat. 2019;45(1):3.
3. MacDonald MJ, Hasan NM, Ansari IUH, Longacre MJ, Kendrick MA, Stoker SW. Discovery of a Genetic Metabolic Cause for Mauriac Syndrome in Type 1 Diabetes. Diabetes. 2016;65(7):2051-9.
4. Alhajjaj AH, Aljishi FK. Mauriac Syndrome Still Exists in Poorly Controlled Type 1 Diabetes: A Report of Two Cases and Literature Review. Cureus. 2021;13(4):e14704.
5. Patita M, Nunes G, Alves De Matos A, Coelho H, Fonseca C, Fonseca J. Mauriac Syndrome: A Rare Hepatic Glycogenosis in Poorly Controlled Type 1 Diabetes. GE Portuguese J Gastroenterol. 2019;26(5):370-4.
6. Lee RGL, Bode HH. Stunted growth and hepatomegaly in diabetes mellitus. J Pediat. 1977;91(1):82-4.
7. Gutch M, Philip R, Saran S, Tyagi R, Gupta K. Re-emergence of a rare syndrome: A case of Mauriac syndrome. Indian J Endocrinol Metabol. 2013;17(1):283.

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