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Berardinelli-Seip syndrome in a Chinese boy with Seipin gene mutation: a case study and literature review of genotype-phenotype

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Objective

Congenital generalized lipodystrophy (CGL), or Berardinelli-Seip syndrome, is a rare and heterogeneous disease of autosomal recessive inheritance characterized by the generalized absence of adipose tissue at birth and severe adverse metabolic consequences. The identified causative genes for CGL include 1-acylglycerol-3-phosphate O-acyltransferase 2 (*AGPAT2*), Berardinelli-Seip congenital lipodystrophy 2 (*BSCL2* or Seipin), Caveolin-1 (*CAV1*) and polymerase I and transcript release factor (*PTRF*). Although more than 60 cases of CGL with different gene mutations have been found in Asian patients, only 7 patients were Chinese. Data are also limited regarding genotype-phenotype analysis in Asian CGL patients. Therefore, we aimed to analyze variations of two identified major causative genes, Seipin and *AGPAT2*, involved in CGL etiology in a mainland Chinese affected family and explore the genotype-phenotype of Berardinelli-Seip syndrome in Asian populations.

Methods

We report a detailed clinical and genetic analysis of a Chinese boy with CGL who was followed from infancy through preschool. Sequences of the entire coding region of Seipin and *AGPAT2* were examined. Phenotypes in various Asian subtypes were compared and the related literature about Berardinelli-Seip syndrome was reviewed.

Results

We identified a homozygous frameshift mutation (c.974-975insG) in the Seipin gene in the CGL-affected boy. His parents were heterozygous for the same mutation. No variation was found in the AGPAT2 gene.

Conclusion

In Asian populations, Seipin and PTRF are the main genes identified to date as being responsible for CGL and Seipin is a major causative gene. Genetic heterogeneity is accompanied by phenotypic heterogeneity.

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