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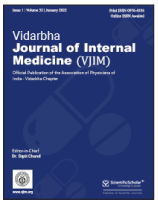

Case Report

Mauriac Syndrome in a Patient of Type 1 Diabetes Mellitus

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ABSTRACT

Mauriac syndrome is a metabolic complication of poor glycaemic control in Type 1 DM patients, unique to children, characterised by growth failure, short stature, delayed puberty, Cushingoid features, and marked hepatomegaly from massive deposition of glycogen. It is probably due to a combination of factors including inadequate glucose uptake and utilisation in the tissues, decreased insulin-like growth factor-1 and growth hormone levels, impaired bioactivity of these hormones, a circulating hormone inhibitor, resistant or defective hormone receptors, insulin deficiency, and poor glycaemic control. This syndrome is rarely seen nowadays because of better treatment modalities.

Keywords: Hepatomegaly, Mauriac Syndrome, and Type 1 Diabetes mellitus

INTRODUCTION

Mauriac syndrome is a metabolic complication of poor glycaemic control in Type 1 DM patients, unique to children, characterised by growth failure, short stature, delayed puberty, Cushingoid features, and marked hepatomegaly from massive deposition of glycogen.[1] We present to you a case of a 14-year-old female with a known history of Type 1 DM and poor glycaemic control admitted with the complaints of excessive loss of weight and abdominal distension for the past 1 month.

CASE REPORT

A 14-year-old female patient, resident of Wardha district, Maharashtra, was brought by relatives to medicine emergency with the complaints of excessive loss of weight, increased intake of food, and abdominal distension for the past 1 month. She was a known case of Type 1 diabetes mellitus, not compliant with insulin doses with no regular follow-up hospital visits. She was previously on injection Mixtard insulin (50:50) 6 units before breakfast and dinner and injection Actrapid insulin 4 units before lunch. There was a history of hospital admission 6 months back due to diabetic ketoacidosis when she was diagnosed with Type 1 diabetes mellitus.

On admission, after general physical examination, she was thin built with a protuberant abdomen, afebrile, conscious, oriented to time, place, and person, pulse rate was 76 beats per min regular, low volume, BP 70 systolic mm of Hg, maintaining 99% oxygen saturation on room air with random blood sugar level 251 mg/dl and urinary ketones small. On inspection, the patient was pale with no cyanosis, icterus, clubbing, oedema feet; jugular venous pressure 7 cm above the sternal angle. On abdominal palpation, she had a soft abdomen with moderate hepatomegaly and mild splenomegaly. On auscultation, a loud second heart sound was heard and breath sounds were decreased bilaterally in basal zones. Rest systemic examination was within normal limits.

Laboratory investigations on 2 March 2022 were haemoglobin 8.5 g/dl, total counts 4500/mm^3, platelets 2.33 lac/mm^3, total serum proteins 6.0 gms/dl, total bilirubin 0.5mg/dl, ALP 276 U/L, AST 960 U/L, ALT 486 U/L, urea 14 mg/dl, creatinine 0.5 mg/dl, triglycerides 149 mg%, total cholesterol 162 mg%, HDL 53 mg% and LDL 79 mg% and on 4 April 2022 were haemoglobin 10.2 g/dl, total counts 5300/mm^3, platelets 1.59 lac/mm^3, total protein 6.3, total bilirubin 0.5 mg/dl, ALP 319 U/L, AST 126 U/L, ALT 198 U/L, urea 17 mg/dl, creatinine 0.4 mg/dl, triglycerides 100 mg%, total cholesterol 154 mg%, HDL 54 mg%, LDL 62 mg% and PT INR 0.93. Her height was 129.5 cm and weight 20 kg with body mass index of 11.92 kg/m^2. Her mid-arm circumference measured 13 cm (below 5th percentile for age, hence, severely undernourished), upper segment 58 cm and lower segment 71 cm with the upper to lower segment ratio 0.81. Her arm

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Received: 26 April 2022 **Accepted:** 16 May 2022 **Published:** 10 August 2022 **DOI:** 10.25259/VJIM_6_2022

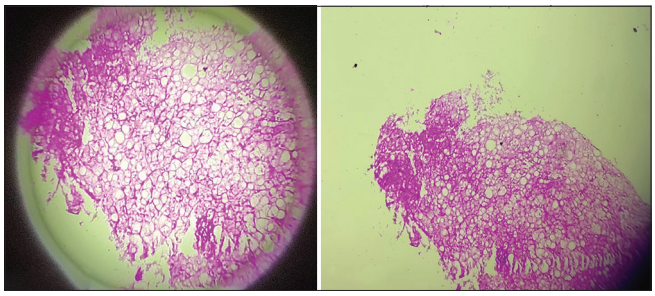

Figure 1: Biopsy slides showing PAS-positive material.

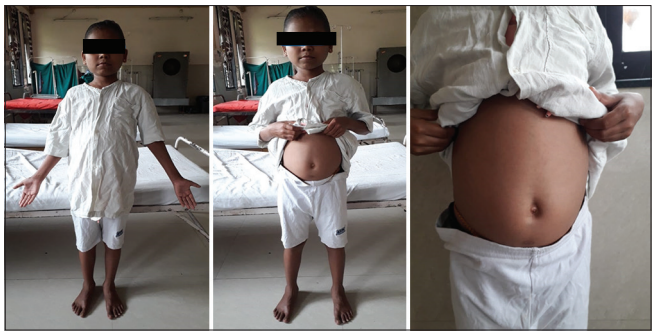

Figure 2: Clinical photograph of the patient.

span was 123 cm. Her waist-to-hip ratio was 1.16. Her bone age was 13.5 years normal for her age.

Her HbA1c was 9.6%. Serology for HIV, HBsAg, and anti-HCV was negative. Morning serum cortisol level was 16.08 ug/dl and serum uric acid level was 3.7 g/dl. Serum calcium was 9.5 mg/dl and phosphorus 3.9 mg/dl. Her thyroid profile was within normal limits and her anti-thyroid peroxidase level was 0.6 IU/ml. Serum amylase level was 142 IU/l. Serum ferritin level was 165.8 ug/l. The solubility test for sickling was positive. Hb electrophoresis revealed AS pattern and her parents were also AS pattern. ECG was suggestive of sinus tachycardia and chest X-ray PA view revealed cardiomegaly. Her USG (abdomen and pelvis) revealed massive hepatomegaly (20 cm) with liver parenchymal disease and mild splenomegaly (12.7 cm) with mild ascites. 2D echocardiographic examination findings showed preserved ejection fraction with evidence of mitral valve prolapse and a thin rim of pericardial effusion. CT head plain revealed normal brain parenchyma, ventricular system, pituitary fossa, and posterior fossa structures. Fundus examination was within normal limits. A liver biopsy panel [Figure 1] revealed enlarged, rarefied hepatocytes with 5–33% of steatosis and PAS-positive diastase-resistant material with mild portal fibrosis and with focal parenchymal extension; the possibility of a storage disorder. During the hospital stay, strict blood sugar monitoring was done and after she was vitally stable, she was discharged to Inj. Basal insulin (NPH) 10 units before breakfast and dinner and Inj. Aspart insulin 10 units before breakfast and 6 units each before lunch and dinner with precautionary measures for episodes of hypoglycaemia. Considering her history of Type 1 diabetes mellitus and hepatomegaly with normal ferritin levels, a provisional diagnosis of Mauriac syndrome was kept [Figure 2].

DISCUSSION

Mauriac syndrome occurs in males and females equally and is more common in adolescence.[2] It is associated with poor control of Type 1 diabetes mellitus in adolescents and may present as obesity, hepatomegaly, Cushingoid facies, and elevated transaminases. It is typically associated with growth failure and delayed pubertal maturation.[3] It is probably due to a combination of factors including inadequate glucose uptake and utilisation in the tissues, decreased insulin-like growth factor-1 and growth hormone levels, impaired bioactivity of these hormones, a circulating hormone inhibitor, resistant or defective hormone receptors, insulin deficiency, poor glycaemic control, concurrent autoimmune diseases, decreased caloric intake and/or eating disorders. The hepatomegaly is not seen in newly diagnosed patients who have been severely insulin-deficient because it appears that periods of supraphysiologic insulin levels are associated with the hepatomegaly.[4]

Autoimmune diseases that are commonly associated include Addison's disease, autoimmune gastritis, celiac disease, and hypothyroidism.[5]

This syndrome is rarely seen nowadays because of better treatment modalities. Before treatment with long-acting insulin, delays in growth and sexual maturity were common but generally modest.[6]

CONCLUSION

Mauriac Syndrome is a rare complication of type 1 diabetes mellitus. Hepatic glycogenosis is an under-recognized cause of abdominal pain and may be confused with non-alcoholic fatty liver disease, with important therapeutic implications and distinct prognosis. The mainstay of treatment for hepatic glycogenosis is strict glycaemic control with an excellent prognosis when achieved.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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How to cite this article: Bagchi N, Nagdeve R, Walke A, Chatterjee S. Mauriac Syndrome in a patient of Type 1 diabetes mellitus. Vidarbha J Intern Med 2022;32:152-4.