Hypochondroplasia

Description

Hypochondroplasia is a form of short-limbed dwarfism. This condition affects the conversion of cartilage into bone (a process called ossification), particularly in the long bones of the arms and legs. Hypochondroplasia is similar to another skeletal disorder called achondroplasia, but the features tend to be milder.

All people with hypochondroplasia have short stature. The adult height for men with this condition ranges from 138 centimeters to 165 centimeters (4 feet, 6 inches to 5 feet, 5 inches). The height range for adult women is 128 centimeters to 151 centimeters (4 feet, 2 inches to 4 feet, 11 inches).

People with hypochondroplasia have short arms and legs and broad, short hands and feet. Other characteristic features include a a large head (macrocephaly), limited range of motion at the elbows, a sway of the lower back (lordosis), and bowed legs. These signs are generally less pronounced than those seen in people with achondroplasia and may not be noticeable until early or middle childhood. Affected individuals have a small increased risk of a seizure disorder known as temporal lobe epilepsy. Some studies have reported that a small percentage of people with hypochondroplasia have mild to moderate intellectual disability or learning problems, but other studies have produced conflicting results.

Frequency

The incidence of hypochondroplasia is unknown. Researchers believe that it may be about as common as achondroplasia, which occurs in 1 in 15,000 to 40,000 newborns. More than 200 people worldwide have been diagnosed with hypochondroplasia.

Causes

The vast majority of cases of hypochondroplasia are caused by variants in the *FGFR3* gene. This gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. Although it remains unclear how *FGFR3* gene variants lead to the features of hypochondroplasia, researchers believe that these genetic changes cause the protein to be overly active. The overactive FGFR3 protein likely interferes with skeletal development and leads to the disturbances in bone growth that are characteristic of this disorder.

Some people with hypochondroplasia do no have a variant in the *FGFR3* gene. Researchers suspect that variants in other genes cause hypochondroplasia in these people, although these genes have not been identified.

Learn more about the gene associated with Hypochondroplasia

- FGFR3

Inheritance

Hypochondroplasia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most people with hypochondroplasia are born to parents who do not have the condition and are of average heights; these cases are caused by new variants in the *FGFR3* gene. In the remaining cases, people with hypochondroplasia inherit an altered *FGFR3* gene from one or two affected parents. Individuals who inherit two altered copies of this gene typically have more severe problems with bone growth than those who inherit a single *FGFR3* variant.

Other Names for This Condition

- HCH
- Hypochondrodysplasia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Hypochondroplasia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0410529/)

Genetic and Rare Diseases Information Center

- Hypochondroplasia (https://rarediseases.info.nih.gov/diseases/6724/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Hypochondroplasia%22)

Catalog of Genes and Diseases from OMIM

- HYPOCHONDROPLASIA; HCH (https://omim.org/entry/146000)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Dwarfism%5BMAJR%5D%29+AND+%28%28hypochondroplasia%5BTIAB%5D%29+OR+%28hch%5BTIAB%5D%29+OR+%28hypochondrodysplasia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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