NxGen MDx

Propionic Acidemia, *PCCA*-Related

What Your Results Mean

Test results indicate that you are a carrier of propionic acidemia, *PCCA*-related. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of propionic acidemia, *PCCA*-related 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.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

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 are carriers for propionic acidemia, *PCCA*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Propionic Acidemia, *PCCA*-Related Explained

What Is Propionic Acidemia, *PCCA*-Related?

Propionic acidemia, PCCA-related is an inherited disorder in which the body is unable to process certain parts of proteins and lipids (fats) properly. It is classified as an organic acid disorder, which is a condition that leads to an abnormal buildup of particular acids known as organic acids. Abnormal levels of organic acids in the blood (organic acidemia), urine (organic aciduria), and tissues can be toxic and can cause serious health problems. In most cases, the features of propionic acidemia become apparent within a few days after birth. The initial symptoms include poor feeding, vomiting, loss of appetite, weak muscle tone (hypotonia), and lack of energy (lethargy). These symptoms sometimes progress to more serious medical problems, including heart abnormalities, seizures, coma, and possibly death. Less commonly, the signs and symptoms of propionic acidemia appear during childhood and may come and go over time. Some affected children experience intellectual disability or delayed development. In children with this later-onset form of the condition, episodes of more serious health problems can be triggered by prolonged periods without food (fasting), fever, or infections.

Prognosis

Without appropriate treatment, the condition may lead to coma or death. Life expectancy is variable; for patients diagnosed before metabolic crisis, preventative treatment often results in a better outcome.

Treatment

There is no cure for propionic acidemia, *PCCA*-related. Individuals with this condition have to follow a specific diet including a low protein intake and specific food formulas (medical foods). Liver transplant is a surgical option that can help decrease the frequency of acute metabolic episodes (decompensation).

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/propionic-acidemia

National Society of Genetic Counselors
https://www.nsgc.org/

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