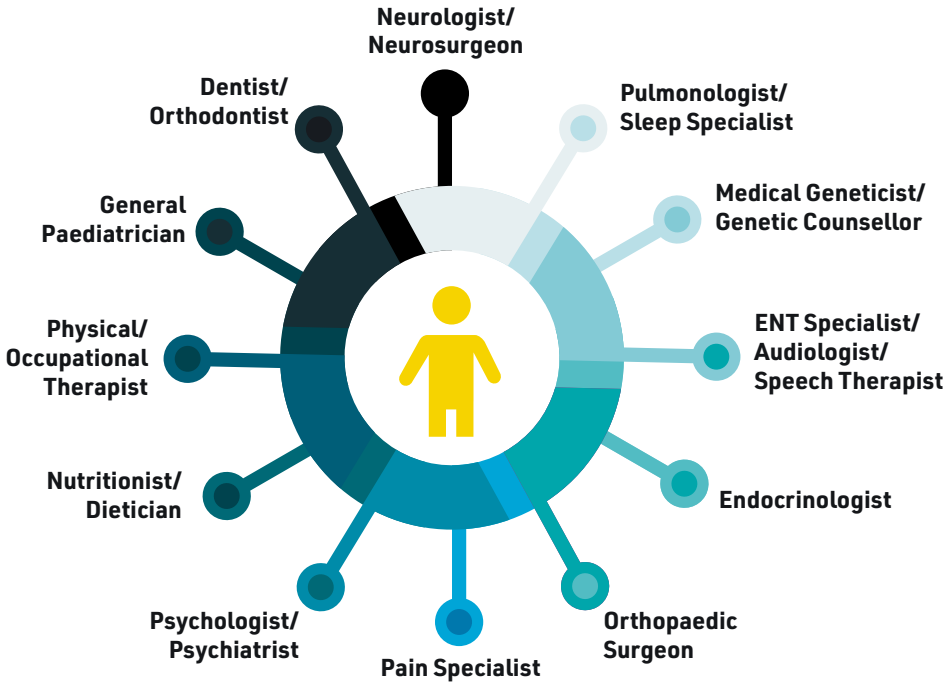
COMPREHENSIVE ACHONDROPLASIA CARE

Do your patients have a multidisciplinary care team?

Impaired bone growth caused by achondroplasia can lead to multisystemic complications.[1] The optimal management of achondroplasia means anticipating specific complications at every stage of development.[1] As **80% of children with achondroplasia are born to parents of average stature**,[2] people with achondroplasia and their caregivers may not be familiar with potential complications that can arise.



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 your patients and their caregivers 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 PATIENTS AND CAREGIVERS TO HELP THEM UNDERSTAND 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. Published online June 24, 2014. **2.** Pauli RM. Achondroplasia: a comprehensive clinical review. *Orphanet J Rare Dis*. 2019;14(1):1. **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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UNDERSTANDING THE MULTIDISCIPLINARY CARE TEAM

Proactive care is essential for people with achondroplasia. This page is designed to be used as a tool to start a conversation between healthcare professionals, patients 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 ACHONDROPLASIA PATIENT WILL NEED TO SEE EVERY SPECIALIST. SOME SPECIALISTS WILL ONLY BE NEEDED AT CERTAIN STAGES OF DEVELOPMENT.[1]

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

FOR MORE INFORMATION, VISIT HCP.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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