

UK consensus guidelines for multidisciplinary care of children and young people with achondroplasia: a modified Delphi process.

The UK Achondroplasia Network · 2026

UK Consensus Guidelines for the Care of Children and Young People with Achondroplasia: Study Guide

This study guide provides a comprehensive overview of the first UK-specific multidisciplinary clinical recommendations for managing achondroplasia (ACH) in children and young people (CYP). These guidelines were developed by the UK Achondroplasia Network using a modified Delphi process involving 25 multidisciplinary experts to ensure standardized, proactive, and equitable care.

Section 1: Key Concepts and Clinical Framework

1. Etiology and Pathophysiology

- **Genetic Basis:** ACH is an autosomal dominant disorder caused by gain-of-function variants in the *fibroblast growth factor receptor 3 (FGFR3)* gene.
- **Mechanism:** The *FGFR3* gene normally inhibits endochondral ossification. The gain-of-function variant results in abnormal bone development and disproportionate short stature.
- **Incidence:** Approximately 1 in 25,000 live births (roughly 26 new diagnoses in the UK in 2023).

2. Clinical Presentation

- **Physical Features:** Disproportionate short stature, rhizomelic (proximal) limb shortening, macrocephaly with frontal bossing, and midface hypoplasia.
- **Inheritance Risk:**
 - **One parent with ACH:** 50% chance of ACH, 50% average stature.
 - **Both parents with ACH:** 50% chance of ACH (heterozygous), 25% chance of average stature, and 25% chance of biallelic variants (lethal in the perinatal period).

3. Developmental Stages and Monitoring Priorities

Developmental Stage	Age Range	Key Complications to Monitor
Infancy	<12 months	Foramen magnum stenosis (FMS), hydrocephalus, sudden death, kyphosis, sleep apnoea.
Preschool	1-4 years	Obstructive sleep apnoea, middle ear infections (OME), hearing deficits, gross motor delay, tibial bowing.
Primary School	5-11 years	Dental crowding/malocclusion, psychosocial difficulties, limb alignment, obesity.
Secondary School	11-18 years	Spinal stenosis, claudication pain, fatigue, social media/body image pressures, transition to adult care.

4. Critical Medical Monitoring Protocols

- **Neuroimaging (MRI):** A routine MRI of the brain and spine is recommended **within 3 months of birth** to screen for FMS and hydrocephalus.
- **Respiratory Assessment:** Respiratory polygraphy/polysomnography should be conducted **by 6 months of age**.
- **Hearing:** Newborn screening followed by a full audiologic examination **by 12 months of age**, with 6-monthly reviews during preschool years.
- **Orthopaedics:** A formal review by a specialist orthopaedic surgeon should occur **by age 7**.
- **Genetics:** Genetic counseling for the individual should be offered at **ages 13-14** to discuss inheritance and reproductive choices.

Section 2: Short-Answer Practice Questions

Q1: At what point in gestation does ACH usually become apparent on a prenatal ultrasound, and what are the characteristic features? Answer: ACH is typically suspected after 25 weeks of gestation. Features include shortened long bones, relative macrocephaly, and a widened proximal femoral diaphysis-metaphysis angle. (Routine 12-week and 20-week scans usually appear normal).

Q2: What specific handling advice should be given to parents of infants with ACH to reduce the risk of neurological injury? Answer: Caregivers should avoid the use of slings, baby bouncers, bouncing toys, and trampolines, as these can increase the risk of injury related to foramen magnum stenosis and contribute to fixed kyphosis.

Q3: Why is respiratory polygraphy preferred over full polysomnography in some clinical settings for ACH? Answer: While polysomnography is the gold standard, polygraphy is more accessible and

easier to use. It effectively assesses the primary concerns in ACH: obstructive apnoea, central apnoea, and hypoventilation, without requiring an electroencephalogram (EEG).

Q4: What is the "critical period" for surgical intervention in lower limb alignment, and why does it exist? *Answer:* The critical period is typically before ages 11–12. Beyond this age, simple surgical interventions like "guided growth" are unlikely to be effective in influencing limb alignment.

Q5: What are the clinical indicators of cervicomedullary compression (FMS) in infants? *Answer:* Indicators include apnoea, hyperreflexia, clonus, delayed milestone acquisition (based on ACH-specific charts), difficulty swallowing, poor weight gain, and irritability.

Q6: What dental and orthodontic issues are common in primary school-aged children with ACH? *Answer:* Due to relative maxillary hypoplasia, children often develop dental crossbites and crowding. A specialist orthodontic review is recommended at approximately 10 years of age.

Q7: How is "shared care" defined in the UK recommendations? *Answer:* It involves a partnership between a specialist regional ACH centre and a named consultant within a local paediatric service (alongside GPs and health visitors) to aid holistic care and local support.

Section 3: Essay Prompts for Deeper Exploration

1. **The Multidisciplinary Approach:** Analyze the composition and necessity of the Multidisciplinary Team (MDT) in the care of children with ACH. Discuss how the integration of specialties—such as neurosurgery, orthopaedics, occupational therapy, and psychology—contributes to reducing preventable morbidity and enhancing the quality of life.
2. **Psychosocial Transitions:** Explore the evolving psychosocial needs of a person with achondroplasia from early childhood through adolescence. Consider the impacts of school entry, the emergence of complex emotions around age seven, and the challenges of social media and independence during the secondary school years.
3. **Advancements in Health Technology:** Discuss the role of emerging therapies (such as vosoritide and other FGFR3-selective inhibitors) in the management of ACH. What are the clinical and ethical considerations for specialist teams when introducing these technologies to families?
4. **Managing the Transition to Adulthood:** Evaluate the recommended process for transferring a young person from paediatric to adult services. Why is it essential that this process begins years before the actual transfer, and what non-medical factors (e.g., employment, housing, benefits) must be addressed?

Section 4: Glossary of Important Terms

- **Achondroplasia (ACH):** The most common form of skeletal dysplasia, characterized by disproportionate short stature and caused by *FGFR3* gene variants.

- **Achondroplasia Foramen Magnum Score (AFMS):** A scoring system used by neuroradiologists to grade the severity of stenosis at the foramen magnum; AFMS 4 typically requires surgical decompression.
- **Cervicomedullary Compression:** Pressure at the junction of the brainstem and spinal cord, often caused by foramen magnum stenosis.
- **Claudication Pain:** Pain in the limbs (common in the lumbar spine in ACH) that occurs during mobilization and is often relieved by squatting or resting; a sign of spinal stenosis.
- **Delphi Process:** A structured communication technique/interactive forecasting method which relies on a panel of experts to reach a consensus.
- **Endochondral Ossification:** The process by which growing cartilage is replaced by bone to form the skeleton.
- **Foramen Magnum Stenosis (FMS):** Abnormal narrowing of the opening at the base of the skull, posing a risk of motor impairment, apnoea, and sudden infant death.
- **Midface Hypoplasia:** Underdevelopment of the middle part of the face, contributing to dental crowding and obstructive sleep apnoea.
- **Otitis Media with Effusion (OME):** A collection of non-infected fluid in the middle ear ("glue ear"), common in ACH due to shorter, more horizontal eustachian tubes.
- **Rhizomelic:** Referring to the shortening of the most proximal segment of a limb (e.g., the humerus in the arm or femur in the leg).
- **Thoracolumbar Gibbus:** A sharp angulation or prominence of the spine, often seen in infants with ACH before they begin walking.
- **Vosoritide:** A C-type natriuretic peptide (CNP) analogue designed to increase stature in children with open growth plates (approved in the US/EU, but not yet licensed in the UK as of the guideline date).