Genetics Uncoded:

NxGen MDx

Facts about Type I Tyrosinemia

What Your Test Results Mean

Carriers typically do not show symptoms of type I tyrosinemia; however, carriers are at an increased risk of having a child with type I tyrosinemia. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Type I Tyrosinemia Explained

Type I tyrosinemia is an inherited metabolic disorder that may present with failure to thrive, diarrhea, vomiting, jaundice, and an increased tendency to bleed. Variants in the FAH gene cause a decrease in a liver enzyme called fumarylacetoacetate hydrolase, which cause a dangerous accumulation of tyrosine or its byproducts that are toxic to the liver, kidneys, and other organs and tissues. Type I tyrosinemia can potentially lead to liver and kidney failure, as well as affect the nervous system. Those with this disorder have an increased risk for liver cancer.

A restriction of tyrosine in the diet is recommended for those affected. A metabolic specialist and dietician typically manage care of individuals with the disorder. Lifespan is not typically decreased in treated individuals; however, death may occur in undetected and untreated individuals early in life.

How the Genetics Work

Type I tyrosinemia is an autosomal recessive disorder caused by variants in the *FAH* gene. In general, individuals have two copies of the *FAH* gene. Carriers of type I tyrosinemia have a variant in one copy of the *FAH* gene while individuals with the disorder have variants in both copies of *FAH* gene, 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.

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