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An unexpected presentation of very severe hypertriglyceridemia in a boy with Coffin-Lowry syndrome: a case report

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Abstract

Background Coffin-Lowry syndrome (CLS) is a rare X-linked condition with intellectual disability, growth retardation, characteristic facies and skeletal anomalies. To date, hypertriglyceridemia has not been reported in literature to be associated with CLS.

Case Presentation Herein, we report a case of very severe hypertriglyceridemia 32 mmol/L (2834 mg/dL) detected incidentally at three months old in an otherwise well boy born late preterm with intrauterine growth restriction, when he presented with lipaemic plasma. He was later diagnosed with CLS. No pathogenic mutations were found for hypertriglyceridemia, and no secondary causes could explain his very severe hypertriglyceridemia.

Conclusions The very severe hypertriglyceridemia in this case may appear to be a serious presentation of an unrecognised clinical feature of CLS, further expanding its phenotype.

Keywords Hypertriglyceridemia, Coffin-Lowry syndrome, Intrauterine growth restriction, Lipaemic plasma

Background

CLS is a rare but increasingly recognised X-linked semi-dominant syndrome characterised by psychomotor and growth retardation, facial dysmorphism, digit abnormalities and progressive skeletal changes resulting from mutations in the RPS6KA3 gene [1]. They may also have neurological, ophthalmological, hearing, dental, cardiovascular and respiratory problems [1, 2]. Despite its multisystemic manifestations, hypertriglyceridemia (HTG) has not been reported in CLS. HTG is defined as triglyceride (TG) concentrations > 150 mg/dL [3]. It is incredibly uncommon in infancy and, if present, should prompt consideration of familial lipoprotein lipase deficiency [3]. Here, we describe a case of very severe HTG in infancy with no identifiable primary nor secondary cause. The child was subsequently diagnosed with CLS.

Case presentation

Our patient's mother was 37 years old at delivery with uncomplicated antenatal history. Our patient was born vigorous via elective lower segment caesarean section at 35 weeks for intrauterine growth restriction (IUGR) with a low birth weight of 2.1 kg. He was admitted for observation in view of prematurity and did not receive any medications. He was given phototherapy for neonatal jaundice and was discharged well on day five of life.

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