# MCN for Neonatology

# West of Scotland

# Neonatal Guideline


# Achondroplasia

## Investigation & Management with Neonates with Suspected Achondroplasia

**Introduction**
This guideline is applicable to neonatal staff working with neonates in the West-of-Scotland managed clinical network. This guideline is intended to provide guidance on the investigation and management of babies born with suspected achondroplasia.

**Objectives**

1. **To identify which initial investigations should be carried out prior to discharge from the NICU.**
2. **To highlight the specialties to whom onward referral should be made in infants who have a clinical or genetic diagnosis of achondroplasia.**
3. **To provide information on follow up for infants with achondroplasia and health surveillance in the first year of life.**
4. **To highlight advice, resources, and support available for parents and carers of infants with achondroplasia.**

**Introduction**

Achondroplasia is the most common form of disproportionate short stature, with reported incidence from 1 in 10,000 to 1 in 30,000 live births and affecting over 250,000 people worldwide. Achondroplasia is caused by a genetic mutation which decreases chondrocyte proliferation with consequent reduction in endochondral bone formation resulting in the observed proximal limb shortening. The main clinical features typically seen in achondroplasia are: disproportionate short limbs when compared to the trunk, macrocephaly with frontal bossing, and 'trident' shaped hands. Typical radiographic findings include squaring of the pelvis, 'chevron shape' on distal femoral epiphyses, and small vertebral pedicles. Diagnosis can be confirmed by genetic testing.

A genetic diagnosis is made by testing for a mutation in the Fibroblast Growth Factor Receptor 3 (FGFR3) gene, with 99% of cases being caused by a Gly380Arg substitution. Around 75% of children with achondroplasia are born to two parents of average stature, representing a de novo mutation. There are several complications associated with achondroplasia, including increased incidence of sleep-related disordered breathing, hearing loss, and narrowing of the foramen magnum. Parents should be given advice and information on these aspects, as well as other aspects of the development and care of children with achondroplasia.

**Management**

1. **Genetic Testing and Counselling**
    - Genetic counselling should be provided for the family to understand the implications, inheritance patterns and potential for recurrence in future pregnancies. This may useful both antenatally and postnatally.

2. **Respiratory Assessment**
    - Infants should have a clinical examination of the respiratory system shortly after delivery, and oxygen saturations checked if any concerns.
    - Prior to discharge, children with achondroplasia should have a car seat challenge to assess for sleep-related breathing disorders (discuss with neonatal team, as per local protocol for car seat challenge.)
    - Where there are concerns related to breathing during sleep, or abnormal car seat challenge, refer to respiratory team for overnight oximetry and respiratory study as an in-patient.
    - In infants with no concerns identified, arrange for overnight oximetry study within the first 3 months and make an outpatient respiratory referral.
    - Continue to monitor for sleep-related disordered breathing, with 6-12 monthly pulse oximetry until age 5 years.
    - Advise parents that most children with achondroplasia snore, and to observe for other features of obstructive sleep apnoea, including intermittent breathing, choking, glottal stops, or deep compensatory sighs.

2. **Imaging**
    - It is recommended to perform a cranial ultrasound for all infants with achondroplasia before discharge to detect any signs of hydrocephalus and assess the ventricles.
    - If ultrasound does not indicate any abnormalities, MRI should be performed within three months to provide a more detailed assessment of the brain. MRI brain and spine should be requested to look for cranio-cervical junction stenosis.
    - If ultrasound indicates any abnormalities such as hydrocephalus, then results may be discussed with neurosurgical team and further imaging arranged.

3. **Growth Advice**
    - Prior to discharge parents should be informed about the expected growth pattern for children with achondroplasia; the importance of regular follow-up with healthcare providers should be emphasised to monitor weight and length.
    - Prior to discharge parents should be provided with the copy of an appropriate achondroplasia growth chart to be added to their red book for monitoring of growth in the community **(see Appendix 1).**
    - Regular follow up every one or two weeks with health visitors should be organised to measure child's head circumference in the first six months of life. Health visitors should be advised to contact neonatal team in the first instance if there are any concerns.

4. **Developmental Advice**
    - Parents should be provided with information about average expected time for developmental milestones for children with achondroplasia and be advised that it is often delayed in comparison to children without achondroplasia **(see Appendix 2).**

- Parents should be provided with guidance on positioning and handling, adaptive equipment, and activities to support their child's development (e.g. choosing appropriate car seat). **(See Appendix 2).**
- Physiotherapy can help to address motor development issues. The referral is expected to be made by the bone clinic based on child's individual needs.

5. **Bone Clinic Referral**
    - A referral to Glasgow Children's Hospital Achondroplasia MDT clinic on discharge is recommended to provide ongoing care.

6. **Hearing Assessment**
    - Ensure universal newborn hearing screening programme is performed within first four weeks and results reviewed, with audiology follow-up arranged if necessary.
    - Formal behavioural audiometric assessment is recommended to be carried out by 9-12 months.
    - It is recommended that formal hearing assessment is carried out routinely for children with achondroplasia, ideally annually, and care established with an ENT surgeon if required.
    - Children residing within NHS Greater Glasgow and Clyde should be referred to audiology to take part in Annual Hearing Screening Programme for Children with Achondroplasia. Local arrangements may exist for children in other health boards.
    - Advise parents on the risk of serous otitis media secondary to short Eustachian tubes and recommend ear examination is indicated in presence of ear pain, or any persistent upper respiratory tract infection.
    - Any child with language delay should have a chronic serous otitis media and resulting conductive hearing loss excluded.

7. **Family Support and Counselling**
    - Families of the infants affected with achondroplasia should be offered guidance and counselling in order to provide them with appropriate emotional and informational support and help to navigate potential challenges
    - Functional difficulties and potential challenges should be discussed, while emphasising that most people with achondroplasia can live self-sufficient lives.
    - The infant's family should be offered psychological and social support to address their concerns and provide them with coping strategies.
    - Involvement in support groups or organizations specialising in achondroplasia should be encouraged (e.g.: https://www.dsauk.org/, https://littlepeopleuk.org/, https://rgauk.org).

**Document Properties**

**Document Title**

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**Implementation and review dates**

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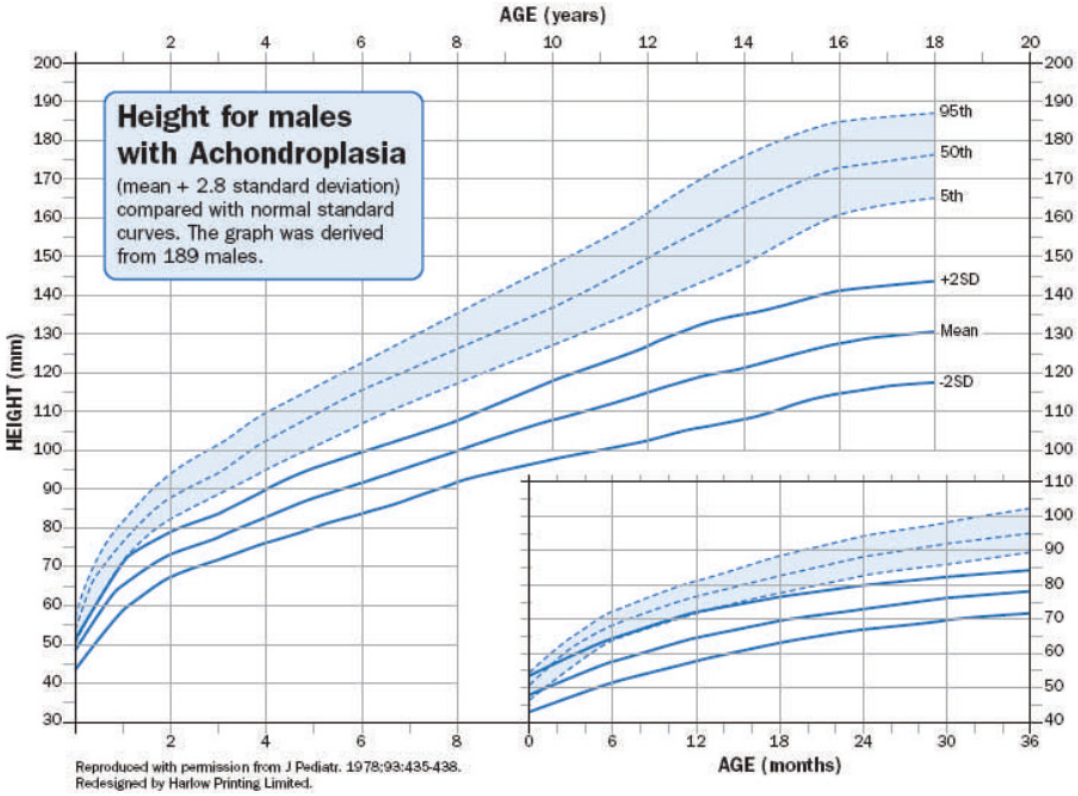
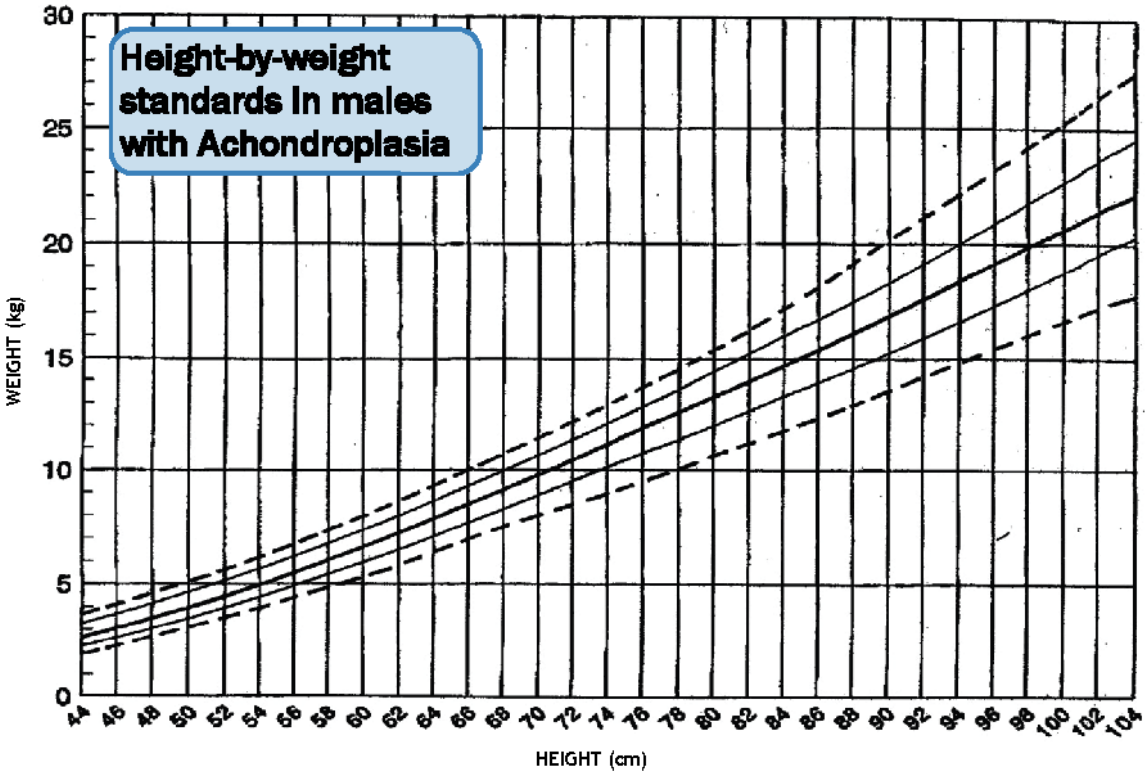
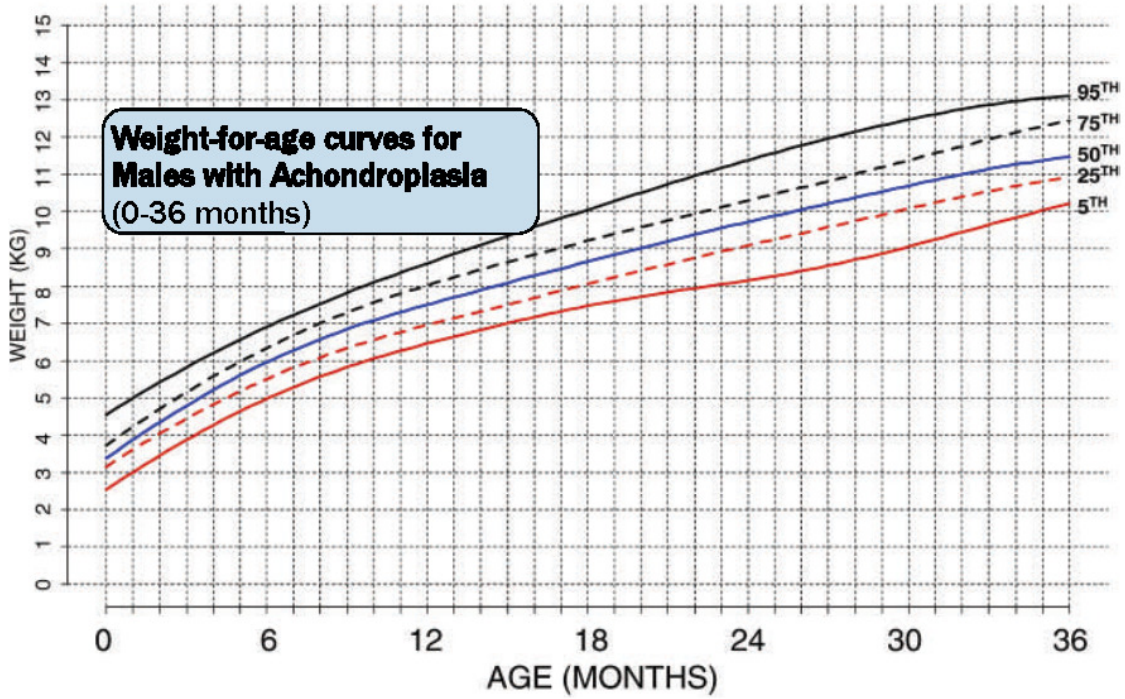
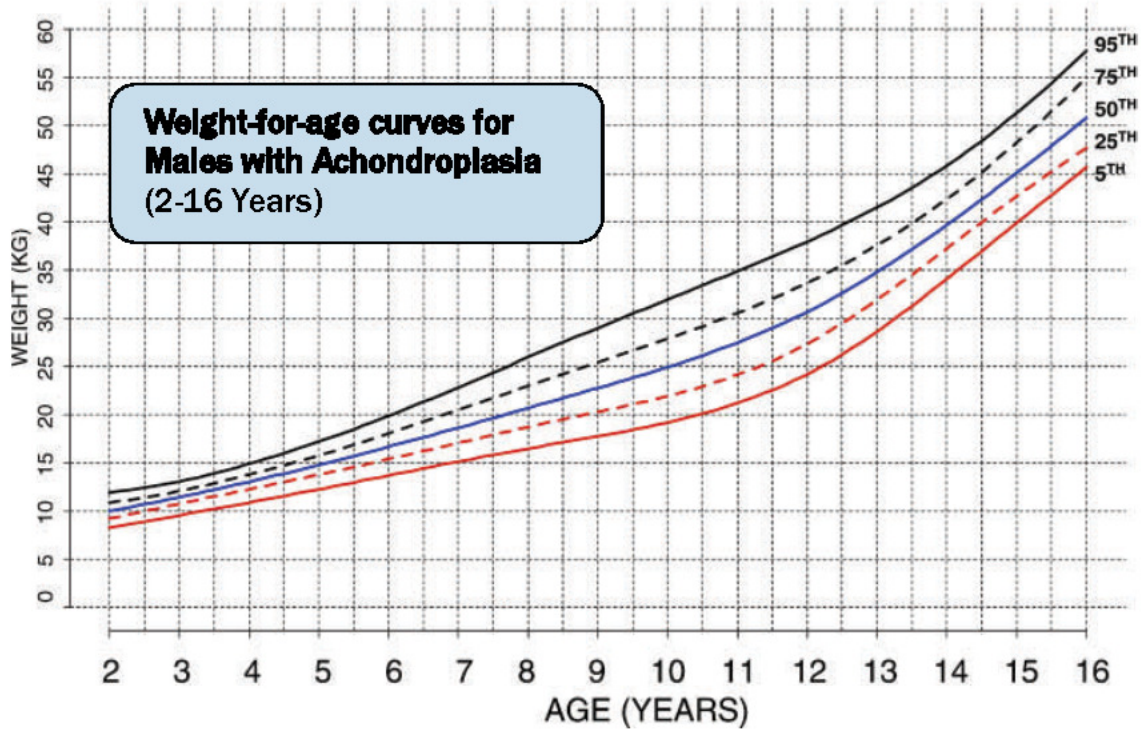
|---|---|---|---|
| Implementation date | 15/5/25 | Next review | 15/5/28 |

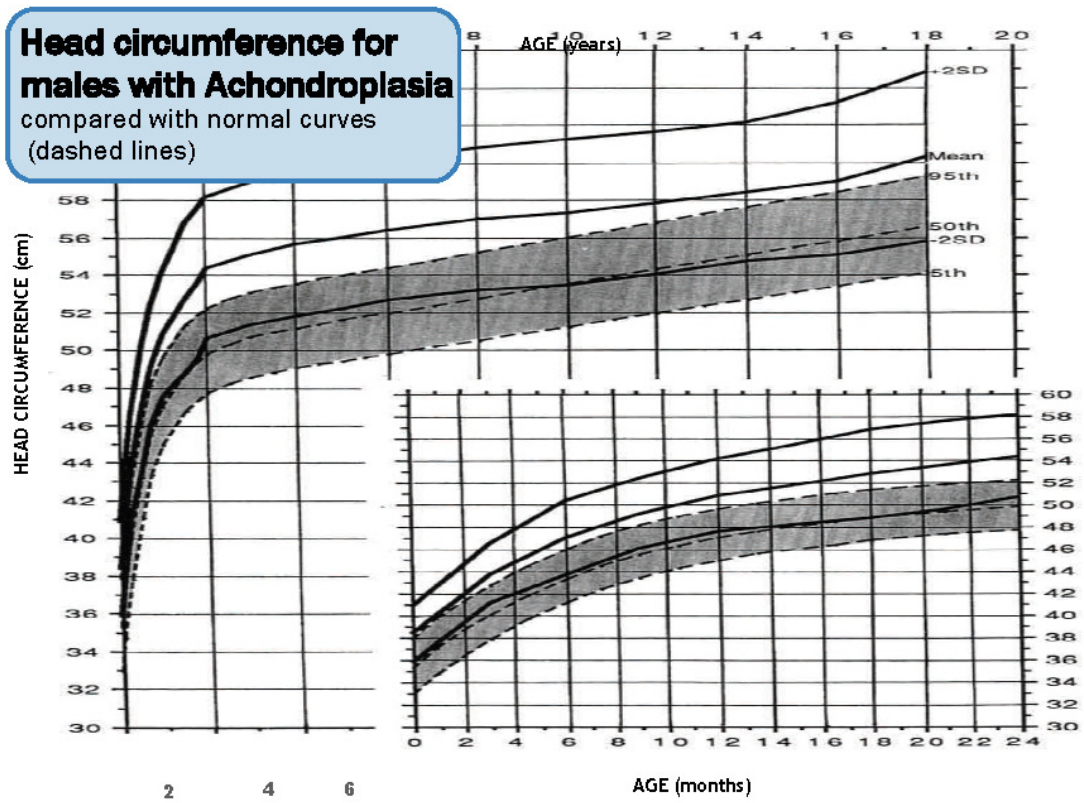
**Appendix 1 – Relevant Growth Charts**



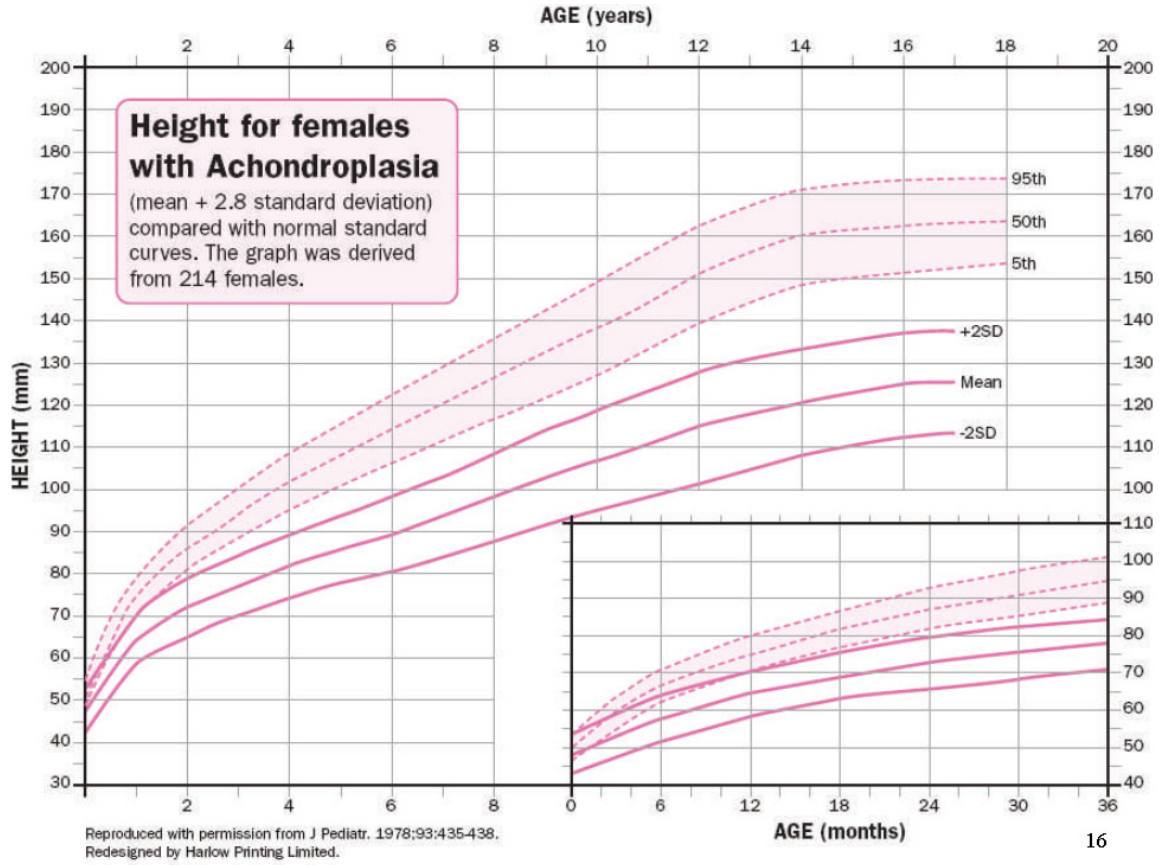








Greenwood SC, Growth References: Third Trimester to Adulthood. 2nd ed. (1998) 107-110 Copyright (1998 Greenwood Genetic Centre) Reprinted with permission of Greenwood Genetic Centre.


Height for females with Achondroplasia
(mean + 2.8 standard deviation) compared with normal standard curves. The graph was derived from 214 females.
95th
50th
5th
+2SD
Mean
-2SD
HEIGHT (mm)
AGE (years)
AGE (months)
Reproduced with permission from J Pediatr. 1978;93:435-438.
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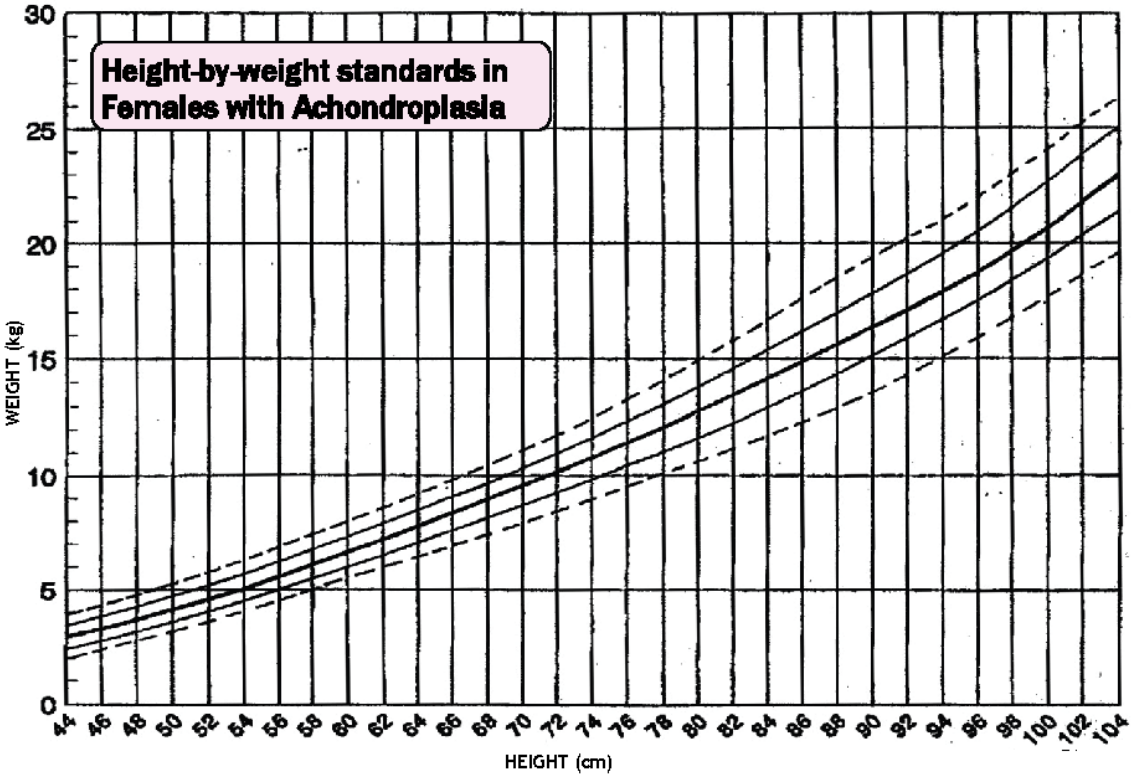
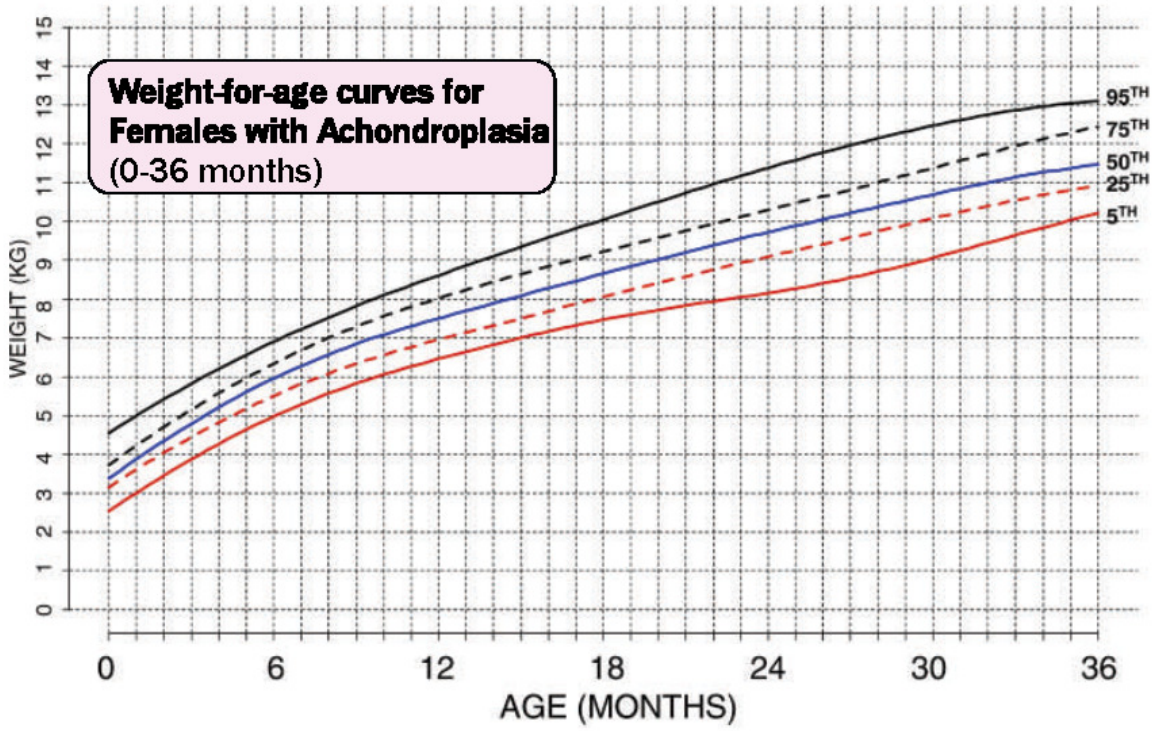
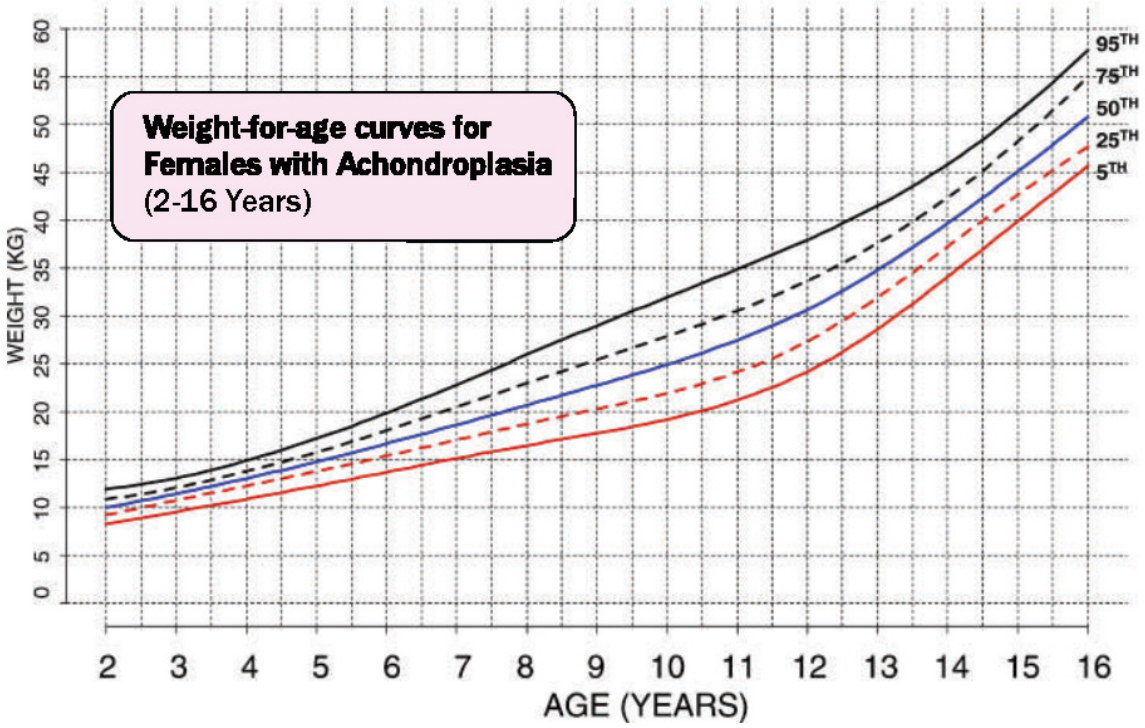
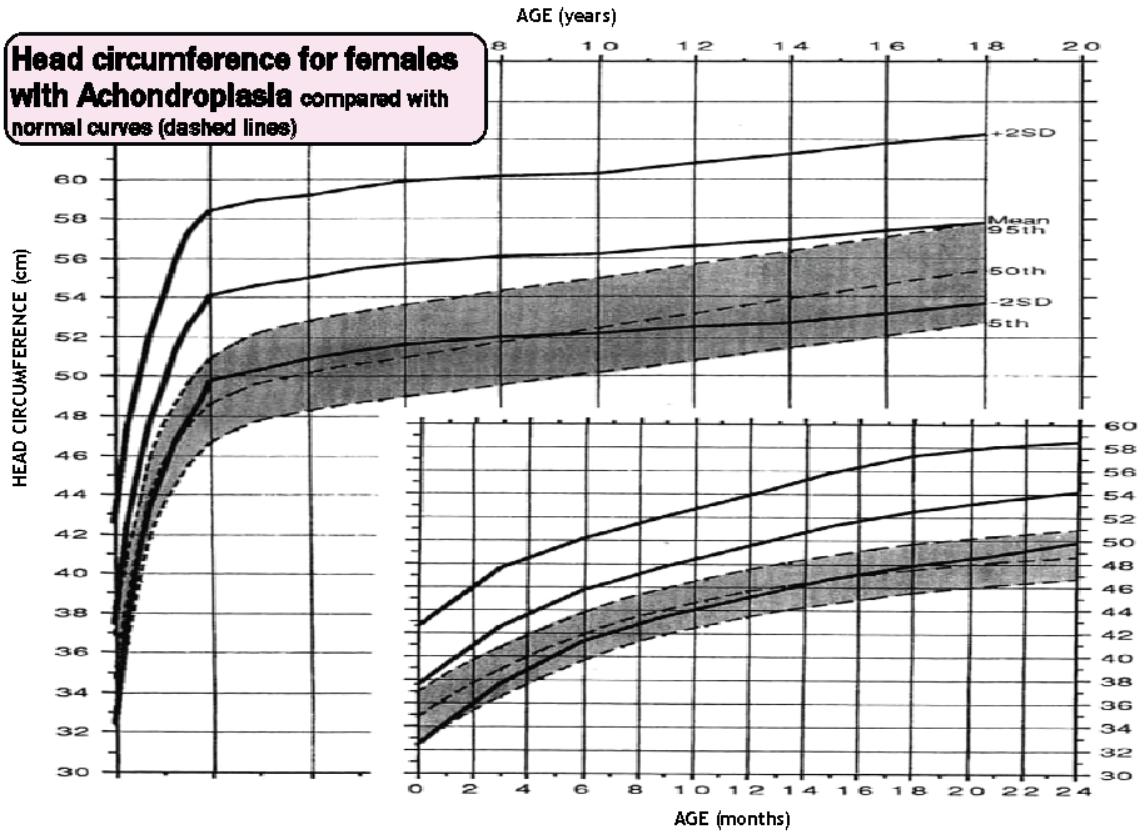
16


Height-by-weight standards in Females with Achondroplasia
WEIGHT (kg)
HEIGHT (cm)



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20

NHS
Greater Glasgow and Clyde

Information about

# Achondroplasia

Royal Hospital for Children

## What Is Achondroplasia?

Achondroplasia is a condition where the bones do not grow as much as usual. The condition affects how some of the bones develop, particularly the limb bones and specifically the upper arms and thighs. About 1 in 25, 000 babies born in the UK has achondroplasia.

## Why does Achondroplasia happen?

Achondroplasia is a genetic condition. You may have heard about genes which are the instructions for how our body develops and works. We have many thousands of genes which have different jobs to do within the body. Our genes come in pairs because we get one from each of our parents. Achondroplasia happens when one copy of a particular gene (called FGFR3) does not work normally. This gene is important in how our bones grow. Achondroplasia most often happens 'out of the blue' in a baby whose parents are of normal height. It is not related to anything you did or didn't do in pregnancy.

If you want more information about this you can talk to a Genetics doctor.

## Support and advice for your new baby:

This information sheet explains the recommended positioning, handling and seating advice (car travel and feeding), for infants with achondroplasia. If you have any further questions or concerns, please speak to a doctor or therapist caring for your child.

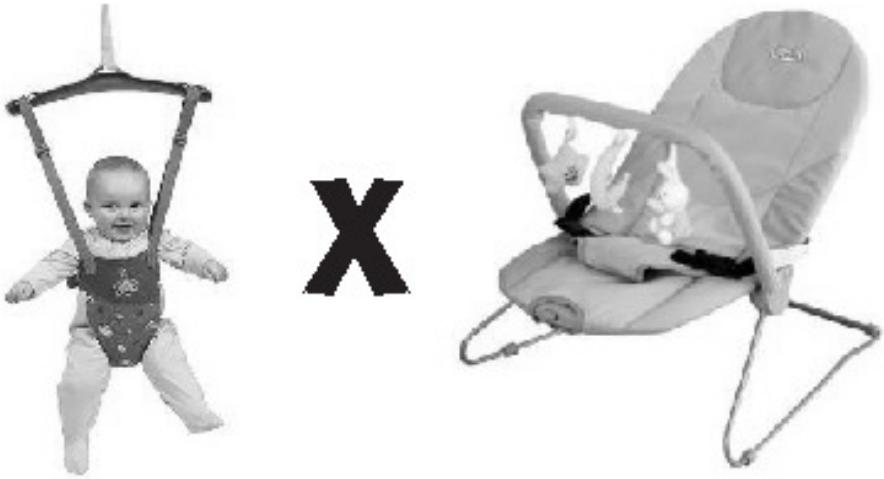
## Positioning and handling in relation to your child's back and spine:

Infants with achondroplasia have a spinal curve called a gibbus which often resolves as their body strength improves and may disappear when they start to walk. The recommended positions when your baby is awake are on their tummy, side or back. These positions help the gibbus to resolve. If sitting with support is encouraged too early the spinal gibbus can be made worse.

**Advice is:**

- When being handled and moved between positions, we advise your baby is held in a supported position with a flat spine, avoiding a 'C' shaped curve.
- When lifting and carrying your baby make sure you support their head, neck and lower back. When winding your baby, avoid placing in a seated position. One example is to support the baby upright against your chest.
- Your baby or infant should not practice or be placed in an unsupported sitting position with a 'C' shaped curve of the spine until they can move themselves into sitting.
- Soft canvas baby rockers and bouncers, baby walkers, carriers and strollers do not provide enough neck and spine support and should not be used.
- Infant bouncers, swings and trampolines should be avoided. Care should be taken with any activity that places a strain on their neck.

## These types of play equipment are not recommended



## How can you support your child to develop?

- Play-Allow your baby or infant to lie flat on a mat on the floor for play. Placing them on a play mat will encourage the development of muscle strength while supporting their back and head. When they have developed enough strength they will be able to lie on their side and to roll. Your baby or infant will benefit from tummy time to help develop the strength in their back for managing their gibbus. To begin with, position a small rolled towel under their chest until they are more tolerant of this position and can lift their head.
- Bath Time - any seating used for bath time should be supported with a 30 degrees reclined from upright with a firm back. This is important to help your baby or infant maintain a flat and elongated spine.
- Feeding - When weaning and feeding, if a high chair is too big for your baby or infant, rolled up towels or inserted cushions or pillows can be used to fill spaces, which will provide them with additional support. Keep your baby to 30 degrees.

- Key transitions - Occupational therapists can assess and support with key transitions such as starting nursery and school. They can provide advice and recommendations on environmental adaptations and any supportive equipment that may be needed.

## Guidelines for Car Seat Use – Infants with Achondroplasia

Providing safe motor vehicle transport for infants is a priority for parents and health professionals worldwide. It is essential to provide adequate head and spine support for children with achondroplasia to minimise the risk of complications that may occur. We recommend the use of a suitable car seat to provide supportive positioning for your child.

## Choosing an appropriate car seat

Correct car seat positioning is vital. Babies with achondroplasia are at risk of breathing difficulties particularly if they fall asleep when upright. Therefore we recommend a lie-flat travel seat and to keep car journeys the shortest time possible and frequent breaks and positional changes. This will also provide support for their spine.

**NB.** Please be aware that NHS staff cannot suggest brands or models of child car seats, as they are unable to endorse products.

## Developmental milestones

Your child's developmental milestones will be different to other children. They may learn to move in different ways due to their body proportions. It may be helpful to refer to developmental charts specific to children with Achondroplasia like the one below:

| Skill | Usual range for children with Achondroplasia | Average for children without Achondroplasia |
|---|---|---|
| Stand alone | 16-29 months | 11-12 months |
| Walk | 14-27 months | 12-14 months |
| Reach | 6-15 months | 3-4 months |
| Pass Objects | 8-14 months | 6 months |
| Bang 2 Objects | 9-14 months | 8-9 months |
| Scribble | 15-30 months | 13-14 months |

Adapted from achondroplasia developmental recording, Ireland. Dongahey, McGill, Zankl, Ware, Johnson, Pacey, Ault, Savarirayan, Sillence, Thompson, Townshend, Johnston 2011.

## Useful sources of Information and support groups

Some families find it helpful to link in with support groups in order to meet and share experiences with other parents, carers and children with achondroplasia and short stature. Supporting organisations that we frequently signpost families to include:

- Little People UK: http://littlepeopleuk.org/
- The Restricted Growth Association: http://rgauk.org/
- Dwarf Sports Association: https://www.dsauk.org/

## Notes

Review Date: July 2025 • 338373 v1.0