

Clinical and mutational features of three Chinese children with congenital generalized lipodystrophy

ABSTRACT

Objective: To investigate the clinical and molecular features of congenital generalized lipodystrophy (CGL) in three Chinese patients with various typical manifestations. **Methods:** Data on clinical symptoms, results of laboratory analyses, and previous treatments in three Chinese patients were collected by a retrospective review of medical records. All coding regions and adjacent exon–intron junction regions of AGPAT2 and BSCL2 genes were amplified by polymerase chain reaction and sequenced. **Results:** Generalized lipodystrophy, acanthosis nigricans, muscular hypertrophy, severe hypertriglyceridemia, and hepatomegaly were features in all three patients. Patient 1 developed diabetes mellitus at the early age of 2 months and he was the youngest CGL patient reported with overt diabetes. Patient 2 was found to have cardiomyopathy when she was aged 6 months. All of the patients were found to have mutations in the BSCL2 gene, but none of these was a novel mutation. We did not find any AGPAT2 mutation in our patients. **Conclusion:** All of our patients exhibited characteristic features of CGL due to mutations in the BSCL2 gene.

Keyword: Lipodystrophy; Hypertriglyceridemia; Diabetes; Cardiomyopathy