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

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ABSTRACT

Background Achondroplasia (ACH), the most common skeletal dysplasia, arises from gain-of-function variants in the *fibroblast growth factor receptor 3* gene. Children with ACH experience lifelong medical, functional and psychosocial challenges requiring coordinated and anticipatory care. Although international guidance exists, the UK lacks national clinical care recommendations specific to its healthcare systems.

Objective To develop UK-specific, multidisciplinary clinical recommendations for the care of children and young people (CYP) with ACH.

Methods The UK Achondroplasia Network developed guidance in stages: stakeholder mapping of the care pathway, integration of contemporary literature with clinical expertise to draft age-specific guidance and Delphi statements, and a modified Delphi process with 25 multidisciplinary experts. The Delphi process involved two voting rounds and an in-person meeting, with consensus defined as ≥80% agreement.

Results In the first Delphi round, all 20 statements achieved consensus; nine achieved 100% agreement. To strengthen consensus, after meeting in person, 17 statements were refined (four were divided into two statements), one created and one removed, resulting in 24 statements for Round 2; all achieved consensus, with 21 reaching 100% agreement. The guidance outlines age-specific monitoring and referral from infancy to adolescence. Recommendations address medical management of complications, psychosocial support, educational planning and transfer to adult care.

Conclusion These are the first UK-specific multidisciplinary recommendations for the care of CYP with ACH. Aligned with international best practices and tailored to UK healthcare systems, they support anticipatory care, promote independence and enhance health and psychosocial outcomes. The guidelines offer a foundation for service planning, standardisation and equitable care.

INTRODUCTION

Achondroplasia (ACH) is the most common form of skeletal dysplasia, with an estimated incidence of approximately 1 in 25 000 live births,[1 2] corresponding to about 26 new diagnoses in the UK in 2023.[3–5] ACH is an autosomal dominant genetic disorder caused by gain-of-function variants in the *fibroblast growth factor receptor 3* (*FGFR3*) gene, which normally inhibits endochondral ossification,[6–8] resulting in abnormal bone development and disproportionate short stature.

Clinically, ACH is characterised by short stature, rhizomelic upper limb shortening, macrocephaly with frontal bossing and midface hypoplasia[6 9–11] and is associated with multisystem complications (online supplemental material 1).[6 9 10] In addition to medical needs, individuals often face functional and psychological challenges that can impact their quality of life. Taken together, comprehensive multidisciplinary care is essential to support the health and well-being of individuals with ACH.[12–15]

Several international guidelines provide frameworks for the diagnosis and management of children with ACH,[10 12 14 16] but their application in clinical practice remains inconsistent,[17] and healthcare systems differ across countries. While national guidance exists in Australia, Italy, Japan and Spain,[18–21] the UK currently lacks standardised clinical recommendations for the care of children and young people (CYP) with ACH. A recent coroner's Regulation 28 report highlighted the need for nationally agreed pathways to reduce preventable morbidity.[13]

In response, the UK Achondroplasia Network has developed cohesive, UK-specific clinical guidance. This manuscript presents the first UK multidisciplinary recommendations for the care of CYP with ACH developed with contributions from all major specialties. These recommendations integrate established international best practices within the context of UK healthcare systems and align with European guiding principles.

We acknowledge that clinical practices vary internationally, and the scope of these recommendations is focused on delivering evidence-based, contextually appropriate guidance for UK healthcare professionals (HCPs). By establishing a consistent and proactive care approach, these guidelines aim to optimise health outcomes and improve the quality

of life for CYP with ACH and by extension, into adulthood, across the UK.

METHODS

The development of the UK guidance for diagnosing, monitoring and managing CYP with ACH was undertaken in three stages: (1) stakeholder collaboration to define the care pathway, (2) development of age-specific guidance informed by literature and expert consensus and (3) a modified Delphi process to finalise recommendations.

1. In 2018, a multidisciplinary expert group of 27 UK HCPs mapped the ACH care pathway from diagnosis to transition, with input from two patient advocacy groups (online supplemental material 2).
2. Following publication of international ACH guidelines,[10 12 14 16] the pathway was revisited in March 2025 by 25 experts from the UK Achondroplasia Network, representing 16 specialties (online supplemental material 2). The clinical lead for each ACH unit in the UK was invited to contribute, including subspecialty team representatives, ensuring geographic and specialty diversity. The aim was to produce a structured, age-specific framework aligned with best practice in the UK.

   To inform the framework, a non-systematic PubMed search was led by the lead authors (March 2025) using combinations of keywords and MeSH terms (eg, 'achondroplasia', 'guidelines', 'management', 'paediatric', 'skeletal dysplasia'). The search was limited to English-language publications and publications in the last 5 years were prioritised; additional references were identified from citations. Where evidence was limited, expert opinion and consensus were used to guide recommendations. Care was segmented into age-specific categories: infancy, preschool, primary school and secondary school ages.
3. A modified Delphi process was conducted to develop UK-specific consensus recommendations, capturing expert opinion transparently to ensure statements reflected evidence and considerations for UK clinical practice.

   The 25 multidisciplinary members who contributed to the age-specific guidance development served as the Delphi panel. Two clinical leads (MSC, TC) drafted an initial set of statements (online supplemental material 3) informed by international guidelines, current literature and clinical experience. Panellists anonymously rated their agreement with each statement using a five-point Likert scale, with consensus pre-defined as ≥80% agreement (ratings of 'strongly agree' or 'agree'). Two rounds of voting were conducted between 15 and 28 April 2025, and a facilitated interim meeting refined statements requiring clarification. This iterative process ensured that all final statements reflected a high level of expert agreement and were grounded in the best available evidence. The guideline was reviewed by four parents of children with ACH to ensure it addressed the needs of children, families and caregivers. Full details of the Delphi process are provided in online supplemental material 4.

CLINICAL GUIDANCE

Diagnosis and perinatal period

Prompt diagnosis and referral to specialist care are essential for planning appropriate support for babies with ACH and their families.[12–14]

Up to 80% of infants with ACH are born to parents of average stature,[7] with the remaining born into families where one or both parents have ACH. As an autosomal dominant condition, each child of a parent with ACH has a 50% chance of inheriting the condition, with equal probability for males and females. If both parents have ACH, there is a 25% chance the child will have average stature, a 50% chance of ACH (inheriting a single *FGFR3* variant) and a 25% chance of inheriting biallelic variants (either homozygous or compound heterozygous), which is typically lethal in the perinatal period.[10 22 23]

Pre-implantation diagnosis

For families with a known history of ACH (whether a parent has ACH or a sibling has been diagnosed), support can begin prior to conception with appropriate genetic counselling to discuss inheritance (eg, parental gonadal mosaicism), the potential for prenatal diagnosis and the possibility of pre-implantation genetic testing, if appropriate (figure 1).[10 22]

Prenatal diagnosis

Suspicion of ACH typically arises after 25 weeks of gestation when ultrasound features (eg, shortened long bones, relative macrocephaly and a widened proximal femoral diaphysis-metaphysis) become apparent.[12 14 23 24] The routine 12-week dating scan and 20-week anomaly scan usually appear normal in fetuses with ACH. In pregnancies without a family history of ACH, ACH may only be suspected during later pregnancy scans performed for other indications, such as fetal growth restriction (eg, small for gestational age) or incidental findings during scans for unrelated indications (eg, low-lying placenta). When prenatal ultrasound features suggestive of ACH are identified, referral to a fetal medicine centre, equipped with advanced scanning capabilities and the potential for invasive testing, is warranted.[25] Diagnosis can be confirmed via non-invasive prenatal diagnosis (NIPD) or invasive genetic testing, with fetal medicine and clinical genetics support, to ensure appropriate counselling, interpretation and follow-up.[10 20]

Prenatal referrals

Obstetric and fetal medicine teams provide ongoing care during pregnancy. The neonatal team should meet the family before birth and be present at delivery, as newborns with ACH may require immediate medical support or resuscitation.[22]

Referral to a specialist team experienced in ACH is recommended as soon as possible after a diagnosis of ACH to maximise clinical and supportive outcomes.[13 14 16 20] In cases of prenatal diagnosis, engagement with a member of the ACH specialist team, which may be the clinical nurse specialist or occupational therapist, should be established before birth. The care plan should be developed collaboratively with the multidisciplinary team (MDT) and local paediatrician, where applicable. Additional components of the care plan may include contacts for the local paediatrician and specialist ACH team, information on routine medical monitoring, psychological support services available, and UK-based support organisations (online supplemental material 5).[10 19 20 23] Anticipatory guidance, including information about appropriate car seats, positioning and handling advice, and basic life support training should be provided before discharge to ensure parents feel informed and prepared for the transition to home care.[6 10 13 22 23]

Pregnancy in mothers with ACH

In pregnancies where the mother has ACH, referral to additional specialists is essential to support good maternal and fetal outcomes.[10 19 23 25] These may include fetal-maternal medicine specialists, obstetric anaesthetists and potentially respiratory

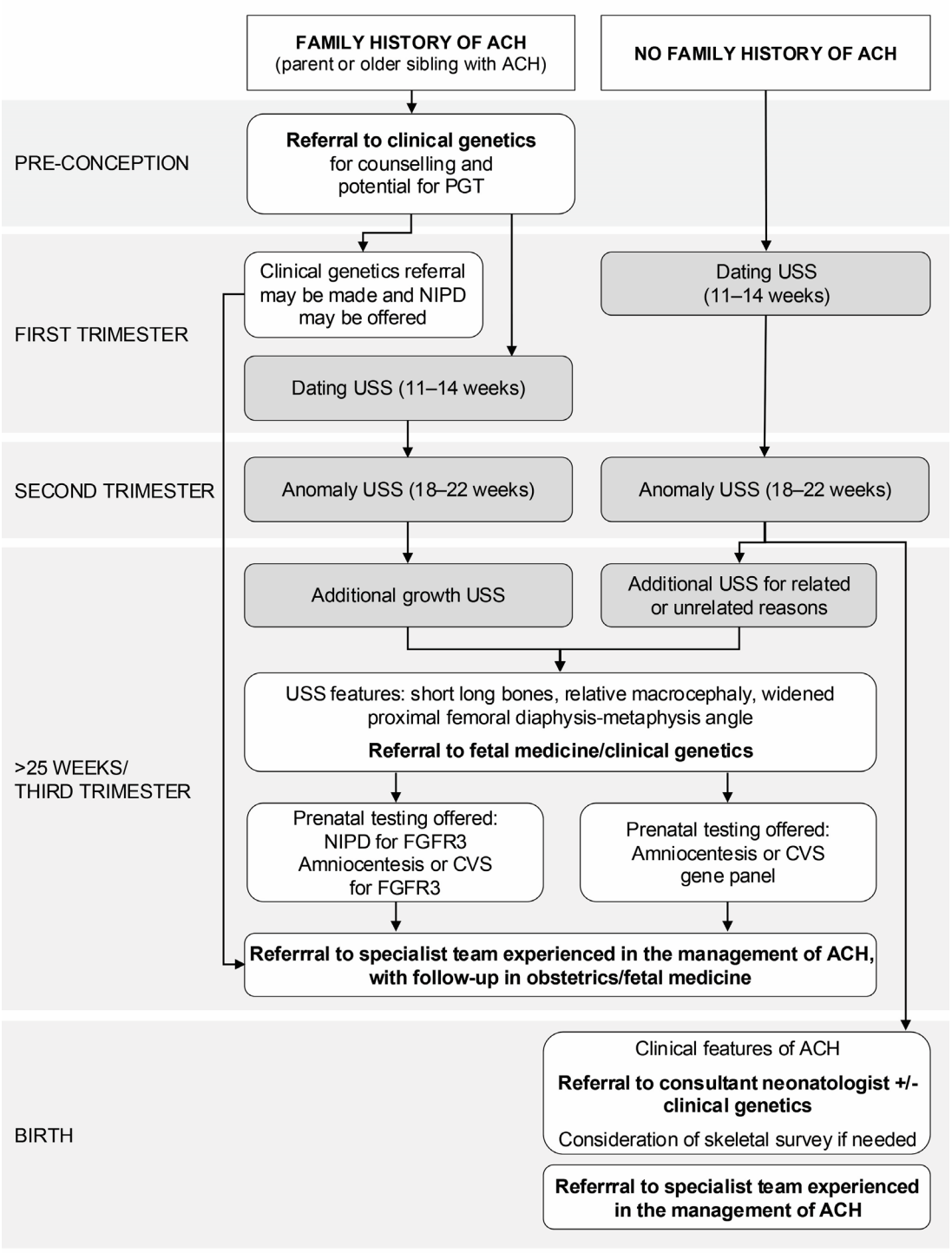

Figure 1 Referral pathways for the diagnosis of achondroplasia. Flow diagram outlining referral pathways in the diagnosis of ACH in pregnancies with and without a family history of ACH. Dark grey boxes denote routine USS during pregnancy. Bold text highlights referrals. ACH, achondroplasia; CVS, chorionic villus sampling; FGFR3, fibroblast growth factor receptor 3; NIPD, non-invasive prenatal diagnosis; PGT, pre-implantation genetic testing; USS, ultrasound scan.

teams. Genetic counselling and clinical genetics support are particularly important if the partner also has a skeletal dysplasia.[10 22 23] Care should be consultant-led in a tertiary centre, with delivery typically via elective caesarean section.[10 19 23 25]

Postnatal diagnosis and referral

For infants without a prenatal diagnosis of ACH, postnatal diagnosis is based on classical clinical and radiological features.[12] While genetic testing may be initiated to confirm the diagnosis, referral to the specialist medical team should not be delayed while awaiting test results.

AGE-RELATED HEALTHCARE SUPPORT

Complications associated with ACH change throughout the lifespan and careful monitoring and timely intervention can help reduce complications, support optimal quality of life and

Table 1 Age-related complications and clinical monitoring frequency in children with achondroplasia

	Development stage	Infancy	Preschool	Primary school	Secondary school
	Age	<12 months	1–4 years	5–11 years	11–18 years
Medical complications	Hydrocephalus				
	Sudden death				
	Cervicomedullary compression				
	Kyphosis				
	Hearing deficit				
	Otitis media				
	Sleep disordered breathing				
	Upper airway obstruction				
	Gross motor delay*				
	Speech delay				
	Pain				
	Tibial bowing				
	Lordosis				
	Malocclusion				
	Obesity				
	Psychosocial difficulties				
	Spinal stenosis				
Medical monitoring (specialist team)	Growth parameters	Birth and at least every 6 months	Every 6–12 months	Annually or as clinically indicated	Every 12–24 months
	Developmental assessment	Birth and at least every 6 months	Every 6–12 months	Every 12–24 months	–
	Neurological assessment	Birth and at least every 6 months	Every 6–12 months	As indicated	As indicated
	Neuroimaging MRI	By 3 months With repeat as needed or per local policy	As indicated	As indicated	As indicated
	Respiratory polysomnography	By 6 months or earlier if clinically indicated	As indicated or per local policy	As indicated	As indicated
	Spinal deformities	At least every 6 months	Every 6–12 months	Every 12–24 months	Annually
	Hearing assessment	Newborn hearing test	Audiology at 1 year old and every 6 months	As indicated	As indicated
	Limb deformities	–	Every 6–12 months	Orthopaedic review by 7 years old or as clinically indicated	As indicated
	Dental assessment	–	Paediatric dentist review at 1 year old	Orthodontic evaluation at approximately 10 years old	
	Psychology assessment	–	As indicated	As indicated	Annually
	Genetic counselling	–	–	–	From age 13/14 years

This table outlines common medical complications and specialist follow-up across four developmental stages: infancy, preschool, primary school and secondary school. In the upper panel, shading highlights the stage when complications are most common. The lower panel presents the recommended frequency of monitoring with the specialist team.
*Compared with achondroplasia-specific norms.

promote long-term well-being (table 1 and online supplemental material 6).

Infancy (<12 months old)

General care principles

Consultations with the specialist centre should occur at least every 6 months until the infant reaches 1 year of age, with shared care with local neonatal, paediatric or community teams between visits. Establishing shared-care relationships at an early stage is important as these are crucial throughout childhood. Each consultation should include assessment of growth, development, musculoskeletal status and neurological function with referral to appropriate specialists as indicated (online supplemental material 6).

Families, general practitioners, health visitors and secondary care teams should be familiar with the acute medical needs of infants with ACH. Signs of apnoea, rapidly increasing head circumference, failure to thrive and developmental delay relative to ACH-specific standards require prompt liaison with the specialist team.

Consultations with specialised services

An initial consultation with a doctor experienced in ACH management should occur as soon as possible following diagnosis.[10] This meeting is important for explaining the diagnosis and likely implications for the child and family, building a relationship with the family, introducing the MDT and establishing the importance of ongoing medical consultations. It is also an opportunity to address questions and gauge interest concerning potential treatments and clinical trials, support the families' process of adjustment to diagnosis, and verify the diagnosis if necessary. An expert in ACH management should explain and

counsel regarding medical therapeutic options available as early as possible. Families should be informed of any clinical trial opportunities as they arise.

Families should receive accessible information about ACH-specific challenges, including advice on safe handling techniques, appropriate positioning and the use of supportive equipment (such as lie-flat car seats and buggies) to ensure the child's safety and comfort.[10 13 18 23] Caregivers should be advised to avoid the use of slings, baby bouncers, bouncing toys and trampolines, as these may increase the risk of neurological injury related to foramen magnum stenosis (FMS) and contribute to the development of fixed kyphosis; further discussion with the specialist team is recommended if there are specific individual needs.[10 18 23 26] Families should also be signposted to ACH support organisations, for valuable resources and peer support.

A comprehensive clinical assessment should be completed, including growth measurements (length, weight and head circumference), developmental assessment and musculoskeletal and neurological examination.[10 23 27] All measurements should be interpreted using ACH-specific reference standards. ACH-specific growth and developmental charts should be used and shared with families for inclusion in the Personal Child Health Record ('red book').[28 29] An MRI scan should be scheduled within the first 3 months of birth to screen for radiological features of FMS and hydrocephalus. Respiratory polygraphy/polysomnography should be conducted by 6 months of age. Ongoing medical monitoring is described in box 1.

Psychosocial support

Early signposting to ACH support organisations can reduce parental isolation and support adjustment. These support networks help foster future resilience in both caregivers and children.[10] Offering psychological support to caregivers at this stage can help provide emotional containment and support their own adjustment, which will in turn help the child's development of positive self-esteem, body image and identity.[10] Families should be encouraged to engage with local authority services and family hub sites providing universal healthcare services.

Preschool (1–4 years old)

The preschool years, spanning ages 1–4 years old, mark an important period in the growth and development of children with ACH. During this period, children progress through significant developmental milestones, work towards independence in self-care and adapt to their unique physical capabilities. Families and HCPs should closely monitor and support these children, addressing emerging challenges and meeting their medical needs (online supplemental material 6). Particular attention should be given to complications such as obstructive sleep apnoea, middle ear infections, hearing difficulties and orthopaedic issues (box 2). Discussions regarding potential medical therapeutic options and clinical trial opportunities should remain ongoing.

Independence and home environment

Preschoolers start to develop their independence, learning how to dress themselves, use the toilet, feed themselves and initiate awareness of increasing their ability to partake in self-care needs and everyday tasks. Specialist paediatric occupational therapists and/or physiotherapists, where available, will collaborate with community occupational therapists to advise and review adaptations required to support independence. They will also help maintain postural management to minimise the development or progression of a gibbus deformity.[10] Suitable seating, along with

Box 1 Key medical monitoring needs in infancy

Foramen magnum stenosis

FMS is the most severe and potentially life-threatening manifestation of ACH and requires early management.[37 38] The smaller misshapen foramen magnum increases the risk of FMS and serious complications such as motor impairment, breathing difficulties and sudden infant death.[37] The risk of sudden infant death in infants with ACH may be as high as 7.5% in the first year of life.[6] All babies should be assessed for FMS in the first few months of life.[6 13 21 37]

We recommend that assessment for FMS should include a routine MRI scan within 3 months of birth, respiratory polygraphy/polysomnography within 6 months of birth (or earlier if signs of apnoea or concerns with the MRI) and a neurologic examination. The MRI should follow recommended protocols and include the brain and spinal cord.[39] A series of scans in the sagittal and axial planes should take no more than 25–35 min and will negate the need for an additional scan of the spine should decompression surgery be indicated. Clinical indicators of compression include apnoea, hyperreflexia and/or clonus, delayed milestone acquisition (as assessed by an ACH-specific milestone chart), difficulty swallowing, poor weight gain and irritability and should be urgently evaluated.[8 10] MRI findings and clinical indicators should be interpreted in conjunction to guide the need for surgical decompression.[40 41] Interpretation of the MRI should be by a paediatric neuroradiologist or neurosurgeon experienced in ACH using the Achondroplasia Foramen Magnum Score (AFMS).[37 40 42] Up to 50% of infants have an AFMS3 or AFMS4 and require additional monitoring or surgical decompression, respectively.[40 43] Repeat scanning is advised within 6–12 months for infants with AFMS3, as 40% progress to AFMS4 within a year.[37] Decisions on foramen magnum decompression should be made on an individual basis by a neurosurgeon experienced in ACH jointly with the medical MDT. Any surgical intervention should be performed by a neurosurgeon and anaesthetist with experience of the procedure in infants and young children with ACH.[8 37]

Growth

Growth parameters should be reviewed at each visit using ACH-specific growth charts as previously discussed. Should there be a rapid increase in head circumference when plotted on the ACH-specific chart, the infant should have an urgent brain and spine MRI and be referred for evaluation by a neurosurgeon with experience in ACH.[10] Obstructive hydrocephalus requiring intervention is seen in around 3% of infants; this is lower than previously thought, due to recognition of benign ventriculomegaly and the improved understanding of cervicomedullary compression.[44]

Development

Due to differences in body structure, gross motor skills may be delayed in infants with ACH compared with infants of average stature. Unique movement patterns (eg, snow ploughing, bear walking) should be monitored using ACH-specific charts.[10 29 45] Caregivers should be educated on these movements to reduce concern and provided with advice on how to support and progress the development of gross motor skills. Delays in development should be reported to the specialist paediatrician for assessment of possible FMS with referral to specialist paediatric occupational therapy and/or specialist paediatric physiotherapy.[10] A joint assessment can help evaluate factors causing delay and strategies for progression.

Continued

Box 1 Continued

Spinal review

Nearly all infants present with a thoracolumbar gibbus, which typically resolves as the infant starts standing and walking.[6 23 46] At each visit, guidance on 'tummy time', truncal support and appropriate seating should be reinforced with the aim of preventing progression of the gibbus.[10 23] Early upright sitting should be avoided until the infant has developed the trunk muscular control to do so independently;[46] however, the impact on the typical weaning/feeding process should be minimised. During feeding, supportive sitting should be reclined from the upright position for gibbus management and only undertaken for short periods. Active management of spinal deformity at a young age can help to reduce the impact of sequelae which contribute to morbidity later in life. Families can be signposted to ACH support organisations for further educational materials.

Neurological assessment

Abnormal neurological assessment, especially suspected cervicomedullary compression, warrants referral to a paediatric neurosurgeon with experience in ACH and an MRI of the brain and spine using a recommended protocol.[39] Results should be discussed in neuroradiology and specialist MDTs to evaluate next steps.

Respiratory/ENT

Up to 80% of children with ACH experience sleep-disordered breathing (eg, central apnoea, upper airway obstruction and laryngomalacia) which can arise from structural changes such as midface hypoplasia and adenotonsillar hypertrophy or central causes such as FMS.[10 47] Respiratory polygraphy should be conducted by 6 months of age and requested by the MDT, or earlier if clinically indicated. Some infants with sleep-disordered breathing may have no obvious symptoms such as snoring or apnoea.

While polysomnography is the gold standard for diagnosing sleep-disordered breathing, electroencephalogram is not necessary in this cohort. In ACH, the primary concern is detecting respiratory issues such as obstructive and central apnoea or hypoventilation, areas that respiratory polygraphy can assess effectively. With its focus on respiratory parameters, greater accessibility and ease of use, respiratory polygraphy offers a practical and appropriate alternative to polysomnography in these children.[47] The approach taken should be discussed with the respiratory paediatrician.

In ACH, sleep apnoea can be multifactorial. Foramen magnum stenosis with brainstem compression often leads to central apnoeas but can also lead to obstructive apnoeas.[48] Foramen magnum compression should be considered and discussion with a neurosurgeon may be helpful. If respiratory polygraphy shows obstruction, referral to a paediatric respiratory specialist and/or paediatric ENT surgeon (depending on local pathways) with experience in ACH is recommended for comprehensive airway assessment. Adenotonsillar hypertrophy can contribute, but in infants (under a year of age) it is rarely the primary or sole cause of obstruction and as such adenotonsillectomy is infrequently required at this age. Other causes of obstruction should be considered, such as midface hypoplasia, small nasal passages, relative macroglossia and thick, soft tissues may also contribute and, if respiratory compromise occurs, nasopharyngeal airway or ventilatory support may be necessary.

Hearing

Continued

Box 1 Continued

Middle ear infections and hearing loss are common in infants and children with ACH and early detection is essential to reduce any potential impacts on speech and language development.[10 15] The middle ear clearance function is often affected in ACH due to a shorter and more horizontal eustachian tube.[49] This dysfunction can cause middle ear issues, including recurrent or chronic otitis media with effusion (OME), which in turn can cause conductive hearing loss. The risk of OME is considered two times higher in children with ACH compared with those without ACH, and the need for intervention is almost 10 times higher.[49] Following newborn screening, a full audiologic examination should be completed by 12 months of age, or earlier if there are concerns. Any identified concerns should prompt referral to ENT teams with experience in managing hearing loss in children with ACH for further evaluation and management.

Lifestyle/weight

Guidance on healthy lifestyles should be emphasised from infancy, throughout life and recommended for the whole family. Maintaining a healthy weight can reduce the long-term risk of joint and back problems.

ACH, achondroplasia; FMS, foramen magnum stenosis; MDT, multidisciplinary team.

other adaptations such as steps and rails in the toilet, may be required.[6]

Preparation for school

Children may attend preschool with the support of a specialist occupational therapist, working in collaboration with the nursery and community occupational therapy teams. This coordinated approach ensures that access arrangements, environmental adaptations and care plans are established before the child begins school (online supplemental material 7).

Preparation for entry to primary school also commences during preschool years (online supplemental material 7). Support from a specialist occupational therapist and/or physiotherapist regarding modifications/adaptations of the proposed school environment, in collaboration with the special educational needs coordinator and community therapy teams, is essential. The need for an individual education and healthcare plan or similar, alongside reasonable adjustments, should also be considered during this period.

Psychosocial support

The well-being of parents/caregivers, including siblings and grandparents, should be evaluated with psychosocial support offered. Caregivers will often feel anxious about a child with ACH starting to attend school, so offering psychological support ahead of their start date can be very beneficial to provide strategies and containment. Signposting to ACH support organisations facilitates peer support, allows sharing of experience of occupational aids and opportunity for social activities.

Primary school (5–11 years old)

Greater socialisation and independence are encouraged during the primary school years through friendships and participation in appropriate activities. Engagement with ACH support organisations can support peer relationships and a sense of belonging. Celebrating children's engagement and achievements across

Box 2 Key medical monitoring needs in the preschool years

Growth and development
Growth and development should continue to be monitored using ACH-specific charts. Families should be provided with recommendations on appropriate physical activities and food portion sizes to support healthy weight maintenance. In preschool children, advice on developmental milestones includes crawling (eg, snow ploughing, bear walking), walking and managing falls.[29] As children progress, the development of speech and social interaction becomes increasingly important. Given the high prevalence of hearing impairment in children with ACH, ongoing hearing monitoring is recommended as deficits can impact speech and language development.[6 10] Referrals to audiology and speech and language therapy (SALT) should be considered where concerns arise,[50] and early intervention with hearing aids should be considered.

Neurological assessment
Routine neurological assessments are required during the preschool years. FMS may progress over time and should be closely monitored until the age of 3 years.[37] If neurological signs or symptoms are present, an MRI should be repeated. Decisions regarding foramen magnum surgery must involve medical input from the MDT and be performed by a paediatric neurosurgeon or spinal surgeon who has experience in the procedure in children with ACH.[37]

Hearing/ENT review
Audiologic evaluations are recommended every 6 months in the preschool years as this is a critical time for speech and language development. Evaluations should start when the child is 9–12 months old, when the child's skills and attention enable engagement with behavioural hearing tests and is a time when potential eustachian tube issues may arise.

Middle ear infections are very common in young children with ACH, affecting 40% to over 80% of individuals by the age of 5 years[51 52] and as many as half of these require long-term/repeated interventions.[52] The need for intervention should be guided by the degree of hearing impairment and not just the presence of middle ear effusion alone. Intervention may be considered if hearing is difficult and speech and language development, balance or behaviour are affected. Paediatric audiologists play a central role in ongoing monitoring. Management should align with local guidance on OME, and long-term follow-up is often necessary.

Children with recurrent otitis media should be referred to paediatric ENT for evaluation and consideration for grommet insertion or prophylactic antibiotics.[50] Management of middle ear effusion involves observations for 3 months, followed by consideration of grommet insertion or hearing aids if persistent hearing loss is confirmed.[10 50] Not all conductive hearing loss is due to middle ear effusions. A congenital abnormality of the middle ear bones (commonly the incus) may lead to permanent conductive hearing loss. In addition, persistent middle ear inflammation from infections can cause acquired ossicular stiffness, leading to permanently reduced sound conduction and worse hearing levels. In such cases, if intervention is required, bone or air conducting hearing aids are likely to be the preferred option.

Care must be taken during myringotomy procedures due to the potential presence of a high or exposed jugular bulb, which poses a risk for serious bleeding if inadvertently incised.[30 50]

Continued

Box 2 Continued

Respiratory review
Airway obstruction in toddlers and school-age children may be due to adenotonsillar hypertrophy, at least in part, but other causes, as mentioned previously, may also contribute. Caregivers must be educated to recognise signs of obstructive sleep apnoea. These signs include neck hyperextension, frequent or permanent snoring, witnessed apnoea, disturbed sleep and mouth breathing.[47]

It is recommended that children should have regular sleep studies, every 6–12 months up to 4 years of age.[47] If there are signs of apnoea, referral is required to a specialist paediatric ENT/respiratory team for further evaluation and polygraphy.[47] Interpretation of the results should be made in conjunction with the MDT.[47]

In toddlers and school-age children, adenotonsillectomy is often helpful but the child should be followed up, as persistent obstructive sleep apnoea after surgery is common. Other causes of airway obstruction, such as foramen magnum compression, should also be considered. Management strategies include adenotonsillectomy and potentially continuous positive airway pressure or bilevel respiratory support.[10 47]

Spinal review
Kyphosis generally continues to resolve during the preschool years[46] but should be monitored. Referral to a specialist spinal surgeon may be indicated in more severe cases, or if neurological symptoms or signs in the lower limbs emerge.

Orthopaedic review
Lower limb deformities become more apparent during the preschool years and can lead to pain and reduced participation in physical activities. Bowing of the knees and lower legs affects up to 50% of children with ACH[6] and over 16% of children have lower extremity surgery by 10 years of age.[53] An early referral to a paediatric orthopaedic surgeon should be made for comprehensive evaluation, including assessment of the limbs and spine, a clinical analysis of gait and radiographic evaluation.[6] For accurate assessment of alignment and to avoid unnecessary radiation, full-length weight-bearing low radiation imaging (such as EOS) should be used where available, rather than traditional radiographic long leg views and certainly instead of isolated joint imaging. Decisions regarding surgery must involve medical input from the MDT and be performed by a paediatric orthopaedic surgeon who has experience in the procedure in children with ACH.[10]

Dental review
All children with ACH should be referred to a paediatric dentist by 1 year of age for assessment and specialist advice on how to maintain good oral health. As for all children, CYP with ACH should be seen regularly by a general dental practitioner to help maintain a low caries risk. Limiting frequency of consumption of refined carbohydrates and use of an age-appropriate fluoridated toothpaste are strategies to be encouraged.

ACH, achondroplasia; CYP, children and young people; FMS, foramen magnum stenosis; MDT, multidisciplinary team; OME, otitis media with effusion.

academic, creative and physical domains supports positive self-esteem and identity development. Ongoing medical monitoring remains important (box 3).

Box 3 Key medical monitoring needs in primary school years

Ongoing reviews
Ongoing monitoring of growth, weight, motor function, hearing, otitis media, sleep-disordered breathing and neurological status should continue as previously described, with appropriate referrals as indicated. Information on maintaining a healthy lifestyle should be reinforced.

Spinal review
Persistent moderate to severe kyphosis into the primary school years warrants referral to a spinal surgeon for specialist evaluation.[46]

Orthopaedic review
Regular paediatric orthopaedic reviews as part of a joint MDT, where possible, should be undertaken as clinically indicated. All children should undergo a formal orthopaedic evaluation ideally during preschool years, but at the latest by the age of 7 years, to ensure specialist assessment, monitoring and appropriate surgical management of limb alignment. There is a critical period, typically before the age of 11–12 years, beyond which simple surgical interventions, such as guided growth, are unlikely to influence lower limb alignment.[46]Any surgical procedures should be carried out by a paediatric orthopaedic surgeon with experience in the procedure in children with ACH.

Dental/orthodontic review
Children with ACH may develop dental crossbites and crowding due to relative maxillary hypoplasia.[6] Referral to a specialist orthodontist is recommended around the age of 10 years old for formal assessment of occlusion. Interceptive management may be indicated for local crossbite correction or maxillary arch development.

ACH, achondroplasia; MDT, multidisciplinary team.

Home environment

As physical needs change, home adaptations may be necessary to support age-appropriate independence.[6] Specialist occupational therapists should liaise with community teams to request needs-led home assessment and recommend modifications and/or adaptations to support daily activities (eg, self-care and the use of foldable bottom-wipers, snack preparation).[30]

Sibling rivalry may emerge, especially if younger siblings surpass the child with ACH in height or physical capabilities; psychological support may help families manage these dynamics.

School

Children with ACH require proactive, ongoing support to promote inclusion and independence at school. This includes monitoring of physical, social, emotional and learning needs, making environmental adjustments and planning for key transitions, particularly to secondary school(online supplemental file 7).[30] A multidisciplinary approach involving educators, HCPs and families supports adaptive skills and promotes inclusion. Encouraging children to problem-solve and adapt tasks for their needs cultivates lifelong strategies for independence and resilience.

Medical monitoring

Medical complications often stabilise during the primary school years; however, monitoring should continue at intervals of 12–24 months, with the option for earlier review by the specialist team if concerns arise (box 3).[10] Discussions regarding stature modifications (eg, limb lengthening) and clinical trial opportunities may be provided.

Psychosocial support

During the primary school years, children are likely to notice differences in their height and appearance and may require support to manage questions or teasing.[31 32] School-based education may be sufficient, but psychology referral should be considered.[30] From around the age of 7 years, brain development enables children to experience more complex emotions and to develop a greater awareness of their ACH as a lifelong condition that they will need to manage. This is a key developmental stage to consider whether a referral to psychology would be beneficial for the young person. Ongoing monitoring of family well-being well-being should continue during this period.[30]

Secondary school (11–18 years old)

Psychosocial support

During secondary school years, short stature may increasingly affect daily functioning as young people seek independence.[31] Emotional well-being, self-image and self-esteem are particularly important, and additional support may be required to assist with social, academic and future employment-related decision-making.[10]

Social media pressures, cyberbullying and concerns about future prospects frequently emerge.[32]

Despite the attenuation of a pubertal growth spurt for individuals with ACH,[28] adolescence remains a critical period of cognitive, emotional and physical development and vital for developing a sense of identity. Understanding the specific psychosocial challenges faced by adolescents with ACH is key to providing appropriate support. Participation in ACH support organisations can foster valuable friendships that often continue into later life.[30 32]

Independence

As young people with ACH progress through secondary school, emphasis should shift towards developing independence, daily living skills and self-advocacy.[30] Discussions should include mobility and transportation, such as learning to drive with vehicle adaptations[6] and future living arrangements.

Personal hygiene is essential for both health and psychosocial well-being, yet physical limitations, such as limited upper limb reach, can make routine self-care more challenging.[6] Girls may require tailored support around menstruation and access to adapted hygiene products.

Occupational therapy input can support adaptation of routines and environments,[6 10] with additional resources available through ACH support organisations.

School

Young people should be supported to participate fully in school life, with reasonable adjustments to accommodate reduced stature and mobility and to enable inclusion in school trips (online supplemental material 7). Examination arrangements should be reviewed early, with consideration given to adjustments including extra time, rest breaks or the use of scribes or laptops. Vocational planning should include guidance on career pathways, workplace accessibility and access to further education, training or employment.

Box 4 Key medical monitoring needs in secondary school

Obesity
ACH-specific weight charts should be used to monitor for overweight and obesity.[54] The risk of obesity increases with age and affects ACH-related complications like joint pain, fatigue and apnoea.[15 23 32 55] A healthy diet and regular physical activity should be promoted to prevent excessive weight gain and encourage healthy habits into adulthood.[10 14 19 21] Further advice may be required from a specialist dietician or physiotherapist in those children struggling to manage their weight effectively.

Respiratory review
Symptoms of obstructive sleep apnoea, as previously outlined, should be regularly monitored. In young people, unidentified obstructive sleep apnoea may contribute to concentration and learning difficulties and behavioural issues, such as daytime sleepiness and irritability.[47] Symptoms should be investigated with sleep studies where clinically indicated, with referrals to respiratory or ENT specialists.

Spinal review
Symptomatic spinal stenosis, particularly of the lumbar spine, is a severe complication that increases in incidence throughout childhood and into adulthood.[9 56] Claudication pain on mobilisation, often relieved by resting in a squatting position, is the most common symptom, though it can be difficult to distinguish from joint pain or fatigue. Other signs may be impaired mobility over a relatively short time span and may include bowel or bladder symptoms.[9 56] An urgent referral for neurological evaluation is required, with consideration of a repeat spinal MRI if symptoms are progressive. If spinal cord compression is identified, referral to a spinal surgeon experienced in the management of individuals with ACH is necessary.[10]

Orthopaedic review
Regular orthopaedic reviews should continue to monitor for limb deformities and chronic pain. The functional and emotional impact of short limbs and short stature should be explored, with appropriate treatment options discussed alongside the specialist ACH team.

Pain and fatigue management
Young people with ACH may commonly experience pain and fatigue symptoms.[15 57] These can negatively affect daily function and participation and so should be evaluated at each appointment.[10 21] Individualised supportive strategies, including pacing, analgesic plan and mobility aids, should be considered to assist the young person in their daily activities and maintain social relationships. Referral to a specialist physiotherapist and/or occupational therapist should be considered. Physiotherapy or occupational therapy intervention should address functional and participation limitations using goal-based approaches, focusing on activities that are important to the young person, or which have been prioritised by them. Personalised exercise programmes may be given by a specialist physiotherapist to address mechanical causes of pain. Input from local therapy teams should be considered if further support is required at home or school.

Dental/orthodontic review
Continued monitoring of dental and occlusal development is recommended during adolescence as the permanent dentition is established. Maxillary retrognathia and class III malocclusion are common. In mild cases, routine orthodontic treatment can

Continued

Box 4 Continued

suffice; however, in severe cases, orthodontics and orthognathic surgery with maxillary advancement will be required during late adolescence.[10]

ACH, achondroplasia.

Medical monitoring

Secondary school pupils may disengage from medical follow-up and avoid adaptations in an effort to fit in with peers.[33] It is important to ensure that young people understand their ACH, have their questions answered and that proposed interventions are aligned with their personal priorities. Encouraging independent participation in healthcare interactions from adolescence helps in the preparation for adult services and self-advocacy (online supplemental material 6).

While many childhood complications of ACH have resolved or are well managed, others (such as spinal stenosis, obesity and sleep apnoea) may emerge or progress.[14] Annual medical reviews are recommended, with follow-up tailored to individual needs (box 4).

Genetic services

From the age of 13–14 years, or where appropriate for the young person, the individual should be offered access to genetic services to discuss inheritance and reproductive choices.[14 21] These are common questions that arise at this age, and access to genetic counselling should be available when the young person feels ready.

Transfer to adult services

The process of transfer of care to adult services should be gradually introduced during adolescence to support autonomy and self-advocacy. This can take several years and should be initiated many years before the actual transfer occurs, with the overarching goal to foster autonomy (online supplemental material 6).

This process should address not only medical care, but also education, employment, housing, managing benefits (eg, for mobility), relationships, peer acceptance and psychosocial wellbeing. Referral to genetic services should be offered, as above.

Support should be available for parents and caregivers, who may be anxious about access to coordinated adult services and handing over autonomy to their young person.

By the age of 18, care should ideally be transferred to an adult centre of expertise where complications can be monitored and referrals made to experienced specialists as needed.[14] In the absence of a dedicated adult service, management and follow-up may be provided by the general practitioner (GP). As GP familiarity with ACH may vary, young people should be informed about the key signs and symptoms that may require medical attention in adulthood. Consultation guides are available through the International Achondroplasia Forum and may be helpful for both individuals with ACH and HCPs.[34]

ANAESTHESIA CONSIDERATIONS

Anatomical features in children with ACH present specific challenges for general anaesthesia, necessitating careful planning with the MDT. General anaesthesia should be administered in a hospital setting by an anaesthetist experienced in managing children with ACH. Best practice recommendations should be followed, including a comprehensive preoperative assessment

Table 2 Second round modified Delphi statements with ≥80% agreement—recommendations for care for children with achondroplasia

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree	Consensus, n=25
A prenatal suspicion of achondroplasia on ultrasound should be referred to specialist maternal care and fetal medicine/clinical genetics teams, with parents offered appropriate counselling and prenatal genetic testing to confirm the diagnosis.	25 (100%)	0	0	0	0	100%
Care for pregnant mothers with achondroplasia and mothers carrying babies with achondroplasia should be consultant-led and managed in a specialist centre, with delivery typically planned via elective caesarean section due to cephalopelvic disproportion in mothers or fetal macrocephaly respectively.	23 (92%)	2 (8%)	0	0	0	100%
Following a diagnosis of achondroplasia, patients should be promptly referred to a team experienced in the postnatal management of achondroplasia. In instances of prenatal diagnosis, engagement should be established before birth.	23 (92%)	2 (8%)	0	0	0	100%
Parents of infants with achondroplasia should be given information about appropriate handling and positioning, car seat advice, and taught basic life support, prior to discharge from the postnatal ward.	22 (88%)	3 (12%)	0	0	0	100%
Children with achondroplasia require multidisciplinary care. The multidisciplinary team should include experienced and relevant achondroplasia healthcare professionals (online supplemental material 8) and have access to specialist tertiary services through established pathways. Allied healthcare professionals such as occupational therapists, physiotherapists, clinical nurse specialists and psychologists should be embedded in the team.	23 (92%)	2 (8%)	0	0	0	100%
Shared care with a named consultant within a local paediatric service is essential to aid holistic care.	24 (96%)	0	1 (4%)	0	0	96%
Due to the severity of complications in the first years of life, infants should be seen by the specialist team at least every 6 months, and preschoolers at least every 6–12 months, with shared care in between with local services.	23 (92%)	2 (8%)	0	0	0	100%
Within 3 months of birth (or diagnosis), all infants with achondroplasia should have MRI of the brain and spine to screen for radiological features of foramen magnum stenosis and hydrocephalus using a recommended protocol.	22 (88%)	3 (12%)	0	0	0	100%
Infants with achondroplasia should have respiratory polysomnography no later than 6 months after birth (or diagnosis), or earlier if apnoea has been reported or there are concerns with the MRI.	21 (84%)	4 (16%)	0	0	0	100%
The decision to undertake foramen magnum decompression surgery or placement of shunts in infancy must be made jointly by medical and neurosurgical teams and the procedure carried out by a neurosurgeon with experience in foramen magnum decompression in infants with achondroplasia. Surgery requires appropriate anaesthetic, neurophysiology and supportive care.	23 (92%)	2 (8%)	0	0	0	100%
Children's growth (height, weight and head circumference) and development should be regularly monitored using achondroplasia-specific charts, which should be provided to parents for their 'red book'. At each visit, the importance of a healthy lifestyle, incorporating appropriate exercise and balanced nutrition, should be reinforced.	24 (96%)	1 (4%)	0	0	0	100%
Families and individuals with achondroplasia should be offered the contact information of achondroplasia-support groups for peer support.	23 (92%)	2 (8%)	0	0	0	100%
Children and young people with achondroplasia require access to specialist physiotherapy and occupational therapy; they will promote independence, activity, participation and access, with consideration to adaptations and equipment required. Shared care with local services is essential.	23 (92%)	2 (8%)	0	0	0	100%
Children and young people with achondroplasia and their families should be offered specialist paediatric psychological support from diagnosis until transition to adult care.	20 (80%)	5 (20%)	0	0	0	100%
Preschoolers with achondroplasia require 6-monthly audiological evaluations starting from 12 months of age. Abnormal audiology findings or clinical concern should be referred to ENT for evaluation and management.	22 (88%)	3 (12%)	0	0	0	100%
A change in neurological function, such as gait disturbance, an acute reduction in walking distance or new loss of bowel or bladder continence, should warrant immediate evaluation and an MRI considered. Urgent consultation with a paediatric neurosurgeon or spinal surgeon may be required.	25 (100%)	0	0	0	0	100%
All children should be reviewed by an orthopaedic surgeon by the age of 7 years old.	23 (92%)	2 (8%)	0	0	0	100%
If orthopaedic or spinal intervention is necessary, the decision to proceed with surgery should be made by a multidisciplinary team, and the operation should be performed by a surgeon who has expertise in treating individuals with achondroplasia. Surgery requires appropriate anaesthetic, neurophysiology and supportive care.	24 (96%)	1 (4%)	0	0	0	100%
Infants with achondroplasia require a dental check by 12 months of age with a paediatric dentist to help prevent early childhood caries.	22 (88%)	3 (12%)	0	0	0	100%
Children with achondroplasia should be seen at approximately 10 years of age by an orthodontist familiar with achondroplasia to assess facial growth.	22 (88%)	2 (8%)	1 (4%)	0	0	96%

Continued

Table 2 Continued

	Strongly agree	Agree	Neutral	Disagree	Strongly disagree	Consensus, n=25
Care of young people should be transferred to adult services led by a doctor with experience in rare bone conditions where available.	21 (84%)	4 (16%)	0	0	0	100%
In preparation for adulthood, young people should be offered referral to genetic services.	23 (92%)	2 (8%)	0	0	0	100%
New medical technologies should be prescribed under the care of a specialist achondroplasia centre. A minimal national dataset is needed for monitoring outcomes—this is in development.	20 (80%)	5 (20%)	0	0	0	100%
All children and young people with achondroplasia, and their families, should be informed about available novel medical technologies, with written information provided, and be informed about research opportunities.	19 (76%)	5 (20%)	1 (4%)	0	0	96%
All Delphi participants (n=25) voted on each statement. Consensus was predefined as ≥80% of participants strongly agreeing or agreeing.						

with particular attention to any history of snoring or sleep-disordered breathing, to help determine the need for extended post-anaesthetic monitoring.[10] For procedures lasting longer than 1 hour, neurophysiology monitoring is recommended to detect early signs of neurological compromise and minimise the risk of injury during general anaesthesia.

HEALTH TECHNOLOGIES AND ACH

Health technologies used in ACH aim to reduce complications and increase stature, thereby enhancing independence and overall quality of life. In 2021, vosoritide, a C-type natriuretic peptide (CNP) analogue, was approved by the European Medicines Agency and the Food and Drug Administration for use in children with ACH who have open growth plates.[16 35] It is not yet licensed for use in the UK. In addition to CNP analogues, several other therapies are currently in clinical trials, including FGFR3-selective tyrosine kinase inhibitors, anti-FGFR3 antibodies, aptamers against FGF2 and soluble FGF3 constructs.[36] These may become available for clinical use in the future.

In the UK, we propose that the introduction of any new medical technology for ACH should be managed by specialist clinical teams who are experienced in the multidisciplinary care of ACH. Technologies which have been approved by NICE for use in England or by equivalent systems for use in other parts of the UK should be discussed with all patients and caregivers as possible options. This discussion should include the known effects and potential adverse effects. Families must be provided with comprehensive and accessible written information about the new technologies to support informed decision making. It is essential that the choice to adopt, or not adopt, a medical technology does not affect the standard of care provided to the child and family.

The UK Achondroplasia Network also recommends that a minimal dataset is nationally agreed and collected among prescribing units to monitor the impact of new technologies. This would support real-world data collection to inform long-term efficacy and safety of technologies within the UK context. Clinical teams must remain up to date with emerging research to ensure families receive current information.

UK EXPERT CONSENSUS RECOMMENDATIONS

Modified Delphi voting was used to develop UK-specific best practice guidelines for CYP with ACH. In the first round of Delphi voting, all 20 statements achieved ≥80% agreement, including nine reaching 100% agreement (online supplemental material 3). Following expert discussion at a face-to-face meeting, 17 statements were refined, four were split into two separate statements, one was removed and one created.

The second round of voting included 24 statements (table 2), all reaching ≥80% agreement, with 21 statements achieving 100% agreement and 3 achieving 96% agreement. Overall agreement strengthened, and these final consensus statements form recommendations for CYP with ACH (table 2).

Based on consensus, literature and expert clinical experience, age-specific monitoring guidance is provided as a reference tool outlining key areas of care referral pathways and anticipatory advice to support the holistic care of CYP with ACH (online supplemental material 6).

CONCLUSION

This manuscript presents the first UK-specific recommendations developed by an MDT for the diagnosis, monitoring and management of CYP with ACH. Developed through expert

consensus and informed by current literature and clinical practice, the guidance addresses the needs of children from infancy through adolescence within the UK healthcare context.

Recognising variation in service, these recommendations reflect an ideal model to inform future service planning and development. ACH-specific regional centres should operate within a shared care framework that maintains local support while enabling timely specialist input and the adoption of new technologies. Overall, the guidance aligns with international best practices[10 12 14 16] and provides age-specific recommendations to improve consistency of care and outcomes for CYP with ACH.

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