Hypochondroplasia

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 large head, 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 with achondroplasia and may not be noticeable until early or middle childhood. 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

About 70 percent of all cases of hypochondroplasia are caused by mutations in the \textit{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 \textit{FGFR3} mutations lead to the features of hypochondroplasia, researchers believe that these genetic changes cause the protein to be overly active. The overactive \textit{FGFR3} protein likely interferes with skeletal development and leads to the disturbances in bone growth that are characteristic of this disorder.

In the absence of a mutation in the \textit{FGFR3} gene, the cause of hypochondroplasia is unknown. Researchers suspect that mutations in other genes are involved, although these genes have not been identified.

Inheritance Pattern

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 have average-size parents; these cases result from a new
mutation in the \textit{FGFR3} gene. In the remaining cases, people with hypochondroplasia have inherited an altered \textit{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 \textit{FGFR3} mutation.

Other Names for This Condition

- HCH
- Hypochondrodysplasia

Diagnosis & Management

Genetic Testing Information

- What is genetic testing? /primer/testing/genetictesting

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov https://clinicaltrials.gov/ct2/results?cond=%22hypochondroplasia%22

Other Diagnosis and Management Resources


Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Lordosis https://medlineplus.gov/ency/article/003278.htm

Genetic and Rare Diseases Information Center

- Hypochondroplasia https://rarediseases.info.nih.gov/diseases/6724/hypochondroplasia
Educational Resources
• KidsHealth from the Nemours Foundation
• MalaCards: hypochondroplasia
  https://www.malacards.org/card/hypochondroplasia
• Orphanet: Hypochondroplasia
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=429

Patient Support and Advocacy Resources
• Human Growth Foundation
  https://www.hgfound.org/
• International Skeletal Dysplasia Registry, UCLA
  https://www.uclahealth.org/ortho/isdr
• Little People of America, Inc.
  https://www.lpaonline.org/
• Little People UK
  https://littlepeopleuk.org/
• National Organization for Rare Disorders
  https://rarediseases.org/rare-diseases/hypochondroplasia/
• Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/dwarfism.html
• The MAGIC Foundation
  https://www.magicfoundation.org/

Clinical Information from GeneReviews
• Hypochondroplasia
  https://www.ncbi.nlm.nih.gov/books/NBK1477

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

Catalog of Genes and Diseases from OMIM
• HYPOCHONDROPLASIA
  http://omim.org/entry/146000
Sources for This Summary


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Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services