



Genetics Uncoded:

# Facts about

## Very Long-Chain Acyl-CoA Dehydrogenase Deficiency



### What Your Test Results Mean

**Carriers typically show no symptoms of very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency; however, carriers are at an increased risk of having a child with VLCAD deficiency.** Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

### ● Very Long-Chain Acyl-CoA Dehydrogenase Deficiency Explained

VLCAD deficiency is an inherited disorder that prevents the body from converting certain fats to energy, particularly during fasting periods. Variants in the *ACADVL* gene result in a build-up of very long-chain fatty acids. Symptoms of VLCAD deficiency usually appear during infancy or early childhood and can include low blood sugar, lack of energy, and muscle weakness. Serious complications include liver abnormalities and life-threatening heart problems. The later the symptoms first develop, the milder the disorder. Affected individuals exhibit the most problems during periods of fasting, illness, and exercise. VLCAD deficiency is usually not fatal as long as it is diagnosed early. Treatment is usually required throughout life for those affected with VLCAD deficiency. Affected individuals are encouraged avoid going long periods without food in order to prevent a metabolic crisis. Management is overseen by a group of metabolic specialists.

### ● How the Genetics Work

VLCAD deficiency is an autosomal recessive metabolic disorder caused by variants in the *ACADVL* gene. In general, individuals have two copies of the *ACADVL* gene. Carriers of VLCAD deficiency have a variant in one copy of the *ACADVL* gene while individuals with VLCAD deficiency have variants in both copies of *ACADVL*, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

### Questions?

Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.