www.jcimcr.org

JCIMCR OPEN ACCESS Journal of Clinical Images and Medical Case Reports

ISSN 2766-7820

Case Report

Open Access, Volume 6

Mauriac syndrome in adolescents: A persisting clinical entity

Marwa Sassioui*; Saloua Elamari; Fatima-Zahra Bellabes; Nada Faquir; Soukaina Laidi; Fatima Marouane

Department of Endocrinology, Diabetology, Metabolic Disease and Nutrition, Mohammed VI University of Health Sciences [UM6SS], 82403, Casablanca, Morocco.

***Corresponding Author: Marwa Sassioui**
Faculty of Medicine Mohammed VI University of Health Sciences [UM6SS] 82403, Casablanca, Morocco.
Email: Sassi-marwa@hotmail.fr

Received: Jun 24, 2025
Accepted: Jul 21, 2025
Published: Jul 28, 2025
Archived: www.jcimcr.org

DOI: www.doi.org/10.52768/2766-7820/3707

Abstract

Mauriac Syndrome (MS) is a rare complication of poorly controlled type 1 diabetes, primarily affecting children and adolescents, with fewer cases reported in adults. It is characterized by delayed growth, delayed puberty, hepatomegaly, and liver dysfunction, resulting from glycogen accumulation in the liver due to diabetic imbalance.

In this case, a 16-year-old patient with poorly controlled type 1 diabetes since age 2 presented with abdominal pain, diarrhea, nausea, and vomiting. Examination revealed severe statural delay, delayed puberty, and hepatomegaly. Laboratory tests showed hyperglycemia, metabolic acidosis, and elevated liver enzymes.

Imaging confirmed hepatomegaly. Viral and immunological studies were negative. During hospitalization, there was an improvement in liver function tests. Liver biopsy findings included hepatocytes with enlarged size and a vegetal appearance, with macrovesicular steatosis suggestive of glycogenolysis. Given the hepatomegaly, hepatic balance disturbances, negative etiological work-up, and association with growth delay and delayed puberty, Mauriac syndrome was considered. Treatment included rehydration and intravenous insulin. The case underscores the importance of recognizing Mauriac syndrome promptly for appropriate management.

Keywords: Mauriac syndrome; Type 1 diabetes; Hepatomegaly; Insulin therapy; Glycogen.

Introduction

Mauriac Syndrome (MS) is a rare complication of poorly controlled type 1 diabetes. It is mainly observed in children and adolescents, and is less common in adults.

It is defined by the presence in a type 1 diabetic of a delay in growth and development and puberty, hepatomegaly and a disturbance in the liver balance. This syndrome is secondary to a diabetic imbalance that causes an accumulation of glycogen in the liver [1].

MS represents a clinical challenge often overshadowed by the primary focus on glycemic control in diabetes management. In poorly controlled type 1 diabetic patients, periods of hyperglycemia followed by occasional hyperinsulinisation and high levels of cortisol as a hypoglycemic counter-regulatory hormone lead to hepatic glycogen storage. In case of hyperglycemia, glucose freely enters the hepatocyte and is also stored as glycogen. On the other hand, deficient insulinisation due to poor glycemic control leads to lipolysis and ketone release. Ketosis activates cortisol synthesis promoting fatty acid release and hyperglycemia [2].

A common finding in these patients is growth retardation and/or hypogonadism secondary to elevated cortisol levels.

In this case, we aim to elucidate the contemporary clinical insights into Mauriac syndrome among adolescents, emphasizing the diagnostic challenges, therapeutic considerations, and long-term implications for patient care.

Case presentation

The patient was 16 years old, known to have Type 1 Diabetes Mellitus (T1DM) since the age of 2 years, poorly followed up with repeated hospitalizations for episodes of ketoacidosis.

He presented three days before his admission to the intensive care unit: Epigastralgia radiating to the right hypochondrium, episodes of glutinous diarrhea, nausea and vomiting, without fever. The clinical examination revealed a somnolent patient, a blood pressure of 128/84 mmHg, a tachycardia of 150 beats/minute. He presented a Kussmaul's polypnea. The examination did not reveal any signs of organ hypoperfusion. In particular, there was no mottling, warm extremities and no oliguria. We noted an abdominal distension with a hepatic arrow superior to 12 cm.

The Height was 136 cm (-4 DS) and the weight was 30 kg. The pubertal stage according to the Tanner classification was P2G2 (Figure 1).

The capillary blood glucose was high above 6 g/l, and the urine dipstick showed a two-cross glycosuria and a three-cross ketonuria by semi-quantitative method. The Arterial blood gas showed metabolic acidosis. The plasma ionogram shows kalemia at 5.16 mmol/l, natremia at 128 mmol/l, urea at 0.38 mmol/l and creatinine at 9.4 µmol/l. the C-reactive protein was at 5.6 mg/l. The glycated hemoglobin was at 12.2%.

The liver balance was disturbed with a hepatic cytolysis: ASAT at 200 UI/l and ALAT at 270 UI/l, an anicteric cholestasis: (γGT at 80 UI/l, alkaline phosphatase =160 UI/l, total bilirubinemia at 2 µmol/l), with no stigma of associated hepatocellular failure (PT=93%).

On electrocardiogram, there was no repolarization abnormality, however troponins were negative at 0.006 µg/L.

We performed also a Viral serologies including Epstein-Barr Virus (EBV), Hepatitis A Virus (HAV), Hepatitis B Virus (HBV), Hepatitis C Virus (HCV), Hepatitis E Virus (HEV), Cytomegalovirus (CMV), and Herpes Simplex Virus (HSV), all was negative. Also, an immunological workup including anti-LKM1, anti-mitochondria and anti-smooth muscle antibodies was also negative.

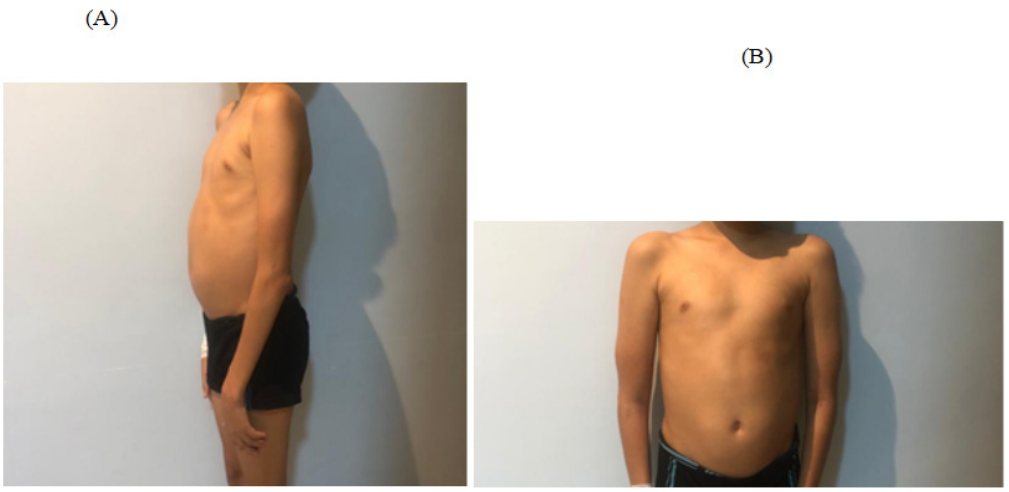

Figure 1: Image showing statural delay in our 16-year-old patient and abdominal distension. **(A)** Image showing statural delay in our 16-year-old patient. **(B)** Image illustrating abdominal distension.

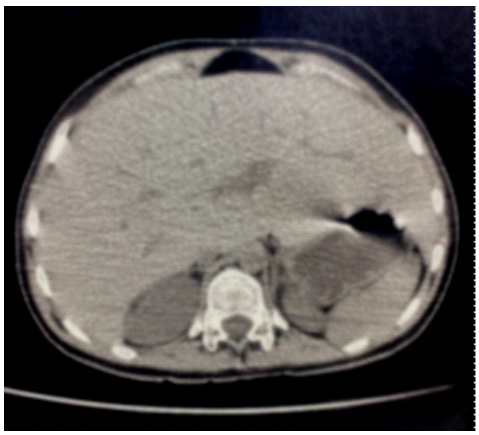

Figure 2: Scannographic image showing the enlargement of the liver which appears homogeneous.

In order to explore growth retardation, we carried out a balance sheet including the bone age which was 13 years and a IGF1 dosage which was normal for the age and a testosterone dosage of the LH which showed the demarcation absence of puberty. the rest of the hypophysogram including cortisol and thyroid hormones were without noticeable abnormalities.

During the hospitalization, we noted an improvement of the liver balance (ASAT: 58.9 IU/L, ALAT: 50.6 IU/L, GGT: 142 IU/L, PAL: 151 IU/L).

The liver biopsy showed hepatocytes with enlarged size and a vegetal appearance, featuring clarified cytoplasm occasionally granular without nuclear inclusion. A discrete macrovesicular steatosis estimated at less than 5% favored glycogenolysis (Figure 3).

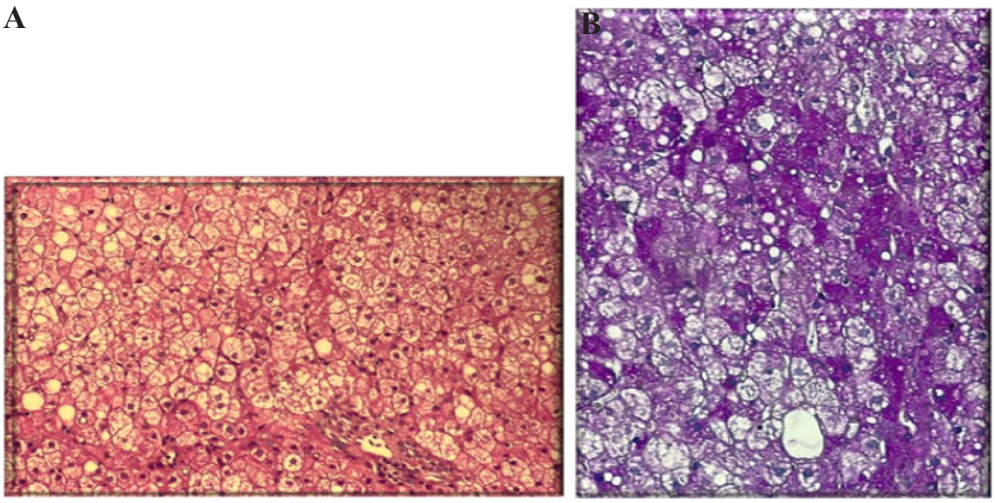

Figure 1: Image sowing the results of a liver biopsy. **(A)** Hepatocytes with clarified and granular cytoplasm (HEX40). **(B)** Positive for PAS staining (negative after diastase (x40).

Table 1: Laboratory data.

Plasma Ionogram	Values
Kalemia	5.16 mmol/l
Natremia	128 mmol/l
Urea	0.38 mmol/l
Creatinine	9.4 µmol/l
Other Laboratory Results	Values
C-reactive protein	5.6 mg/l
Glycated hemoglobin	12.20%
Liver Function Tests	Values
ASAT (SGOT)	200 UI/l
ALAT (SGPT)	270 UI/l
γGT	80 UI/l
Alkaline phosphatase	160 UI/l
Total bilirubinemia	2 µmol/l
Prothrombin Time (PT)	93%

An esophagogastroduodenoscopy was performed with duodenal biopsies, which did not reveal any evidence of celiac disease or autoimmune gastritis.

In view of the hepatomegaly, the disturbances of the hepatic balance, the negativity of the etiological work-up and the association with a delay in the growth and development of puberty in our patient who presented a chronic imbalance, no signs of celiac disease or Addison's disease, we evoke Mauriac's syndrome.

The treatment is mainly based on Insulin therapy and education of the patient and his parents on glycemic balance which can lead to complete remission of clinical, biological and histological abnormalities.

Discussion

Mauriac syndrome was first described by Mauriac in 1930, in children with Diabetes Mellitus Type 1 (DM1) presenting with stature-weight retardation, hepatomegaly, and elevated transaminases [2]. Mauriac syndrome occurs primarily in children and adolescents, but is almost absent in those who have passed the pubertal period.

Adolescence is a critical period of development recognized by changes in responsibilities and identity construction. This period is more complex for adolescents diagnosed with type 1 diabetes, who must struggle with intensive medical regimens, regular clinic appointments, and daily monitoring of blood glucose levels [3]. Although new insulin analogues and educational techniques allow for better diabetes self-management and glycemic control and thus improved quality of life, many adolescents with type 1 diabetes achieve suboptimal glycemic control. At the extreme, a Mauriac syndrome may still be seen in adolescents.

The actual cause is unknown, but it is plausibly a combination of factors:

- Inadequate glucose uptake and utilization in the tissues.
- Decreased insulin-like growth factor (IGF1) and growth hormone levels, altered bioactivity of these hormones,
- Insulin deficiency
- Poor glycemic control,
- Concomitant autoimmune diseases: include Addison's disease, autoimmune gastritis, celiac disease and hypothyroidism.
- Decreased caloric intake and/or eating disorders [4].

The diagnosis of Mauriac syndrome is suspected in the presence of: Hepatomegaly, delayed weight gain and hepatic cytolysis. The associated biological abnormalities are disturbances of the hepatic balance (elevation of transaminases and Gamma GT, alkaline phosphatases) and dyslipidemia with most often hypertriglyceridemia and sometimes hypercholesterolemia.

The differential diagnosis were the toxic causes, viral hepatitis, Wilson's disease, autoimmune hepatitis, celiac disease, minor glycogenosis. The clinical approach allows to orientate towards one or the other diagnosis, the paraclinical explorations confirm the clinical suspicion and allow to establish the diagnosis. Table 2 summarizes the explorations that allow to confirm the different differential diagnoses.

Diagnostic confirmation relies mainly on blood tests and liver tests which are essential to rule out most of these conditions. Some authors have highlighted the elevation of serum lactate as a possible biomarker of hepatic glycogenosis in young

Table 2: The explorations that allow to confirm the different differential diagnoses [10].

Differentiel diagnostics	Investigations
Toxic hepatitis	Investigation, urinary toxicity tests
Viral hepatitis	Viral hepatitis serology (A, B, C, E), EBV, CMV (parvovirus B19, HHV6) serology
Auto-immune hepatitis	Anti-LKM1, anti-smooth muscle, anti-nuclear, anti-cytosol, anti-actin antibodies, Serum protein electrophoresis
Celiac disease	Anti-transglutaminase IgA
Alpha 1 antitrypsin deficiency	Protein electrophoreses
Wilson's disease	Cupremia, ceruloplasmin, 24h cupruria, ophthalmologic examination
Endocrine pathology, (if growth retardation or puberty)	Thyroid check-up, cortisolemia, somatotropic or gonadotropic axis exploration, IGF1 dosage.
Myopathy, glycogenosis III	Determination of CPK

diabetic patients [5]. Ultrasound examination of the liver is a simple and useful procedure to obtain information about the size and characteristics of the liver tissue, but it is not a gold standard examination. Radiological imaging studies such as CT and Magnetic Resonance Imaging MRI to establish the diagnosis could depend on interval changes in liver density, but to date their sensitivity and specificity are not established for this condition [6]. Murata et al. reported the potential use of gradient-double echo magnetic resonance imaging sequence of the liver as a noninvasive and useful tool for the diagnosis of HG in distinguishing from nonalcoholic fatty liver disease [7]. The gold standard examination for the diagnosis of HG is liver biopsy [8], to rule out autoimmune hepatitis.

However, some authors do not recommend a biopsy if the liver balance is normalized with good glycemic control [9]. The evolution is usually favorable with good glycemic control, and the liver damage usually disappears in two to four weeks.

Conclusion

The discovery of hepatomegaly with delayed growth and weight in a diabetic patient should raise several diagnoses, including Mauriac syndrome, which, although rare, should not be overlooked by the practitioner, especially in cases of unbalanced diabetes.

This diagnosis must lead to a closer follow-up, in order to balance the patient's diabetes, which will allow him to recover both biologically and clinically. Appropriate insulin dosing and optimal glycemic control can improve hepatomegaly, hepatic glycogenosis and delayed pubertal maturation. The patient and family should be informed about compliance with insulin therapy and be involved in a therapeutic education program.

Declarations

Data availability: Data not available due to ethical restrictions due to the nature of this research, participants of this study did not agree for their data to be shared publicly, so supporting data is not available.

Conflicts of interest: The authors do not declare conflicts of interest.

Funding statement: The work doesn't receive specific funding.

References

1. Trifi A, Abdellatif S, Ben Ismail K, Touil Y, Daly F, et al. Glycogénose hépatique: Une complication rare du diabète déséquilibré (à propos d'un cas). Méd Intensive Réa. 2017; 26: 335-338.

2. Ohouana RLM, Belmejdoub G. Le Syndrome De Mauriac: A propos d'un cas. Health Sciences And Disease. 2022; 23(3).

3. Dias J, Martins S, Carvalho S, Marques O, Antunes A. Mauriac syndrome still exists. Endocrinología y Nutrition. 2013; 60(5): 245 248.

4. Madhu SV, Jain R, Kant S, Prakash V. Mauriac syndrome: A rare complication of type 1 diabetes mellitus. Indian J Endocrinol Metab. 2013; 17(4): 764 765.

5. Lombardo F, Passanisi S, Gasbarro A, Tuccari G, Ieni A, et al. Hepatomegaly and type 1 diabetes: A clinical case of Mauriac's syndrome. Ital J Pediatr. 2019; 45(1): 3.

6. Sweetser S, Kraichely RE. The bright liver of glycogenic hepatopathy. Hepatology. 2010; 51(2): 711 2.

7. Murata F, Horie I, Ando T, Isomoto E, Hayashi H, et al. A case of glycogenic hepatopathy developed in a patient with new-onset fulminant type 1 diabetes: the role of image modalities in diagnosing hepatic glycogen deposition including gradient-dual-echo MRI. Endocr J. 2012; 59(8): 669 76.

8. Hudacko RM, Manoukian AV, Schneider SH, Fyfe B. Clinical resolution of glycogenic hepatopathy following improved glycemic control. J Diabetes Complicat. 2008; 22: 329-30.

9. Giordano S, Martocchia A, Toussan L, Stefanelli M, Pastore F, et al. Diagnosis of hepatic glycogenosis in poorly controlled type 1 diabetes mel- litus. World J Diabetes. 2014; 5: 882-888.