and 2% children respectively. Amongst patients with GHD, 60.7% had wild type (GHRH/GHR), 19.2% were heterozygous (GHRH/GHRd) and 20.1% were homozygous (GHRHd), whereas for idiopathic short stature they were 67.5%, 14.5% and 18% respectively.

**Conclusions:** The diagnosis could be attained in 85% of cases. Growth hormone deficiency and celiac disease contribute significantly even though majority is normal variants. Also, genotyping done would help in prediction of response to recombinant GH therapy in a resource constraint resulting in appropriation of finances which could be utilized for a higher priority area.

**Keywords:** Short stature; growth hormone receptor

---

**AB143. Berardinelli-Seip congenital lipodystrophy and its diagnostic implications**

Premala Muthukumarasamy, Meow Keong Thong

Genetics and Metabolism Unit, Department of Paediatrics, University Malaya Medical Centre, Malaysia

**Abstract:** Berardinelli-Seip congenital lipodystrophy (BSCLD) (OMIM#269700) is a rare autosomal recessive disorder characterized by marked paucity of adipose tissue, a muscular habitus, insulin resistance, hypertriglyceridemia and hepatic steatosis. It is an anabolic syndrome due to the deficiency of leptin, a lipid modulator, leading to the inappropriate mobilization of fat. Four types of BSCLD have been described based on the site of genetic mutation. Type 2 accounts for an earlier onset, more severe complications and intellectual disability. This is due to the lower leptin levels and the loss of functional seipin in type 2 BSCLD. We report the evolving phenotype of type 2 BSCLD in a boy clinically diagnosed at age 5 months and confirmed by molecular genetic study which revealed a pathogenic mutation in the BSCL2 gene: c.782dupG, p.Lle262Hisfs*12 homozygous.

He is the firstborn of non-consanguineous parents with no significant family history. He has a muscular habitus with a distinct facies, hypertrichosis, absence of subcutaneous fat and hepatomegaly. Investigations revealed hypertriglyceridemia and insulin resistance. A conventional approach to management with dietary fat restriction successfully controlled his biochemical parameters, thus preventing catastrophic complications such as systemic arterial hypertension, heart failure and pancreatitis. Genetic counseling was conferred and prenatal testing recommended for future pregnancies. On follow-up, he had a muscular habitus with hepatomegaly and global developmental delay. Despite early correction of his biochemical parameters, an abdominal ultrasound revealed early onset fatty liver disease and an echocardiogram showed a thickened interventricular septum suggestive of early hypertrophic obstructive cardiomyopathy (HOCM). A fasting serum leptin done was low at 0.8 ng/mL (normal range, 2.0-5.6 ng/mL) confirming leptin deficiency. A leptin analog is now available as an adjunct to mollify the metabolic complications of BSCLD. However, it is yet to be available in developing nations and whether it is able to assuage progressive hepatic cholestasis and HOCM remains a field to venture. This report highlights the importance of early recognition and confirmation by molecular diagnosis to direct appropriate surveillance of complications. This also enables early genetic counseling and prenatal testing for future pregnancies. We describe the evolving phenotype, pertinent investigations and the conventional approach to the management of BSCLD.

**Keywords:** Autosomal recessive; generalized lipodystrophy; hypertriglyceridemia; insulin resistance; leptin

---
