Considerations for the transition from paediatric to adult care in achondroplasia

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Disclosures

- Receives honoraria from BioMarin, QED, Ascendis, Novo Nordisk, Sanofi

AAP Guidelines and consensus statement:
What is recommended?

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CONSENSUS STATEMENT

Check for updates

International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia

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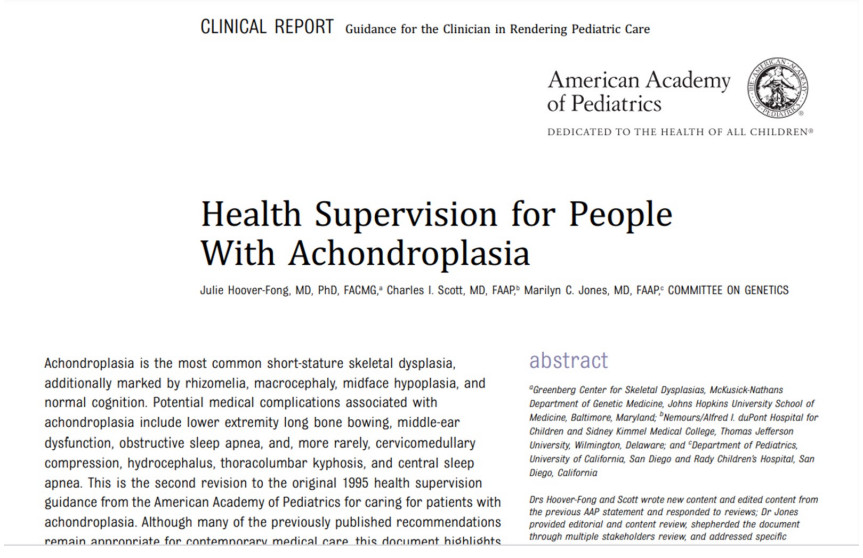
CLINICAL REPORT Guidance for the Clinician in Rendering Pediatric Care

American Academy of Pediatrics

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Health Supervision for People With Achondroplasia

Julie Hoover-Fong, MD, PhD, FACMG,[a] Charles I. Scott, MD, FAAP,[b] Marilyn C. Jones, MD, FAAP,[c] COMMITTEE ON GENETICS

abstract

Achondroplasia is the most common short-stature skeletal dysplasia, additionally marked by rhizomelia, macrocephaly, midface hypoplasia, and normal cognition. Potential medical complications associated with achondroplasia include lower extremity long bone bowing, middle-ear dysfunction, obstructive sleep apnea, and, more rarely, cervicomedullary compression, hydrocephalus, thoracolumbar kyphosis, and central sleep apnea. This is the second revision to the original 1995 health supervision guidance from the American Academy of Pediatrics for caring for patients with achondroplasia. Although many of the previously published recommendations remain appropriate for contemporary medical care, this document highlights

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Drs Hoover-Fong and Scott wrote new content and edited content from the previous AAP statement and responded to reviews; Dr Jones provided editorial and content review, shepherded the document through multiple stakeholders review, and addressed specific

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to physiotherapists and/or occupational therapists wit
skills in this area might assist with the timely provisio
of equipment to maximize independence.

Obesity is a major health problem in achondr
plasia necessitating an early yet complex clinic
management[69,74]. Anticipatory care should be directe
at identifying children and adolescents who are at hig
risk of developing obesity. Adolescents with achondr
plasia should be provided with information regardin
appropriate exercise and/or sports by health pract
tioners with experience in working with individua
with achondroplasia to minimize the risks of injury
complications. Physical activities will help with moo
mental wellbeing, maintaining an appropriate weig
for height, musculoskeletal range and promoting soci
inclusion[62].

Some adolescents might avoid using additional
modified equipment at school due to concerns that
might cause others to tease them or ask questions th
they are not comfortable in answering[3,26]. Their fam
ily should be asked if any concerns get in the way
accessing support and then be offered support to he
overcome these obstacles.

We present the following recommendations regardin
the care of achondroplasia in adolescents.

Recommendation 67. Regular follow-up of adolescen
with achondroplasia is recommended, preferably by
medical practitioner or allied health team experience
in the management of this age group[8,13,27].

Recommendation 68. The effects that pain has on moo
self-care, education and leisure activities in adolescen
with achondroplasia should be specifically evaluated an
monitored.

Recommendation 69. Adolescents with symptom
of spinal stenosis should be referred to a spinal se
vice with experience in managing individuals wit
achondroplasia[26,75].

Recommendation 70. The need for adaptive equip
ment, mobility devices or environmental modifica
tions required in order to maximize independen
should be assessed regularly in adolescents wit
achondroplasia[8,13,27].

Recommendation 71. Overweight and obesity issue
are common in adolescents with achondroplasi
Monitoring of weight using condition-specific growt
and BMI charts and education regarding healthy ea
ing should be provided at each follow-up appointmen
involving the wider family.

Recommendation 72. Adolescents with achondroplas
should be encouraged to maintain an active lifestyle[62,76].

Recommendation 73. Adolescents with achondropl
sia might benefit from the support of dedicated pr
fessionals to assist with adjustment to their conditio
and the development of coping strategies for schoo
employment and social environments[8,13].

Management in adulthood

Although most individuals with achondroplasia lead ordinary lives, the diagnosis might have considerable effects on their physical and mental health[20,23,25,26,78]. Achondroplasia-related medical complications might occur throughout the entire lifespan, necessitating regular follow-up and management. Symptoms of spinal stenosis and OSA are important to diagnose and manage appropriately[20,79–81]. Recurrent ear infections in childhood and the craniofacial anatomy in achondroplasia results in a risk of impaired hearing in adulthood[82,83]. Regular blood pressure monitoring is important, as is preventing weight gain by keeping a healthy diet and regular physical activity[84,85]. Several studies have reported a high prevalence of pain and declined physical health in adults with achondroplasia that affects daily functioning[19,20,22]. People with achondroplasia do not seem to be at increased risk of malignancy compared with the rest of the population[15,24,58]. However, they should participate in appropriate screening programmes (for example, mammography and cervical smear) as recommended for the general population.

The need of relevant helping aids and adaptations at work or at home should be considered to promote optimal participation. Assessment of psychosocial well-being and the potential need for additional support should form part of routine health care for all not just for those with achondroplasia. However, several studies have demonstrated that adults with achondroplasia tend to score lower in quality of life questionnaires for both physical and mental domains than the general population[19,22,23,25,26]. Appropriate support should be made available to adults with achondroplasia and should be provided in a culturally sensitive way.

We provide the following recommendations for the care of adults with achondroplasia.

Recommendation 74. Ongoing back pain, combined with neurological symptoms, such as claudication, spasticity, reduced walking distance, or bladder and/or bowel dysfunction, might be related to spinal stenosis in adults with achondroplasia and an MRI scan of the entire spine should be considered[20,80,86].

Recommendation 75. In the presence of symptomatic spinal stenosis in adults with achondroplasia, where imaging demonstrates evidence of spinal cord impingement or compression, a prompt referral to a spine centre experienced in the management of spinal stenosis in achondroplasia should be considered[20,75].

Recommendation 76. In adults with achondroplasia presenting with symptoms suggestive of OSA, an overnight sleep study should be performed[83].

Recommendation 77. Blood pressure should be regularly monitored in adults with achondroplasia using a cuff that fits the arm appropriately. Blood pressure measurement on the forearm is an option when elbow contractures or rhizomelia prevents measurement at the upper arm[84,87].

Recommendation 78. Adults with achondroplasia might be at increased risk of early-onset hearing loss. There should be a low threshold for assessment of symptomatic individuals and consideration of routine screening at an earlier age than in the general population[83,88].

Recommendation 79. Pain should be monitored longitudinally in adults with achondroplasia at each medical check-up. The effect that pain has upon mood, self-care, education, employment and leisure activities in individuals with achondroplasia should be specifically examined and monitored using patient-reported outcome scales such as the Brief Pain Inventory[19,20,22,26].

Recommendation 80. Adults with achondroplasia should have routine health checks as advised for the general population in their community.

Recommendation 81. Older adolescents and adults with achondroplasia should be offered genetic counselling to provide information on reproductive options.

Recommendation 82. Adults with achondroplasia should be offered psychosocial support as part of their routine health care, which may include formal assessment by a trained professional[23,25].

Recommendation 83. Anaesthesia for people with achondroplasia should be performed by staff experienced with achondroplasia and follow consensus recommendations[12].

Recommendation 84. Aids and adaptations, including car adaptations, are required for adults with achondroplasia[20].

Management by specialty area

Access to specialized care is a major challenge in all regions of the world where clinicians with expertise in the management of achondroplasia are unavailable. The consensus statements related to specialty areas therefore focus on the respective key medical issues of concern. They include practical considerations regarding the types and frequency of assessments and interventions in the specialty to provide a global and sustainable approach to care for all individuals with achondroplasia. Relevant literature and evidence have been referenced wherever these exist. The authors consider it paramount that 'non-expert' health-care professionals (for example, those with limited or no previous experience in managing patients with achondroplasia) seek expert assistance and guidance or refer their patients to such experts to facilitate the best outcomes.

Spine

A thoracolumbar kyphosis is commonly seen in infants with achondroplasia; incidence has been reported to be as high as 90%, decreasing once the child starts mobilizing[14,34,66,89]. Several authors have proposed that development of a fixed thoracolumbar kyphosis might be exacerbated by prolonged periods of positioning with full spinal flexion[2,90]. The effects of trunk hypotonia, increased head weight and increased ligamentous laxity combine with the effect of gravity to promote a slumped sitting position, which might contribute to increased anterior wedging of the vertebra and narrowing of the spinal canal[34,37,89]. In the great majority of patients, the kyphotic deformity resolves spontaneously; however, brace therapy can be considered for children with marked hypotonia and/or motor delay to prevent progression. In situations where fusion and stabilization of the spine are being considered after decompression spanning more than five spinal levels, the effect of the consequent reduction in lumbar spine mobility on activities of daily living in these patients must be considered, which is further exacerbated by short arms and (often) limited elbow extension[75]. This author panel agrees that thoracolumbar kyphosis is often seen during infancy in individuals with achondroplasia but resolves without intervention or treatment in the majority of infants when they start to walk[34,64,66].

The author panel reached consensus over the following recommendations relating to spine care in individuals with achondroplasia.

Recommendation 85. Thoracolumbar stenosis in individuals with achondroplasia might lead to signs or symptoms of neurogenic claudication. This condition can be mitigated by conservative interventions such as weight loss and physical therapy[27]. However, if conservative management fails, surgery might be beneficial[37,91].

Recommendation 86. Thoracolumbar kyphosis is very common in infants with achondroplasia. All patients should be assessed clinically and, if thoracolumbar kyphosis is pronounced, patients should receive radiographs at baseline and subsequent radiographs as clinically indicated if the kyphosis is progressive[3,34,37,64,66,91].

Recommendation 87. At the time of initial surgical decompression of the spine, fusion and stabilization should be performed in skeletally immature patients with achondroplasia given the propensity for development of post-laminectomy kyphosis due to continued spinal growth[63,75].

Recommendation 88. In order to prevent worsening of kyphosis after surgery, fusion and stabilization are recommended in skeletally mature patients with achondroplasia undergoing spinal decompression spanning more than five levels, crossing a junctional area and in patients with unfavourable sagittal alignment, including thoracolumbar kyphosis[75,90].

Recommendation 89. Spinal canal stenosis can lead to signs of myelopathy when it occurs in the cervical and thoracic spine. MRI is recommended for patients with achondroplasia who present with neurological symptoms such as weakness, impaired locomotor and/or fine motor activity, changes in bladder and bowel continence, or pathological reflexes such as hyper-reflexia and clonus[20,75,92].

TABLE 1 Health Supervision for People With Achondroplasia

	Prepregnancy and Short-Stature Parents	Prenatal and Short- and Average-Stature Parents	Birth to 1 mo	1 mo to 1 y	1–5 y	5–13 y	13–21	Adult
Diagnosis								
Physical examination	X	X of fetus	X	X	—	—	—	—
Imaging	X radiographs	X ultrasonography of fetus	X	—	—	—	—	—
Molecular testing	X	X of fetus	X	—	—	—	—	X
Genetic counseling								
Review natural history	X of potential offspring	X	X	X	X	X	X	X
Recurrence risk and genetics	X	X	X	X	X	X	X	X
Delivery mode and location	X	X	—	—	—	—	—	X
Support group(s), family support	X	X	X	X	X	X	X	X
Desired pregnancy?	—	X	X	—	—	—	—	X
Medical evaluation								
Growth (height or length, weight, occipitofrontal circumference)	—	X	X	X	X	X	X	X
Physical examination	—	—	X	X	X	X	X	X
Neurologic examination	—	—	X	X	X	X	X	X
Development	—	—	X	X	X	X	—	—
Neuroimaging	—	—	X	X if new diagnosis	X as indicated	X as indicated	X as indicate	X as indicated
Polysomnography	—	—	X	X if new diagnosis	X as indicated	X as indicated	X as indicate	X as indicated
Hearing assessment	—	—	X	X	X	X	X	X
Radiography for kyphosis, genu varus, bowing	—	—	—	X	X as indicated	X as indicated	X as indicate	X as indicated
Anticipation or guidance								
Warning signs of severe complications	—	—	X	X	X	X	X	X
Car seats	—	X for hospital discharge	X	X	X	X	—	—
Achondroplasia-specific development	—	—	X	X	X	—	X	—
Jugular bulb dehiscence warning	—	—	—	X	X	X	X	X
Supplemental security income inclusion	—	—	—	X	X	X	X	X
Accommodations	—	—	—	—	X	X	X	X
Obesity, exercise, diet	—	—	—	—	X	X	X	X
Driving	—	—	—	—	—	—	X	X
College	—	—	—	—	—	—	X	X
Job training	—	—	—	—	—	—	X	X

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What is the paediatric model?

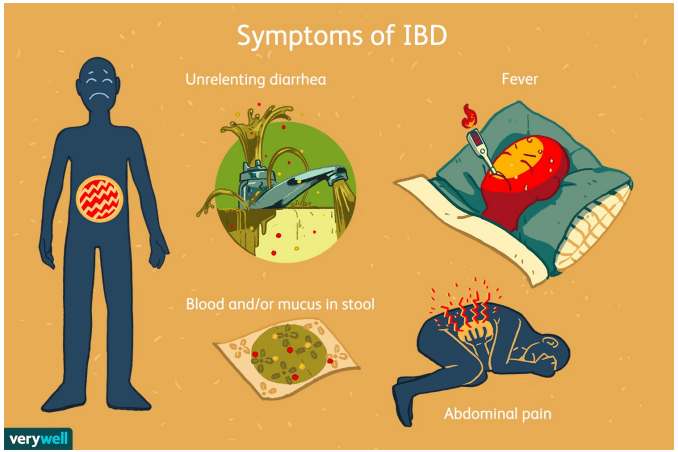
Box 2 Complications of achondroplasia (%) in childhood

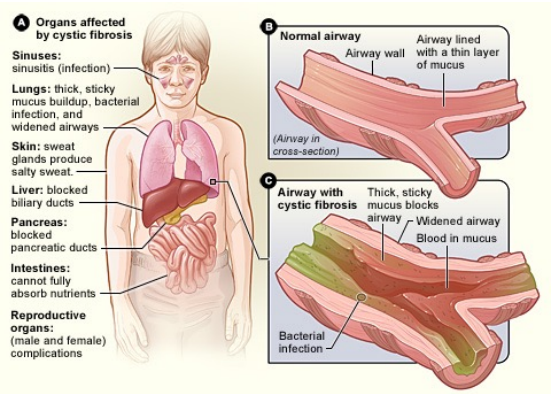
- Neurological
  - Foramen magnum compression (5–10%)
  - Craniocervical instability (very rare)
  - Symptomatic hydrocephalus (6%)
- Orthopaedic
  - Progressive, unresolving thoracolumbar kyphosis
  - Decreased range of movement, elbows and hips
  - Tibial bowing (10%)
  - Symptomatic lumbar spinal stenosis (20%)
- ENT
  - Recurrent otitis media (89%)
  - Adenotonsillar hypertrophy (25%)
- Dental
  - Dental overcrowding (>50%)
- Respiratory
  - Sleep apnoea (75%)
- Growth
  - Short stature
  - Increased body mass index
- Development
  - Comparative motor delay
  - Speech delay (25%)
  - Conductive hearing loss (40%)
- Activities of daily living
  - Restricted through short stature, rhizomelic shortening of upper limbs
- Pyschosocial impact for child and family

Dedicated achondroplasia clinic

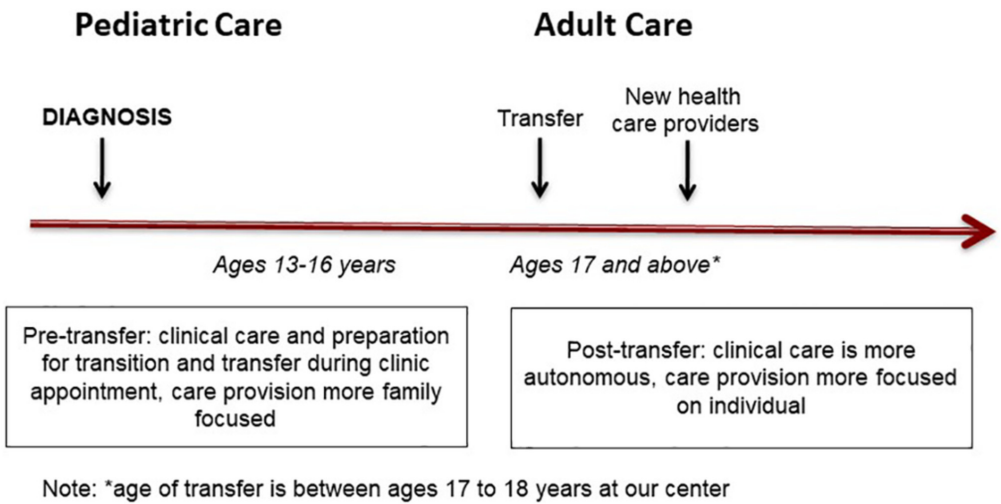
- Paediatric endocrinologist (MC)
- Clinical Geneticist (MI)
- Clinical Nurse Specialist (RG-C)
- Occupational therapist (JM)
- Physiotherapy (KP)
- Paediatric radiologist (CL,EK)
- Paediatric respiratory (MF,PG)
- 'Directory' of specialists – eg dentistry (MH), neurosurgery(DT), ENT(VP)
- Psychology (2018)

Examples of other settings around transition



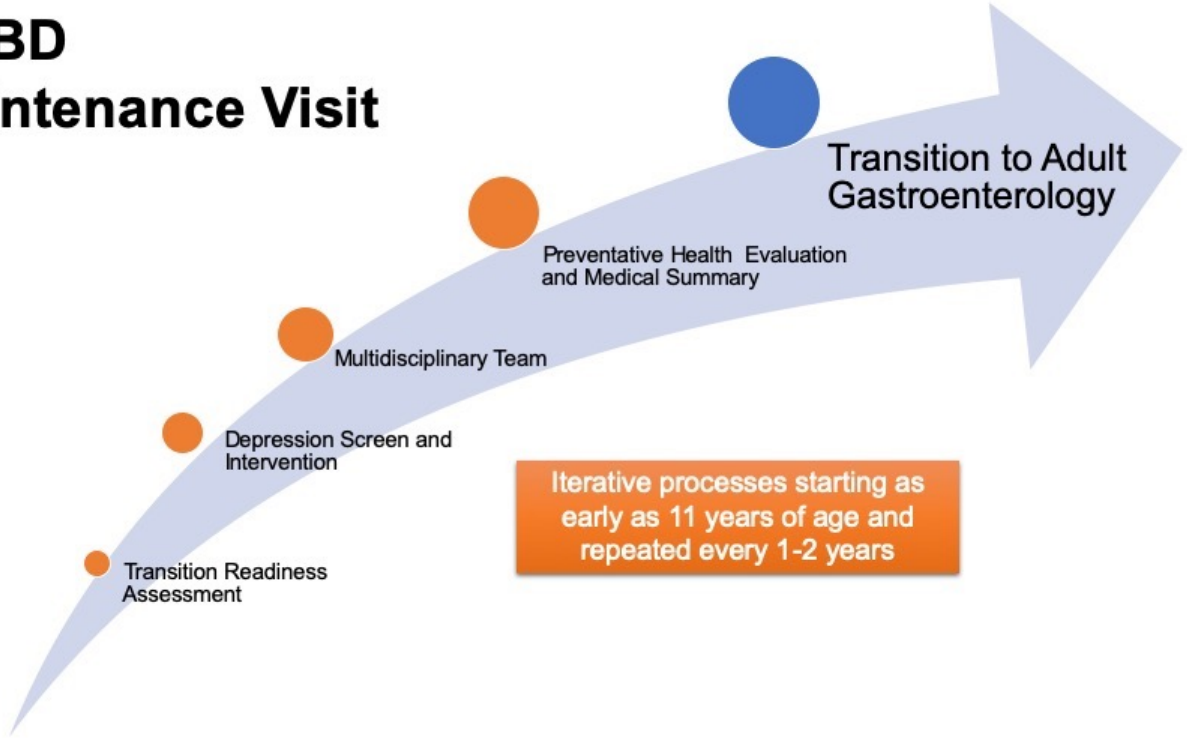


Youth with diabetes and their parents' perspectives on transition care from pediatric to adult diabetes care services: A qualitative study



Health Science Reports, Volume: 3, Issue: 3, First published: 06 August 2020, DOI: (10.1002/hsr2.181)

Pediatric IBD
Health Maintenance Visit



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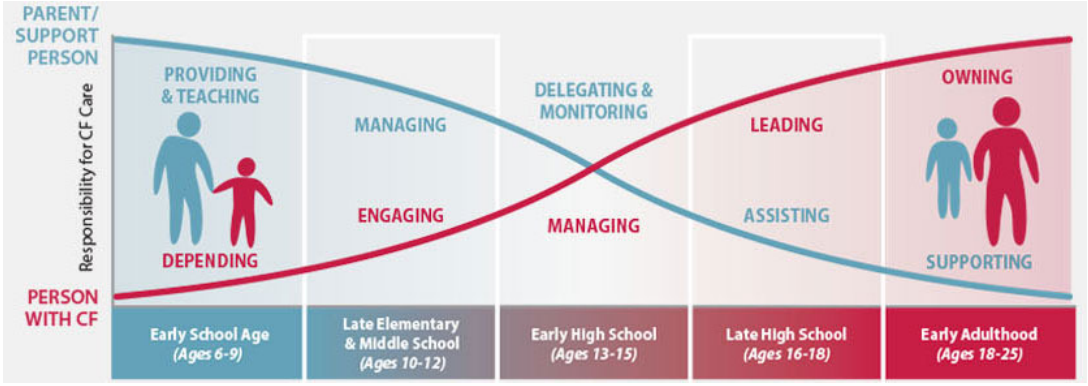
Clinical IBD 2013

Attitudes and Experiences of Adolescents in an Innovative IBD Transition Service

Kay Greveson

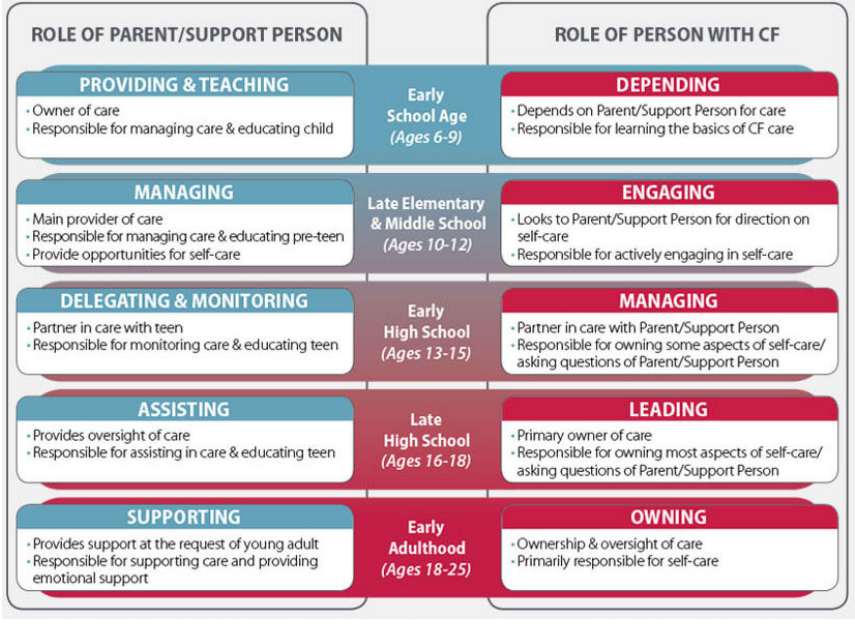
Table 1. Transition models

Handover clinic	One-off direct transfer—a single step from paediatric to adult clinic with little face-to-face communication
Parallel clinic	Paediatric and adult clinics run at the same time but independently, allowing for communication between teams
Transition clinic	Adolescent patients seen by both teams so that all involved develop familiarity and transfer of expertise



Transition in CF

CFR.I.S.E.



Best state to be in at transition – consider:

- Mid to late childhood
- Sleep disordered breathing
- Weight
- Mental health
- Spinal stenosis
- Pain management
- Shunt
- Informed

Gap in the care pathway

- Paediatric MDT v. GP

v.

Suggested ways to fill that gap

- Put ‘patients’ in control of their own health
- Produce a checklist to facilitate this
    - App based
- Educate people with Ach and GPs
    - Educational videos on EAF website
    - Simple language 101 documents (e.g. SDMC)
    - Link to app
- Develop a country-specific directory of specialists
    - Iterative
- Discussion/support forum
    - NHS 111

Resources from Skeletal Dysplasia Management Consortium

Directory of specialists

- Who
- Where
- When to refer
- Create a virtual MDT

Directory of specialists

When to refer	Who and when
Symptoms described: snoring, pauses in breathing during sleep, daytime somnolence	Sleep specialist
Pain (claudication), pins and needles, numbness, incontinence	Spine neurosurgeon
Weight gain	Nutritionist/dietician

Thank you

European Achondroplasia Forum