

Medical care is important to prevent and manage serious health problems in people with achondroplasia

What is achondroplasia?

Achondroplasia is a bone growth condition that prevents bone from replacing cartilage (smooth, firm tissue) during the growth process. It is the most common type of dwarfism.

Achondroplasia affects almost all bones in a person’s body and causes them to have:

- Short arms and legs compared to their torso
- Short-stature (height)
- Large head size
- Upper jaw, cheekbones, and eye bones that have not grown as much as the rest of their face – this can make their eyes seem very large

It does not affect thinking or level of intelligence, but people with achondroplasia may complete less years of education due to social challenges, such as being bullied.¹⁻²

What causes achondroplasia?










Achondroplasia is caused by a change in a gene that controls bone growth (genes are pieces of DNA that tell cells in your body what to do). The change leads to extra signals that **slow** bone growth. This prevents bone from replacing cartilage.



In most people with achondroplasia, the gene change happens on its own, instead of being passed down from a parent.

What health problems does achondroplasia cause?

It is important to watch for health problems caused by achondroplasia. Some problems need health care right away and some will go away on their own over time. The table below shows some of the common health problems people with achondroplasia may have during their life:^{1, 3-5}

Health problem	When it often happens
 Squeezing at the base of the skull that presses on the brainstem and spinal cord	Birth to age 4
 Ear infections	Birth to age 14
 Breathing problems when sleeping	Birth through adulthood
 Leg bowing and elbow stiffness	Age 2 through adulthood
 Long-lasting pain	Age 5 through adulthood
 Spinal cord problems	Age 5 through adulthood
 Obesity	Age 5 through adulthood
 Teeth and dental problems	Age 5 through adulthood
 Mental and emotional health problems, such as lower self-esteem, depression, and anxiety	Age 5 through adulthood

Why is it important to prevent and treat health problems caused by achondroplasia?

Health care providers focus on treating symptoms and other health problems caused by achondroplasia. It's vital for people with achondroplasia to get health care from a team of specialists to prevent, find, and treat health problems. This health care may include:

Surgeries, such as:



- Neurosurgery to correct problems like fluid on the brain or pressure on the spinal cord
- Orthopaedic surgery to correct problems with bones or add length in the arms or legs to help movement
- Ear, nose, and throat (ENT) surgery to remove the tonsils to help with sleep apnea

Having tubes put in the ears to prevent repeat ear infections



Care from a cardiologist (heart doctor) or pulmonologist (lung doctor) to treat any heart and lung problems



Care from an orthodontist to correct teeth or mouth problems



Therapy, support, and guidance from a range of providers, including physical and occupational therapists (PTs and OTs) and mental health providers



Regular care from a team of specialists can help people with achondroplasia live independently.

Specialized care can benefit patients and society

Specialized care can make it possible for people with achondroplasia to live independently and have equal opportunities, including to:³⁻⁷

- Carry out daily activities on their own, such as to move around, drive, and use the bathroom
- Carry out school activities
- Help lessen pain
- Get and keep a job



Learn more, get support, and advocate!

Visit achondroplasia.com

References

1. Pauli RM. Achondroplasia: a comprehensive clinical review. *Orphanet Journal of Rare Diseases* 2019;14:1.
2. 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 Aug 1;120A(4):447-58. doi: 10.1002/ajmg.a.20127. PMID: 12884421.
3. BioMarin. 111-501 (LIAISE) Observational Study Report. 2020.
4. Fernández Arregui. Social stigmatization in achondroplasia. Available at: https://www.fundacionalpe.org/images/alpe/library/social_stigmatization_english.pdf. 2010. Last accessed: April 2021.
5. Jennings SE, Ditro CP, Bober MB, et al. Prevalence of mental health conditions and pain in adults with skeletal dysplasia. *Quality of Life Research* 2019;28:1457-1464.
6. Roizen N, Ekwo E, Gosselink C. Comparison of education and occupation of adults with achondroplasia with same-sex sibs. *American Journal of Medical Genetics* 1990;35:257-260.
7. Pan W, Guillen-Navarro E, Queisser-Wahrendorf A, et al. Lifetime impact of achondroplasia on health-related quality of life (HR-QoL) and healthcare resource use: Interim results from a multinational study. *ACMG Annual Clinical Genetics*