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## Mauriac Syndrome in a Young Child with Diabetes

Mauriac syndrome is a rare complication of type 1 diabetes mellitus, usually reported in adolescents with poor glycemic control. It is characterized by hepatomegaly due to glycogen deposition, growth failure and delayed puberty [1]. Often these children have cushingoid facies, elevated liver enzymes and dyslipidemia in the form of increased cholesterol and triglyceride level in blood [2].

A two-year-old girl, diagnosed with type 1 diabetes six months back, presented with severe diabetic ketoacidosis. She had been advised split-mix regime but had poor compliance with the treatment. On examination, she had tachypnea, growth failure (weight and height <-2 SD for age), cushingoid facies, distended abdomen, and hepatomegaly (span 10 cm).

Investigations showed elevated blood glucose of 506 mg/dL, severe ketoacidosis (blood pH 6.98) ketonuria, HbA1c 11.5g/dL, high serum triglycerides (207mg/dL) and serum total cholesterol levels (192 mg/dL), elevated hepatic transaminase (AST 183 U/L, ALT 196 U/L), and normal antinuclear antibodies, thyroid function tests and anti-tissue transglutaminase levels. Ultrasound of abdomen showed normal echotexture of liver and normal intrahepatic biliary redicles. Ketoacidosis was managed as per standard protocol, and she was discharged on subcutaneous isophane and regular insulin. There was normalization of hepatomegaly and elevated liver enzymes after two months and liver biopsy was deferred. Mauriac syndrome was considered as the most probable explanation for liver dysfunction in this child.

There are anecdotal case reports of mutation in *PHKG2*, which is the catalytic subunit of the enzyme

glycogen phosphorylase kinase. Hepatomegaly is a cardinal feature of Mauriac syndrome present in the majority, which occurs due to hepatic glycogen deposition due to the facilitated glucose diffusion across the hepatocytes [3]. The possible mechanisms for growth failure in Mauriac syndrome are inadequate tissue glucose availability, reduced circulating IGF-I level, and a relative growth hormone-resistant state [4]. The cushingoid features probably occur due to secondary hyperadrenocorticism. Short acting insulin regimens and brittle glycemic control are predisposing factors in these children for this rare complication. Most children with Mauriac syndrome are reported during adolescence. To the best of our knowledge, the youngest case of this syndrome reported earlier was aged three years [5]. Clinicians should suspect it in any child with uncontrolled diabetes and hepatic dysfunction. Identification of this syndrome indicates poor disease control, which can guide further management.

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