

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:

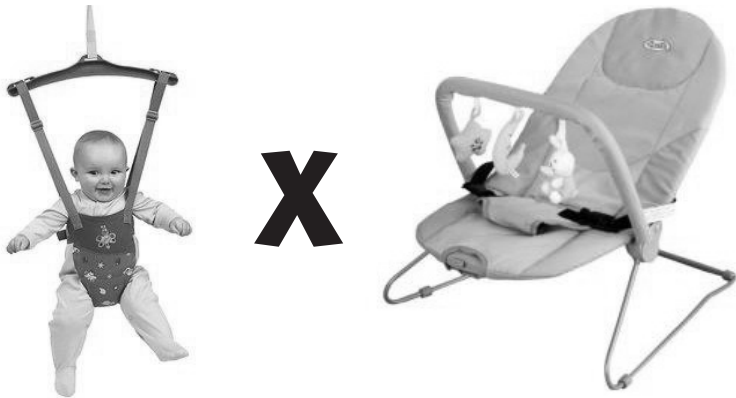
Infants with achondroplasia have a spinal curve called a gibbus which often resolves as their body strength improves and may disappear when they start to walk. The recommended positions when your baby is awake are on their tummy, side or back. These positions help the gibbus to resolve. If sitting with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

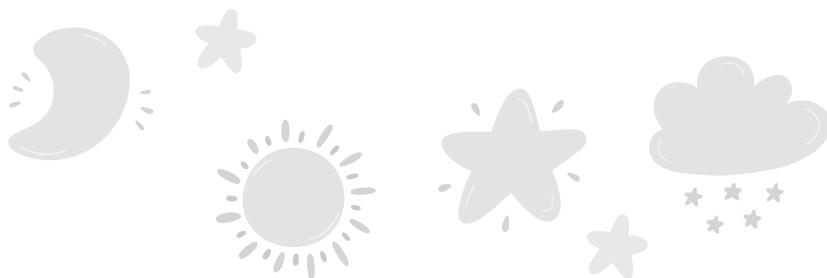
Guidelines for Car Seat Use – Infants with Achondroplasia

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/>

