Inheritest® CARRIER SCREEN

Cartilage-hair hypoplasia

What is cartilage-hair hypoplasia?

Cartilage-hair hypoplasia is an inherited disease characterized by abnormal bone growth, causing short stature and other skeletal anomalies, blood and immune problems, and fine, sparse, light-colored hair. Symptoms associated with cartilage-hair hypoplasia are due to the disruption of normal cellular function, primarily affecting the skeletal and immune systems.[1,2]

Cartilage-hair hypoplasia belongs to a group of disorders known as cartilage-hair hypoplasia–anauxetic dysplasia spectrum disorders, and is also known as McKusick metaphyseal chondrodysplasia syndrome.[1,2]

What are the symptoms of cartilage-hair hypoplasia and what treatment is available?

Cartilage-hair hypoplasia is a disease that varies in severity, even within families. Some signs and symptoms of cartilage-hair hypoplasia are usually apparent at birth and may include:[2]

- Unusually short limbs, short stature, and other skeletal problems
- Hypotrichosis (light, sparse, fine silky hair)
- Hypermobility (increased flexibility of joints), except for the elbows
- Macrocytic anemia (enlarged red blood cells in lower than normal amounts)
- Recurrent infections due to immunodeficiency
- Gastrointestinal problems
- Increased risk for certain cancers

Some affected individuals have normal hair and are diagnosed as having metaphyseal dysplasia without hypotrichosis.[2]

There is no cure for cartilage-hair hypoplasia. Treatment includes supportive care for symptoms and may include blood transfusions for severe anemia, bone surgery, and antibiotics for infections. For individuals with recurrent severe infections, bone marrow transplant may be considered.[2] Due to the increased risk for infections and cancers, life expectancy may be decreased.[3]

How is cartilage-hair hypoplasia inherited?

Cartilage-hair hypoplasia is an autosomal recessive disease caused by mutations in the *RMRP* gene.[1] An individual who inherits one *RMRP* gene mutation is a carrier and is not expected to have related health problems. An individual who inherits two copies of an *RMRP* gene mutation, one from each parent, is expected to be affected with cartilage-hair hypoplasia.[4]

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for cartilage-hair hypoplasia?

Cartilage-hair hypoplasia is a rare disorder most frequently reported in the Old Order Amish and Finnish populations but occurring worldwide. The carrier frequency in the Old Order Amish is estimated to be 1 in 19 with a calculated incidence of approximately 1 in 1400.[3] Among the Finnish, the carrier frequency is estimated to be 1 in 76, with a calculated incidence of approximately 1 in 23,000.[3]

Having a relative who is a carrier or is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Genetics Home Reference: http://ghr.nlm.nih.gov/condition/cartilage-hair-hypoplasia

References

1. Cartilage Hair Hypoplasia. *Genetics Home Reference.* Available at: http://ghr.nlm.nih.gov/condition/cartilage-hair-hypoplasia. Accessed on: March 29, 2012.
2. Thiel C. Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders. *GeneReviews.* Available at: http://www.ncbi.nlm.nih.gov/books/NBK84550/ . Accessed on: March 29, 2012.
3. Ridanpää. M *et al.* The major mutation in the RMRP gene causing CHH among the Amish is the same as that found in most Finnish cases. *Am J Med Genet C Semin Med Genet.* 2003;121C(1):81-3.
4. Ridanpää. M *et al.* Genetic changes in the RNA components of RNase MRP and RNase P in Schmid metahpyseal chondrodsyplasia *J Med Genet* 2033;40:741-746.