Clarity Genetics

Genetics Uncoded:

Facts about Hypophosphatasia

What Your Test Results Mean

Some carriers of hypophosphatasia will show symptoms of the disorder while others remain asymptomatic. Whether a carrier has symptoms of the disease or not, they are at an increased risk to have a child with hypophosphatasia. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Hypophosphatasia Explained

Hypophosphatasia is an inherited disorder that is characterized by weak and soft bones that result in skeletal abnormalities, short limbs, abnormally shaped chest, poor feeding, failure to gain weight, respiratory problems, hypercalcemia, and kidney problems. These problems can be life-threatening. Symptoms appear any time from before birth to adulthood, depending on the severity of the disease. 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. Affected children experience short stature with bowed legs or knock knees, enlarged wrist and ankle joints, and an abnormal skull shape. Currently, there is no approved therapy for hypophosphatasia. Current management of the disease involves relieving symptoms, maintaining calcium balance, and using surgical interventions when needed.

How the Genetics Work

Hypophosphatasia is a genetic disorder caused by pathogenic variants in the *ALPL* gene. In general, individuals have two copies of the *ALPL* gene. Some individuals with one variant in the *ALPL* gene may have mild symptoms of the disease; however, most affected individuals have two variants in their *ALPL* genes. Risk for an individual with one variant in the *ALPL* gene to have a child with the disorder is 50%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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