Cartilage-Hair Hypoplasia – Anauxetic Dysplasia Spectrum Disorders

What Your Results Mean

Test results indicate that you are a carrier of cartilage-hair hypoplasia – anauxetic dysplasia spectrum disorders. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of cartilage-hair hypoplasia – anauxetic dysplasia spectrum disorders 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 are carriers for cartilage-hair hypoplasia – anauxetic dysplasia spectrum disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

Cartilage-Hair Hypoplasia – Anauxetic Dysplasia Spectrum Disorders Explained

What is Cartilage-Hair Hypoplasia – Anauxetic Dysplasia Spectrum Disorders?

Cartilage-hair hypoplasia is an inherited condition that affects the skeletal and immune systems. Symptoms include short-limbed dwarfism, light-colored and sparse hair, anemia, and immune problems that can lead to multiple infections. Affected individuals may also have light-colored skin (hypopigmentation), malformed nails, and dental abnormalities. They are also at an increased risk of developing cancers such as skin cancer, leukemia, and lymphoma. The extent of immunodeficiency can vary from mild to severe in individuals.

Prognosis

Prognosis is dependent on the presence and severity of immunodeficiency. Many individuals can live a normal lifespan; however, this ultimately depends on the severity of immunodeficiency and avoidance of opportunistic infections, particularly in childhood.

Treatment

Management is focused on protecting against infection by means of vaccinations and managing infections when they occur. Bone marrow transplant has been used successfully to manage individuals with severe immunodeficiency. Growth hormones can be considered for some individuals.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/cartilage-hair-hypoplasia
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/mckusick-type-metaphyseal-chondrodysplasia/
National Society of Genetic Counselors
https://www.nsgc.org/

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