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Two Novel Heterozygous *MCCC1* Mutations in a Neonate with Asymptomatic 3-methylcrotonyl-coenzyme A Carboxylase Deficiency

Isolated 3-methylcrotonyl-coenzyme A carboxylase deficiency is a rare metabolic disorder inherited as an autosomal recessive trait [1]. We report features of patient with asymptomatic 3-methylcrotonyl-coenzyme A carboxylase deficiency and two novel heterozygous *MCCC1* mutations.

The proband, a girl, was born in the 41th week of gestation with a weight of 2.85 kg to non-consanguineous parents. On the fourth day of life, tandem mass spectrometry was performed, revealing that the level of 3-hydroxyisovaleryl-carnitine (C5-OH) was high (2.74 μM; reference range <0.6 μM). C5-OH levels were repeated in dried blood spot, and it was again high (3.23 ìM). Due to persistently elevated level of C5-OH, blood samples from the proband, her parents and her elder brother were tested for genetic- and mutation-analysis, after informed consent. Genetic analysis of the proband showed two heterozygous novel missense mutations (*MCCC1* NW_020166.3: c.1570G>C (p.D524H) and *MCCC1* NW_020166.3: c.601C>T (p.P201S)) in the *MCCC1* gene. The father was heterozygous for c.601C>T (p.P201S) and the mother was heterozygous for c.1570G>C (p.D524H). The elder brother was heterozygous for c.601C>T (p.P201S). At 11 months of age, the proband was still asymptomatic, and with normal growth and development.

In general, most newborns with isolated 3-methylcrotonyl-coenzyme A carboxylase deficiency detected on screening appear clinically normal and healthy. Features such as vomiting, ketosis, poor oral intake, irritability, lethargy and hypotonia are reported in up to 15% of patients; majority (92.5%) of infants show completely appropriate age-matched development [2].

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