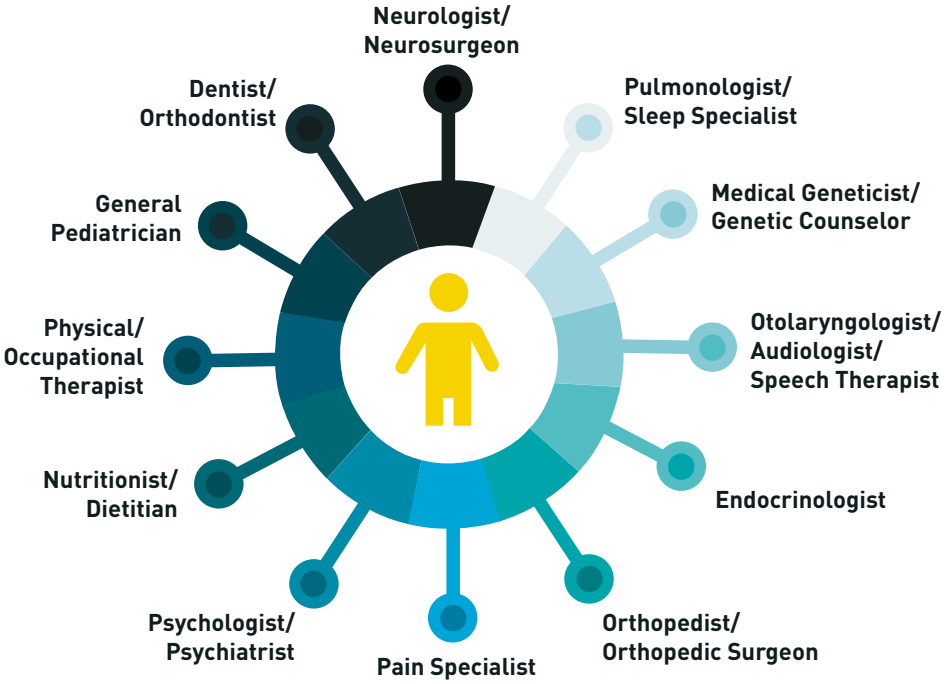
COMPREHENSIVE ACHONDROPLASIA CARE

Do your patients have a multidisciplinary care team?

Achondroplasia is caused by impaired bone growth, which can lead to multisystemic complications.[1] Effective management of achondroplasia means anticipating specific complications at every stage of development.[1] Because 80% of children with achondroplasia are born to parents of average stature,[2] they may be less familiar with the specialists they can turn to when potential complications arise, compared to parents or caregivers who have achondroplasia themselves.



Setting the proper expectations for long-term management, including assembling a skilled care network, is essential.[1,3]

A tool for proactive management

Use the second page of this document to start a conversation with caregivers and their child about:

- Understanding common, and potentially serious, complications
- Topics for caregivers to discuss with specialists
- Developing a multidisciplinary care team
- Providing referrals as needed

USE THE FOLLOWING PAGE TO WORK WITH CAREGIVERS TO HELP THEM ESTABLISH A MULTIDISCIPLINARY TEAM OF SPECIALISTS

References: 1. Ireland PJ, Pacey V, Zankl A, Edwards P, Johnston LM, Savarirayan R. Optimal management of complications associated with achondroplasia. *Appl Clin Genet*. 2014;7:117-125. **2.** Hecht JT, Bodensteiner JB, Butler IJ. Neurologic manifestations of achondroplasia. *Handb Clin Neurol*. 2014;119:551-563. **3.** Hoover-Fong J, Scott CI, Jones MC; Committee on Genetics. Health supervision for people with achondroplasia. *Pediatrics*. 2020;145(6):e20201010.

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TOGETHER, WE CAN CREATE A COMPLETE CARE TEAM

Proactive care is essential for children with achondroplasia. This page is designed to be used as a tool to start a conversation between healthcare professionals, children, and their caregivers about which doctors they are currently seeing, whom they need referrals for, and which specialists they may need to see in the future.

THE CARE TEAM SHOULD BE TAILORED TO THE INDIVIDUAL. NOT EVERY CHILD WILL NEED TO SEE EVERY SPECIALIST THROUGHOUT DEVELOPMENT.[1]

Specialist[1-4]	Discussion Topics[1,3-6]	Name and Contact Info
Medical Geneticist/ Genetic Counselor	Diagnosis, what to expect from infancy to adulthood, ongoing care, and family planning considerations	
Neurologist/ Neurosurgeon	Disproportionate head size or shape, signs of sleep apnea, poor weight gain, or neurological symptoms	
Pulmonologist/ Sleep Specialist	Breathing problems, signs of sleep apnea and sleep apnea treatment	
Otolaryngologist/ Audiologist/Speech Therapist	Recurrent ear infections, any noticeable speech delays, or hearing loss	
Endocrinologist	Achondroplasia-specific growth monitoring and developmental milestones	
Orthopedic Surgeon	Difficulty walking, excessive inward or outward curvature of the spine, bowing of the legs, or chronic pain with movement	
Psychologist/ Psychiatrist	Signs of self-esteem issues, social adaptation difficulties, or depression	
Nutritionist/Dietitian	Healthy weight management and nutritional planning	
Physical/Occupational Therapist	Hip mobility, ability to partake in physical activity, and possible environmental adaptations to promote independence	
Dentist/Orthodontist	Misalignment of bite and/or teeth and narrow palate	

FOR MORE INFORMATION, VISIT ACHONDROPLASIA.COM

References: 1. Hoover-Fong J, Scott CI, Jones MC; Committee on Genetics. Health supervision for people with achondroplasia. *Pediatrics*. 2020;145(6):e20201010. **2.** Unger S, Bonafé L, Gouze E. Current care and investigational therapies in achondroplasia. *Curr Osteoporos Rep*. 2017;15(2):53-60. **3.** Wright MJ, Irving MD. Clinical management of achondroplasia. *Arch Dis Child*. 2012;97(2):129-134. **4.** Hunter AG, Bankier A, Rogers JG, Sillence D, Scott CI Jr. Medical complications of achondroplasia: a multicentre patient review. *J Med Genet*. 1998;35(9):705-712. **5.** Gollust SE, Thompson RE, Gooding HC, Biesecker BB. Living with achondroplasia in an average-sized world: an assessment of quality of life. *Am J Med Genet A*. 2003;120A(4):447-458. **6.** Jennings SE, Ditro CP, Bober MB, et al. Prevalence of mental health conditions and pain in adults with skeletal dysplasia. *Qual Life Res*. 2019;28(6):1457-1464.

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