ACHONDROPLASIA (0-5 YEARS OF AGE): THERAPY GUIDELINES

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 normal 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 this risks, which is outlined in this document.

CHANGE SUMMARY

- N/A – new document.

READ ACKNOWLEDGEMENT

- Any Health professional providing clinical care to under 5's with Achondroplasia needs to read and be aware of this whole document

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1 Introduction and Background

These guidelines have been developed in order to assist community therapists when treating babies and toddlers with achondroplasia. The guidelines have been tailored for use with the patients of the Connective Tissue Dysplasia (CTD) Clinic at the Children's Hospital at Westmead.

The CTD Clinic Rehabilitation stream consists of; a rehabilitation physician, physiotherapist and occupational therapist.

The team aims to review patients with achondroplasia every 4 months in the first year of life, then every 6 months until 3 years of age, then yearly following that until starting school.

Patients are also encouraged to have local therapy support as the CTD clinic therapists are able to provide a consultative service only.

Please do not hesitate to contact any of the CTD 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. Children with achondroplasia have normal sitting height (trunk length), a large head and shortened long bones, particularly the humerus and femur. Average adult heights are approximately 130cm (males) and 126cm (females). Intelligence is generally normal, and all children will be expected to participate in normal education pathways.

Children with achondroplasia will usually present with the following easily observable musculoskeletal features:

- limited elbow extension (still within functional limits)
- limited supination of the forearm
- short hands with stubby fingers
- short, broad, flat feet
- hypermobility of the fingers, wrists, feet, knees and spine

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 the normal size, so the fit inside the canal is quite tight, from the foramen magnum to the end of the spine. Pressure on the brainstem or spinal cord may cause symptoms of spinal canal stenosis or high cervical myelopathy.

Hip flexion contractures or poor core stability result in a hyperlordotic 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) – oromotor dysfunction including difficulties swallowing and articulating, central sleep apnoea, hyperreflexia, hypertonia or clonus

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 ligamentous laxity of their spine. This allows a thoracolumbar kyphus to develop in the presence of muscle weakness (refer to fig 1). Ongoing muscle weakness may 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

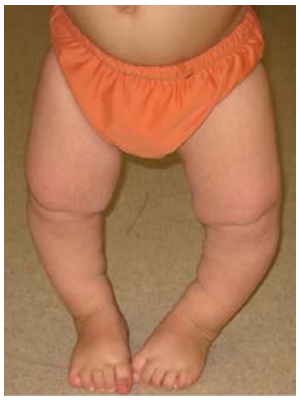
The elbow positioning is bony. No attempt should be made to increase range by splinting. Avoid lifting the child by pulling on the arms.

2.1.4 Lower limb deformity

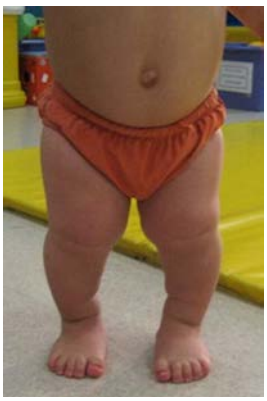
Children with achondroplasia commonly develop 'genu varum' (bow legs) as a result of their ligamentous laxity around the knee joint. This is normal and unless severe, will almost entirely resolve over time with increases in strength (refer to Fig 2).

Some children may develop tibial bowing, possibly including a leg length discrepancy, and may require surgical correction of the deformity. Ilizarov correction is most commonly used, and follows the usual postoperative course of management. Surgery is rarely indicated in the first 5 years of life.

Fig 2: Genu Valgum



Genu Valgum at rest



Laxity able to be corrected

2.1.5 Joint pain and fatigue

Joint pain and fatigue are common in the hyper-mobile joints of children with achondroplasia, particularly in the knees during walking and sporting activities, and in the hands during writing and fine motor tasks. Pain and fatigue problems are sometimes present in toddler years, although they tend to become more of an issue with the increasing school demands over time.

2.1.6 Hearing

The Eustachian tube is shorter and more horizontal, within the short skull base. Children with achondroplasia have a tendency toward middle ear infections in the first five to six years of life. 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 SWISH 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 (infant) is useful. 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.

2.1.8 Hydrocephalus

As a baby with achondroplasia has a large head, hydrocephalus is often suspected. Medical monitoring of head growth on achondroplasia specific growth charts, and baseline head ultrasound should be performed. 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 hypermobile neck requires extra back and neck extensor strength, often resulting in a delay in obtaining head control. Most babies with achondroplasia are also hypotonic, so it takes even longer to gain strength and 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 mobilize 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:

- Able to lift head up in prone by 5 months
- Able to roll by 7months
- Commence pulling to stand and stand independently by 16 months
- Achieve independent transition into sitting by 17 months
- Walk with hands held by 19 months
- Stand independently by 20 months
- Walk independently by 23 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.

2.3 Fine motor development

The acquisition of fine motor skills in babies with achondroplasia is similar to that of other children. However, babies and toddlers with achondroplasia often have difficulties in grasping large objects due to their typically short, stumpy fingers. Limited elbow extension and supination, combined with hyper-mobile wrist and finger joints can also create difficulties with some fine motor skills. The following progress is considered to be within normal limits for these children:

- Reach for an object by 6 months
- Transfer objects between hands by 7 months
- Bang two objects together by 9 months
- Scribble with a crayon by 18 months
- Build a 2 block tower by 18 months
- Draw a circle by 33 months

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 hypotonia, hearing problems and restricted floor play and handling (for back care). The following developmental progress is currently considered to be within normal limits for these children:

- Smiles at 1-3 months
- Babbles at 6-12 months
- Specific words for example mama, dada at 8 -15 months
- Understands "peek-a-boo" at 8 -14 months
- Identifies body parts at 13 - 24 months
- Two word phrases at 20 - 31 months

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, hypotonia and small airways can create difficulties and noisy breathing when bottle feeding and children with achondroplasia will tend to have smaller volumes of feeds due to their smaller stomach. However, it must always be remembered that poor oromotor dysfunction can also be a sign of high spinal cord compression.

Children with achondroplasia are also reported to have difficulties with coughing and gagging, spillage from the mouth and new textures.

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

2.6 Role of the therapy team

Physiotherapists, Occupational Therapists and Speech Pathologists are able to assist in the management of achondroplasia, in conjunction with experienced medical practitioners, to maximize children's quality of life and development, while minimizing potential musculoskeletal problems that may result from the condition.

All children with achondroplasia have spinal canal stenosis. The therapists' role is to implement early preventative strategies such as appropriate seating, handling and play activities to reduce the likelihood of the spinal canal stenosis becoming symptomatic. At the same time, it is essential that therapists assist families in providing opportunities to maximize their child's developmental potential.

Following the first few years, preparing parents, families, day-care centres and preschools to have an understanding of the condition and full access to facilities and equipment for the achondroplasia child, becomes the primary focus of therapy.

3 Musculoskeletal Management

3.1 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.

Recommendations:

- Independent sitting and standing should not be encouraged until the child has sufficient back strength to actively maintain an extended upright posture.
- Development of trunk extensor muscle strength should be actively encouraged with prone activities (tummy time). Wedges may be used if required.
- 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. Baby slings and pouches are not recommended due to insufficient support of the thoracolumbar spine.
- If sitting for brief periods on carers' laps, for example while bringing up wind after a feed, firm support should be provided to the baby's lumbar spine to prevent a kyphotic sitting posture. "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.
- Sitting should initially be restricted to short periods of 10-20 minutes when the child is awake and alert in an infant seat with a firm back support (eg Frazer Chair) reclined back to 30 degrees from the ground. This is usually commenced around 2-3 months of age, and the angle of recline 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. 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 to at least 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.

Fig 4.

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

3.2 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 hyper-mobile neck is essential to minimise the risk of central apnoea, high cervical myelopathy and spinal canal stenosis.

Recommendations:

Educate all carers on providing appropriate support to the baby's head and neck during handling, to protect the cervical spine.

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

Avoid hyperextension of the neck when the baby is prone.

Carrying pouches and mechanical swings are not recommended due to the uncontrolled head movement.

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 "normal" 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. 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.

4.1 Management

In the first couple of years, prevention of musculoskeletal deformity 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 but just a little later. 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 weightbearing 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 tumbleform with a table around to allow for 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 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, Preschool & School Preparation & Management

Due to the short stature and propensity for fatigue in the achondroplasia child, difficulties present both at preschool and school in remaining independent and fully participating with their peers. Pre-planning for preschool and school transitions should always be considered. A pre-school or school visit and written recommendations are recommended.

The possibility of wheelchair or crutches use if lower limb surgery may be indicated should be considered. The following issues need to be addressed:

5.1 Physical Access

Children with achondroplasia easily fatigue and may have difficulty keeping up with their peers over longer distances. Furthermore, their short stature can prevent them from accessing handles, lockers, furniture and hooks set at a standard height. Appropriate recommendations for schooling include:

- Location of classrooms close to amenities
- Location of classrooms on the ground floor to minimize steps
- Any hooks for bag storage outside classrooms to be of an appropriate reach height or a small step provided
- Equipment required to be accessed during class time needs to be accessible for the child. It may need to be stored on a lower shelf, or small steps provided.
- Children who experience hip or knee pain may have difficulty getting on and off the floor. Provision of a small chair for the child to sit on when the other children are partaking in floor activities is recommended in this situation.
- Handles on doors may need to be lowered, or a chain extender attached, to allow the child to independently open and close doors. This is especially important in the event of an emergency.
- Bubblers may need to be lowered, or have a small step available, to allow for independent access.
- Boys urinals need to be accessible – either a floor urinal or a small step provided
- Toilets may require a small step, or a toilet throne grab rail, to be provided to allow access to flush mechanisms and easy transfers on and off
- Small step may need to be provided to access wash basins and paper towel dispensers
- Taps must be able to be turned on and off easily by small hands. Lever type taps may be required.
- Assistance with toileting may be required during the first year of school until independent skills are obtained.
- If walking long distances is required, such as on excursions or to sporting ovals, extra rest breaks and/or extra time should be provided to minimize fatigue.
- Access to the counter at the school canteen should be considered. A small step may need to be provided.

6 Seating

Appropriate spinal and foot support should be provided at all times while maintaining the child at the same height as their peers (achondroplasia children have near normal sitting height). A footrest may need to be provided due to their shortened lower limbs. A cushion to support behind their back may also be required to reduce the seat depth. In general, specialized seating is not required.

While seating in the classroom must be appropriately modified, seating in the outdoor playground should also be considered to allow the child to interact with their peers, while also maintaining appropriate postural support. A small step may need to be provided, or outdoor benches lowered.

7 School Performance

Difficulties with accuracy and speed of fine motor tasks, particularly writing, cutting and craft, are common in children with achondroplasia. Performance of these tasks should be monitored at school to allow for modification or equipment prescription if necessary.

Fatigue, short stature and restrictions on some gross motor activities for prevention of long-term musculoskeletal problems, may result in difficulties in participating in physical activities. Modifications may be required. It is essential to the social development of the child that they are able to participate with their peers.

Maintenance and improvements in cardiovascular fitness, weight control and skill acquisition are also gained by full participation in physical activities.

8 References

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