Isovaleric Acidemia

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Isovaleric acidemia is a rare disorder in which the body is unable to properly break down a particular protein building block (amino acid). The condition is classified as an organic acid disorder, which is a condition that leads to an abnormal buildup of particular acids known as organic acids. Abnormal levels of organic acids in the blood (organic acidemia), urine (organic aciduria), and tissues can be toxic and can cause serious health problems.

genetic conditions

1. Introduction

Normally, the body breaks down proteins from food into smaller parts called amino acids. Amino acids can be further processed to provide energy for growth and development. People with isovaleric acidemia have inadequate levels of an enzyme that helps break down a particular amino acid called leucine.

Health problems related to isovaleric acidemia range from very mild to life-threatening. In severe cases, the features of isovaleric acidemia become apparent within a few days after birth. The initial symptoms include poor feeding, vomiting, seizures, and lack of energy (lethargy). These symptoms sometimes progress to more serious medical problems, including seizures, coma, and possibly death. A characteristic sign of isovaleric acidemia is a distinctive odor of sweaty feet during acute illness. This odor is caused by the buildup of a compound called isovaleric acid in affected individuals.

In other cases, the signs and symptoms of isovaleric acidemia appear during childhood and may come and go over time. Children with this condition may fail to gain weight and grow at the expected rate (failure to thrive) and often have delayed development. In these children, episodes of more serious health problems can be triggered by prolonged periods without food (fasting), infections, or eating an increased amount of protein-rich foods.

Some people with gene mutations that cause isovaleric acidemia are asymptomatic, which means they never experience any signs or symptoms of the condition.

2. Frequency

Isovaleric acidemia is estimated to affect at least 1 in 250,000 people in the United States.

3. Causes

Mutations in the *IVD* gene cause isovaleric acidemia. The *IVD* gene provides instructions for making an enzyme that plays an essential role in breaking down proteins from the diet. Specifically, this enzyme helps process the amino acid leucine, which is part of many proteins. If a mutation in the *IVD* gene reduces or eliminates the activity of this enzyme, the body is unable to break down leucine properly. As a result, an organic acid called isovaleric acid and related compounds build up to harmful levels in the body. This buildup damages the brain and nervous system, causing serious health problems.

3.1. The gene associated with Isovaleric acidemia

- IVD

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- Isovaleric acid-CoA dehydrogenase deficiency
- Isovaleryl-CoA dehydrogenase deficiency
- IVA
- IVD deficiency

References

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