# Achondroplasia

## Description

Achondroplasia is the most common form of short-limbed dwarfism. The word achondroplasia means "without cartilage formation." Cartilage is a tough but flexible tissue that makes up much of the skeleton during early development. However, in people with achondroplasia the problem is not forming cartilage but converting it to bone (a process called ossification), particularly in the long bones of the arms and legs. Achondroplasia is similar to another skeletal disorder called hypochondroplasia, but the features of achondroplasia tend to be more severe.

All people with achondroplasia have short stature. Without treatment, the average height of an adult male with achondroplasia is 131 centimeters (4 feet, 4 inches), and the average height for adult females is 124 centimeters (4 feet, 1 inch). Characteristic features of achondroplasia include an average-size trunk, short arms and legs with particularly short upper arms and thighs, limited range of motion at the elbows, and an enlarged head (macrocephaly) with a prominent forehead. Fingers are typically short and the ring finger and middle finger may diverge, giving the hand a three-pronged ( trident) appearance.

Health problems commonly associated with achondroplasia include obesity and recurrent ear infections. People with achondroplasia are generally of normal intelligence. In childhood, individuals with the condition usually develop a pronounced and permanent sway of the lower back (lordosis) and bowed legs. Some affected people also develop abnormal front-to-back curvature of the spine (kyphosis) and back pain.

As affected individuals age, they may experience a potentially serious complication of achondroplasia called spinal stenosis. Spinal stenosis is a narrowing of the spinal canal that can pinch (compress) the upper part of the spinal cord. Spinal stenosis causes with pain, tingling, and weakness in the legs that can make walking difficult. An uncommon but serious complication of achondroplasia in early childhood is stenosis of the hole at the base of the skull where the spinal cord comes out of brain (foramen magnum). This complication can cause compression of the brain stem, which can lead to pauses in breathing during sleep (sleep apnea) or a condition known as hydrocephalus. Hydrocephalus is a buildup of fluid in the brain that can lead to increased head size and related brain abnormalities.

## Frequency

Achondroplasia is the most common type of short-limbed dwarfism. The condition occurs in 1 in 15,000 to 40,000 newborns.

## Causes

Variants (also called mutations) in the *FGFR3* gene cause achondroplasia. The *FGFR3* gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. Two specific variants in the *FGFR3* gene are responsible for almost all cases of achondroplasia. These variants cause the FGFR3 protein to be overly active, which interferes with skeletal development and leads to the disturbances in bone growth seen with this disorder.

Learn more about the gene associated with Achondroplasia

- FGFR3

## Inheritance

Achondroplasia is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. About 80 percent of people with achondroplasia 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 achondroplasia inherit an altered *FGFR3* gene from one or two affected parents.

Individuals who inherit two altered copies of the *FGFR3* gene typically have a severe form of achondroplasia that causes extreme shortening of the bones and an underdeveloped rib cage. These individuals are usually stillborn or die shortly after birth from respiratory failure.

## Other Names for This Condition

- ACH
- Achondroplastic dwarfism
- Dwarf, achondroplastic

## Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Achondroplasia (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0001080/)

Genetic and Rare Diseases Information Center

- Achondroplasia (https://rarediseases.info.nih.gov/diseases/8173/index)

Patient Support and Advocacy Resources

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

Clinical Trials

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

Catalog of Genes and Diseases from OMIM

- ACHONDROPLASIA; ACH (https://omim.org/entry/100800)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Achondroplasia%5BMAJR%5D%29+AND+%28achondroplasia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

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