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CASE REPORT

Persistent hyperlactataemia: a case report of Mauriac syndrome at Chris Hani Baragwanath Academic Hospital, South Africa

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Mauriac syndrome is a rare and potentially reversible complication of poorly controlled type 1 diabetes mellitus. This case report details the presentation of a patient at Chris Hani Baragwanath Academic Hospital, South Africa, who, over 8 years, presented with clinical features of Mauriac syndrome and persistent hyperlactatemia. The case highlights the association between socioeconomic factors and poor glycaemic control in resource limited settings, emphasising the importance of early recognition and multidisciplinary management. A limited number of described cases were found in the literature, but to the present authors' knowledge, no such cases have been published from South Africa. Evolving insights into the pathophysiology, prevalence, and reversibility of this syndrome are discussed, as well as highlighting the association with hyperlactataemia. While considered rare, Mauriac syndrome may be under-recognised and this underscores the need for tailored interventions to improve diabetes care and prevent long-term complications in vulnerable populations.

Keywords: hyperlactataemia, Mauriac syndrome

Introduction

Mauriac syndrome, a complication of uncontrolled type 1 diabetes, presents as a combination of growth retardation, delayed puberty, Cushingoid features, hepatomegaly, and poor glycaemic control. While the prevalence of this syndrome has decreased with advancement in insulin and glucose monitoring, cases continue to emerge, particularly in populations facing socioeconomic challenges.[1]

We present the case of a 16-year-old male patient with uncontrolled type 1 diabetes with features consistent with Mauriac syndrome and persistent hyperlactatemia. This report explores the clinical presentation, diagnostic challenges, and management of the patient until his unfortunate demise. We integrate insights from existing literature to contextualise the syndrome's relevance, especially in resource-constrained healthcare settings.

Case presentation

The case is of a patient who resided in Soweto, a large township in the south of Johannesburg. He lived with his mother in a house with access to basic amenities.

The patient was known to the Endocrine Clinic with type 1 diabetes mellitus diagnosed at the age of 9. His first admission to Chris Hani Baragwanath Academic Hospital was in 2019, at age 11, when he presented in diabetic ketoacidosis (DKA) secondary to poor compliance with medication due to poor socioeconomic circumstances and difficult access to healthcare.

The patient was admitted again that year for DKA. Following resolution of the DKA, his blood glucose exhibited wide variations, ranging from 3.2 to 26.7 mmol/L, despite an attempt at tight glucose control. It was noticed that he had hepatomegaly, but no jaundice. Blood results revealed elevated alanine transaminase, aspartate transaminase, and gamma-glutamyl transferase levels with no features of acute or chronic liver disease. Diagnostic tests such as cytomegalovirus, Epstein–Barr virus, and viral hepatitis serology, caeruloplasmin, and antibodies for autoimmune hepatitis were all negative. Ultrasonography revealed a homogeneous, enlarged right liver lobe with no focal lesions.

There were many further admissions for recurrent DKA. In October 2021, an abdominal ultrasound showed gross hepatomegaly of the right lobe of the liver with a bright echo pattern, with no focal liver lesions. On this admission he had persistent hyperlactataemia. A liver biopsy revealed hepatocytes that showed extensive feathery degeneration, large droplet steatosis of approximately 10% of hepatocytes, and glycogenated nuclei. On later admissions it was noted the patient had short stature for his age and delay in onset of puberty. Lactate levels were persistently elevated even when not in DKA (Figure 1). His HbA1c level remained persistently elevated; the last recorded one before his demise was 10.8%.

On his last admission, he presented in DKA and with a left lower limb deep vein thrombosis, presumably a complication from previous femoral central line insertion and reduced mobility. He was treated in the Medical Acute Care Unit after he developed bilateral pulmonary emboli. His clinical condition deteriorated, requiring inotropic support, endotracheal intubation, and ventilation; he was transferred to the Intensive Care Unit, where he spent approximately 4 weeks.

In ICU, he developed a ventilator-associated pneumonia complicated by acute respiratory distress syndrome and acute kidney injury requiring continuous renal replacement therapy. The patient then, unfortunately, died due to refractory multi-organ failure.

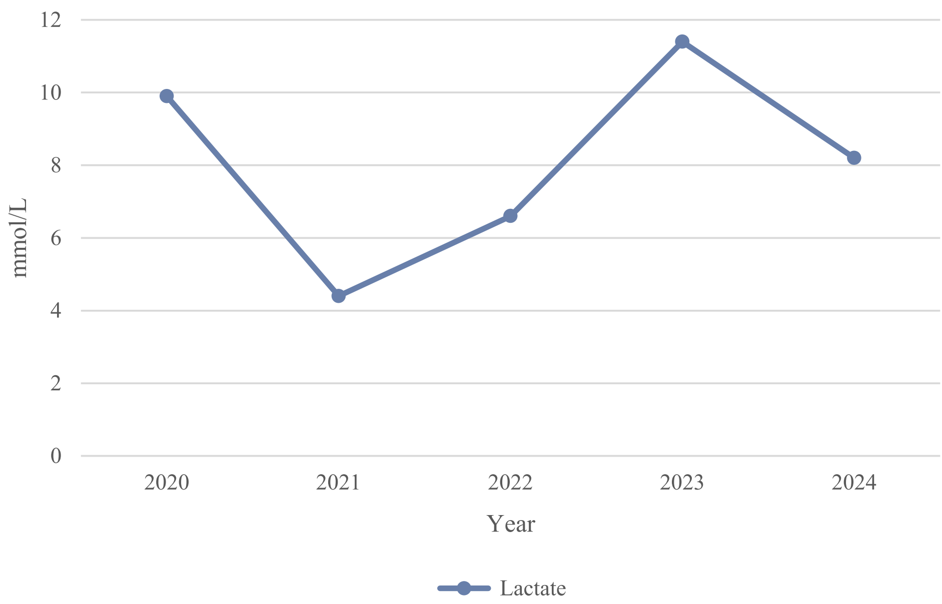

Figure 1: Lactate trend (2020–2024).

Discussion

Mauriac syndrome is considered a rare complication of type 1 diabetes, first described by French physician Pierre Mauriac in 1930.[2] The exact pathophysiology is not yet fully understood; it is postulated to be due to impaired tissue uptake, decreased insulin-like growth factor-1, decreased growth hormone levels, and increased cortisol levels.[3] To our knowledge, there are no published diagnostic criteria for Mauriac syndrome. It is characterised by growth impairment, hepatomegaly, and delayed puberty in adolescents with poorly controlled type 1 diabetes.[3] Cushingoid features include moon facies and a protuberant abdomen. Such patients often have dyslipidaemia, transaminase elevation, and hepatomegaly.[3] Mauriac syndrome is described as infrequent in modern times, likely due to advancement in insulin therapy and glucose monitoring reducing its prevalence in better-resourced areas.[4,5] There are limited data on the prevalence of this condition in lower income countries.[2]

Glycogen accumulation in hepatocytes is a hallmark of Mauriac syndrome. Its cause is attributed to excessive insulin doses and recurrent DKA episodes.[2,6] Prolonged hyperglycaemia causes hepatic glycogen deposition; large amounts of exogenous insulin drive glycogen synthesis and decrease gluconeogenesis and glycogenolysis.[7,8] Episodes of DKA compound the problem because treatment requires high amounts of insulin and dextrose, promoting glycogen synthesis.

Typical histological findings of glycogenic hepatopathy include marked glycogen accumulation in hepatocytes, which causes swelling with pale, clear cytoplasm. There is usually mild to no steatosis and no significant inflammatory infiltrate or fibrosis. Liver architecture is maintained, and on electron microscopy abundant glycogen granules are found within the cytoplasm.[2,9]

Mauriac syndrome is associated with persistent hyperlactatemia, particularly during episodes of DKA and persisting beyond its resolution, a phenomenon that was clearly demonstrated in this case.[6] In a paediatric study, over half of participants with Mauriac syndrome had elevated lactate levels despite no signs of illness or DKA.[9] The mechanism of hyperlactatemia in Mauriac syndrome is poorly understood.[10] A reduction in gluconeogenesis in the liver may raise lactate levels in the body. Hyperlactataemia could also be explained by impaired conversion of pyruvate to glucose.[6]

Mauriac syndrome is potentially reversible and liver abnormalities may improve with optimised blood glucose control.[2] There is a documented case of reversal of Mauriac syndrome in a patient admitted to the paediatric ICU with multiple DKA episodes.[11] The patient was managed with optimised insulin therapy, diabetic education, nutritional support, and close monitoring. Following intervention, her liver enzymes normalised, hepatomegaly resolved, and growth improved. This case emphasises that early recognition and multidisciplinary management can prevent both short-term complications like lactic acidosis and long-term consequences such as hepatic dysfunction and growth impairment.

Poor diabetes management often reflects socioeconomic disparities in healthcare, as shown by a published case of a patient with poor glycaemic control due to financial constraints preventing access to insulin.[12] The report highlights how children and adolescents in low-resource settings struggle with healthcare access, insulin affordability, and low health literacy, making them susceptible to Mauriac syndrome. The authors advocate for public health interventions to make diabetes care more accessible and affordable. Difficult socioeconomic circumstances and lack of family support played a significant part in our patient's illness.

Conclusion

Our case highlights the importance of recognising the clinical presentation of Mauriac syndrome in our healthcare setting and being aware of the presence of persistent hyperlactatemia as a complication. Early identification in vulnerable populations could prevent long-term complications such as hepatic dysfunction and growth impairment.

Mauriac syndrome remains a relevant clinical entity in resource-constrained settings where poor glycaemic control and healthcare inequities prevail. While the syndrome's reversibility offers hope for affected patients, systemic changes are needed to reduce the prevalence of such complications in underprivileged populations. Numerous areas in South Africa have socioeconomic barriers such as poor social support, low economic means, limited healthcare access, and low health literacy, significantly impacting diabetes management.

Future research into genetic and environmental associations of Mauriac syndrome will hopefully enhance understanding and improve diagnostic and therapeutic strategies.

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