Genetics Uncoded:
FACTS ABOUT ISOVALERIC ACIDEMIA

AKA | Isovaleric acid-CoA dehydrogenase deficiency, Isovaleryl-CoA dehydrogenase deficiency, IVA, IVD Deficiency

What Your Test Results Mean
Testing results indicate that you are a carrier of isovaleric acidemia. Although carriers do not typically exhibit symptoms of the disease, all carriers are at an increased risk for having a child born with isovaleric acidemia. Testing your partner and seeing a genetic counselor for further information on your risk assessment is recommended.

Isovaleric Acidemia Explained
Isovaleric academia is an inherited metabolic condition characterized by the inability of the body to break down the amino acid leucine due to having 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 is 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 academia is an autosomal recessive disorder caused by mutations in the IVD. Carriers of isovaleric academia have a mutation in one copy of the IVD gene while individuals with the disorder have mutations in both copies their gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps
Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?
Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.