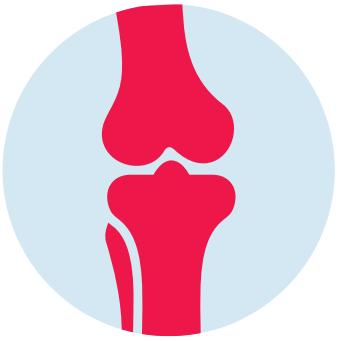
EU-ACH-00212
August 2021

ACHONDROPLASIA

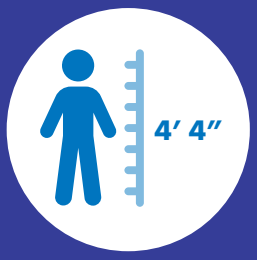


What is Achondroplasia?

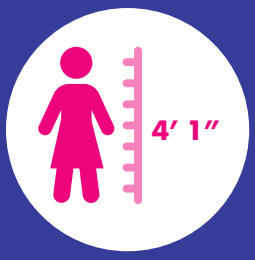
Achondroplasia is a genetic condition in which endochronal bone growth is impaired, and the most common form of disproportionate short stature.[1] Achondroplasia is caused by a change in the fibroblast growth factor receptor 3 (FGFR3), which slows down the formation of bone in the cartilage of the epiphyes.*[2]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

*growth plates

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 *FGFR3* 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]

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]

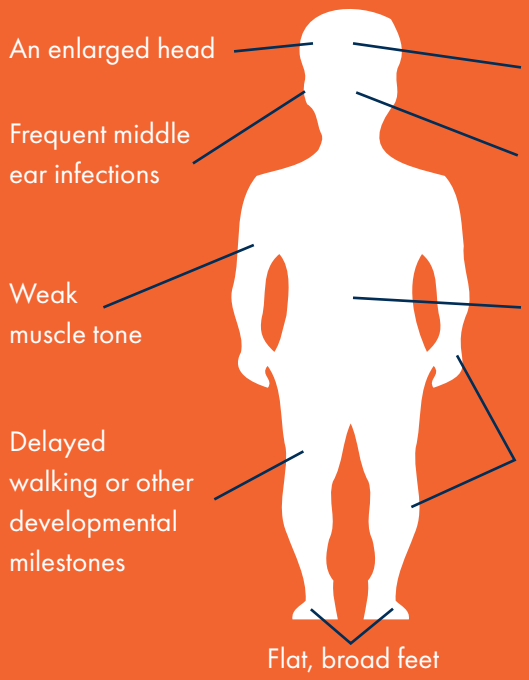
Symptoms & Managing Complications

Achondroplasia is characterised by distinctive features including disproportionate 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 (apnoea), 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
- Orthopaedic 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[7]

experience chronic back pain by age 50[2]

experience chronic leg pain by adulthood[8]

1. About Achondroplasia. Genome.gov. https://www.genome.gov/Genetic-Disorders/Achondroplasia. Published July 15, 2016. Accessed July 6, 2021.
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