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My46 Trait Profile

Hypochondroplasia

Hypochondroplasia is a form of short-limbed dwarfism. Most of the time this condition is caused by a single mutation in the FGFR3 gene that makes a protein called fibroblast growth factor receptor 3.

Characteristics of Hypochondroplasia

Hypochondroplasia is a mild form of short-limbed dwarfism – a condition of short stature. Individuals with this condition are shorter than average and their limbs (arms and legs) are shortened in relation to the length of the body or trunk (disproportionate short stature). The average adult height in an individual with hypochondroplasia ranges from 4 feet, 2 inches to 5 feet, 5 inches, depending on whether they are male or female and how tall parents and family members are. Individuals with hypochondroplasia may have short hands and feet, limited bending of the elbow (limitation of elbow extension), mildly flexible joints, and a large head. Individuals with hypochondroplasia may also have lumbar lordosis (inner curve of the lower spine), bowed legs, mild to moderate intellectual disability, and/or learning disabilities, Much less common features include scoliosis (curvature of the spine) and epilepsy (seizures). Hypochondroplasia is similar to another condition of short-limbed dwarfism called achondroplasia (see trait profile) though the features of hypochondroplasia tend to be milder.

Diagnosis/Testing

Hypochondroplasia has commonly been diagnosed based on physical characteristics and X-ray findings. Sometimes it is difficult to make the diagnosis in children under three years of age as the short stature may be mild and difficult to recognize. The majority (~70%) of individuals with hypochondroplasia 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 slow down growth excessively. Clinical genetic testing is available and is often used to confirm a clinical diagnosis of hypochondroplasia. Prenatal diagnosis is available for couples in which one or both parents have hypochondroplasia. Hypochondroplasia is not typically detected by ultrasound during pregnancy.

Management/Surveillance

The majority of individuals with hypochondroplasia do not have significant medical complications related to their condition. However, because some of the physical features of the condition are similar to individuals who have achondroplasia, a more serious form of short stature, health care supervision guidelines for achondroplasia may also be followed for individuals with hypochondroplasia. Management of hypochondroplasia is best provided by a multi-disciplinary team including genetics, orthopedics, neurology and/or neurosurgery, ear, nose, and throat specialists, as well as physical, occupational and speech therapies.

Common issues in children with achondroplasia, and sometimes hypochondroplasia, include difficulty breathing during sleep (apnea), kyphosis (an outward curve of the mid-spine) and lordosis (an inward curve of the lower spine), leg bowing, recurrent ear infections, and speech and motor delay. The most critical issue for babies with achondroplasia, and possibly hypochondroplasia, is that the opening at the base of the skull may be significantly narrowed. This can cause impingement or compression of the brainstem and/or hydrocephalus (increased fluid in the brain) and may require surgery in a small number of children. As a result, it is important that young children be monitored closely for signs of

these problems.

Additionally, intellectual disability or learning disability is thought to occur more frequently in hypochondroplasia than achondroplasia or the general population. As such, developmental intervention, assessment for learning disabilities, and special education may be appropriate, based on an individual's needs.

The need for adaptations or accommodations in the home, school, workplace, vehicles and other places may be necessary for individuals with hypochondroplasia because of the short stature and limited reach.

Mode of inheritance

Hypochondroplasia is inherited in an autosomal dominant pattern. This means inheriting one FGFR3 mutation is enough for an individual to be affected and show signs of hypochondroplasia. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child. The majority of individuals with a diagnosis of hypochondroplasia have the condition as a result of a de novo mutation.

Risk to family members

The risk to family members depends on whether or not the individual with hypochondroplasia has a parent affected with the same condition. For a couple in which one of the parents has hypochondroplasia, the risk of having a child with hypochondroplasia is 50% with each pregnancy. For an average-sized couple who have had one child with hypochondroplasia, the risk of other siblings being affected is very low.

For couples in which both parents have hypochondroplasia, there is a 25% chance with each pregnancy that the baby will not receive an FGFR3 mutation and have average stature (height). There is a 50% chance with each pregnancy that the baby will have hypochondroplasia. There is a 25% chance with each pregnancy that the child will inherit two FGFR3 mutations and may have a more severe form of the condition.

Special considerations

Hypochondroplasia is considered a mild disorder of stature and many affected individuals do not consider themselves disabled. However, some individuals may consider short stature a significant physical, emotional and social disability and the social aspect of having dwarfism can be as or more significant than the medical and adaptive aspects. Little People of America (LPA; see link below) is the support organization for individuals with 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/dwarfism know that they are not alone in their circumstances.

Resources

Little People of America
http://www.lpaonline.org
The Child Growth Foundation (UK)
http://www.childgrowthfoundation.org/
Genetics Home Reference: Hypochondroplasia
http://ghr.nlm.nih.gov/condition/hypochondroplasia

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