



# Facts about Isovaleric Acidemia



## What Your Test Results Mean

**Carriers show no symptoms of isovaleric academia and are not at risk to develop symptoms of the disorder.** Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

## ● Isovaleric Acidemia Explained

Isovaleric acidemia is an inherited metabolic condition characterized by the body's inability to break down the amino acid leucine due to inadequate levels of the enzyme isovaleryl-CoA dehydrogenase. Due to the lack of this enzyme, proteins cannot be processed properly and organic acids, including isovaleric acid, build up in the body, which can cause a variety of health problems.

Initial symptoms may be present shortly after birth and can include vomiting, poor feeding, and lethargy. The symptoms have the ability to progress to further medical concerns such as seizures, coma, and ultimately potentially causing death. The build up of isovaleric acid causes a distinct characteristic of the disorder—the odor of sweaty feet during acute illness. Other medical problems that characterize this disorder are the failure to grow and gain weight at a normal rate (failure to thrive), as well as exhibiting delayed development. Treatment consists of restricting protein in the diet, more commonly protein that contains the amino acid leucine. Avoiding protein-rich foods, infection, and fasting (going for long periods without eating) can help prevent symptoms of the disease.

## ● How the Genetics Work

Isovaleric acidemia is an autosomal recessive disorder caused by pathogenic variants in the *IVD* gene. Carriers of isovaleric acidemia have a single variant in one copy of the *IVD* gene while individuals with the disorder have variants in both copies of the *IVD* gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

**Questions?** Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.