

Achondroplasia

Achondroplasia is a rare genetic condition that leads to well-characterized skeletal dysplasia and serious muscular, neurological, and cardiorespiratory complications.¹⁻⁸

Etiology

Achondroplasia arises from a systemic variant in the fibroblast growth receptor 3 (FGFR3) gene.^{4,8}

Epidemiology

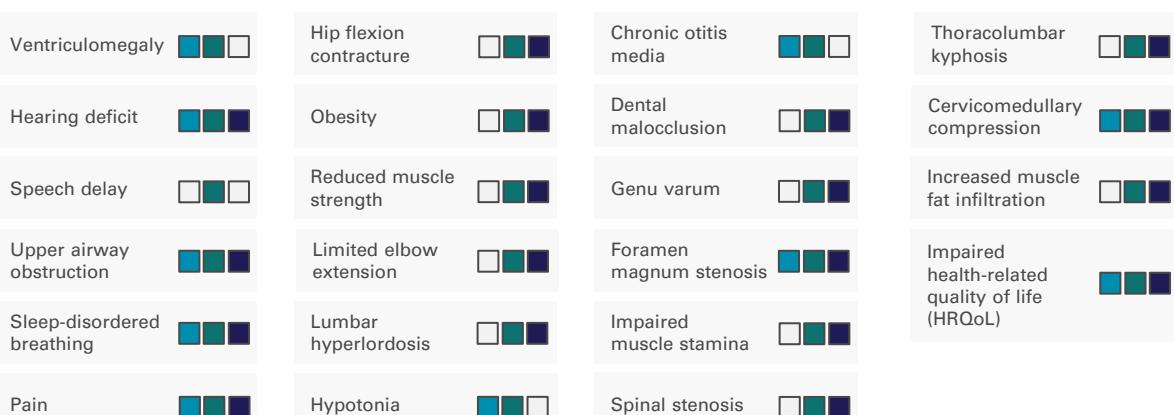
- Achondroplasia affects more than 250,000 people worldwide, varying geographically.^{2,5}
- Achondroplasia is inherited as an autosomal dominant condition with 100% penetrance; however, approximately 80% of affected individuals have parents without achondroplasia.^{4,6}

Clinical Presentation

Clinical manifestations are associated with significant, potentially life-threatening complications over the lifetime of an individual.¹⁴ Clinical manifestations and medical complications have detrimental effects on QoL, physical functioning, and psychosocial function.^{2,5,6,15}

Medical complications may vary across different stages of life.^{3,6,16-19}

Life stage impacted Infancy Childhood and adolescence Adulthood

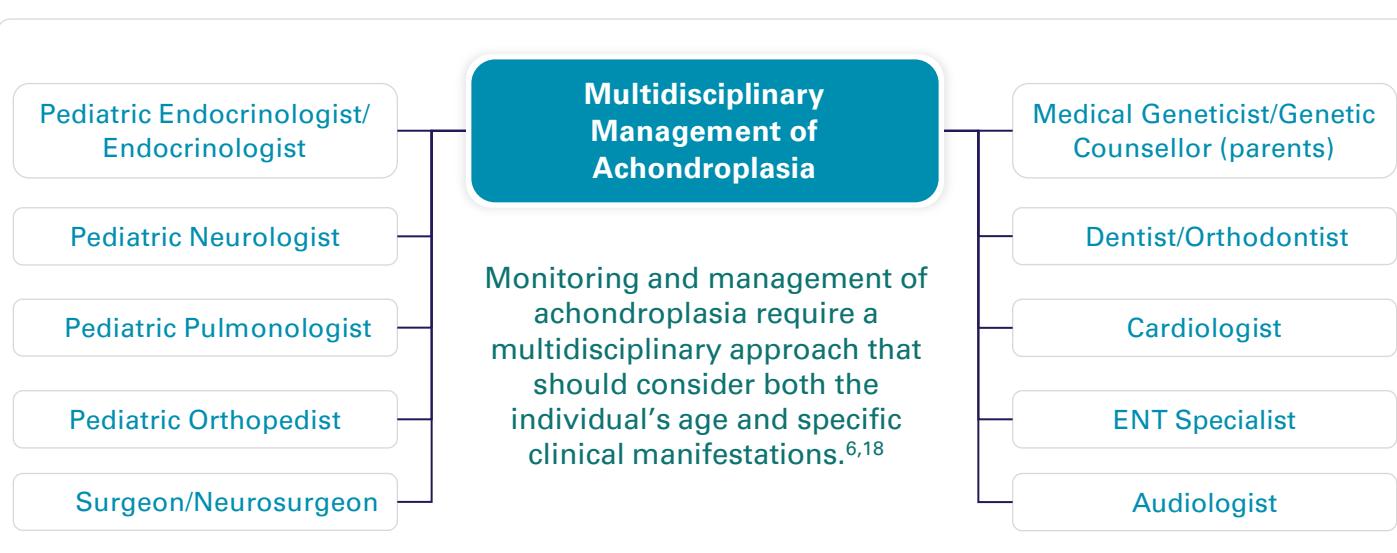
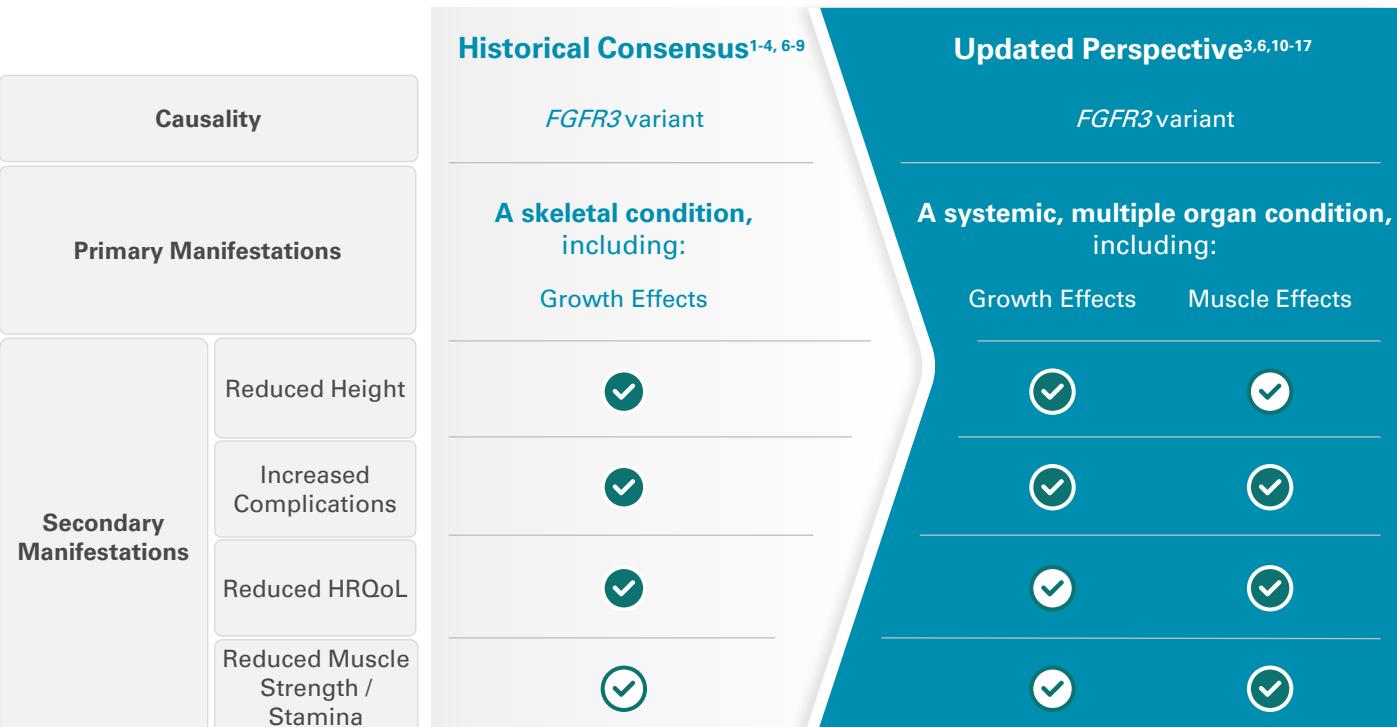


1. Ireland PJ, et al. *Appl Clin Genet* 2014; 2. Horton WA, et al. *Lancet*. 2007; 3. Sims DT, et al. *J Appl Physiol*. 2018; 4. Pauli RM. *Orphanet J Rare Dis* 2019; 5. Baujat G, et al. *Best Pract Res Clin Rheumatol*. 2008; 6. Savarirayan R, et al. *Nat Rev Endocrinol*. 2022; 7. Cormier-Daire V, et al. *Orphanet J Rare Dis*. 2022; 8. Cormier-Daire V, et al. *Orphanet J Rare Dis*. 2021; 9. Chen LJ, et al. *J Formos Med Assoc*. 2020; 10. Sallout B, et al. *Ann Saudi Med*. 2015; 11. Stoll C, Alembik Y, Dott B, Roth MP. *Eur J Med Genet*. 2022; 12. Tofts L, t al. *Am. J. Med. Genet. A*. 2021; 13. Coi A, et al. *American Journal of Medical Genetics*. ISSN 1552-4825; 14. McGraw SA, et al. *Adv Ther* 2022; 15. Constantines C, et al. *Disabil Rehabil* 2022; 16. Hoover-Fong J, et al. *Bone* 2021; 17. de Vries OM, et al. *Am J Med Genet A*. 2021; 18. Sims DT, et al. *Front Physiol*. 2018; 19. Rintz E, et al. *Int J Mol Sci*, 2022; 23(11).

Achondroplasia

Updated Perspective

Historically, achondroplasia has been viewed primarily as a skeletal condition; however, *FGFR3* is expressed in multiple tissues throughout the body, resulting in a wide range of medical complications.¹⁻⁵



ENT: Ears, Nose, Throat; FGFR3: Fibroblast Growth Factor Receptor 3; HRQoL: Health-related Quality of Life

1. Ireland PJ, et al. *Appl Clin Genet* 2014; 2. Horton WA, et al. *Lancet*. 2007; 3. Sims DT, et al. *J Appl Physiol*. 2018; 4. Pauli RM. *Orphanet J Rare Dis* 2019; 5. Murton MC, et al. *Adv Ther* 2023; 6. Savarirayan R, et al. *Nat Rev Endocrinol*. 2022; 7. Wrobel W, et al. *Int J Mol Sci* 2021; 8. Rintz E, et al. *Int J Mol Sci* 2022; 9. Krejci P, et al. *PLoS One* 2008; 10. Baujat G, et al. *Best Pract Res Clin Rheumatol* 2008; 11. Hoover-Fong J, et al. *Bone*. 2021; 12. de Vries OM, et al. *Am J Med Genet A*. 2021; 13. Sims DT, et al. *Front Physiol*. 2018; 14. Takken T, et al. *J Pediatr*. 2007; 15. Reynolds KK, et al. *Am J Med Genet*. 2001; 16. Sogos V, et al. *Arch Biochem Biophys*. 1973; 18. Bodensteiner JB. *Curr Neurol Neurosci Rep*. 2019