Facts about Hypophosphatiasia

Hypophosphatiasia Explained

Hypophosphatiasia 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 hypophosphatiasia 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 hypophosphatiasia. Current management of the disease involves relieving symptoms, maintaining calcium balance, and using surgical interventions when needed.

How the Genetics Work

Hypophosphatiasia 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 1-855-776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.