NxGen MDx

Phenylalanine Hydroxylase Deficiency

What Your Results Mean

Test results indicate that you are a carrier of phenylalanine hydroxylase deficiency. Carriers typically show no symptoms. Risk for current or future pregnancies is dependent on your partner's carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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 phenylalanine hydroxylase deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Phenylalanine Hydroxylase Deficiency Explained

What is Phenylalanine Hydroxylase Deficiency?

Phenylalanine hydroxylase deficiency, otherwise known as phenylketonuria (PKU), is an inherited condition that affects the body's ability to process the amino acid phenylalanine from food. In affected individuals, phenylalanine obtained from the diet accumulates in the body. If not treated, the excess amount of phenylalanine can cause severe intellectual disabilities. The disease can be divided into several categories based on the amount of enzyme deficiency: classic phenylketonuria (PKU), variant PKU, and non-PKU hyperphenylalaninemia (non-PKU HPA). Symptoms can range from mild to severe. Classic PKU is the most severe form and affected individuals without dietary treatment experience seizures, delayed development, behavioral problems, and psychiatric disorders. Individuals with the mild forms of PKU have a lower risk of severe intellectual disability when left untreated.

Prognosis

Prognosis with treatment is generally favorable. Adhering to a PKU diet immediately after birth can prevent the neurological and psychiatric symptoms of this disease. Treatment is recommended for life; however, the diet is often difficult to follow during adolescence and adulthood. Without treatment, the average lifespan is about 56 years.

Treatment

Treatment involves following a phenylalanine-restricted diet. Affected individuals must maintain a low-protein diet consisting primarily of vegetables, fruits, and low-protein forms of certain foods. A phenylalanine-free essential amino acid formula is required for individuals to ensure their protein needs are met. Keeping blood phenylalanine levels within safe limits helps to prevent intellectual disabilities and ensure normal growth and development.

Resources

Children's PKU Network
http://www.pkunetwork.org/Childrens_PKU_Network/Home.html
March of Dimes
https://www.marchofdimes.org/complications/phenylketonuria-in-your-baby.aspx
National PKU News
https://pkunews.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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