Case Report

The forgotten face of Type 1 diabetes: A rare case of Mauriac syndrome in an adolescent male

Avi Singh[1], Mohd Laraib[2], Deepak Sharma[3], Saurabh Srivastava[4]

From [1]Fellow Diabetes, [2]PGJR, [3]Assistant Professor, [4]Professor, Department of Medicine, Government Institute of Medical Sciences, Greater Noida, Uttar Pradesh, India

ABSTRACT

Mauriac syndrome is a rare but clinically significant complication of poorly controlled Type 1 diabetes mellitus (T1DM), characterized by growth retardation, delayed puberty, hepatomegaly, delayed skeletal maturation, and impaired sexual maturation. We report the case of an 18-year-old male with T1DM diagnosed at 6 years of age who presented with severe short stature, delayed puberty, hepatomegaly, splenomegaly, and markedly delayed skeletal maturation. At presentation, he was receiving a basal-bolus insulin regimen, and glycated hemoglobin was 8.0%. Clinical examination revealed height below the third percentile, Tanner stage II pubertal development, hepatomegaly, and mild splenomegaly. Hormonal evaluation demonstrated hypogonadotropic hypogonadism, while radiographic assessment of skeletal maturity revealed a bone age of approximately 9 years despite a chronological age of 18 years. The diagnosis of Mauriac syndrome was established from the combination of long-standing T1DM, severe growth retardation, delayed puberty, delayed skeletal maturation, hepatomegaly, and exclusion of other endocrine and hepatic disorders. This case emphasizes that Mauriac syndrome should remain an important differential diagnosis in adolescents with T1DM presenting with growth failure despite apparently satisfactory contemporary glycemic control.

Key words: Bone age, Delayed puberty, Glycogenic hepatopathy, Growth retardation, Mauriac syndrome, Type 1 diabetes mellitus

Mauriac syndrome is a rare but well-recognized complication of poorly controlled type 1 diabetes mellitus (T1DM), first described by Mauriac in 1930 [1]. The syndrome is classically characterized by growth retardation, delayed puberty, hepatomegaly, delayed skeletal maturation, and occasionally cushingoid features [1,2]. Before the advent of intensive insulin therapy, Mauriac syndrome represented a severe manifestation of chronic metabolic decompensation in children with T1DM. However, improvements in insulin analogues, continuous glucose monitoring, structured diabetes education, and multidisciplinary diabetes care have led to a marked decline in its incidence [2,3]. The pathogenesis of Mauriac syndrome is complex and multifactorial. Chronic insulin deficiency during critical periods of growth results in impaired hepatic production of insulin-like growth factor-1 (IGF-1), despite normal or elevated growth hormone concentrations, thereby disrupting linear growth and skeletal maturation [4,5]. Persistent metabolic derangement also suppresses the hypothalamic–pituitary–gonadal axis, contributing to delayed puberty and hypogonadotropic hypogonadism [5]. Hepatomegaly, one of the hallmark manifestations of the syndrome, is commonly attributed to glycogenic hepatopathy, a reversible condition caused by excessive glycogen accumulation within hepatocytes secondary to recurrent fluctuations between hyperglycemia and exogenous insulin administration [6]. The diagnosis of Mauriac syndrome remains primarily clinical and is supported by careful anthropometric assessment, pubertal staging, endocrine evaluation, radiographic assessment of bone age, and appropriate imaging studies after excluding other causes of growth failure and hepatomegaly [7]. Although hepatomegaly is a classical feature, splenomegaly is uncommon and should prompt evaluation for alternative causes, including portal hypertension, chronic liver disease, hematological disorders, and infiltrative conditions before being attributed to Mauriac syndrome [8].

We report a classical presentation of Mauriac syndrome in an adolescent male with long-standing T1DM who presented with severe growth retardation,

Access this article online	
Received - 04 July 2026 Initial Review - 20 July 2026 Accepted - 04 August 2026	Quick Response code
DOI: ***	

Correspondence to: Dr. Avi Singh, Government Institute Of Medical Sciences, Greater Noida. E-mail: avi1310in@gmail.com

delayed puberty, marked skeletal maturation delay, hypogonadotropic hypogonadism, and hepatosplenomegaly. This case highlights that the cumulative historical burden of poor glycemic control, rather than a single contemporary glycated hemoglobin (HbA1c) value, may determine the development of this rare complication and underscores the importance of early recognition to prevent irreversible impairment of growth and pubertal development.

CASE PRESENTATION

An 18-year-old male presented to the Endocrinology outpatient department with complaints of short stature and delayed pubertal development. He had been diagnosed with T1DM at the age of 6 years and had been receiving insulin therapy for the preceding 12 years. Detailed history obtained from the patient and his parents revealed prolonged periods of poor glycemic control during childhood and early adolescence due to irregular insulin administration, socioeconomic constraints, inadequate diabetes education, and inconsistent medical follow-up. There was no history of recurrent diabetic ketoacidosis, chronic diarrhea, prolonged corticosteroid therapy, chronic systemic illness, recurrent infections, or significant weight loss unrelated to diabetes.

At presentation, the patient was receiving a basal-bolus insulin regimen comprising regular insulin (4 units before breakfast, lunch, and dinner) and insulin glargine (8 units at bedtime). Self-monitoring of blood glucose over the preceding several months suggested relatively improved glycemic control. HbA1c at presentation was 8.0%, although previous medical records documented persistently poor metabolic control during childhood. The patient was born at term following an uncomplicated pregnancy with normal birth weight and had achieved developmental milestones appropriately. There was no family history of diabetes mellitus, delayed puberty, short stature, chronic liver disease, endocrine disorders, or inherited metabolic diseases. The mother's height was 143.5 cm, and the father's height was 162 cm.

On examination, the patient appeared considerably younger than his chronological age. His height was 160 cm (below the 3rd percentile for age according to the Centers for Disease Control and Prevention growth charts), weight was approximately 32 kg, and body mass index was below the normal range for age and sex (Fig. 1). Blood pressure was 108/72 mmHg, pulse rate was 78 beats/min, respiratory rate was 16 breaths/min, and oxygen saturation was 98% on room air. Secondary sexual characteristics were markedly delayed. Tanner staging corresponded to stage II, with sparse pubic hair, minimal axillary and facial hair, and bilateral testicular volume of approximately 4–5 mL, consistent with delayed pubertal development. No gynecomastia, dysmorphic features, cushingoid facies, moon facies, or buffalo hump were noted. Abdominal examination revealed hepatomegaly, with the liver palpable approximately 5–6 cm below

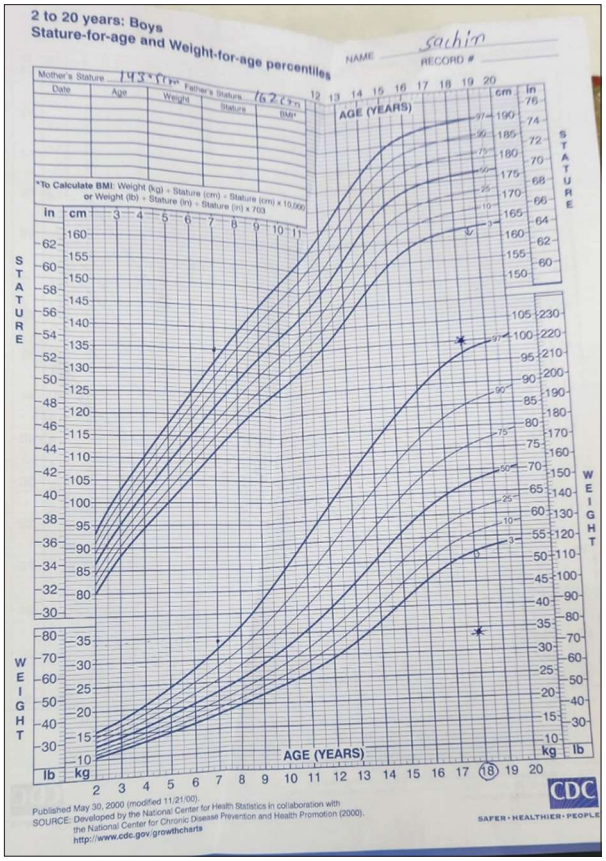

Figure 1: The Centers for Disease Control and Prevention growth chart

the right costal margin, and mild splenomegaly, with the spleen palpable approximately 2 cm below the left costal margin. There was no ascites, jaundice, pedal edema, spider angiomas, palmar erythema, or other clinical features suggestive of chronic liver disease or portal hypertension. Cardiovascular, respiratory, and neurological examinations were unremarkable.

Laboratory investigations demonstrated a fasting plasma glucose of 128 mg/dL, postprandial plasma glucose of 182 mg/dL, and HbA1c of 8.0%. Complete blood count, renal function tests, serum electrolytes, and thyroid function tests were within normal limits. Liver function tests revealed mild elevation of serum transaminases with preserved synthetic liver function. Endocrine evaluation showed markedly reduced serum testosterone levels with inappropriately low luteinizing hormone (LH) and follicle-stimulating hormone (FSH) concentrations, consistent with hypogonadotropic hypogonadism. Serum prolactin levels were within normal limits.

Abdominal ultrasonography demonstrated hepatomegaly and mild splenomegaly with preserved hepatic echotexture and no evidence of portal hypertension, cirrhosis, biliary obstruction, or focal hepatic lesions. Magnetic resonance imaging, transient elastography (FibroScan), and liver biopsy were not performed. Therefore, although glycogenic hepatopathy could not be confirmed histologically or radiologically, it was considered the most probable cause of hepatomegaly based on the characteristic clinical presentation, prolonged history of poorly controlled T1DM, mild

transaminase elevation, and absence of alternative causes of chronic liver disease.

Radiographic assessment of skeletal maturity was performed using radiographs of the left hand, wrist, and elbow. Comparison with the Greulich and Pyle atlas demonstrated markedly delayed skeletal maturation, corresponding to a bone age of approximately 9 years despite a chronological age of 18 years. Persistent open epiphyseal growth plates and delayed ossification centers were evident (Fig. 2).

The differential diagnosis included constitutional delay of growth and puberty, growth hormone deficiency, hypothyroidism, celiac disease, chronic systemic illness, glycogen storage disorders, chronic liver disease, and isolated hypogonadotropic hypogonadism. However, the coexistence of long-standing poorly controlled T1DM, severe growth retardation, delayed puberty, markedly delayed bone age, hypogonadotropic hypogonadism, and hepatomegaly strongly supported the diagnosis of classical Mauriac syndrome. Although splenomegaly is not a typical manifestation, the absence of clinical or ultrasonographic evidence of portal hypertension or chronic liver disease made other common causes less likely.

DISCUSSION

Mauriac syndrome is a rare but classical complication of poorly controlled T1DM, first described by Mauriac in 1930 [1]. Although considered uncommon in the era of intensive insulin therapy, sporadic cases continue to be reported worldwide, particularly in patients with prolonged inadequate glycemic control during childhood and adolescence [2,3]. The present case demonstrates the classical phenotype of Mauriac syndrome, characterized by severe growth retardation, delayed puberty, markedly delayed skeletal maturation, hepatomegaly, and hypogonadotropic hypogonadism.

The pathophysiology of Mauriac syndrome is multifactorial and primarily related to prolonged insulin deficiency during critical periods of growth. Chronic hyperglycemia impairs hepatic production of IGF-1 despite normal or elevated growth hormone concentrations, resulting in functional growth hormone resistance and impaired linear growth [4,5]. Persistent metabolic derangement also disrupts the hypothalamic-pituitary-gonadal axis, leading to delayed pubertal development and hypogonadotropic hypogonadism [5]. The markedly delayed bone age and low gonadotropin levels observed in our patient are consistent with these mechanisms.

Growth retardation remains one of the most striking manifestations of Mauriac syndrome. Although our patient's current HbA1c was 8.0%, review of previous medical records and history revealed prolonged periods of inadequate glycemic control during childhood. This observation emphasizes that the development of Mauriac syndrome reflects the cumulative burden of chronic metabolic dysregulation rather than a single contemporary HbA1c measurement. Similar observations have been reported in recent case reports, where patients developed classical manifestations despite partial improvement in glycemic control before presentation [3,7].

Delayed puberty is another hallmark feature of Mauriac syndrome and is believed to result from chronic suppression of the hypothalamic-pituitary-gonadal axis secondary to prolonged metabolic stress and nutritional compromise [5,9]. In the present case, Tanner stage II sexual maturation, reduced serum testosterone concentrations, and inappropriately low LH and FSH levels were consistent with hypogonadotropic hypogonadism. Early recognition is important because pubertal progression may improve following sustained metabolic optimization before epiphyseal fusion occurs [10].

Hepatomegaly is one of the cardinal manifestations of Mauriac syndrome and is most commonly attributed to glycogenic hepatopathy, a condition characterized by excessive glycogen accumulation within hepatocytes resulting from recurrent fluctuations between hyperglycemia and exogenous insulin administration [6]. Glycogenic hepatopathy is generally considered a benign and potentially reversible condition with improvement in glycemic control. However, definitive diagnosis requires liver biopsy demonstrating glycogen accumulation or characteristic findings on advanced imaging modalities such as gradient dual-echo magnetic resonance imaging. As neither liver biopsy, magnetic resonance imaging,

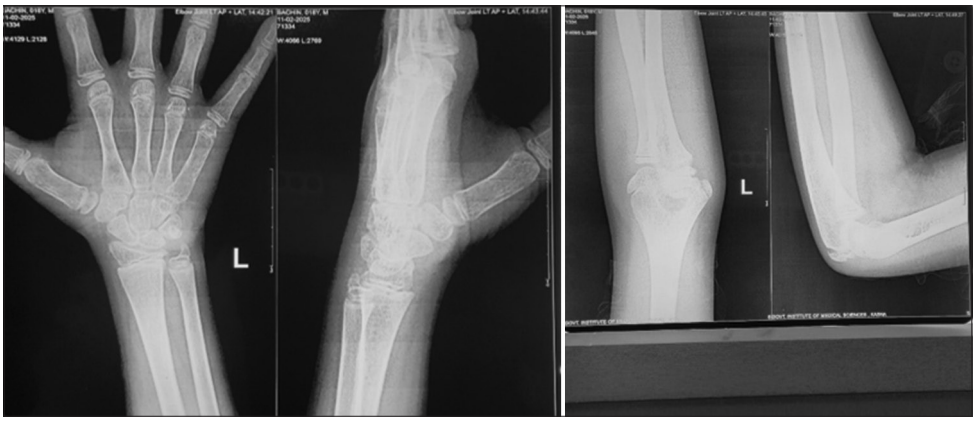

Figure 2: X-rays of the left hand with wrist and elbow joint

nor transient elastography (FibroScan) was performed in our patient, glycogenic hepatopathy could not be confirmed conclusively. Therefore, hepatomegaly was considered most likely secondary to probable glycogenic hepatopathy, based on the characteristic clinical context, ultrasonographic findings, mild elevation of liver enzymes, and absence of clinical or imaging features suggestive of chronic liver disease. This represents an important limitation of the present report.

The presence of splenomegaly in our patient deserves special consideration. Unlike hepatomegaly, splenomegaly is not a classical feature of Mauriac syndrome and has been described only in isolated case reports [8,11]. The underlying mechanism remains uncertain, although chronic hepatic involvement, altered portal hemodynamics without overt portal hypertension, or long-standing metabolic disturbances have been proposed. Importantly, alternative causes, including portal hypertension, chronic liver disease, hematological disorders, infiltrative diseases, and chronic infections, should be carefully excluded before attributing splenomegaly to Mauriac syndrome. In our patient, preserved hepatic architecture on ultrasonography and the absence of portal hypertension, cytopenias, constitutional symptoms, or clinical evidence of chronic liver disease made these alternative diagnoses less likely.

The diagnosis of Mauriac syndrome is essentially clinical and should be based on the characteristic constellation of long-standing T1DM, growth failure, delayed puberty, hepatomegaly, delayed skeletal maturation, and supportive endocrine evaluation after excluding other causes of growth retardation [7]. The principal differential diagnoses include constitutional delay of growth and puberty, growth hormone deficiency, hypothyroidism, celiac disease, glycogen storage disorders, chronic systemic illnesses, chronic liver disease, and isolated hypogonadotropic hypogonadism [12]. In the present case, the coexistence of long-standing T1DM with severe growth retardation, delayed bone age, hypogonadotropic hypogonadism, and hepatomegaly strongly supported the diagnosis of Mauriac syndrome.

Management primarily focuses on sustained optimization of glycemic control through intensive insulin therapy, structured diabetes education, nutritional rehabilitation, and close endocrinology follow-up [13]. Advances in continuous glucose monitoring and insulin pump therapy have substantially reduced the incidence of Mauriac syndrome and may facilitate recovery of hepatic enlargement, pubertal progression, and growth velocity when instituted before epiphyseal closure [3,14]. Although catch-up growth may be limited in patients with longstanding disease, timely intervention remains essential to prevent further endocrine and metabolic complications.

Our case reinforces several important clinical messages. First, Mauriac syndrome, although rare, continues to occur in developing countries where prolonged suboptimal glycemic control remains common. Second, a relatively acceptable HbA1c at presentation should not exclude the diagnosis, as historical glycemic burden is often more relevant than current metabolic status. Third, careful anthropometric assessment, Tanner staging, bone age estimation, and endocrine evaluation remain indispensable in adolescents with T1DM presenting with growth failure.

The present report has certain limitations. Advanced imaging modalities such as magnetic resonance imaging or transient elastography (FibroScan) and liver biopsy were not performed; therefore, glycogenic hepatopathy could not be confirmed histologically or radiologically. Similarly, assessment of serum IGF-1 and growth hormone axis was not available. Nevertheless, the characteristic clinical phenotype, endocrine findings, markedly delayed bone age, and exclusion of common alternative diagnoses strongly supported the diagnosis of Mauriac syndrome.

CONCLUSION

Mauriac syndrome remains a rare but clinically significant complication of poorly controlled T1DM despite major advances in diabetes management. This case demonstrates that the syndrome should continue to be considered in adolescents with long-standing T1DM who present with severe growth retardation, delayed puberty, and hepatomegaly, even when contemporary glycemic control appears relatively satisfactory. The presence of markedly delayed skeletal maturation and hypogonadotropic hypogonadism further strengthened the diagnosis in our patient.

REFERENCES

1. Mauriac P. Large abdomen, hepatomegaly, and growth disorders in diabetic children treated with insulin for several years. Gaz Hebd Sci Med Bordeaux 1930;26:402-10.
2. Kim MS, Quintos JB. Mauriac syndrome: Growth failure and type 1 diabetes mellitus. Pediatr Endocrinol Rev 2008;5: 989-93.
3. 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:e14704.
4. Acerini CL, Savage MO. Growth and puberty in children with type 1 diabetes mellitus. Horm Res 2001;56 Suppl 1:28-33.
5. Merimee TJ, Fineberg SE. Growth hormone, insulin-like growth factors and diabetes mellitus. Endocr Rev 1986;7:34-44.
6. Sherigar JM, Castro J, Yin YM, Guss D, Mohanty SR. Glycogenic hepatopathy: A narrative review. World J Hepatol 2018;10:172-85.
7. Kocova M, Milenkova L. Old syndrome-new approach: Mauriac syndrome treated with continuous insulin delivery. SAGE Open Med Case Rep 2018;6:2050313X18785510.
8. Pinto MJ, Melo N, Flores L, Cunha F. Mauriac syndrome: A rare complication of Type 1 diabetes mellitus. Eur J Case Rep Intern Med 2018;5:000969.
9. Soliman AT, De Sanctis V, Elalaily R. Nutrition and pubertal development. Indian J Endocrinol Metab 2014;18 Suppl 1: S39-47.
10. Dunger DB, Edge JA. Predicting and preventing complications in type 1 diabetes. Arch Dis Child 1997;77:458-62.

11. 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 Port J Gastroenterol 2019;26:370-4.
12. Greulich WW, Pyle SI. Radiographic Atlas of Skeletal Development of the Hand and Wrist. 2nd ed. Stanford, CA: Stanford University Press; 1959.
13. American Diabetes Association Professional Practice Committee. 13. Children and adolescents: Standards of care in diabetes-2025. Diabetes Care 2025;48 Suppl 1:S273-99.
14. Chiang JL, Maahs DM, Garvey KC, Hood KK, Laffel LM, Weinzimer SA, *et al*. Type 1 diabetes in children and adolescents: A position statement by the American diabetes association. Diabetes Care 2018;41:2026-44.

Funding: Nil; Conflicts of interest: Nil.

How to cite this article: Singh A, Laraib M, Sharma D, Srivastava S. The Forgotten face of type 1 diabetes: A rare case of Mauriac syndrome in an adolescent male. Indian J Case Reports. 2026; August 11 [Epub ahead of print].