A Pediatric Case with Congenital Generalized Lipodystrophy

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Congenital generalized lipodystrophy, also known as Berardinelli-Seip congenital lipodystrophy (BSCL), is a rare autosomal recessive disease. This condition is characterized by the absence of adipose tissue in the body. Patients have severe disturbances in carbohydrate and lipid metabolism due to the absence of adipose tissue as well as cardiomyopathy, increased growth velocity, bone disorders, cirrhosis, hyperandrogenism and mild mental retardation in some cases. This case report presents the genotype, phenotype and follow-up data of a patient who had been diagnosed with BSCL at the age of 5.5 years.

His birth weight was 1500 g and his parents were first-degree cousins. He had progeroid facial features at birth. Elevated aspartate aminotransferase (AST) and alanine aminotransferase (ALT) levels were found in the first six months after birth. Being monitored with preliminary diagnosis of lipodystrophy in other centers since the age of 1 year, the case was referred to our hospital after diagnosis of cirrhosis during the follow-up. Physical examination of the case with mild mental retardation revealed weight of 33.7 kg [standard deviation score (SDS): +4.5], height of 133 cm (SDS: +3.4 SDS), generalized absence of adipose tissue, acanthosis nigricans, hypertrichosis and hepatosplenomegaly. Pre-pubertal patient's penis size was found to be 9 cm (>90th percentile). The laboratory assessment revealed the following: AST: 85 U/L, ALT: 214 U/L, alkaline phosphatase: 365 U/L, gamma-glutamyltransferase: 85 U/L, total cholesterol: 261 mg/dL, triglycerid: 185 mg/dL, low-density lipoprotein: 177 mg/dL, high-density lipoprotein: 47 mg/dL. Serum leptin was found to be very low-0.1 ng/mL (N: 0.5-3.2). Fasting blood glucose was 87 mg/dL, fasting insulin was 44.8 mIU/mL, homeostasis model assessment of insulin resistance was 9.5 and bone age was found to be 10 years. His echocardiography was normal. A homozygous IVS4+1 G>A mutation in the *BSCL2* gene was detected in the patient; the same mutation was heterozygous in his parents. The patient was followed up with treatment including omega-3, ursodeoxycholic acid, medium-chain triglyceride oil; however, metformin was added to treatment later due to persistence of insulin resistance. Leptin analog (metreleptin) treatment was planned for the patient with stable cirrhosis whose insulin resistance was still persistent despite the treatment.

Key words: Congenital generalized lipodystrophy, Berardinelli-Seip congenital lipodystrophy, *BSCL2* gene