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My46 Trait Profile

Achondroplasia

Achondroplasia is the most common condition causing disproportionate short stature. It is caused by mutations in the FGFR3 gene that makes a protein called fibroblast growth factor receptor 3.

Characteristics of Achondroplasia

Achondroplasia is the most common type of dwarfism – a condition characterized by short stature. Achondroplasia causes significant short stature with the limbs being significantly shortened in relation to the length of the body. The average adult height is about 4 feet 4 inches for males, and 4 feet 1 inch for females. Individuals with achondroplasia have a large head with a prominent forehead and an underdeveloped midface with a depressed nasal bridge. A trident configuration (split between the 3rd and 4th fingers) of the hands is common as is a small chest. Bowed legs and an inward curve of the lower spine (lumbar lordosis) develop in childhood. Intelligence is usually normal in individuals with achondroplasia.

Diagnosis/Testing

Achondroplasia has commonly been diagnosed based on physical characteristics and x-ray findings. Almost all individuals with achondroplasia have a change or mutation in the FGFR3 gene. This gene makes a growth factor receptor protein that is part of the growth regulation process. Its role is to slow down growth. It is thought that mutations in the FGFR3 gene turn on the receptor and therefore excessively slow down growth.

Management/Surveillance

Management of achondroplasia is best provided by a multispecialty team, including genetics, orthopedics, neurology and/or neurosurgery, ear, nose and throat providers, as well as physical, occupational and speech therapies.

Common issues in childhood are difficulty breathing during sleep (e.g., apnea), kyphosis (an outward curve of the mid-spine), leg bowing, recurrent ear infections, speech and motor delay. The most critical issue for babies with achondroplasia is that the opening at the base of the skull is significantly narrowed. This can cause impingement or compression of the brainstem and/or hydrocephalus. A very small number of children require surgical intervention, but it is critical that all children, ages 0-2, be monitored very closely for signs of these problems. Medical issues that adults with achondroplasia may face include compression of the spinal cord in the lumbar region, sleep apnea, and obesity.

The need for adaptations or accommodations in the home, school, workplace, vehicles and other places is an ongoing necessity for people with achondroplasia due to the significant short stature.

Mode of inheritance

Achondroplasia is inherited in an autosomal dominant pattern. This means that inheriting one FGFR3 mutation is enough for an individual to be affected and show signs of achondroplasia. The mutation can be inherited from an affected parent, however 80% of the time, it occurs brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on whether or not the individual with achondroplasia has a parent with achondroplasia. If a parent also has achondroplasia, the risk of having a child with achondroplasia is 50% with each

pregnancy. If a parent does not have achondroplasia, the risk of other siblings being affected is very low.

For couples in which both parents have achondroplasia, there is a 25% chance with each pregnancy that their offspring will not receive the mutation and be average statured. There is a 50% chance with each pregnancy that their offspring will have achondroplasia. There is a 25% chance with each pregnancy that their offspring will inherit two achondroplasia mutations (one from each parent) and the baby will have a very severe form of achondroplasia in which the baby usually passes away in the newborn period.

Special considerations

The social aspect of having dwarfism can be as or more significant than the medical and adaptive aspects. Being short-statured and orthopedically disabled carries great stigma in our society. When out in public, individuals with achondroplasia are confronted with stares, comments, teasing, and ignorance. Children need assistance to develop coping mechanisms and reactions to these intrusions. Little People of America (LPA; see link below) is the support organization for all people of short stature and their families. This organization can be extremely helpful in assisting with social adjustment and, more than anything else, letting people with short stature know that they are not alone in their circumstances.

Resources

Little People of America
http://www.lpaonline.org/
Genetics Home Reference: Achondroplasia
http://ghr.nlm.nih.gov/condition/achondroplasia
Kaiser Permanente Genetics Northern California: Achondroplasia
http://mydoctor.kaiserpermanente.org/ncal/specialty/genetics/resources/conditions/achondroplasia.jsp
NHGRI: Learning About Achondroplasia
http://www.genome.gov/19517823

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Created: 09/2013 **Created by:** Ericka Okenfuss, MS, CGC

Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC