

Achondroplasia care: Standards across Europe

This document is intended to provide an overview of the standards of care for people living with achondroplasia across Europe.

What is achondroplasia?

- Achondroplasia is a rare genetic condition that **affects how the majority of the bones in the body grow**, occurring in about **one in every 25,000 live births**.ⁱ
- While the most visible effect of achondroplasia may be **short stature, impaired bone growth can have a serious impact on health** – complications can include foramen magnum compression, sleep apnoea, bowed legs, mid-face hypoplasia, permanent sway of the lower back, spinal stenosis and recurrent ear infections.ⁱⁱⁱ

Diagnosing achondroplasia

- Diagnosing achondroplasia primarily takes place prenatally or within 1 month of birth. There is some variation between countries, with France and the UK diagnosing most cases prenatally, and Spain and Italy confirming suspicions at Day 0.
- The diagnostic pathway for achondroplasia is largely consistent between countries, beginning with an ultrasound, which can be enough to diagnose achondroplasia prenatally, supported by CT scans where needed and available. Signs of achondroplasia should then prompt further ultrasound investigations and confirmation of diagnosis through single gene testing, but this is not available or widely used in all countries.
- After birth, genetic testing is the best way to confirm a clinical diagnosis, but the role of postnatal testing is not standardised across countries.
- Accurate diagnosis and timely referral enables an experienced multi-disciplinary team to best support the families and to plan appropriate management for the child. However, there is often a delay, with referral taking more than 2 months in 1/3 of cases.
- The delays and variable times of referral can be related to: the structure of the local healthcare organisations, lack of awareness of comorbidities among non-experts, and the varied resources and facilities between countries.

Achondroplasia standard of care

- There is currently a lack of consensus around the optimal management of achondroplasia, and **significant regional variation exists around Europe**, which can depend on areas of local expertise.
- This variation is apparent from the time of diagnosis and is dependent upon the structure of the local healthcare system, the involvement of advocacy support groups, and different cultural backgrounds.
- Healthcare management of achondroplasia in Europe can often be reactive, with many specialist HCPs involved in care required, due to the complications that may arise from the condition.
- Parents of children with achondroplasia can subsequently face a significant upheaval, managing a number of medical appointments and surgeries with different healthcare specialists. This can require travel over long distances to access care.
- Many children do not have access to coordinated care through a multidisciplinary care team (MDT). As a result, it can be a challenge for parents to access preventative care for their children from HCPs, with many having to rely on management of complications associated with achondroplasia as they arise.
- In some countries, such as Italy and Spain, **limb lengthening** is considered a potential treatment to increase both height and reach.
- The decision to undergo limb lengthening can be very difficult for families and children/adolescents with achondroplasia. It is a **big family commitment** to undergo this procedure, often with significant travel and time investment required.

Achondroplasia caregiver survey

In a surveyⁱⁱⁱ of European parents of children with achondroplasia, **over half** had seen more than **five different healthcare specialists** in the past two years.

Over a quarter of parents must travel more than **60 miles** to see their child's primary physicianⁱⁱⁱ

Across Europe, there is great variety in the specialism of people with achondroplasia's primary physician.ⁱⁱⁱ



41% have a clinical geneticist as their primary physician



8% Endocrinologist



18% Paediatrician



5% Obstetrician










22% Orthopaedist



6% Rehabilitation specialist

Care variance across Europe

		UK	GER	FR	ESP	ITA
 National management guidelines	Currently in development	●			●	●
	National management guidelines exist		●	●		
 Patient Advocacy Groups	Currently play an active role in developing care pathways		●		●	●
	MDT approach established	●			●	●
 Multidisciplinary team approach	MDT approach not established		●	●		
	Regional centres of excellence established		●		●	●
 Centres of excellence	Regional centres of excellence not established	●		●		
	Specialist paediatrician (endocrinologist or geneticist)		●		●	
 Speciality of physician coordinating care	Clinical geneticist			●		
	Either a paediatrician or endocrinologist					●
	It varies - clinical geneticist, paediatric endocrinologist or paediatric respiratory care	●				
	Very common					●
 Limb lengthening	Common		●		●	
	Rare	●		●		
	Framework exists			●		
 Framework of care to support transition from child to adult services	Framework does not exist	●	●		●	●

Burden of care

- The burden of care of achondroplasia can have a detrimental effect on people living with achondroplasia and their families.ⁱⁱⁱ
- Over half of parents of children with achondroplasia in Europe said that their child's achondroplasia had an effect on their own emotional wellbeing (54%).ⁱⁱⁱ
- While almost two thirds (65%) had never been offered psychosocial support since their child was diagnosed with achondroplasia.ⁱⁱⁱ

Our call to action

- We would like to see a consistent approach to achondroplasia care across Europe.
- We recommend the development and roll-out of clear guidelines and management pathways to lessen the significant burden of care for those with achondroplasia.
- We recommend that all people living with achondroplasia deserve a dedicated multidisciplinary team for the management of their condition, particularly when transitioning from childhood to adulthood where co-ordinated care often stops.

Glossary

Foramen magnum compression

Typically, the brain stem and spinal cord pass through the foramen magnum, an opening at the base of the skull. Foramen magnum stenosis happens when the opening narrows, potentially putting pressure on the brain or spinal cord.^{iv}

Sleep apnoea

Temporary cessation of breathing that occurs most often during sleep. Untreated obstructive sleep apnoea and central sleep apnoea may both have serious developmental consequences in children with achondroplasia.ⁱ

Bowed legs

A leg bowed outward at or below the knee, also known as genu varum; this bowed effect can contribute to back pain and affect mobility.ⁱ

Mid-face hypoplasia

Where some bones in the middle of the face such as the upper jaw, cheekbones and eye sockets do not grow as much as the rest of the face which can cause sleep apnoea.^v

Permanent sway of the lower back

Most children with achondroplasia develop an exaggerated curve in their lower back when they begin to stand and walk. When this is pronounced, there may be an increased incidence of pain and spinal stenosis as well as a noticeable impact on mobility.ⁱ

Spinal stenosis

A narrowing of the space within the spine, which can put pressure on the nerves that travel through the spine and can subsequently cause issues such as persistent leg weakness, co-ordination challenges, changes in gait, and development of bladder or bowel incontinence.ⁱ

Multidisciplinary team (MDT)

A multidisciplinary team involves various healthcare professionals, from a range of specialisms, working together to deliver comprehensive patient care. In achondroplasia, this could involve paediatricians, clinical genetics, orthopaedics and physiotherapy. This can consolidate the care provided to help avoid multiple hospital appointments and maintain patient safety.

[Learn more at achondroplasia.com](https://www.achondroplasia.com)

References

ⁱ Pauli R M 'Achondroplasia: a comprehensive clinical review' Orphanet J Rare Dis. 2019;14:1.

ⁱⁱ Ireland P J et al. 'Optimal management of complications associated with achondroplasia' Applied Clinical Genetics. 2014;7:117-125.

ⁱⁱⁱ Achondroplasia Caregiver Survey – A global perspective on diagnostic pathways, healthcare management and personal impact from carers of children with achondroplasia -- Presented at the ACMG Annual Clinical Genetics Meeting: April 13–16, 2021

^{iv} <https://www.cedars-sinai.org/programs/neurology-neurosurgery/clinical/pediatrics/neurosurgery/skeletal-dysplasia/foramen-magnum-stenosis.html>

^v <https://www.stlouischildrens.org/conditions-treatments/plastic-surgery/midfacial-hypoplasia#:~:text=What%20is%20midface%20hypoplasia%3F,the%20rest%20of%20the%20face.>