State of Connecticut
Genetics Newborn Screening Program
Health Professional Fact Sheet

Short-Chain ACYL-CoA Dehydrogenase Deficiency (SCADD)

Introduction

Short chain acyl-CoA Dehydrogenase Deficiency (SCADD) is a rare autosomal recessive disorder of fatty acid oxidation. Only 20 SCAD deficient patients have been described. The majority became symptomatic in the first week-3 months of life, 4 before 24 months and two presented as adults. About 50% of patients have hypotonia and developmental delay. Variable symptoms are present in patients presenting early, including seizures, respiratory distress, acidosis, poor feeding, vomiting or failure to thrive, and one death. Only one patient had hypoglycemia. Acute symptoms in most patients did not recur. Both adults presented with proximal muscle weakness associated with periods of pain, nausea/vomiting and shortness of breath. Ethymalonic acid and methylsuccinate are elevated in the urine.

Clinical Features

SCAD deficiency has been identified in only a few patients with highly variable clinical and laboratory findings. For more information on case reports see The Metabolic and Molecular Bases of Inherited Disease, Charles R. Scriver, Arthur L. Beaudet, William S. Sly, David Valle, and Barton Childs, February 2001, McGraw-Hill Companies, 8TH Edition or the "Online Mendelian Inheritance in Man website http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM (#606885 and #210470).

Diagnosis

Newborn screening—Tandem mass spectrometry C4
Confirmation—a second sample may be requested or follow up testing will be done at the Metabolic Treatment Center at Yale or UCONN Genetics.

Treatment

Common fatty acid oxidation therapies including low fat diet, carnitine and riboflavin supplements have not improved hypotonia or developmental delay. One patient with cyclic vomiting improved on frequent feeding.

- Avoid fasting.
- Feed at regular intervals during the day and limit overnight fasting
- Should not go without food intake longer than 4 hours for the first 4 months of life; 6 hours for ages 4-8 months; and no longer than 8 hours thereafter
- Restriction of dietary fat is controversial but it is reasonable during intercurrent infections. The Metabolic Treatment Center will set a patient's diet prescription that determines the optimum percentage of fat, carbohydrate, and protein.
- If the child is vomiting or refuses to eat, (s)he needs to be taken to an emergency room for IV administration of 10% dextrose.
- Infants and children with SCADD should have regularly scheduled visits at the Metabolic Treatment Center.
- The parents usually have an emergency protocol with them. This protocol, provided by the Metabolic Treatment Center, contains basic information about the disorder, necessary diagnostic investigations and guidelines for treatment.

- It is also important for the primary care provider and the Metabolic Treatment Center to develop an on-going collaborative relationship in caring for these patients.

Situations that risk metabolic decompensation

Metabolic decompensation can be triggered by the catabolic processes that occur in the course of an infection, after an immunization or with a prolonged period of fasting.

Illness

- Any illness can potentially lead to metabolic decompensation.
- Prevention and/or early intervention is of particular importance.
- Care should be coordinated by the Metabolic Treatment Center.

Immunization

- Immunizations must be kept current.

Surgical/surgical procedures

- Discuss any plans for surgical and dental procedures with the Metabolic Treatment Center.

Growth and development

- SCADD can cause failure to thrive.
- It is crucial to closely monitor all growth parameters on a regular basis.
- In cases with neurological deficits, the child should be referred to an early intervention program and developmental progress closely monitored by both the metabolic team and the primary care provider.

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