

A large, stylized orange grid graphic that resembles a globe or a sphere, composed of thick, hand-drawn lines. It is positioned in the upper half of the page, partially overlapping the dark grey banner.

Navigating diagnosis, monitoring, and early treatment in achondroplasia: Multidisciplinary insights from specialist obstetricians and paediatricians

Practice aid for the management of people living with achondroplasia

For more information, visit: www.touchendocrinologyime.org

An international consensus emphasizes MDT management and timely referrals¹



In infancy¹



Many complications
common within the first 2 years of life
benefit from early intervention¹

e.g.

- Sleep-disordered breathing
- Foramen magnum stenosis
- Developmental delay



Specialist and local healthcare teams working in parallel should regularly evaluate infants in the first year of life¹



Monitor every ≥ 2 –4 months, then every 3–6 months as necessary¹



AT DIAGNOSIS:¹

- Refer to a skeletal dysplasia centre

MONITORING:¹

- Regular MDT follow-up led by a clinician with achondroplasia expertise
- Close monitoring in the first 2 years of life is important

MDT and referral needs in infancy¹



**NEUROLOGY/
IMAGING**

- Evaluate for cervicomedullary compression and foramen magnum stenosis
- Concerning signs or symptoms require urgent evaluation by a paediatric neurosurgeon



**RESPIRATORY/
SLEEP MEDICINE**

- Inform parents of typical signs of sleep apnoea
- Polysomnography study should be performed within the first year of life



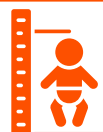
**AUDIOLOGY/
ENT**

- Recurrent and chronic otitis media are common and may cause hearing loss
- Audiology screening at least annually
- Early referral to ENT should be considered



Region-specific guidelines for the management of achondroplasia are available^{2–6}

Achondroplasia-specific growth charts can help tailor MDT management



Regular monitoring of growth and development in children and adolescents with achondroplasia using achondroplasia-specific screening tools is recommended¹



Integrating growth charts into MDT management

- **Monitor infant growth** at each medical check-up using appropriate, specific tools¹
- Provide parents with **achondroplasia-specific charts** and a growth parameters register to support management
- **Measures should include:**¹
Head circumference, height, weight and height:weight ratio



Points to consider with growth monitoring in achondroplasia



Head circumference

Rapid growth with signs of hydrocephalus or cervicomedullary compression may indicate need for neurosurgical evaluation¹



BMI

Values can be distorted leading to increased perception of excess weight and risk of being overweight/obese⁷



Milestone attainment

Delays in milestone attainment compared to achondroplasia standards should be investigated¹



Region-specific growth charts for infants and children with achondroplasia are available⁷⁻¹²

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[Argentina](#)⁸



[Europe](#)¹⁰



[USA](#)¹²



[Australia](#)⁹



[Japan](#)¹¹



[Development and clinical application](#)⁷

Treatment goals can be supported with MDT care and early pharmacotherapy



In the absence of published data on the use of pharmacotherapies in conjunction with other medical interventions, effective patient/parent–physician conversations are important during treatment of achondroplasia¹³



Treatment goals*^{13,14}

- Increase height attainment
- Improve physical functionality
- Support ADL
- Greater independence
- Improve QoL into adulthood
- Minimize stigma
- Improve social life
- Prevent disease-related complications



Treatment choices are a balance between achieving treatment goals and respect for personal identity



Approach to MDT care^{1,13,15}

Clinical genetics/paediatrics/endocrinology

Neurosurgery

**Core
MDT¹³**

Respiratory

Orthopaedics

Rehabilitation

ENT

Psychosocial support

Audiology

PCP

Provide management and follow-up if adult MDT unavailable¹⁵



Meaningful communication and guidance for families about medical and psychosocial aspects of living with achondroplasia should be a key goal for HCPs involved in treatment and care¹



Early pharmacotherapy¹⁶⁻¹⁹

CNP analogues in paediatric populations with achondroplasia and open epiphyses are approved (**vosoritide**)^{16,17} or in development (**navepegritide**)¹⁸



FGFR3 inhibitors (e.g. **infigratinib**) are in development in paediatric cohorts¹⁹

As pharmacotherapies become more widely used, sharing experiences among HCPs involved in achondroplasia management will be important¹³

*Goals listed are based on clinical practice perspectives of physicians with expertise in achondroplasia, and qualitative assessment of outcomes important to people with achondroplasia and their families.

Abbreviations and references

Abbreviations

ADL, activities of daily living; BMI, body mass index; CNP, C-type natriuretic peptide; ENT, ear, nose and throat; FGFR3, fibroblast growth factor receptor 3; HCP, healthcare professional; MDT, multidisciplinary team; PCP, primary care physician; QoL, quality of life.

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