Facts about Cartilage-Hair Hypoplasia – Anauxetic Dysplasia Spectrum Disorders

CHH-AD Explained

CHH-AD spectrum disorders are inherited conditions that affect the skeletal and immune systems. The CHH-AD spectrum includes the milder cartilage-hair hypoplasia and metaphyseal dysplasia without hypotrichosis, and the more severe Anauxetic Dysplasia. Symptoms include short-limbed dwarfism, light-colored, sparse hair, and immune problems. Prognosis is dependent on the presence and severity of immunodeficiency. Management is focused on protecting affected individuals from infection by means of vaccinations and managing infections when they occur. Bone marrow transplant has been used successfully to manage individuals with severe immunodeficiency.

How the Genetics Work

CHH-AD spectrum disorders are a group of autosomal recessive disorders caused by variants in the RMRP gene. In general, individuals have two copies of the RMRP gene. Carriers of cartilage-hair hypoplasia have a single variant in one copy of the RMRP gene while individuals with cartilage-hair hypoplasia 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.