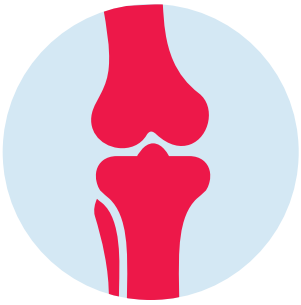
ACHONDROPLASIA

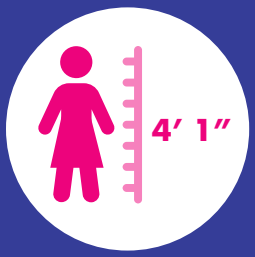


What is Achondroplasia?

Achondroplasia is a rare genetic bone growth disorder and the most common form of dwarfism and disproportionate short stature.[1] Achondroplasia is caused by a change in the fibroblast growth factor receptor 3 *(FGFR3)* gene, which slows down the formation of bone in the cartilage of the growth plate.[2] This impairs growth in almost all bones in the body.

Average Achondroplasia Adult Height

A male with achondroplasia will reach an average adult height of approximately 4 feet, 4 inches.



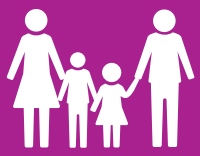
A female with achondroplasia will reach an average adult height of about 4 feet, 1 inch.[3]

What Causes Achondroplasia?

In achondroplasia, a change in the structure of the *FGFR3* gene causes the body's cartilage cells, called chondrocytes, to continuously send out signals to slow bone growth. Because *FGFR3* receptors are overactive, the signals to slow bone growth are stronger than the signals that tell bones to grow. As a result, the cells in the cartilage have trouble lining up to form new bone, causing slowed bone growth.

How is Achondroplasia Diagnosed?

Achondroplasia may be diagnosed before birth by fetal ultrasound. DNA testing can also be used to identify a change in the *FGFR3* gene to confirm fetal ultasound results or clinical diagnosis. The condition may also be diagnosed after birth through a physical exam.[4]



Over 80%
of individuals with achondroplasia have parents of average height and are born with achondroplasia as a result of a new FGFR3 gene change in their family.[1]

Achondroplasia impacts endochondral bone growth, which is the form of bone growth that replaces cartilage with bone tissue as children grow.

More than 90%
of bones in the body can be affected.

1/25,000 births
Achondroplasia is a rare disease and the most common form of skeletal dysplasia, occurring in one in every 25,000 births.[5]

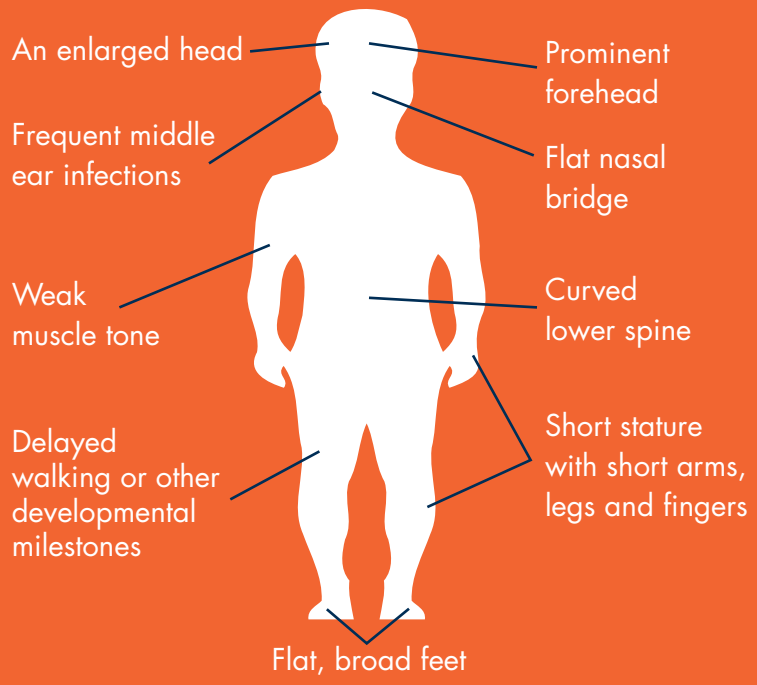
Symptoms & Managing Complications

Achondroplasia is characterized by distinctive features including short stature, curvature of the spine and an enlarged head (macrocephaly). These characteristics may lead to health challenges including reduced breathing for short periods of time (apnea), upper airway obstruction, obesity, hearing loss and dental problems.[6] In addition, adults may develop bowed legs and lower back problems that can lead to difficulty walking.

Options to help alleviate clinical complications of achondroplasia may include:

- Surgery to remove the tonsils or adenoids to help with difficulty breathing
- Ear tubes to help treat ear infections
- Orthopedic management to correct bone problems that cause pain and affect mobility
- Visiting the orthodontist for problems with the teeth or mouth, like misaligned teeth, a narrow palate, open bite, or underbite

Clinical features of achondroplasia may include[2]:



Living with Achondroplasia

Achondroplasia does not affect cognitive development and individuals have an average lifespan; however, they must be vigilant to monitor for possible complications and manage health challenges that occur.[1] Because of the effects throughout the body, many different specialties should participate in the care of people with achondroplasia for optimal outcomes. A child with achondroplasia may begin to see certain specialists at different times throughout life. Some will follow the child over their lives, while others will be more important at certain ages.

6x increased risk of sudden infant death due to foramen magnum stenosis

70% experience chronic back pain by age 50

50% experience chronic leg pain by adulthood

1. About Achondroplasia. Genome.gov. https://www.genome.gov/Genetic-Disorders/Achondroplasia. Published July 15, 2016. Accessed July 6, 2021.
2. Hoover-Fong J, Cheung MS, Fano V, et al. Lifetime impact of achondroplasia: Current evidence and perspectives on the natural history. *ScienceDirect*. https://www.sciencedirect.com/science/article/pii/S875632822100034X?via%3Dihub Published February 3, 2021. Accessed July 7, 2021.
3. Achondroplasia: Medline Plus Genetics. *MedlinePlus*. https://medlineplus.gov/genetics/condition/achondroplasia/. Published August 18, 2020. Accessed July 7, 2021.
4. Hoover-Fong J, Scott CI, Jones MC. Health Supervision for People With Achondroplasia. *American Academy of Pediatrics*. https://pediatrics.aappublications.org/content/145/6/e20201010. Published June, 2020. Accessed July 7, 2021.
5. Al-Saleem A, Al-Jobair A. Achondroplasia: Craniofacial manifestations and considerations in dental management. *The Saudi Dental Journal*. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3804960/. Published October 2010. Accessed July 6, 2021.
6. Hoover-Fong JE, Alade AY, Hashmi SS, et al. Achondroplasia Natural History Study (CLARITY): a multicenter retrospective cohort study of achondroplasia in the United States. *Nature News*. https://www.nature.com/articles/s41436-021-01165-2. Published May 18, 2021. Accessed July 7, 2021.

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