Short Chain Acyl-CoA Dehydrogenase Deficiency

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Short chain Acyl-CoA dehydrogenase deficiency (SCADD) is a rare fatty acid oxidative disorder (FAOD) inherited as an autosomal recessive pattern. Patients could present with a variety of symptoms, such as hypoglycemia, metabolic acidosis, cyclical vomiting, myopathy and developmental delay or asymptomatic individual. Since the introduction neonatal screening programs, it has been found that majority of the patients with SCADD are asymptomatic at the time of diagnosis.

We present this atypical, yet symptomatic case of a patient with confirmed SCADD who presented at the age of 11 months with recurrent chest infections, vomiting and cyanotic episodes. The diagnosis was confirmed with increased ethylmalonic acid (EMA) in urine and molecular genetic analysis, which identified a pathogenic variant c.529T>C (p. Trp177Arg) in axon 5 of ACADS gene (OMIM 606885; chromosome 12q24.31). The patient was managed with dietary modifications and referral to a metabolic specialist. Parents were referred for genetic counseling regarding future conception.

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