Facts about Phenylalanine Hydroxylase Deficiency

Phenylalanine Hydroxylase Deficiency Explained

Phenylalanine hydroxylase deficiency is an inherited condition that affects the body’s ability to process the amino acid phenylalanine from food. There are three different categories of phenylalanine hydroxylase deficiency based on their severity. Classical phenylketonuria (PKU) is the most severe form. In this form, the enzyme phenylalanine hydroxylase is completely absent, requiring a protein-restricted diet. In variant PKU, the enzyme level is severely reduced. Children with this variant show milder symptoms but are still at risk if not on a protein-restricted diet. Finally, individuals with the milder non-PKU hyperphenylalaninemia may be able to tolerate a normal diet. Affected individuals who are diagnosed immediately and adhere to their diet can live a normal, healthy life. With early diagnosis being so essential, all states screen newborns for phenylalanine hydroxylase deficiency, usually within 24 hours of birth.

How the Genetics Work

Phenylalanine hydroxylase deficiency is an autosomal recessive disorder caused by variants in the PAH gene. In general, individuals have two copies of the PAH gene. Carriers of phenylalanine hydroxylase deficiency have a single variant in one copy of the PAH gene while individuals with phenylalanine hydroxylase deficiency 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.