ACHONDROPLASIA

AN INFORMATION SHEET FOR PARENTS

What is Achondroplasia?

Achondroplasia is a condition where the bones do not grow as much as usual. People with achondroplasia are usually healthy but short. About 1 in 25,000 babies born in the UK has achondroplasia. Sometimes scans during pregnancy will have noticed that the baby’s bones are shorter than usual. Often, once a baby is born a problem with their bones is suspected. X-rays and a genetic test will usually be done to make the diagnosis.

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 and is no one’s fault. If you want more information about this you can talk to a Genetics doctor.

How will Achondroplasia affect my child?

Children and adults with achondroplasia are shorter than average. This may not be obvious until children go to nursery and you notice they are smaller than their friends. Just as height is different in people who don’t have achondroplasia, it varies in adults with achondroplasia. The average height for adult women with achondroplasia is 124cm (4 feet 1 inch) and in men 131cm (4 feet 4 inches) but may vary from about 110cm - 143cm (3 feet 8 inches - 4 feet 9 inches).

Most people with achondroplasia are healthy but occasionally complications may occur. It will be important for your child to be monitored for these so they can be treated.

How will my child be monitored?

Growth - Your child’s growth will be monitored and there are special growth charts for children with achondroplasia which should be used. Healthy eating and regular exercise are important for all children, including those with achondroplasia, especially in later childhood.

Bones and Joints - Children with achondroplasia are often quite loose jointed (also called hypermobility) but this usually gets better as they get older. Your Physiotherapist and Occupational Therapist can advise about this.

Breathing - Often babies and children with achondroplasia snore. If this is very loud or you notice that your child has irregular breathing when asleep, you should tell your doctors. A special study of breathing during sleep will be done routinely to check that there are no problems.

Hearing - Ear infections and ‘glue ear’ where there is extra fluid in the inside of the ear are more common in children with achondroplasia. If you have any worries about your child’s hearing, tell your doctors and a hearing test can be arranged.
**Teeth** - Teeth may be overcrowded so it is important to see a dentist regularly.

**Development**

Children with achondroplasia take a little longer to do things like sit up and walk. This is nothing to worry about and happens because their head is larger in proportion to the rest of their body and it takes longer for them to develop their strength.

People with achondroplasia have normal intelligence.

The table below shows the ages at which children with achondroplasia usually do things like sit up and walk.

<table>
<thead>
<tr>
<th>Usual range for children with Achondroplasia</th>
<th>Skill</th>
<th>Average for children without Achondroplasia</th>
</tr>
</thead>
<tbody>
<tr>
<td>9-20 months</td>
<td>Sit alone</td>
<td>5-6 months</td>
</tr>
<tr>
<td>16-29 months</td>
<td>Stand alone</td>
<td>11-12 months</td>
</tr>
<tr>
<td>14-27 months</td>
<td>Walk</td>
<td>12 months</td>
</tr>
<tr>
<td>6-15 months</td>
<td>Reach</td>
<td>3-4 months</td>
</tr>
<tr>
<td>8-14 months</td>
<td>Pass Objects</td>
<td>6 months</td>
</tr>
<tr>
<td>9-14 months</td>
<td>Bang 2 objects</td>
<td>8-9 months</td>
</tr>
<tr>
<td>15-30 months</td>
<td>Scribble</td>
<td>13.5 months</td>
</tr>
</tbody>
</table>

**Is there anything different in caring for a baby with Achondroplasia?**

**Lifting & Holding** - As with all newborn babies, it is important that you support your baby’s head and neck when caring for them. Babies with achondroplasia need their head to be supported for longer than babies without achondroplasia.

They will slowly develop muscle strength in their trunk and neck to do this without your support but, until this time, please make sure that you provide the extra support needed.

**Seating** - Avoid letting your baby sit unsupported or in a ‘C’shape position. We need good muscle strength in the trunk and neck in order to sit up.

Bouncing or canvas cradles do not provide enough support and are not recommended. Well-structured supportive seats which allow your baby to lie in a semi-reclined position can be used.
Car seats should be rear facing for as long as possible and should support the back and neck. Your therapist will advise on a good supportive seating position if necessary.

**Play** - Allow your baby to lie flat on a mat on the floor for play. Placing him on the mat will encourage the development of muscle strength while supporting his back and head. When he has developed enough strength he will be able to lie on his side and to roll. When he is able to support his head, he can lie on his tummy for play.

Your child's Occupational Therapist can provide further guidance as needed.

**Additional Support** -

Support and information are available from family support groups including:

- [Short Stature Scotland](#)
- [UK Restricted Growth Association](#)