Dihydropyrimidine Dehydrogenase Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of dihydropyrimidine dehydrogenase deficiency. Carriers are not expected to show symptoms of dihydropyrimidine dehydrogenase deficiency. However some carriers, such as yourself, may experience toxic reactions to fluoropyrimidine drugs, which are used to treat cancer. You and your partner or donor would both have to be carriers of dihydropyrimidine dehydrogenase deficiency for there to be an increased chance to have a child with symptoms. This is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

We recommend that you share and discuss this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for dihydropyrimidine dehydrogenase deficiency, each of your children have a 25%, or 1 in 4, chance to have the condition.

Dihydropyrimidine Dehydrogenase Deficiency Explained

What is Dihydropyrimidine Dehydrogenase Deficiency?

Dihydropyrimidine dehydrogenase deficiency is an inherited condition that prevents the body from breaking down the nucleotides thymine and uracil. The presentation of this condition can vary greatly in severity. Most affected individuals have no symptoms, but some can have mild-to-severe neurological problems that may include seizures, intellectual disability, delayed motor skills, and/or autism. Affected individuals 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 dihydropyrimidine dehydrogenase deficiency.

Prognosis

Prognosis is not well-defined as the severity of the condition is highly variable.

Treatment

There is no cure or specific treatment for dihydropyrimidine dehydrogenase deficiency, but symptoms such as seizures can be addressed if they arise.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/dihydropyrimidine-dehydrogenase-deficiency
Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/19/dihydropyrimidine-dehydrogenase-deficiency
National Society of Genetic Counselors
https://www.nsgc.org/

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