

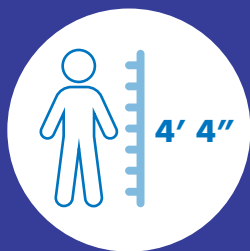
# ACHONDROPLASIA



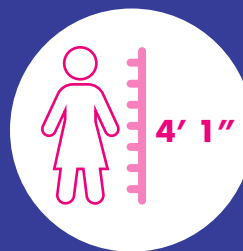
## What is Achondroplasia?

Achondroplasia is a rare genetic bone growth disorder and the most common form of dwarfism and disproportionate short stature.<sup>1</sup> 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.<sup>2</sup> This impairs growth in almost all bones in the body.<sup>1</sup>

## Average Achondroplasia Adult Height



A male with achondroplasia will reach an average adult height of approximately 4 feet, 4 inches.<sup>3</sup>



A female with achondroplasia will reach an average adult height of about 4 feet, 1 inch.<sup>3</sup>

## 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.<sup>1</sup>

## 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 ultrasound results or clinical diagnosis. The condition may also be diagnosed after birth through a physical exam.<sup>4</sup>



## 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.<sup>1</sup>



Achondroplasia impacts endochondral bone growth, which is the form of bone growth that replaces cartilage with bone tissue as children grow.<sup>5</sup>





## More than 90%

of bones in the body can be affected.<sup>6,7</sup>

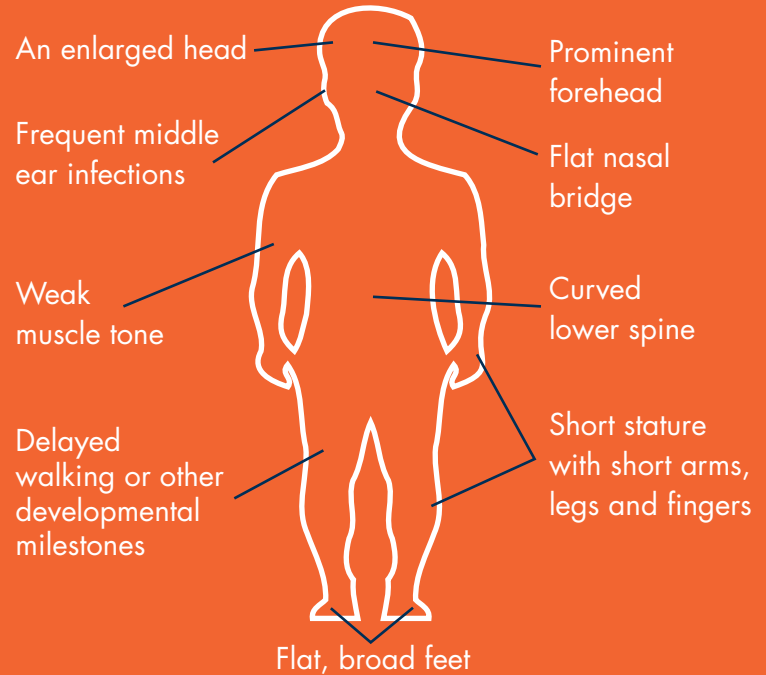
## Symptoms & Managing Complications

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.<sup>8</sup> 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<sup>2</sup>:



## 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 may occur.<sup>1</sup> 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.<sup>9</sup>



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



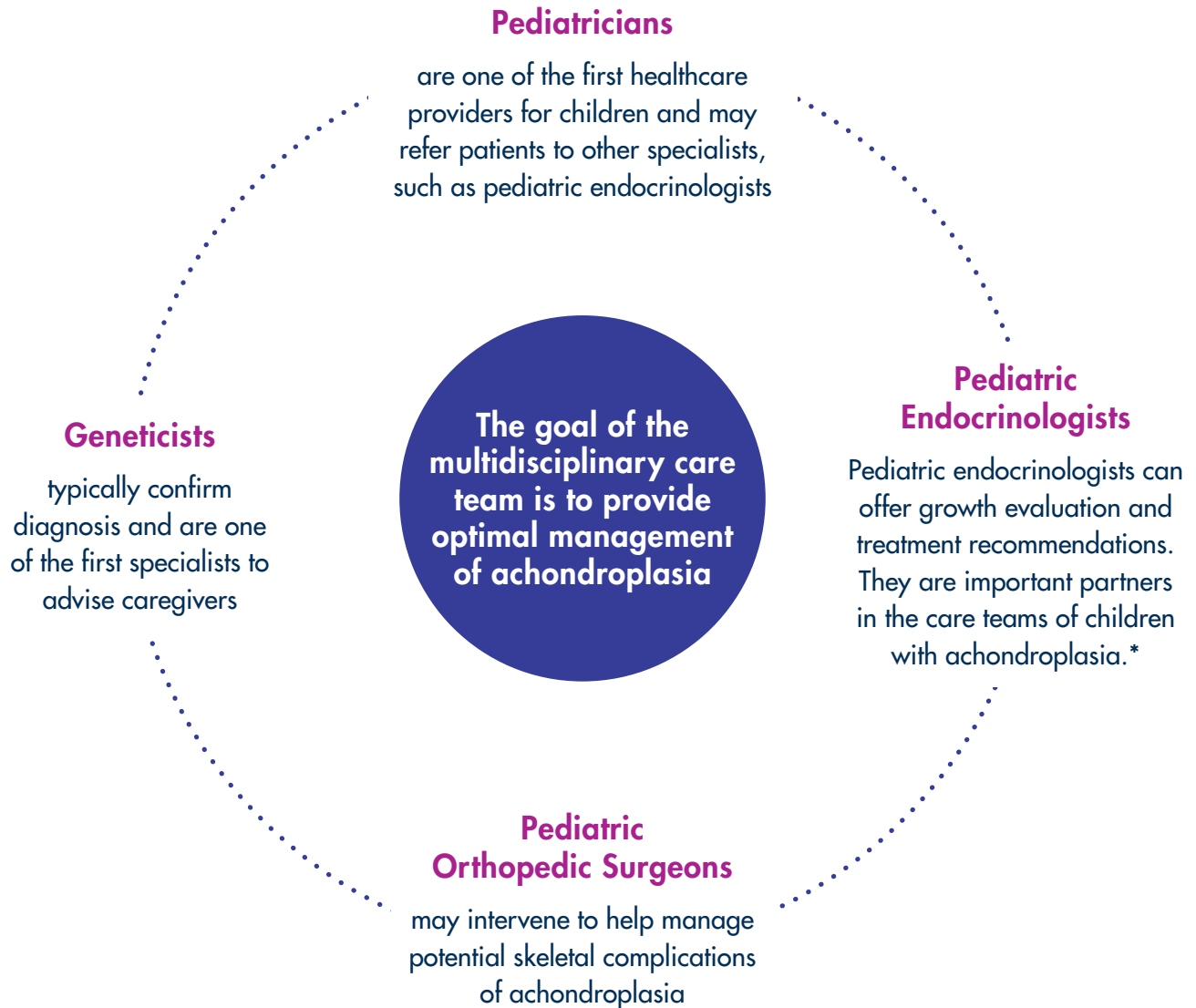
experience chronic back pain by age 50



experience chronic leg pain by adulthood

## Children with Achondroplasia May Require a Multidisciplinary Care Team

Achondroplasia affects every child differently, and their care team may vary as a result.\* Some of the specialists who may play a role in the care of children with achondroplasia include:



\*Due to the diverse and multisystemic characteristics of achondroplasia, each child's full healthcare team may vary.

1. About Achondroplasia. Genome.gov. <https://www.genome.gov/Genetic-Disorders/Achondroplasia>. Published July 15, 2016. Accessed October 2023.
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 October 2023.
3. Achondroplasia: Medline Plus Genetics. *MedlinePlus*. <https://medlineplus.gov/genetics/condition/achondroplasia/>. Published August 18, 2020. Accessed October 2023.
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 October 2023.
5. McDonald EJ, De Jesus O. Achondroplasia. *StatPearls*. <https://www.ncbi.nlm.nih.gov/books/NBK559263/>. Published January 2023. Accessed October 2023.
6. Clarke B. Normal bone anatomy and physiology. *Clinical Journal of the American Society of Nephrology*. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3152283/>. Published November 2008. Accessed October 2023.
7. Berendsen AD, Olsen BR. Bone development. *Bone*. <https://www.sciencedirect.com/science/article/abs/pii/S8756328215001489>. Published November 2015. Accessed October 2023.
8. Pauli, R. Achondroplasia: a comprehensive clinical review. *Orphanet Journal of Rare Diseases*. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6318916/>. Published January 3, 2019. Accessed October 2023.
9. Lin T, Ho C, et al. Lower back pain heralding cauda equina syndrome in a patient with achondroplasia. *Cureus*. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8938231/>. Published February 2022. Accessed October 2023.