Genetics Uncoded:

NxGen MDx

Facts about
Cartilage-Hair Hypoplasia – Anauxetic Dysplasia Spectrum Disorders

What Your Test Results Mean

Carriers of cartilage-hair hypoplasia – anauxetic dysplasia (CHH-AD) spectrum disorders typically show no symptoms; however, carriers are at an increased risk of having a child with CHH-AD. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

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.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504

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