Acyl-CoA Dehydrogenase, Short-Chain, Deficiency of; ACADSD

Alternative Name
ACADSD
ACADS Deficiency
Lipid-Storage Myopathy Secondary to Short-Chain Acyl-CoA Dehydrogenase Deficiency
SCADH Deficiency
SCAD Deficiency

Record Category
Disease phenotype

WHO-ICD
Endocrine, nutritional and metabolic diseases
Metabolic disorders

Incidence per 100,000 Live Births
NA

OMIM Number
201470

Mode of Inheritance
Autosomal recessive

Gene Map Locus
12q24.31

Description
Short-chain acyl CoA deficiency (SCAD) is an autosomal recessive disorder of fatty acid beta-oxidation. Affected individuals can have variable presentations ranging from acidosis and neurological impairment to merely myopathy. Some patients may have vomiting, hypoglycemia and microcephaly. The symptoms may be triggered by viral infections and may present in adulthood. Management includes avoidance of fasting, following a special diet, which is low in fat and high in carbohydrates. Medications include L-carnitine and Riboflavin.

Molecular Genetics
SCAD deficiency is caused by mutations in the ACADS gene. This gene is located on the long arm of chromosome 12. ACADS codes for short-chain acyl CoA dehydrogenase, which metabolize short chain fatty acids. Mutations in this gene impair the enzyme function and thus, fatty acids are not converted into energy. The ACADS gene is 13 kb long and it contains 10 exons. Several mutations in ACADS have been described.

Epidemiology in the Arab World
Saudi Arabia
Moammar et al., (2010) reviewed all patients diagnosed with inborn errors of metabolism (IEM) from 1983 to 2008 at Saudi Aramco medical facilities in the Eastern province of Saudi Arabia. During the study period, 165530 Saudi infants were born, of whom a total of 248 newborns were diagnosed with 55 IEM. Affected patients were evaluated based on clinical manifestations or family history of similar illness and/or unexplained neonatal deaths. Almost all patients were born to consanguineous parents. Fatty acid oxidation disorders were diagnosed in 18/248 patients (7%) who presented with enzymatic deficiency. Among them, four cases from two families were found to have SCAD deficiency. The estimated incidence of this condition was 2 in 100,000 live births.
The authors concluded that data obtained from this study underestimate the true figures of various IEM in the region. Therefore, there is an urgent need for centralized newborn screening program that utilizes tandem mass spectrometry, and offers genetic counseling for these families.

References
Moammar H, Cheriyan G, Mathew R, Al-Sannaa N. Incidence and patterns of inborn errors of metabolism in the Eastern Province of Saudi Arabia, 1983-2008. Ann Saudi Med. 2010 Jul-Aug;30(4):271-7. PMID:20622343

Related CTGA Records
External Links

Contributor:
Balobaid, Ameera: 14.09.2016