ACHONDROPLASIA: PRACTICE GUIDELINES FOR ALLIED HEALTH PROFESSIONALS

PRACTICE GUIDELINE©

DOCUMENT SUMMARY/KEY POINTS

- This guideline has been written to assist the interdisciplinary team in providing care to infants and young children with Achondroplasia, it will also serve as a resource for collaborating community therapists
- Children with Achondroplasia have different patterns of growth and development to average statured people and this document outlines the normal progress expected for a child with Achondroplasia
- Children with Achondroplasia have unique risks of adverse musculoskeletal and neurological outcomes and need specific management to reduce these risks, which is outlined in this document.
- Children with rare skeletal dysplasia are also at risk of spinal deformity and subsequent neurological compromise, parts of this practice guideline may be recommended by their medical specialist to assist with their care on a case by case basis.

Approved by:	SCHN Policy, Procedure and Guideline Committee	
Date Effective:	1st July 2021	**Review Period**: 3 years
Team Leader:	Staff Specialist	**Area/Dept**: Kids Rehab

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CHANGE SUMMARY

- Due for mandatory review; only minor changes.

READ ACKNOWLEDGEMENT

- Any Health professional providing clinical care to babies and infants with Achondroplasia needs to read and be aware of this whole document
- This document is NOT APPROPRIATE for parents and families – please see separate factsheet

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1 Introduction and Background

These guidelines have been developed to assist health professionals when treating babies and children with Achondroplasia. The complex Musculoskeletal Rehabilitation team consists of; a rehabilitation physician, physiotherapist and occupational therapist, and is part of the specialist referral centre clinic based at the Children's Hospital at Westmead. This group are also treated and provided with therapy at other hospitals and sites within NSW health

The team aims to review patients with achondroplasia soon after birth then every 3 months in the first year of life, every 6 months until 3 years of age, then at least yearly following that until starting school.

The clinic works in collaboration with local medical and therapy teams to deliver best outcomes for the children, with as much care as close to home as possible. We encourage all the children and families to enrol in the NDIS to access community therapies and support.

Please do not hesitate to contact any of the team with any questions, queries or feedback.

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2 What is Achondroplasia?

Achondroplasia is a congenital skeletal dysplasia affecting approximately 1 in every 25,000 births, resulting in disproportionate short stature. It is caused by a genetic mutation in the FGFR3 gene which is active in the growth plates (epiphyses). The dominant gain of function mutation essentially tries to make the growth plate work faster which it cannot do resulting in disruption of the typical growth plate function and a slowing of growth velocity. There are emerging therapeutics targeting the molecular causes which are expected to become available in the next few years (1).

There is a current comprehensive review of the condition openly available (2. Pauli et al 2019) as well as updated medical guidelines (3. Hoover-Fong 2020). Children with achondroplasia have normal sitting height (trunk length), a large head, spinal canal stenosis and shortened long bones, particularly the humerus and femur. Other skeletal anomalies include: restricted elbow extension, short hands and fingers, short broad and flat feet. Hypermobility and ligament laxity of the fingers, wrists, feet, knees and spine are usually present.

Average adult heights are approximately 130cm (males) and 126cm (females). Intelligence is generally within the typical range, and all children will be expected to participate in mainstream educational pathways.

2.1 What musculoskeletal and medical issues need to be considered when treating a child with achondroplasia?

2.1.1 Spinal Canal Stenosis

Children with achondroplasia have a small posterior cranial fossa and narrowing of the spinal canal (1/2 – 1/3 of the size of their non-affected peers) as a result of shortened pedicles and thickened laminae. The brainstem and spinal cord are average size, so the fit inside the canal is quite tight, particularly at the foramen magnum. Pressure on the brainstem or spinal cord may cause symptoms of spinal canal stenosis or high cervical myelopathy, this manifest as upper motor neurone signs in the lower limbs, and lower motor neurone signs in the upper limbs, as well as unexpected trunk weakness.

Hip flexion contractures and/or poor core stability result in a lordotic lumbar posture which further reduces the space in the spinal canal for the spinal cord, and may worsen spinal canal stenosis. Increased weight for height will also increase the lumbar lordosis and place greater stress on the already narrowed spinal canal.

Symptoms may include:

High stenosis (brainstem or high cervical level) – central sleep apnoea, oromotor dysfunction including difficulties swallowing and articulating, central sleep apnoea, hyperreflexia, hypertonia or clonus in the lower limbs.

Low stenosis (thoracic or lumbar levels) – pain, altered sensation, weakness or intermittent claudication in lower limbs, bladder and bowel dysfunction, hyporeflexia

2.1.2 Thoracolumbar Kyphus

Anterior wedging of thoracolumbar vertebrae can develop in children with achondroplasia due to the bone dysplasia and ligamentous laxity in their spine. This allows a thoracolumbar kyphus to develop in the presence of muscle weakness (refer to fig 1). Early upright sitting, where the weight of the large head is transmitted through relatively soft vertebral bodies with delayed maturation can result in this kyphus becoming fixed, further exacerbating the spinal canal stenosis as the cord is stretched over the longer kyphotic length. Wedging is usually preventable.

Fig 1.Thoracolumbar Kyphus
http://www.nemours.org/service/medical/orthopedics/dysplasia/achondroplasia/characteristic.html

2.1.3 Elbow Flexion

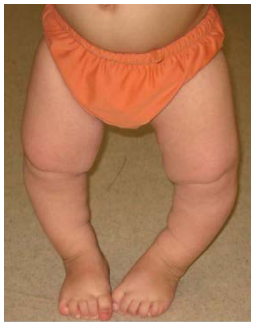
The elbow positioning is structural. No attempt should be made to increase range by splinting and/or casting.

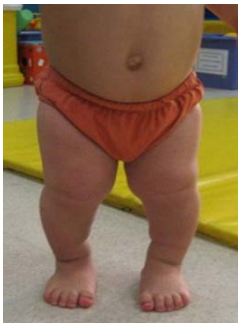
2.1.4 Lower limb deformity

Children with achondroplasia commonly develop ‘genu varum’ (bow legs) as a result of uneven bony growth at the epiphyses and ligamentous laxity around the knee joint. This needs specialist review to ascertain whether it is positional or bony (refer to Fig 2).

Some children may develop tibial bowing, possibly including a leg length discrepancy, and may require surgical correction of the deformity. Surgery is rarely indicated in the first 5 years of life. Specialist orthopaedic consultation is available through the CMSK clinic.

Fig 2: Genu Varum – at rest and after asking the child to "stand up straight" illustrating the contribution of ligamentous laxity





2.1.5 Joint pain and fatigue

Activity related joint pain and physical fatigue are common in children with achondroplasia, particularly in the knees during walking and sporting activities, and in the hands during writing and fine motor tasks. Muscle length is normal and the relatively long muscles cannot work as efficiently with a short lever arm. Pain and fatigue tend to become more of an issue with the increasing school demands over time.

2.1.6 Hearing

The Eustachian tube is shorter, narrower and more horizontal, within the short skull base. Children with achondroplasia commonly have middle ear infections in the first five to six years of life and over half will need grommets. Any infections must be recognized and treated promptly, and hearing should be checked on a regular basis to minimize any hearing loss. Children with an abnormal newborn hearing test should be referred under those guidelines and all children should have a repeat hearing test at 1 year old.

2.1.7 Sleep Apnoea

Children with achondroplasia are at an increased risk of obstructive sleep apnoea due to their small airways, and central sleep apnoea due to possible spinal canal stenosis and brainstem compression. Early referral to a paediatric sleep unit is recommended and an early daytime study (first 4-6 weeks of life) is the first step. Sleep studies should be performed regularly to monitor for this and appropriate weight for height should be maintained. Children with untreated sleep apnoea are likely to be easily fatigued and slower in their early development, older children may present with attention and concentration difficulties. Community health staff should take additional care that safe sleep guidelines are followed due to the increased risk of SUDI in this population.

2.1.8 Ventriculomegaly and Hydrocephalus

As a baby with achondroplasia has a large head, hydrocephalus is often suspected but will generally be ventriculomegaly with no increase in intracranial pressure. Medical monitoring of head growth on achondroplasia specific growth charts is important. Surgical treatment is rarely necessary but may be indicated if the child has significant abnormal neurological symptoms.

2.2 Gross motor development

Babies with achondroplasia are likely to have a delay in gross motor skill acquisition compared to other children. Balancing a large head on a small neck requires extra back and neck extensor strength, often resulting in a delay in obtaining head control. Current treatment aimed at preventing spinal deformity specifically delays independent and upright sitting, further contributing to gross motor delay. Children with achondroplasia also use unconventional methods to mobilise independently when young, often due to their shortened limbs, and do not follow “normal” developmental progress. For example, children with achondroplasia almost never play with their feet, as they are unable to reach them with their shortened limbs.

Fig 3

Snow-ploughing

Reverse snow-ploughing

Bear walking

Push to sit through prone

Move to sit through side-lying

Recent research on Australian children with achondroplasia, combined with our clinical experience leads us to consider the following progress to be within normal limits for these children. However infants with motor delay (compared to normal peers) must be screened at regular (4-6 monthly) intervals by their medical specialist to ensure the delay is not due to emerging neurosurgical issues:

- Able to lift head up in prone by 6 months
- Able to roll by 7months
- Walk with hands held by 21 months
- Stand independently by 24 months
- Walk independently by 26 months

Children with achondroplasia also have very different movement patterns. When moving independently on the floor, traditional 4-point crawling is uncommon. Commando crawling is the most common form of movement for these children, although snow ploughing, reverse snow ploughing and bear walking are also common. All of these movements are normal and safe for children to perform (refer to Fig 3 above).

Transitional movements between positions often use very different movement patterns also. When moving from lying into sitting, most children with achondroplasia will lie prone, abduct their legs to almost 180º, and then push their trunks up into sitting. When pulling to stand, many children with achondroplasia will come up through half kneeling, with one leg abducted, and weight bear through a hyper-extended knee to reach standing. When moving from standing to sitting, many children will either just drop backwards or abduct their hips to get to the floor. All of these movement patterns are normal and safe for children with achondroplasia. Progress in early milestones is carefully monitored by the specialist multidisciplinary team at CHW and babies should only be placed in sitting in a Fraser chair at first and not encouraged into sitting on the floor and/or standing activities unless medical clearance has been specifically communicated via the CHW team.

2.3 Fine motor development

The acquisition of fine motor skills in babies with achondroplasia can be similar to that of other children. However, babies and toddlers with achondroplasia often have difficulties in grasping large objects due to their typically short fingers. Limited elbow extension and supination, combined with lax wrist and finger joints can also create difficulties with some fine motor skills and delay their ability to carry out some self-care activities at the same rate as their peers. Acquisition of tool use such as using crayons, scissors and cutlery as well as prewriting skills in the preschool years may also be delayed.

2.4 Communication skills

Babies and toddlers with achondroplasia may have a speech delay and difficulties with their expressive communication. There are many possible causes for this delay, including hearing problems, restricted floor play and delayed seating (for back care). Speech should be

developing within normal limits, and a specialist opinion is needed if this is not the case for an individual child.

Please note:

13% of achondroplasia toddlers are late talkers (Less than fifty words and no phrases by 24 months). However, we hope that with better early intervention, this will improve.

2.5 Feeding behaviour

Children with achondroplasia tend to also have a delay in their feeding habits when compared to other children. Respiratory difficulties, relatively large tongue and small airways can create difficulties and noisy breathing when bottle feeding and children with achondroplasia will tend to have smaller volumes of feeds. However, it must always be remembered that poor oromotor dysfunction can also be a sign of high spinal cord compression including difficulties with coughing and gagging, spillage from the mouth and difficulty with new textures. Babies with achondroplasia would be expected to double their birth weight in the first 12 months rather than triple per average statured children, feed volumes will be smaller for demand fed babies who should have their growth monitored on condition specific charts (Tofts et al AJMG)

The following developmental progress is currently considered to be within normal limits for these children:

- Manage smooth solids at 4-7 months
- Manage mashed solids at 6-12 months
- Perform finger feeding 8-18 months
- Drink from a cup 8-22 months
- Self feed with a spoon 15-27 months

3 Musculoskeletal Management

Provide parents and carers with the ‘Positioning and Handling of babies with Achondroplasia’ handout developed by the complex Musculoskeletal Therapists. This provides illustrations of correct positioning and handling and outlines precautions. (Handout link: https://www.schn.health.nsw.gov.au/fact-sheets/achondroplasia-positioning-and-handling-of-babies-with-achondroplasia)

3.1 Protection of the Newborn Cervical Spine

Due to the large head size, head control usually takes a longer time to develop in babies with Achondroplasia. Protection of the neck is essential to minimise the risk of high cervical spinal cord damage from spinal canal stenosis.

Recommendations and Precautions:

Educate all carers on providing appropriate support to the baby's head and neck during handling, to protect the cervical spine. Use 'Positioning and Handling of babies with Achondroplasia' handout (Handout link: https://www.schn.health.nsw.gov.au/fact-sheets/achondroplasia-positioning-and-handling-of-babies-with-achondroplasia).

Avoid extreme flexion and extension of the cervical spine to minimise the risk of high cervical myelopathy from spinal canal stenosis.

Do not lift the baby up by the arms or pull-to-sit, with an unsupported head.

Carrying pouches and mechanical swings are not recommended due to the uncontrolled head movement and risk of central and obstructive apnoea.

3.2 Prevention of Fixed Thoracolumbar Kyphus

Although a fixed thoracolumbar kyphus due to anterior wedging of vertebrae was once extremely common, experience in our clinic and overseas indicates that by delaying upright sitting and promoting trunk extensor muscle strength, most cases of fixed deformity may be prevented. 90% of infants with Achondroplasia will have a flexible kyphosis in the first year of life, most resolve with this programme. In our centre about 5% of the group require thoracolumbar bracing compared to 30% in centres overseas who do not follow this protocol.

Recommendations and Precautions:

- Independent sitting and standing should not be encouraged until the child has sufficient back strength to actively maintain an extended upright posture, and is able to attain sitting and standing independently.
- Development of trunk extensor muscle strength should be actively encouraged with prone activities (tummy time). Wedges, therapy balls, rolled up towels etc. may be used.
- When babies are awake they should be positioned in prone, side lying or supine (prone is the preferred position to develop extensor muscle strength).
- Firm thoracolumbar support should be provided in reclined sitting and during handling until the child is able to sit independently. Flexed postures in seating should be avoided and a normal lumbar lordosis and spinal curve should try to be maintained.
- Babies should not be held or carried for more than a few minutes in a "curled-up" position and upright positions should be avoided. Baby slings and pouches are not recommended due to insufficient support of the thoracolumbar spine and neck and risk of the increase risk of apnoea.
- "Burping" against the carers shoulder is a good position as the baby's back is easy to support properly, especially if the carer is encouraged to lean back against pillows/cushions. Rubbing rather than patting is preferred when burping baby. Time in upright position should be limited.
- Sitting in a reclined seat is usually commenced around 4 months of age. This should be restricted to short periods of time and only used when the child is awake and alert. An infant seat with a firm back support such as the Frazer Chair is recommended. Initially

the chair should be reclined back to 30 degrees from the ground. This angle can be gradually increased as strength and age allows.

- Car capsules, baby car seats and strollers should all have a firm back and be reclined as much as possible initially. Babies are best managed in a bassinet attachment for their prams. The seat or stroller can be placed in a more upright position over time as the baby gains better strength and head control.
- Seats to assist with bathing should all have a firm back and be reclined 30 degrees from the ground initially.
- Seats to assist with feeding eg high chair, should all have a firm back and be reclined to 60 degrees from the horizontal during feeding, until the child is able to sit independently with good trunk control.
- Aquatic physiotherapy activities may also help build muscle strength from approximately 6 months onwards (depending on head control and respiratory status).
- Baby bouncers, baby rockers, Jolly Jumpers, baby walkers and baby swings are not recommended due to extra force travelling through the unsupported spine.

Seating in a Fraser chair: foam is used to help position the Achondroplasia child safely, & to support back & neck.

3.3 Management of Elbow & Forearm Deformity

Limitations in elbow extension and supination result from bony changes (possible posterior humeral bowing and radial head dislocations). Most children with achondroplasia will continue to have functional range.

Recommendations:

- Monitor elbow joint range.
- Do not cast or attempt stretching to change the deformity. Reduced range is due to bony changes and will not be improved with casting or stretches.

4 Promotion of Early Development

As outlined above, children with achondroplasia do not follow typical developmental sequences and patterns. Standardised developmental assessments should not be used in this patient group. Wherever possible, parents should be provided with reassurance that their child's movement patterns are safe and normal for Achondroplasia. Penny Ireland's Developmental Screening Form is useful to monitor development compared to the Achondroplasia population and to provide reassurance to parents. Education and support in promoting early language skills, along with encouraging opportunities for fine motor skill practise, while still maintaining positioning and handling to ensure best possible spine care, is important for families. The accompanying parent factsheet has language stimulation suggestions for families.

4.1 Management

In the first couple of years, prevention of musculoskeletal deformity and resultant neurological compromise needs to take priority over "normal" gross motor development. However, fine motor, speech and language development and stimulation should continue as per "normal".

Provide education and support to parents on the need for musculoskeletal prevention to be paramount at this stage. Explain that their child will walk and sit like other children when they are ready, which will be a little later than average statured children. Many parents find this very difficult to accept, particularly when attending playgroups or around other children of a similar age.

Do not push or encourage the acquisition of sitting, standing or weight bearing positions until trunk strength is sufficient to prevent kyphotic posture.

Encourage speech and language development – encourage parents to talk, sing, laugh and interact face to face with their baby. Provide a stimulating physical environment but allow children to develop at their own pace.

Assist the family in finding appropriate small, lighter toys, for their baby's small hands to manage.

Provide opportunities for fine motor developmental play eg sitting reclined in a Fraser chair with a table around to allow for the fine motor play a child would normally do in an independent sitting position. Crayons or pencils for drawing or blocks for play should be easily held in their small hypermobile hands.

Encourage floor play, especially prone time, from an early age. Supine, side lying and prone should all be used for play, along with reclined seating.

Reassure parents when their child performs unconventional means of transitioning and moving.

Once the child has sufficient head control, aquatic physiotherapy may be commenced to provide another environment for strengthening and promoting motor skills. Supported sitting and standing can be trained in neck deep water if spinal posture is appropriately maintained.

Always encourage age-appropriate play and language. Remember that the achondroplasia child often looks much smaller and younger than they really are. Assist families not to 'baby' their child but provide age appropriate activities for them. Provide advice on how to play and communicate with their child.

5 Childcare Preparation

Due to the risk of spinal complications childcare staff need to be educated correct positioning and handling and restrictions of the infant with Achondroplasia. Please contact the Complex Musculoskeletal Team at The Children's Hospital at Westmead for advice.

6 References

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