Hypophosphatasia

What Your Results Mean

Test results indicate that you are a carrier of hypophosphatasia. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of hypophosphatasia 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 or donor are carriers for hypophosphatasia, each of your children has a 1 in 4 (25%) chance to have the condition.

Hypophosphatasia Explained

What is Hypophosphatasia?

Hypophosphatasia is an inherited disorder characterized by weak and soft bones that results in skeletal abnormalities, short limbs, abnormally shaped chest, poor feeding, failure to gain weight, respiratory problems, hypercalcemia, and kidney problems. These symptoms can be life-threatening and appear any time from before birth to adulthood. There are six clinical forms of hypophosphatasia, and the severity of symptoms varies depending on the type. Affected children experience short stature with bowed legs or knock knees, enlarged wrist and ankle joints, and an abnormal skull shape. Adult-onset hypophosphatasia is characterized by softening of the bones. In adults, recurrent fractures in the foot and thigh bones can lead to chronic pain.

Prognosis

Prognosis varies by the type of hypophosphatasia. For example, the perinatal form is lethal, and the infantile form is fatal in half of individuals. Lifespan is expected to be normal for individuals with the adult and odontohypophosphatasic forms.

Treatment

Treatment is supportive and focuses on decreasing the morbidity associated with hypophosphatasia.

Resources

Hypophosphatasia.com
http://www.hypophosphatasia.com/
National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/hypophosphatasia/
National Society of Genetic Counselors
https://www.nsgc.org/

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