Condition: Medium chain-acyl coA dehydrogenase deficiency (MCAD)

Inheritance:

Autosomal recessive.

Genetic etiology:

Mutation in gene *ACADM*, which encodes medium chain acyl CoA dehydrogenase, involved in beta oxidation of medium chain fatty acids. Other fatty acid oxidation defects affect metabolism of longer chain fatty acids.

Frequency:

Approximately 1/5000 – 1/17,000.

Clinical features:

Time of presentation is usually in infancy, but can be later in life. During times of fasting or stress due to illness a crisis may occur with hypoglycemia, metabolic acidosis, elevated liver enzymes, hyperammonemia, hyperuricemia, and lack of ketones in the urine. This can lead to coma and death.

Management:

Management focused on avoidance of hypoglycemia and restriction of medium chain triglyceride intake.

Genetic counseling:

Based on autosomal recessive inheritance, with genetic testing available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.