Genetics Uncoded:

Facts about Hereditary Thymine-Uraciluria

What Your Test Results Mean

Carriers typically show no symptoms of hereditary thymine-uraciluria; however, carriers are at an increased risk of having a child with hereditary thymine-uraciluria. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Hereditary Thymine-Uraciluria Explained

Hereditary thymine-uraciluria is an inherited condition that prevents the body from breaking down the nucleotides thymine and uracil. This disease can vary greatly in severity; most affected individuals have no symptoms, while a few have mild to severe neurological problems that may include seizures, intellectual disability, delayed motor skills, and/or autism. Regardless of the individual's symptoms, every affected person must avoid the chemotherapy drug 5-fluorouracil and other fluoropyrimidines. Because the body cannot break down these drugs, the fluoropyrimidines are toxic and life threatening to people with hereditary thymine-uraciluria. There is no cure or specific treatment for hereditary thymine-uraciluria, but symptoms such as seizures can be addressed as they arise.

How the Genetics Work

Hereditary thymine-uraciluria is an autosomal recessive disorder caused by pathogenic variants in the *DPYD* gene. In general, individuals have two copies of the *DPYD* gene. Carriers of hereditary thymine-uraciluria have a single variant in one copy of the *DPYD* gene while individuals with hereditary thymine-uraciluria have variants in both copies of their genes, 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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