2, 4-Dienoyl-CoA Reductase Deficiency

Background

One patient has been reported with 2, 4-Dienoyl-CoA Reductase Deficiency. This enzyme is necessary for the degradation of unsaturated fatty acids having even numbered double bonds.

Clinical

The patient was born with a small body habitus, a short trunk, arms and fingers, and microcephaly. She was readmitted to the hospital on day two of life with symptoms of sepsis, hypotonia, decreased feeding and intermittent vomiting. A low carnitine level was found in her plasma. She responded poorly to treatment in the hospital, and later developed respiratory acidosis and died at four months of age.

Testing

Newborn screening using tandem mass spectrometry may reveal C10:2 acylcarnitine as a pathognomonic finding. Urine organic acid analysis was normal in the one patient and plasma amino acids showed elevated lysine. The enzyme deficiency can be demon-strated in liver and muscle tissue.

Treatment

Suggested treatment for 2, 4-Dienoyl-CoA Reductase Deficiency involves feeding the patient a formula containing fat derived from medium-chain triglycerides (MCT), administering pharmacologic doses of carnitine, and avoiding fasting.

Because the diagnosis and therapy of metabolic disorders like this is complex, the pediatrician is advised to manage the patient in close collaboration with a consulting pediatric metabolic disease specialist. It is recommended that parents travel with a letter of treatment guidelines from the patient's physician.

Inheritance

This disorder most often follows an autosomal recessive inheritance pattern. With recessive disorders, affected patients usually have two copies of a disease gene (or mutation) in order to show symptoms. People with only one copy of the disease gene (called carriers) generally do not show signs or symptoms of the condition but can pass the disease gene to their children. When both parents are carriers of the disease gene for a particular disorder, there is a 25% chance with each pregnancy that they will have a child affected with the disorder.

References

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Roe, C.R. and Ding, J. Mitochondrial Fatty Acid Disorders. In, The Metabolic and Molecular Basis of Inherited Disease. 8th Edition, 2001. Scriver, Beaudet, et al. McGraw-Hill. Chapter 101, pg. 2297 - 2326.