

Review Checklist

Achondroplasia



CHILDREN (0—16 years)

GROWTH

Record **height and head circumference** on growth charts for children with achondroplasia. Check weight is appropriate for height. **Failure to thrive** may indicate serious respiratory problems/possible **foramen magnum compression**

URGENT REFERRAL for specialist assessment if **head circumference excessively large or crossing centile lines, or failure to thrive.**

HEARING

There may be **hearing problems**. Routine screening is not sufficient. Children should be screened at birth by an audiologist and reviewed annually until puberty. There is increased frequency of **Otitis Media and glue ear.**

Persistent/Recurrent earache or any loss of hearing should be investigated. REFER to ENT consultant.

SKELETON

Check the spine at each review - **thoracolumbar kyphosis, scoliosis, lordosis, lumbar gibbus** - monitor closely during entire growth period, and especially at puberty and during adolescent growth spurts. Emphasise avoiding unsupported sitting in infancy.

REFER to PAEDIATRIC ORTHOPAEDIC SURGEON if there is **fixed lumbar gibbus, kyphosis or scoliosis, Excessive bowing/unstable gait, or persistent knee pain.**

BREATHING

There is a predisposition to respiratory Difficulties including **obstructive & central sleep apnoea**. Indicators may include: **intermittent breathing, deep sighing, choking, sleep disturbance, enuresis, daytime sleepiness and developmental delay.**

24 hour pulse oximetry in the first year should be carried out. Re- refer if symptomatic.

NEUROLOGICAL

Benign Ventriculomegaly is common. Serious cervical cord compression occurs in a minority. All children should be imaged between 6 & 12 months. Before & after this time - only if symptomatic. Symptoms to review include **delayed developmental or motor control, respiratory symptoms and hyperreflexia.**

URGENT REFERRAL to Neurologist if **reflexes excessively brisk or asymmetric, if there is ankle clonus, extreme hypotonia or early hand preference.**

DEVELOPMENT

Review development against values for achondroplasia—problems may include **speech difficulties**, which may or may not be linked to hearing loss. **Motor development** will be delayed.

REFER for learning assessment if there is any **speech delay. REFER to neurologist** if **outside normal ranges for achondroplasia.**

EDUCATION & BEHAVIOUR

Is the child's school being made aware of special needs and/or adaptations? Are these needs being met? Transition between primary and secondary school can be a difficult time.

Advice and help with school integration/adaptation is Available from the Restricted Growth Association. www.restrictedgrowth.org.uk

DENTAL

Maxillary hypoplasia and prominent jawbone may lead to **overcrowding and malocclusion**. Is the child registered with a dentist who is aware of special concerns in relation to **sedation and anaesthesia (see overleaf)? Orthodontic assessment** should be carried out by age 5.

REFER to orthodontist/ dental hospital as appropriate.