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Berardinelli–Seip Congenital Lipodystrophy: A Rare Case Report

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Abstract

Berardinelli–Seip Congenital Lipodystrophy (BSCL) is a rare autosomal recessive disorder characterized by near total absence of adipose tissue, severe insulin resistance, dyslipidemia, and hepatic involvement. We report a prepubertal child presenting with thin build, generalized loss of subcutaneous fat, prominent bones, distended abdomen, hepatomegaly, and delayed secondary sexual characteristics. Anthropometry revealed severe wasting with BMI 11.96 kg/m² and MUAC 13 cm. Laboratory evaluation revealed markedly elevated fasting and postprandial blood glucose (19.2 and 20.8 mmol/L), HbA1c 11.8%, glycosuria, hypertriglyceridemia (388 mg/dl), and mild transaminitis, consistent with poorly controlled diabetes and hepatic steatosis. C-peptide level was low normal with fasting insulin level was low normal for the degree of hyperglycemia, indicating insulin resistance and partial β-cell dysfunction. Other routine parameters, including renal function, thyroid profile, and electrolytes were normal. Echocardiogram revealed atrial septal defect(ASD) secundum. Based on the constellation of generalized lipodystrophy, hepatomegaly, insulin-resistant diabetes, and dyslipidemia, a diagnosis of Berardinelli-Seip Congenital Lipodystrophy was made. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S61]*

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