Medical and Clinical Research: Open Access

Research Article

Diabetic Ketoacidosis Can Reveal Mauriac Syndrome: A Case Report

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ABSTRACT

Background: Mauriac syndrome is a rare but potentially reversible complication of poorly controlled type 1 diabetes mellitus in children and adolescents. It is characterized by hepatomegaly, growth impairment, delayed puberty, and marked elevation of liver enzymes secondary to hepatic glycogen accumulation. We report a case in which diabetic ketoacidosis (DKA) revealed Mauriac syndrome.

Case Presentation: A 15-year-old girl with type 1 diabetes diagnosed at the age of 3 years was admitted for her third episode of DKA related to persistent poor treatment adherence. Since early adolescence, psychosocial difficulties had resulted in recurrent hyperglycemia, repeated DKA episodes, and failure of multiple therapeutic strategies, including continuous glucose monitoring, insulin pump therapy, and psychological support. Clinical examination revealed severe short stature (<−3 SD), Cushingoid features, delayed pubertal maturation, and painless hepatomegaly. Laboratory investigations showed markedly elevated liver enzymes (AST 122 IU/L, ALT 102 IU/L, GGT 244 IU/L), HbA1c of 15%, and exclusion of infectious, autoimmune, and other hepatic disorders. Abdominal ultrasound confirmed homogeneous hepatomegaly without focal lesions. Liver biopsy was not performed. Standard DKA management followed by intensive basal-bolus insulin therapy resulted in rapid normalization of glycemic control, complete resolution of hepatomegaly, and normalization of liver enzymes within two weeks, strongly supporting the diagnosis of Mauriac syndrome.

Discussion: This case highlights Mauriac syndrome as an uncommon but important differential diagnosis in adolescents with poorly controlled type 1 diabetes presenting with DKA and liver dysfunction. Although liver biopsy remains the diagnostic gold standard, the combination of characteristic clinical features, exclusion of alternative etiologies, and complete reversibility following restoration of glycemic control strongly supports the diagnosis. Early recognition is essential because the condition is reversible with optimized insulin therapy and sustained treatment adherence.

Conclusion: Mauriac syndrome should be considered in any child or adolescent with poorly controlled type 1 diabetes presenting with DKA, hepatomegaly, and elevated liver enzymes, particularly when associated with growth or pubertal delay. Prompt restoration of stable glycemic control and multidisciplinary interventions addressing therapeutic adherence are critical to prevent recurrence and improve long-term outcomes.

Keywords

Mauriac syndrome, Diabetic ketoacidosis, Type 1 diabetes mellitus, Hepatic glycogenosis, Hepatomegaly, Treatment adherence.

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Received: May 28, 2026; **Accepted:** June 29, 2026; **Published:** July 10, 2026

Citation: Lufuluabo Serge, Ntalu Nkatu Christophe, Mangyanda kinkembo Laurent, Mbuila Nseka Célestin, Nsibu Ndosimao Célestin, et al. Diabetic Ketoacidosis Can Reveal Mauriac Syndrome: A Case Report. Med Clin Res Open Access. 2026; 7(1):1-4.

Introduction

Two main groups of complications are usually observed in the course of diabetes mellitus: earlier, more frequent microangiopathies and macroangiopathies.

Other rare complications can also accompany the disease. These complications are observed exclusively in children and adolescents whose diabetes, primarily type 1, is poorly controlled. Among these complications is Mauriac syndrome (MS) or Hepatic Glycogen Storage Disease (HGD). This condition is clinically characterized by failure to thrive, delayed puberty, hepatomegaly, and, biologically, hepatic cytolysis with very high liver markers. A definitive diagnosis is made by histology; however, in its absence, clinical and biological signs, as well as the response to normoglycemic treatment, suggest the disease.

We report here the case of a 15-year-old girl, followed in the Pediatric Department of the Delafontaine Hospital Center in Saint-Denis, France, for type 1 diabetes (T1D) diagnosed at the age of 3. At 15, she was hospitalized for diabetic ketoacidosis.

Case Report

This concerns a 15-year-old girl who was hospitalized in the intensive care unit of the CHSD (Centre Hospitalier Sud-Ouest) for a third diabetic ketoacidosis episode due to poor treatment adherence.

Her medical history indicates that her diabetes was diagnosed at age 3 with diabetic ketoacidosis following a three-week-long cardinal syndrome. The teenager experienced menarche at age 14 and was four months amenorrheic on the day of her hospitalization. Glycemic control was satisfactory at the beginning of her illness thanks to multiple daily insulin injections. However, starting at age 11, due to psychosocial difficulties leading to a kind of denial of the illness and poor treatment adherence, she regularly experienced several episodes of hyperglycemia and hypoglycemia, requiring recurrent hospitalizations for diabetic ketoacidosis complications. The attempt to use continuous glucose monitoring sensors and an external insulin pump proved ineffective, forcing a return to the initial regimen of multiple daily injections and finger-prick blood glucose monitoring, but without success. The psychological support implemented also failed to have the desired effect on treatment adherence.

On admission, the patient presented with asthenia, signs of dehydration, non-sensitive and anicteric hepatomegaly, short stature (< 3 SD) (Photo 2), and a Cushingoid facies (Photo 2). The Tarner stage was S4 P4 A2. Laboratory tests revealed hepatic cytolysis with very high markers (AST: 122 IU/L; ALT: 102 IU/L; GGT: 244 IU/L) but with a low total bilirubin level of 5 µmol/L. Viral serologies were negative, including those for HAV, HBV, HCV, HEV, CMV, and EBV. Tests for antinuclear, anti-tissue, and anti-LKM1 antibodies were negative. Other laboratory tests showed an HbA1c level of 15% and alkaline phosphatase of 128 IU/L. The plasma beta-hCG level was negative. The IgA levels were normal.

Abdominal and pelvic ultrasound revealed homogeneous hepatomegaly with a liver span of 250 mm in the midclavicular region, without focal nodular lesions (Photo 3), and showed no pelvic abnormalities.

Therapeutic management focused on correcting fluid and electrolyte imbalances through IV hydration combined with intravenous insulin therapy via syringe pump (IVSE), followed by insulin pen therapy according to the conventional basal-bolus regimen. The outcome was favorable after ten days, with normalization of blood glucose levels, resolution of the hepatomegaly, and normalization of liver markers. The adolescent was thus discharged from the hospital after two weeks, with measures implemented to prevent recurrence.

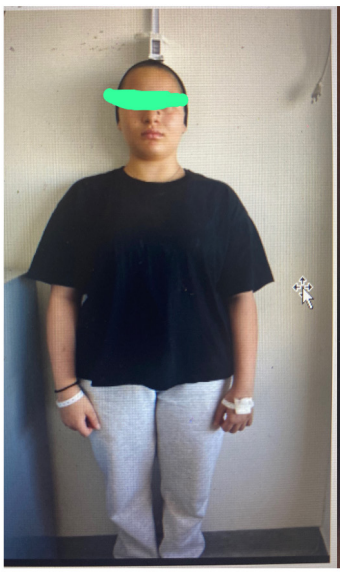

Photo 1

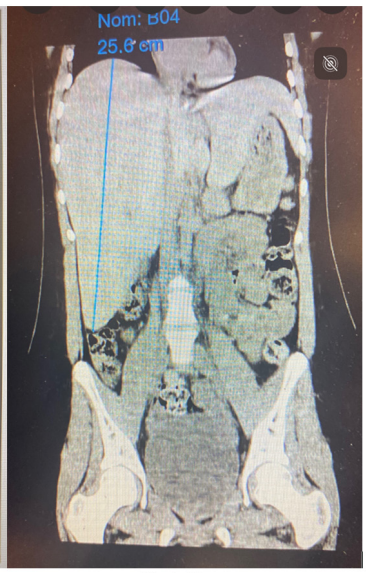

Photo 2

Discussion

Metabolic syndrome (MS) is a very rare complication observed in poorly controlled type 1 diabetes.

It was first described in 1930 by Mauriac [2] in adolescents with poorly controlled type 1 diabetes who presented with a clinical profile dominated by short stature, hepatomegaly, and laboratory findings revealing liver cytolysis more than ten times the normal level.

It primarily affects children and adolescents and is virtually nonexistent in adults. It does not affect individuals of either sex.

Currently, it has become rare thanks to the prescription of new long-acting insulins and the use of new technologies such as continuous glucose monitoring sensors and external insulin pumps in open or closed loop configurations.

The mechanism leading to the hepatomegaly and cytolysis observed in hepatic glycogenosis results from a combined effect of frequent hyperglycemia and the overuse of insulin to correct this hyperglycemia. Indeed, the overuse of insulin leads to excess glucose

storage in hepatocytes through excessive and passive transport of blood glucose to hepatocytes via the GLUT2 glucose transporter. In hepatocytes, glucose is converted to glucose-6-phosphate by the enzyme glucokinase. The glucose-6-phosphate thus trapped in the hepatocytes is converted into glycogen. This massive storage of glycogen in hepatocytes explains the hepatomegaly with cytolysis [1] (Figure 1).

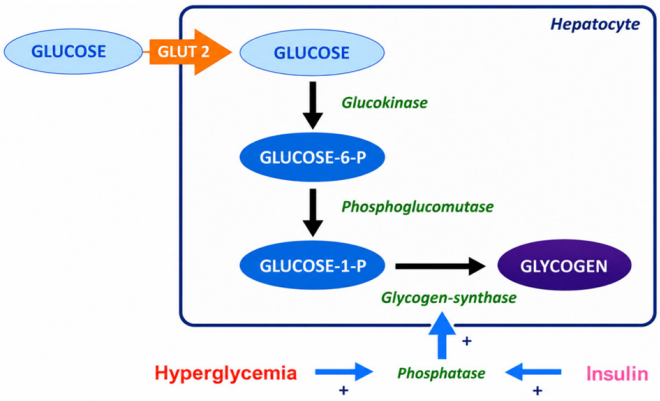

Figure 1: Mechanism of hepatomegaly and cytolysis [1]. GLUT2 : Glucose Transporter Type 2; Glucose-6-P : Glucose - 6 -phosphate ; Glucose-1-P : Glucose - 1'phosphate.

In our patient, this presentation appeared in the context of social conflict and anorexia nervosa, which led to deviations in treatment, resulting in ketoacidotic complications and thus the use of high doses of insulin.

The pathogenesis of growth and pubertal delay is not clearly understood. It is thought to be multifactorial: growth retardation is believed to be a consequence of insulin deficiency due to its anabolic (growth-promoting) effect, which would promote hepatic production of IGF-1 (insulin-like growth factor) under the influence of growth hormone (GH). This results in hepatic resistance to GH with decreased IGF-1 production and, consequently, growth retardation and bone retardation. As for delayed puberty, it is thought to be due to an alteration of the gonadotropin with an excess of corticosteroid hormones leading to hypercortisolism, which in turn leads to hypogonadotropic hypogonadism with a decrease in secondary sex hormones, resulting in delayed puberty [2]. In our young patient, the short stature was isolated, whereas it was associated with weight failure and lower limb edema in the study by M. Ohouana et al. [3].

The key examination for confirming the diagnosis of Mauriac syndrome, or hepatic glycogen storage disease, is a liver biopsy. Histology reveals an appearance of glycogen overload [4] and Figure 2. In its absence, as in our patient, clinical, biological, and anamnestic findings may suffice to suggest this diagnosis. This presents as ketoacidosis in a young diabetic patient on insulin, with an enlarged liver and significant abnormalities in liver markers, associated with short stature and sometimes delayed puberty. Furthermore, the normalization of liver markers and the regression of hepatomegaly provide additional support.

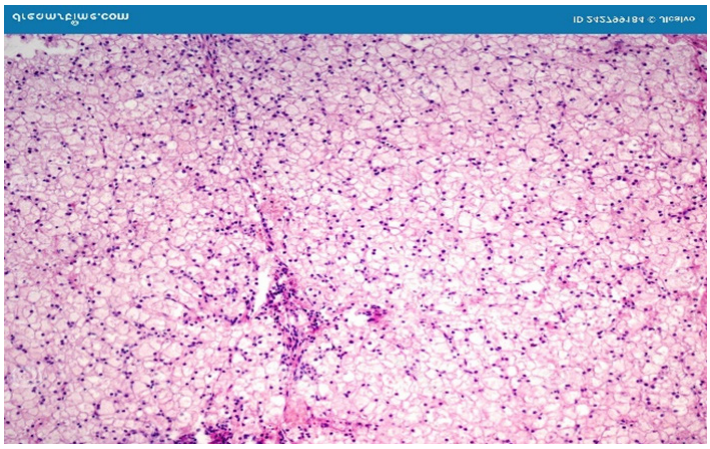

Figure 2: Liver histology [4].

Once ketoacidosis is corrected, the implementation of a normoglycemic insulin regimen and the establishment of regular glycemic control measures are sufficient to correct cytolysis and improve hepatomegaly.

In their observations, Medhioub et al. [4] emphasize the importance of therapeutic adherence in Mauriac syndrome. They report the case of a 19-year-old woman in whom improved glycemic control led to a regression of hepatomegaly with normalization of liver function tests, but who, unfortunately, due to poor adherence to home therapy, experienced a relapse. Thus, in this type of situation, functional insulin therapy using an external insulin pump coupled with open-loop or closed-loop sensors allows for sustained glycemic control and thus avoids this complication. However, all these therapeutic measures must be accompanied by psychological support for the child.

Conclusion and Recommandations

Although hepatic glycogen storage disease is a rare pathological entity, it should be considered in any young diabetic with ketoacidosis presenting with a clinical picture dominated by hepatomegaly associated with pubertal abnormalities, with or without short stature, as well as abnormal liver biomarkers.

Diagnostic certainty relies on histology; however, in its absence, rapid improvement of biological parameters and regression of hepatomegaly under normoglycemic insulin therapy may be sufficient to confirm this diagnosis.

Glycemic stability and good adherence to treatment within the framework of patient education are key preventive elements to avoid the onset or recurrence of this complication. Thus, clinical investigation for hepatomegaly, combined with its biomarkers, allows for the early detection of metabolic syndrome and ensures therapeutic compliance in diabetic adolescents.

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