

Achondroplasia

Achondroplasia is the most common form of disproportionate short stature (DSS). It is also known as dwarfism and restricted growth. Achondroplasia changes how the bones develop which affects the way children grow, particularly their upper arms and thighs.

Many people with achondroplasia do not have serious health problems and lead a relatively normal life. However some health problems can occur as a result.

This information explains:

- Causes
- Characteristics
- Investigations
- Advice and recommendations about positioning and handling babies
- Advice about choosing the right car seat

Causes

Achondroplasia is a genetic condition caused by a change in the FGFR3 gene.

It doesn't always run in the family. In 80 out of 100 cases the child is the first in the family to have it. If a parent has achondroplasia, around 50 out of 100 cases their child will inherit the condition.

Characteristics of achondroplasia

The characteristics of achondroplasia will vary from child to child. Typically children have an average sized body, but short arms, legs and fingers.

Characteristics could include:

- A larger head and a more prominent forehead
- A sunken bridge of the nose
- Changes in their back
- Possible overcrowding of teeth
- Joints are very flexible, except for the elbows, which may not fully straighten.

Complications

Most children can lead a normal life. However they may develop some health problems and these can occur at different times.

These include:

- Children with achondroplasia are born with a kink or curve (kyphosis or gibbus) seen in the lower part of their spine. This can worsen if positioning and handling guidance is not followed when they are a baby – please see guidance on the next page
- As they get older, back problems might occur lower down in the spine if a complication called spinal stenosis develops.
- Bowed legs (tibial bowing)
- Glue ear

Investigations

MRI scan

All infants and children with achondroplasia have an MRI scan of the brain and spine.

Sleep study

We will carry out a detailed sleep study in hospital to look for foramen magnum stenosis. Some infants with achondroplasia have compression of the spinal cord at the top of the neck. This is caused by a narrowing of a channel in the base of the skull. This is called foramen magnum stenosis. It is a serious condition that can cause breathing difficulties and sudden infant death if not detected early enough.

If it is found, your child will be referred to a specialist neurosurgeon for a review to see if they need surgical treatment.

Growth and development charts

Your child's development milestones will be different to others. They may learn to move in different ways due to their body proportions. We will give you charts to complete to record their growth and development. These charts are specific to children with achondroplasia.

Our team of doctors, nurses and therapists will monitor your child on a regular basis and link your child with their community based team.

Advice and recommendations for positioning and handling your baby

Children with achondroplasia have a kink or curve in their lower spine called a gibbus. This often improves as they gain body strength and may disappear when they start to walk.

Do not encourage your child to sit upright too early, even with support, as this can make the spinal gibbus worse. Therefore lots of floor play is recommended until your child is strong enough to sit without support.

While your baby is gaining strength, the recommended positions when they are awake are on their tummy, side or back. These positions can help the gibbus to resolve on its own.

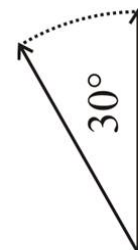
'Tummy time' is recommended for your baby as it will help strengthen their back. It might take time for them to get used to this position and to be able to lift their head, so a small towel can be placed under their chest as it will give them some support. Practicing tummy time frequently but for very short periods of time is recommended to start with.

When being handled and moved between positions, your baby should be held in a supported position with a flat spine, avoiding a 'C' shaped curve. When lifting and carrying them, make sure you support their head, neck and lower back. When your baby needs winding hold them upright against your chest and avoid putting them in a seated position.

Activity that puts a strain on the neck should be avoided, so please do not use infant bouncers, swings and trampolines. Do not use soft canvas baby rocker or bouncer chairs, carriers, strollers or baby walkers as they do not provide enough neck and spine support.

Seating for feeding and bathing

When your baby reaches the weaning stage, seating that reclines to 30 degrees (30°) from upright should be used. Make sure there is firm support for their back. This is important to help your child maintain a flat and elongated spine. If a high chair is too big for your child, rolled up towels, cushions or pillows can be used to fill spaces to give them extra support. Seating should only be used at mealtimes for up to 20 minutes and not for any other activity until your baby can sit up by themselves.



At bath time, please support your baby to lie flat in the bath. If seating is used, please ensure that it reclines to 30 degrees (30°) from upright as well – please do not leave your baby unattended near or in water.

First aid

Our staff will give you important information to help you care for your child at home. Additionally we recommend that you are familiar with basic life support training.

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 restricted growth. You might find these organisations helpful.

Little People UK, <http://littlepeopleuk.org/>

The Restricted Growth Association <http://rgauk.org/>

Dwarf Sports Association <https://www.dsauk.org/>

[NHS.uk](https://www.nhs.uk) has more information about the condition, symptoms and treatments.

Car seats

During car journeys babies with achondroplasia must travel in a lie-flat car seat. There is a significant risk of breathing difficulty occurring if they fall asleep when they are upright. Sleeping in a flat position helps to avoid this and provides support for the spine.

A lie-flat car seat will be required when you and your baby are discharged from hospital.

Guidelines for lie-flat car seats

- Always follow the fitting instructions as directed by the manufacturer.
- Our staff are unable to help parents with fitting car seats.
- Make sure that the 5-point harness is not twisted and it is positioned no more than 2cm below your child's shoulders. The harness should be snug and be positioned in your child's pelvic area. It should not be in contact with their stomach.
- Try to avoid car journeys of more than 30 minutes.
- If possible, have an adult sit in the back of the car with your child to help with repositioning as needed.

Useful sources of information

Our staff cannot suggest brands or models of child car seats, as they are unable to endorse products. You may find helpful information on these web sites.

- Child Car seats. Available at: <https://www.childcarseats.org.uk/types-of-seat/lie-flat-seats/>
- RoSPA Road Safety Research. Available at: <https://www.childcarseats.org.uk/media/1028/carrying-premature-and-low-birth-weight-babies-evidence-review.pdf>
- In car Safety centre – provides support for car seat selection. Available at: <https://incarsafetycentre.co.uk/>

Contact us

If you have any questions or concerns about achondroplasia or any of the information in this leaflet, please contact the Clinical Nurse Specialists **tel:** 020 7188 7188 ext 56129, Monday to Friday, 9am to 5pm. Out of hours, please contact the General Paediatric Registrar on call via the switchboard on 0207 188 7188.

For more information leaflets on conditions, procedures, treatments and services offered at our hospitals, please visit **web:** www.evelinalondon.nhs.uk/leaflets

Evelina London Medicines Helpline

If you have any questions or concerns about your child's medicines, please speak to the staff caring for them or contact our helpline, **tel:** 020 7188 3003, Monday to Friday, 10am-5pm, **email:** letstalkmedicines@gstt.nhs.uk

Your comments and concerns

For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS), **tel:** 020 7188 8801, **email:** pals@gstt.nhs.uk. To make a complaint, contact the complaints department, **tel:** 020 7188 3514, **email:** complaints2@gstt.nhs.uk

Language and Accessible Support Services

If you need an interpreter or information about your care in a different language or format, please get in touch, **tel:** 020 7188 8815, **email:** languagesupport@gstt.nhs.uk

NHS 111

Offers medical help and advice from fully trained advisers supported by experienced nurses and paramedics. Available over the phone 24 hours a day, **tel** 111, **web:** 111.nhs.uk

NHS website

Online information and guidance on all aspects of health and healthcare, to help you take control of your health and wellbeing, **web:** www.nhs.uk

Get involved and have your say: become a member of the Trust

Members of Guy's and St Thomas' NHS Foundation Trust contribute to the organisation on a voluntary basis. We count on them for feedback, local knowledge and support. Membership is free and it is up to you how much you get involved. To find out more, please get in touch, **tel:** 0800 731 0319, **email:** members@gstt.nhs.uk, **web:** www.guysandstthomas.nhs.uk/membership

Was this leaflet useful?

We want to make sure the information you receive is helpful to you. If you have any comments about this leaflet, we would be happy to hear from you, fill in our simple online form, **web:** www.guysandstthomas.nhs.uk/leaflets, or **email:** patientinformationteam@gstt.nhs.uk

Leaflet number: 5307/VER1

Date published: June 2022

Review date: June 2025

© 2022 Guy's and St Thomas' NHS Foundation Trust
A list of sources is available on request