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My46 Trait Profile

Very long-chain acyl-CoA dehydrogenase deficiency

Very long-chain acyl-CoA dehydrogenase deficiency is a metabolic disorder caused by an inability to break down certain fats. It is caused by mutations in the ACADVL gene that makes the very long-chain acyl-coenzyme A dehydrogenase enzyme.

Characteristics of Very long-chain acyl-CoA dehydrogenase deficiency

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency is a rare metabolic condition in which the body cannot breakdown certain fats (i.e. very long chain fatty acids). The condition is characterized by three different forms that occur at different ages.

The early-onset (infancy) form is the most severe and includes failure of the heart and multiple other organs, low blood sugar (hypoglycemia), and weak muscles. Some symptoms of severe illness may be better recognized as extreme sleepiness (lethargy), behavior changes, irritable mood, and poor appetite. The form that appears in childhood includes hypoglycemia and enlargement of the liver. The late-onset (adult) form includes muscle cramps and/or pain.

In general, all individuals with VLCAD deficiency have trouble converting certain fats to energy. This is most commonly an issue during times without food, or fasting. Intelligence is usually normal in individuals with VLCAD deficiency who are treated appropriately. Without treatment, however, the effects can be quite severe and can result in death if the features are seen at a young age.

Diagnosis/Testing

Patients with VLCAD deficiency have a change or mutation in both copies of a gene called ACADVL. This gene makes an enzyme called VLCAD that breaks down very long chain fats. VLCAD is responsible for converting fat into energy for the body to use. Mutations in the ACADVL gene are thought to interfere with fat break down and the ability of the body to gain the proper nutrition from the fat that is consumed and stored in the body.

Management/Surveillance

Management of VLCAD deficiency often includes eating and drinking at regular intervals throughout the day and maintaining a diet low in fat and high in carbohydrates. The medical team should be contacted as soon as any illness occurs. Most individuals with VLCAD also need to avoid long periods of exercise.

Mode of inheritance

VLCAD deficiency is inherited in an autosomal recessive pattern. This means that an individual has to inherit two ACADVL mutations (i.e., one from each parent) to be affected with VLCAD deficiency. If both parents are carriers of a ACADVL mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with VLCAD deficiency. Babies born in the United States are screened for VLCAD deficiency by newborn screening.

Risk to family members

Parents of a child with VLCAD deficiency are carriers of VLCAD deficiency. If a sibling of a child with VLCAD deficiency is unaffected, he/she has a 2 in 3 (or 66%) chance of being a carrier of VLCAD deficiency.

Special considerations

Individuals with VLCAD deficiency can live long and healthy lives if treated appropriately. The severity of the condition generally depends on the type(s) of mutations present in the ACADVL genes. If no enzyme is made, the individual will be more severely affected. If at least a small amount of enzyme is made, the individual will be less severely affected.

Resources

Genetics Home Reference: VLCAD deficiency
http://ghr.nlm.nih.gov/condition/very-long-chain-acyl-coa-dehydrogenase-deficiency
Office of Rare Diseases Research: VLCAD deficiency
http://rarediseases.info.nih.gov/GARD/Condition/5508/VLCAD_deficiency.aspx
Medical Home Portal: VLCADD
http://www.medicalhomeportal.org/diagnoses-and-conditions/vlcadd/description

References

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