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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

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An international consensus emphasizes MDT management and timely referrals[1]

In infancy[1]

Many complications common within the first 2 years of life benefit from early intervention[1]

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 life[1]

Monitor every ≥2–4 months, then every 3–6 months as necessary[1]

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 infancy[1]

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]

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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 recommended[1]

Integrating growth charts into MDT management

- **Monitor infant growth** at each medical check-up using appropriate, specific tools[1]
- 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 evaluation[1]

BMI

Values can be distorted leading to increased perception of excess weight and risk of being overweight/obese[7]

Milestone attainment

Delays in milestone attainment compared to achondroplasia standards should be investigated[1]

Region-specific growth charts for infants and children with achondroplasia are available[7–12]

Click to access

- Argentina[8]
- Australia[9]
- Europe[10]
- Japan[11]
- USA[12]
- 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 achondroplasia[13]

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 MDT[13]** | Respiratory

Orthopaedics | Rehabilitation

ENT | Psychosocial support | Audiology

PCP — Provide management and follow-up if adult MDT unavailable[15]

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 care[1]

Early pharmacotherapy[16-19]

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 cohorts[19]

As pharmacotherapies become more widely used, sharing experiences among HCPs involved in achondroplasia management will be important[13]

*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.

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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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