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Successful Anesthesia Management in a patient with Berardinelli-Seip Congenital Lipodystrophy (BSCL) scheduled for Open Reduction and Internal Fixation of Femur

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We are presenting our case of the patient with Berardinelli-seip congenital lipodystrophy(BSCL), characterized by hyperinsulinemia caused by insulin resistance combined with lipodystrophy and acromegaloid features(8). This patient had portal vein thrombosis, prognathosis, hypertrophic obstructive cardiomyopathy, LVOT obstruction, hyperthyroidism, hypertension and diabetes melitus. He underwent open reduction and internal fixation(ORIF) with titanium elastic plates under spinal anesthesia.

Introduction: Berardinelli-Seip Congenital Lipodystrophy (BSCL), or Congenital Generalized Lipodystrophy, is a rare genetic disorder characterized by a severe deficiency in body fat, first identified in Brazil in 1954. Affecting approximately 1 in 12 million people worldwide, BSCL is inherited in an autosomal recessive manner, often linked to parental consanguinity. The lack of adipose tissue leads to significant metabolic disruptions, including high serum triglycerides, insulin resistance, and early-onset type 2 diabetes, with severe steatosis affecting organs like the liver and muscles. Major symptoms include lipoatrophy of the trunk, limbs, and face, hepatomegaly, and acromegaloid features, while additional issues may include hypertrophic cardiomyopathy and intellectual impairment. Treatment options are limited, though recombinant leptin has shown potential. Limited case reports on anesthesia for BSCL patients highlight the need for careful management due to risks like delayed emergence from anesthesia.

Case Presentation: A 12-year-old boy with a history of developmental delays, hypertension, diabetes mellitus, and hyperthyroidism presented for ORIF of a fractured left femur. His conditions were managed with insulin (Lantus and Novorapid), metformin, propranolol, and thyroxine. He also had a history of portal vein thrombosis, though documentation was missing. Physical exam findings included prognathic jaw, macroglossia, hypertrophy of palatine tonsils, acanthosis nigricans, frontal bossing, a midsystolic murmur, and a distended abdomen. Echocardiography revealed hypertrophic obstructive cardiomyopathy, severe LVH, and mitral regurgitation with a 67% ejection fraction. Lab results showed thrombocytopenia (platelets at 88,000), elevated bilirubin (2.75 mg/dl), and an INR of 1.66. Intraoperatively, he received preoperative FFP, subarachnoid block, and propofol infusion. Surgery lasted 90 minutes with minimal blood loss. He recovered well post-operatively, was fully alert, and was discharged with stable vitals after 5 hours.

Biography

Dr. Sami Ur Rehman, affiliated with Doctors Hospital and Medical Centre in Lahore, Pakistan, is a respected medical professional known for his expertise and dedication to advancing healthcare. With extensive experience in [specific specialties, e.g., Orthopedics, Cardiology], Dr. Rehman combines clinical excellence with a compassionate approach to patient care. His contributions extend beyond direct patient treatment, as he actively engages in medical research and advancements, solidifying his reputation as a leader in his field..

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