What is Achondroplasia?

Restricted Growth Association

Working to benefit people with restricted growth and their families

This booklet has been written to provide information and support for parents of children with achondroplasia, people with the condition, their families, friends, teachers and health care professionals.

Dr Will Christian is a paediatrician who himself has achondroplasia (restricted growth).

RGA acknowledges the help of the many members and their families who have contributed their experiences towards this booklet. Their contributions were compiled by RGA Information Officer, Ros Smith.

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'I have spent 14 years, since my son was born with achondroplasia, trying to gain this level of knowledge about his condition to be able to communicate with doctors and other health professionals and to help him to have the best possible chances in life. I hope that this booklet ensures that other people do not have the same struggle for information and understanding.'

The contents of this booklet are believed to be correct at the time of publication. However, knowledge about achondroplasia is growing all the time, especially in the areas of genetics and orthopaedics. We encourage you to find out about new developments from the RGA and other sources, some of which are listed at the end of this booklet.

What is Achondroplasia?

by Will Christian MBBS, BSc

'When we had learnt to enjoy our child for who he was, we found we had the most beautiful, happiest baby in the world.

When your baby is diagnosed with achondroplasia, you do not really know what it means. You think only of all the problems they will face in future years. All the possible difficulties are rolled into one giant mountain instead of a series of small molehills. You need to learn to take one day at a time and then his personality will shine through.'

Achondroplasia is a rare condition but it is one of the most common types of restricted growth with disproportionate stature (in this case this term refers to people with an average size body but shorter limbs). As with many causes of short stature, achondroplasia is mainly a physical condition - people with the condition have a normal range of intelligence and occupy positions in all sectors of society.

When the initial shock of the diagnosis has settled, many parents are anxious to know what their child's future will be. Achondroplasia is a condition that covers a wide range of heights and abilities. Most people with achondroplasia live normal, happy lives although there may be, of course, some physical limitations.

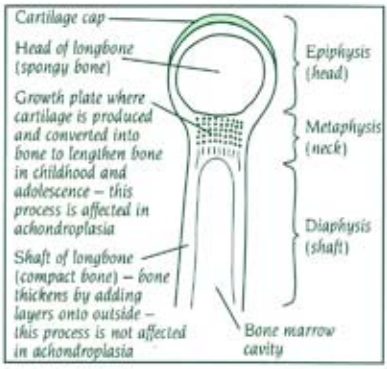
Many practical difficulties can be overcome with a little imagination. Clothes can be altered, and cars/bicycles can be adapted (for more information see the RGA **Lifestyles** series), and there is no reason why someone with achondroplasia should not participate in most activities. All children (and adults!), including those with achondroplasia, will find their own limits and boundaries, and it is important that, within reason, these are not imposed upon them by others or by society.

'The most helpful things for us were meeting someone who had achondroplasia, who showed so obviously that a full and dignified life was possible, and knowing that there were others in the same boat as ourselves, who understood.'

What does achondroplasia mean?

Achondroplasia literally means 'no cartilage growth' but in reality it refers to poor cartilage growth. It is a genetic condition, beginning when a baby is still in the womb and continuing throughout life.

In the growing baby and child bone forms from cartilage. Cartilage is produced at the ends of the long bones in an area called the growth plate. In people with achondroplasia, the growth plate works more slowly than normal. This means that only a small amount of cartilage is produced in the arms and legs for conversion into bone. The result is short limbs. The tissues (muscle/blood vessels, etc.) around the limbs are not affected by the growth plate, and continue to grow. This is why people with achondroplasia tend to have much bulkier legs and arms. It is as if the muscles were waiting for the bone to finish growing.



Development of long bones

The average height for both men and women with achondroplasia is between 1m 10cm (3ft 6in) and 1m 40cm (4ft 6in).

However, the variation amongst individuals is great, and some may be shorter or taller than this.

Indeed, paediatricians and GPs should use growth charts designed specifically for children with achondroplasia to predict their growth rates and expected heights.

The charts below, along with others showing head circumference, growth velocity and upper-lower segment ratios, can be obtained from the RGA *(address at the end of this book).*

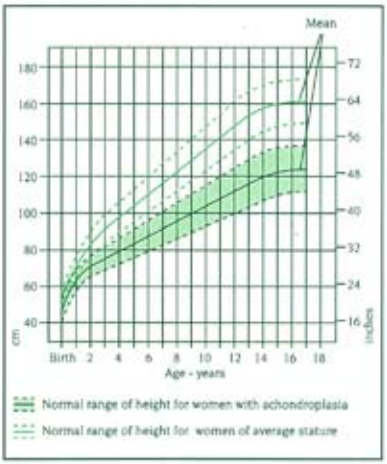


Diagram of height chart of females with achondroplasia

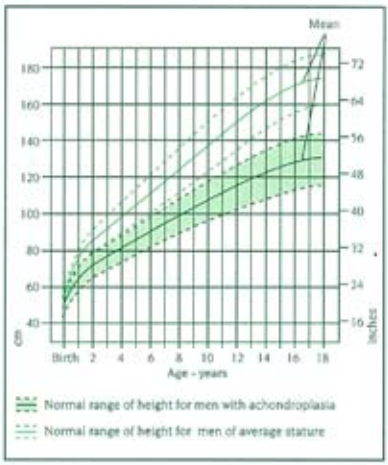


Diagram of height chart of males with achondroplasia

Other effects of achondroplasia

People with achondroplasia are as varied in appearance as people without achondroplasia, although as a group they may share certain physical characteristics. These include:

- a near-normal body length with short arms and legs
- a large head with prominent forehead and flattened bridge of nose
- an increased curvature of the lower spine (lumbar *lordosis**)
- bowing of lower legs
- possible crowded or crooked teeth with a protruding chin
- short, broad, flat feet and short hands with possible separation between middle and ring fingers (the so-called 'trident' hand)
- a tendency to have exceptionally flexible joints.

Robert and Philip are twins

Babies with achondroplasia may develop motor skills and mobility more slowly than normal, because of the combination of a heavier head with shorter arms and legs. They also may be more floppy than babies without achondroplasia (*hypotonia*). No one knows the exact reason for this but it is normal and resolves with time. Talking may also be delayed whilst the child concentrates on becoming mobile, but this is no reflection of intelligence. Ultimately, development is within the expected range, as the following quote from a mother shows:

* Words in italics are found in the glossary

'It was tempting to compare my baby with his older sister who actually walked and talked quite early. But Ben was always happy and contented, and now he never stops talking!'

Possible complications

There are some medical complications that are often associated with achondroplasia. However, it can be difficult for a person of short stature to know whether or not a particular symptom is related to their condition. Unless the family doctor is familiar with achondroplasia (which is unlikely) he or she may be equally uncertain.

Structural difficulties

In children, the skull is made up of large bone plates loosely joined together allowing room for expansion as the brain grows. In children with growth disorders, there may be problems with the rate and co-ordination of growth of these bone plates, leading to distortion of head growth. An example of this is the prominent forehead typical of achondroplasia, which on its own is no cause for concern.

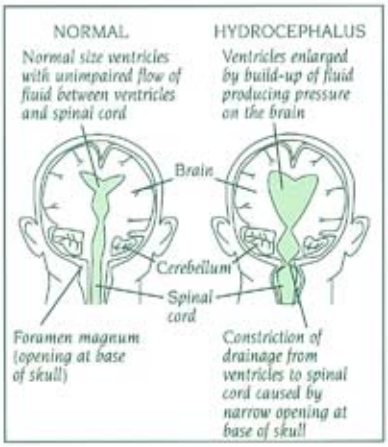
Abnormalities of skull growth can also result in a small jaw and overcrowded teeth. If the jaw grows disproportionately, as it does in achondroplasia, there may then be problems with upper and lower teeth not meeting together properly ('malocclusion') - this can make chewing very difficult. Some teeth may need to be removed and dental work may be necessary - as it is for many young people.

Hydrocephalus

This is a rare but serious complication which may occur in young children with achondroplasia. *Hydrocephalus* (literally 'water in the brain') occurs when there is a build-up of pressure in the fluid that lies within the spaces of the brain (the ventricles). Normally this fluid flows from the

ventricles and down through the spinal cord. In achondroplasia this pathway may become blocked at the level of the junction between the skull and the spinal column (because of a small *foramen magnum*), obstructing the flow of fluid between the brain and the spinal cord.

If this pressure becomes too high, it can cause persistent headaches (sometimes the first sign of developing hydrocephalus), enlargement of the head and even loss of intellectual ability. It is very important for this condition to be picked up at an early stage, when it can be treated effectively; it is therefore essential that your doctor measures the circumference of your child's head at regular intervals (e.g. monthly during the first year of life and then every two months during the second year) so that any increase in size can be picked up. Ultrasound scans may be required if the head circumference increases abnormally, and these can reveal



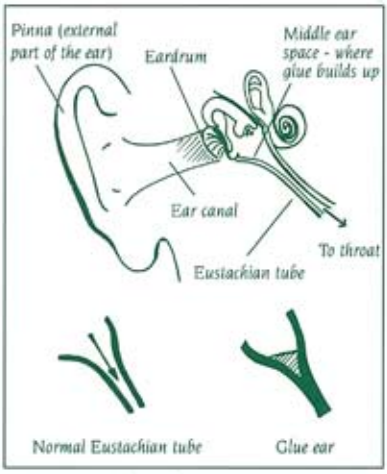
Occurrence of hydrocephalus

whether the skull is expanding more rapidly than is necessary for brain growth.

It is important to remember that people with achondroplasia normally have a larger-than-average head size anyway. Regular measurements of head circumference help to distinguish between this and hydrocephalus. In severe cases of hydrocephalus, insertion of a small drainage tube, called a shunt, into the ventricles may be necessary to relieve the pressure.

Hearing

Babies with achondroplasia often have hearing complications due to structural problems around the inside of the ear. The *Eustachian tube*, which equalises pressure between the middle ear and the outside world, is often narrower in achondroplasia and can become easily blocked. This leads to a build-up of fluid (which may be of the consistency of glue) behind the eardrum.



A *narrow Eustachian tube*

This condition, known as 'glue ear', is a common childhood problem. Unfortunately, it can occasionally result in significant deafness and affects up to $\frac{1}{3}$ of all children.

Glue ear can easily be treated with *grommets* (little tubes that sit in the eardrum and allow the pressure to equalise between the middle and outer ear) and will improve as the child grows older. However, it does mean that regular hearing checks are especially important in young children.

Speaking

As a result of hearing difficulties or problems with the shape of the mouth and face, some children may be slow to learn to speak or their speech may be difficult to understand. Improved hearing or dental work may result in better speech, but some children may need the help of a speech therapist, who should be made aware of the effects of achondroplasia as outlined above.

Breathing

The same problems that lead to overcrowding of the facial features of people with achondroplasia may also lead to respiratory (breathing) problems. These may be especially obvious when the person is asleep, and the soft tissues around the entrance to the windpipe are relaxed and floppy. At its mildest, this can result in noisy breathing and snoring at night and people may sleep with the head tilted right back and the mouth open. If severe, it may actually lead to chronic airway problems. This is because the windpipe is partially obstructed and breathing is difficult. Removing tonsils and adenoids can sometimes improve the situation.

'Michael used to snore like a trooper. But he had his tonsils and adenoids removed at eleven months and the improvement was dramatic.'

Very rarely, young children with achondroplasia have problems with the messages from the brain to the lungs, which are intended to stimulate breathing. This occurs when the hole in the bottom of the skull where the spinal cord leaves the brain (the foramen magnum) is narrower than it should be, causing problems through compression. In some cases, surgery is required to relieve this pressure. Fortunately this situation is rare, but it can be very serious. If you suspect your child is having problems with breathing/sleeping, contact your doctor as a matter of urgency.

All of the problems mentioned above can occasionally lead to a worrying condition in both children and adults known as *sleep apnoea*, where the individual stops breathing for short periods during sleep. This can be a serious problem, often resulting in daytime tiredness and 'cat-napping', and you should tell your doctor about it.

Losing weight may help with this condition and the use of a CPAP (Continuous Positive Airway Pressure) device can have quite dramatic results. More information on this subject can be found in two articles that are available from the RGA:

Tasker, R. (1995) *Respiratory problems in children with achondroplasia;*

Elliott, M. (1997) *Respiratory problems in adults during sleep.*

'It has always been a joke in my family about my snoring, how it could be heard all over the house, and how easily I could fall asleep almost anywhere. I've been using the CPAP machine for five months now and I won't pretend it doesn't take some getting used to, but I do feel so different. I feel much more wide awake, and I don't fight sleep every time I sit quietly or read or watch television or in meetings at work.'

Anaesthetic complications

People with achondroplasia who require a general anaesthetic should make a senior anaesthetist aware of their condition well in advance of the actual operation. It is important to make the anaesthetist aware of any potential difficulties they may face, such as:

- previous anaesthetic complications
- a narrow windpipe
- limited neck movements
- chronic lung disease
- previous adverse reactions to anaesthetics
- spinal problems.

The back

People with achondroplasia are especially prone to back problems. The curvature of the spine is often more exaggerated in the lower back (lumbar *lordosis* – see picture on the next page) and it is also common for the spinal canal (the space in the backbone that carries the spinal cord from the brain to the body) to

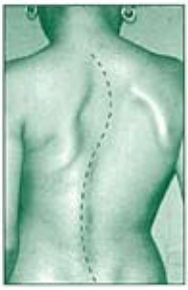
be narrower. This narrowing is called spinal *stenosis*. It is present along the whole length of the backbone, but its effects are most noticeable in the lower back and sometimes in the region of the neck. The combination of increased curvature and narrowness means that the spinal nerves are more prone to compression or squeezing. This may result in a tingling, numb sensation in the arms or legs or sciatica-like pains. It is best to seek medical advice. Symptoms may initially be alleviated by physiotherapy and the measures outlined below. If this does not work then it is possible to have surgery to release pressure from the back of the spinal canal – this procedure is known as *laminectomy*.

The spine may also curve in other places. In the condition known as *kyphosis*, there is a hunched back or outward kink at the junction between the upper and lower back. Sometimes this is present at birth. If it does not correct naturally as the child begins to walk, urgent assessment and treatment, either through bracing or surgery, may be necessary.

'Matthew has been in a brace since he was 18 months old and has now overcome kyphosis.'

Lordosis

Kyphosis



Scoliosis

Scoliosis refers to a sideways curve in the back. This is less common in achondroplasia, but it is best to be alert to its possibility as early

detection leads to more effective treatment. Again, bracing may be necessary.

Preventing back problems

Back problems can sometimes be prevented or lessened by:

- dietary measures to prevent obesity (in fact, many problems can be avoided or lessened by getting into good eating habits at an early age)
- regular gentle and supported exercise, such as swimming (note that children with grommets may be able to swim with specially made ear plugs available from the ENT department of the local hospital)
- avoiding high-risk tasks, such as lifting heavy objects, forward rolls if there is compression in the cervical (neck) area and poor lifting technique.

SUGGESTED WEIGHTS FOR ADULTS WITH ACHONDROPLASIA

HEIGHT ft/in	cms	MALE stones/lbs	kgs	FEMALE stones/lbs	kgs
3'0"	91	4st.2-5st.6	26.3-34.4	3st.10-4st.6	23.6-28.1
3'1"	93	4st.6-5st.8	28.1-35.4	4st.0-4st.10	25.4-29.9
3'2"	96	4st.7-5st.9	28.6-35.8	4st.2-4st.12	26.3-30.8
3'3"	99	4st.9-5st.12	29.5-37.2	4st.4-5st.0	27.2-31.7
3'4"	101	4st.11-5st.13	30.4-37.6	4st.5-5st.2	27.6-32.6
3'5"	104	4st.12-6st.1	30.8-38.5	4st.7-5st.4	28.6-33.5
3'6"	106	5st.0-6st.2	31.7-39.0	4st.9-5st.6	29.5-34.5
3'7"	109	5st.3-6st.4	33.1-39.9	4st.12-5st.10	30.8-36.2
3'8"	111	5st.5-6st.6	34.0-40.8	4st.13-5st.11	31.3-36.7
3'9"	114	5st.6-6st.8	34.5-41.7	5st.2-5st.13	32.6-37.6
3'10"	116	5st.10-6st.9	35.3-42.1	5st.3-6st.2	33.1-39.0
3'11"	119	5st.12-6st.12	36.2-43.5	5st.5-6st.3	34.1-39.5
4'0"	121	6st.0-7st.0	37.2-44.4	5st.8-6st.6	35.3-40.8
4'1"	124	5st.13-7st.2	38.4-45.3	5st.10-6st.8	36.2-41.7
4'2"	127	6st.2-7st.4	39.0-46.2	5st.12-6st.10	37.2-42.6
4'3"	129	6st.4-7.6	39.9-47.1	6st.1-6st.12	38.5-43.5
4'4"	132	6st.6-7st.7	40.8-47.6	6st.2-7st.0	39.0-44.4
4'5"	134	6st.8-7st.9	41.7-48.5	6st.4-7st.2	39.9-45.3
4'6"	137	6st.10-7st.10	42.6-48.9	6st.6-7st.4	40.8-46.2
4'7"	139	6st.12-7st.11	43.5-49.4	6st.8-7st.6	41.7-47.1
4'8"	142	7st.0-8st.0	44.4-50.8	6st.11-7st.10	43.1-48.9
4'9"	144	7st.2-8st.3	45.3-52.1	6st.13-7st.12	43.9-49.9
4'10"	147	7st.4-8st.4	46.2-52.6	7st.1-8st.0	44.9-50.8

For men and women without shoes or clothes, based on a graph by Dr Judy Hall

In young children, it is important to make sure their back is always well supported. It is therefore best to avoid unsupported sitting and/or any activity that involves being in a curved position for any length of time. In addition, baby walkers/ jumpers/backpacks or similar items that do not have adequate back support should be avoided.

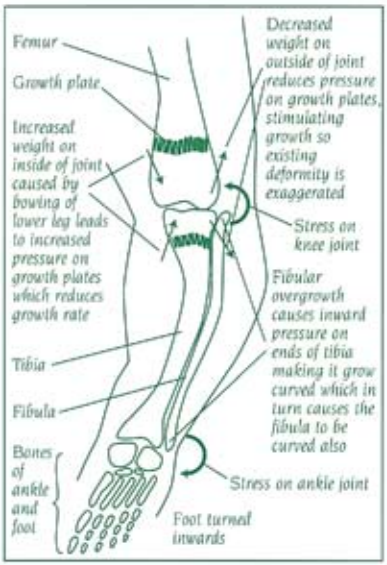
Further advice can be obtained from your local paediatric occupational/physiotherapist.

Limbs, joints and hips

Most people with achondroplasia cannot fully extend their arms at the elbow. This is due to the way that the joint is shaped at the elbow and movement may become more limited as the person grows older.

The lower legs may be bowed as a result of fibular overgrowth. The thinner fibula grows more quickly than the thicker tibia, causing the tibia to become curved and the knee and ankle joint to be distorted. If severe, this can be corrected by orthopaedic surgery, either using fixators or metal plates.

Many people with achondroplasia are affected by hip problems. Curvature of the spine causes the pelvis to be tilted, which in turn reduces free movement of the hip joint. Again, if this severe, it can be corrected by surgically re-aligning the femur.



The development of bowing in the lower legs

Medical opinion on the treatment of these problems changes as more is learned about the various conditions. It is important to equip yourself with as much knowledge as possible and to find a doctor you feel happy with. The RGA will be able to help you contact specialists with the appropriate experience. More information can be found in the **Lifestyles** booklet *Limb Surgery*.

Childbirth

Women with achondroplasia are just as capable of having babies as women without achondroplasia, but need to be closely monitored throughout pregnancy. Narrowing of the pelvic bones means that a planned caesarean section is necessary. You will find more information in the **Lifestyles** booklet entitled *Having a Baby*.

Living with achondroplasia

There is a great deal you can do to make it easier to live with achondroplasia – don't feel that you are entirely in the hands of doctors and health professionals. There are plenty of practical steps you can take which do make a difference.

There may be some difficulties associated with having shorter arms and legs and back problems. Extender sticks and steps can help with reaching, while 'bottom wipers' can be contrived or purchased to aid personal hygiene (there is more information in the **Lifestyles** booklet on *Personal Hygiene*).

Children may need a bit of time in the morning to limber up before they are able to undertake activities which require them to twist, bend and reach. You can contact the RGA office for information on a wide range of gadgets.

Take the lead

One of the greatest problems for many people with achondroplasia is overcoming the prejudice of people who are frightened by anybody who is different.

Parents need to take the lead in discussing their child's height. Children and adults need to learn that the huge diversity that exists amongst people's appearances is a good thing, and that variation in height is just one part of the richness of humanity. Grandparents and other family members, teachers, doctors and friends will take their lead from parents. If parents are able to discuss the issues that their child may face, other people will become more knowledgeable and better able to offer the right kind of support at the right time. It may also help to discuss possible strategies for dealing with inquisitive or insensitive people at school or in the future, and anticipate the questions or comments that may arise.

One of the most important things a parent can do to overcome the problems we have considered here is simply to accept their child for who they are and realise that they are unique, with the same individuality and capacity for love as any other child. All of the attributes of good parenting such as listening, education, love, setting a good example and appropriate discipline apply.

It is just as important not to be over-protective. Children need to develop good self-esteem based on their relationship with their parents and with other children. Young people should be encouraged to

develop friendships, become involved with school activities (including sport, within the limits discussed above) and gain a good all-round education that will help them in the future. There is more on this issue in several of the **Lifestyles** booklets, but especially *Going to School* and *The Teenage Years*.

'I found it really difficult to talk about Angela and didn't really like hearing my wife talk to friends and professionals about the baby's condition. But when I went to my first RGA convention and saw lots of other children playing, and taking advantage of all the opportunities, I realised that my own attitude would have to change if Angela was to grow up with a positive outlook on herself and the world.'

Parents also need to take the lead in obtaining the most appropriate help and advice for their child. GPs often need to have things explained to them – achondroplasia is a rare condition and many doctors have not encountered it before. Parents may need to ask for referral to appropriate specialists such as orthopaedic surgeons, chest physicians, ENT surgeons, speech therapists, physiotherapists or other specialist clinics. RGA is assembling the names of specialists with appropriate experience, but in the meantime parents need to know who to ask and what to ask for. You must become the expert.

Some years ago the following quotation would have summed up the experience of many parents of a baby newly diagnosed with achondroplasia:

'When we were told of our son's diagnosis, it was in an age before the concept of an equal relationship between doctor and patient existed. We had absolutely no support. Nobody could tell us much about our son's condition, none of our friends understood, and even our own parents seemed to think it was somehow our fault. We simply had nowhere to turn to and we just needed someone to say, "it isn't the end of the world".'

Now, however, there are other possibilities:

'I recently met another mother with a baby the same age as mine who is also affected by achondroplasia. It was wonderful to be able to share experiences and compare notes. Although we live a long way apart, we telephone each other often and are able to offer mutual support which is not available anywhere else.'

What causes achondroplasia?

All aspects of an individual are controlled by genetic makeup. Genetic material or DNA provides a blueprint for each person and is contained in every single cell in the body. *Genes* govern the more obvious things like the colour of skin and hair and the shape of the nose, but also extend to the things we can't see, such as the rate of growth, personality, tendencies to certain diseases and conditions. Sometimes a genetic problem arises which affects the aspect of the person controlled by a particular gene.

Achondroplasia is caused by a change in one very specific gene. This gene was discovered in 1994. It was found to lie within *chromosome* 4, and it is the code for an extremely important receptor, the fibroblast growth factor receptor-3 (FGFR-3). The FGFR-3 is present in many cells in the body

and its full function is not yet known. However, it is known that a problem with this receptor causes abnormal cartilage and bone development and is responsible for the changes that occur in achondroplasia. For example, we now know that in childhood, the FGFR-3 is normally switched off, which allows the cartilage to grow. In children with achondroplasia, the receptor is very active and this is thought to inhibit cartilage growth.

How is achondroplasia inherited ?

No one knows exactly how many people have achondroplasia because the condition does not have to be registered. Estimates suggest that between 25 and 30 children are born each year in Great Britain with achondroplasia.

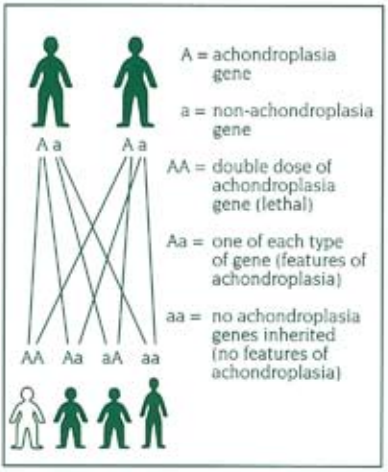
Most of these children are born to parents of average stature. In these cases a spontaneous change (mutation) of the FGFR-3 gene has occurred in either the mother's (egg) or father's (sperm) genetic contribution to their child some time prior to conception. The odds of this happening are thought to be about one in 25000.

Once the genetic change has occurred, a person with achondroplasia can pass the condition on to his or her children. However, every individual with achondroplasia carries two copies of the FGFR-3 gene, one normal and one causing achondroplasia. So if one parent is already affected by achondroplasia (and the other parent is not), their children have a 50% chance of being affected. This is because achondroplasia is an example of a 'dominant' condition where the effects of the gene causing achondroplasia override the effects of the normal gene inherited from the other parent.

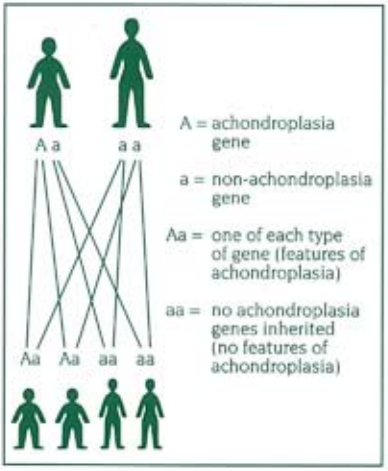
If both parents are affected (i.e. each has one copy of the gene causing achondroplasia), the chances of passing on the

condition to their children is increased. Such a couple has a one in four (25%) chance of having a child of average stature. There is a one in two chance (50%) that a child would inherit one copy of the achondroplasia gene from either parent and a one in four (25%) chance that the baby would inherit the achondroplasia gene from both parents. In this last situation, the child will not survive beyond birth. Where there is a risk of passing on a 'double dose', parents may wish to seek genetic counselling, and a test may be offered to detect the double-dose embryo whilst still in the womb.

This is best illustrated in the following diagrams:



Both parents with achondroplasia



One parent with achondroplasia

'We wanted our baby to be small like us.'

Diagnosis and treatment

Genetic testing

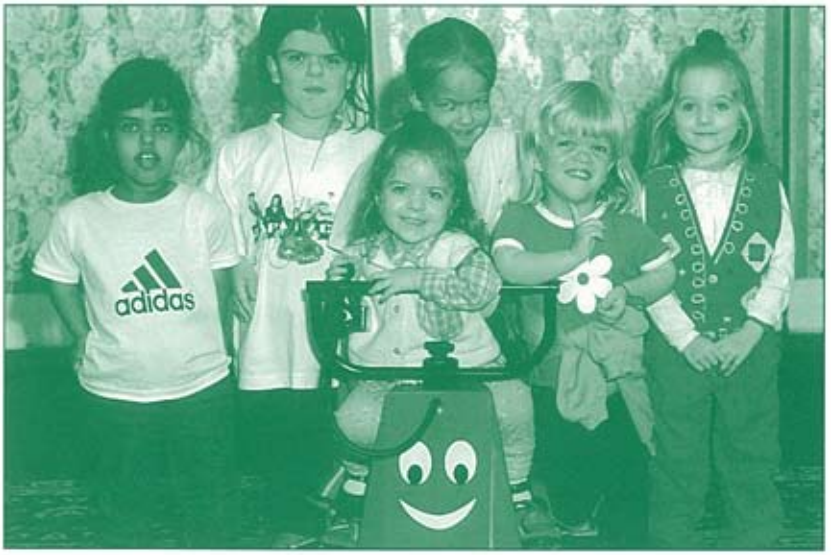
The fact that the gene for achondroplasia has now been discovered means that a test is now possible to diagnose the condition before birth. This test is available where both parents have achondroplasia, and allows detection of the 1 in 4 cases where the baby carries a double dose of the achondroplasia gene and would therefore not live after birth. It could potentially be used to confirm a scan diagnosis. The test is not used in routine screening because the incidence of achondroplasia is so low. Indeed, many would argue that the introduction of a screening program for achondroplasia where neither parent has the condition demeans individuals with the condition and ignores the fact that the majority of people with achondroplasia live active, healthy fulfilling lives.

How is achondroplasia diagnosed ?

The diagnosis of achondroplasia, like other conditions, is based on clinical suspicions and supported by investigations. Achondroplasia is occasionally diagnosed before birth on the basis of ultrasound scans showing disproportionately short limbs. However, diagnosis at this stage can usually only be made with certainty when one or both parents have this condition, as there are other conditions with a similar appearance. After birth the diagnosis can be confirmed on the clinical appearance (such as saddle-shaped nose, short arms and legs and head shape as outlined above) and on X-ray appearance.

Achondroplasia has certain characteristic X-ray features which doctors will look for:

- short limbs and ribs versus trunk length
- 'ball-in-socket' *epiphysis* (a specific appearance of the ends of the long bones)
- distinctive features of the pelvic shape
- hands: fingers widely opposed and equal length ('trident' hands)
- skull: enlarged vault and jaw, small midface and foramen magnum
- spine: narrow diameter with inward curving rear surface (*lordosis*) and spinal canal only $\frac{1}{2}$ normal depth
- decreased distance between the pedicles of the lumbar vertebrae
- vertebrae in thoracolumbar (mid-back) region less well formed (hypoplastic) resulting in a 'bullet-nose' shape.



Treatment

There is currently no single treatment that will result in a baby with achondroplasia attaining a 'normal' physique/appearance by the time they are fully grown. There is no 'magic bullet' that targets the overactive FGFR-3 receptor. Trials are ongoing with growth hormone that indicate some success in improving initial growth rate, but the increase in final height may not be significant.

The best possible treatment consists of a sensible diet, regular gentle exercise and routine check-ups by the paediatrician throughout childhood to identify and treat any of the complications of achondroplasia as soon as they arise. Most of these complications and their prevention or treatment have been mentioned above.

Surgery can be used to lengthen the legs (and increasingly the arms) of people with achondroplasia. However, this remains a controversial issue amongst affected people. On the one side, there is the joy and satisfaction resulting from a gain of several inches in height. On the other side, it is not without complications, and the process can often be long and painful. Surgery is no magic wand and should be considered only when the affected youngster has had an opportunity to develop good self-esteem and a positive self-image. This is one of the reasons why the RGA encourages its young members to socialise together and develop positive attitudes. Unfortunately, this surgical procedure is sometimes offered by doctors who have no concept of the psycho-social issues facing the young person. Further information can be found in the **Lifestyles** booklet *Limb Surgery*.

Sources of information

Research is ongoing into the genetics, the effects and treatment of Achondroplasia.

In this country the largest organisation for parents, their children and other individuals with achondroplasia is the Restricted Growth Association. The RGA is a self-help organisation concerned with the welfare of people of restricted growth. The majority of members have achondroplasia. Members maintain contact through publications, meetings, conferences and social activities. There is a support network for families and medical information is provided. The RGA is also compiling a list of specialists who have expertise in symptoms that are peculiar to achondroplasia. Our address is:

Restricted Growth Association
PO Box 8, *Countesthorpe*,
Leicestershire LE8 5ZS
Tel: 0116 2478913

Another organisation in the UK is the Child Growth Foundation. This campaigns for the screening of growth in children and funds research into the treatment of growth disorders. Their address is :

Child Growth Foundation
2 *Mayfield Avenue*, *Chiswick*,
London W4 1PW
Tel: 0181 995 0257

The Skeletal Dysplasia Group (for medically or scientifically qualified people) aims to find out more about developmental disorders of skeletal growth. They have a list of publications, which are available directly from:

The Skeletal Dysplasia Group for Teaching and Research
2 *Dale Close*, *Oxford* OX1 1TU

The Dwarf Athletic Association makes regular sporting opportunities available to people of short stature. Their address is:

The Dwarf Athletic Association
C/o 44 *Middlecroft Road*,
Stourton Grange, *Leeds* LS10 4QZ
Tel: 0113 2703791

If you have access to the Internet, several organisations and individuals have created web pages dedicated to providing information on conditions with restricted growth. One such group is The Little People of America, the American equivalent of the RGA. Their web page can be found at http://www.bfs.ucsd.edu/dwarfism/lpa.htm

The following sources have been useful in compiling the **What is...** series of booklets :

Greenberg Center for Skeletal Dysplasias - Web site: http://www.med.jhu.edu/Greenberg.Center/Greenbrg.htm

American Academy of Pediatrics. (1995) 'Health Supervision for Children with Achondroplasia'. *Pediatrics*, Mar 95, 443-451.

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Horton W.A., Rotter J.I., Rimoin D.L., Scott C.I. and Hall J.G. (1978) 'Standard Growth Curves for Achondroplasia'. *Journal of Pediatrics*, 93, 435-8.

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Jones K.L. (1988) *Smith's Recognizable Patterns of Human Malformation*. 4th Edition. WB Saunders, Philadelphia.

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Online *Mendelian Inheritance In Man* - Web site: http://www3.ncbi.nlm.nih.gov/Omim/

Rimoin D.L. and Lachman R.S. (1990) 'The Chondrodysplasias'. *Principles and Practice of Medical Genetics*. Eds A.E.H. Emery and D.L. Rimoin; London: Churchill Livingstone, 2nd edition.

Rousseau F., Bonaventure J., Legeai-Mallet L., Schmidt H., Weissenbach J., Maroteaux P., Munnich A. and Le Merrer M. (1995). 'Clinical and genetic heterogeneity of Hypochondroplasia'. *J. Med. Genet*, 33: 749-752.

Skeletal Dysplasia Group for Teaching and Research - Occasional Publications 5b, 6b, 8b and 9 - Hall M. Hurst J., Slaney S. and Wynne-Davies , R. (1998).

Wynne-Davies R., Hall C.M. and Appley A.G. (1985) *Atlas of Skeletal Dysplasias*. Churchill Livingstone, Edinburgh.

List of publications available from the Restricted Growth Association

What is . . .?

Achondroplasia

Hypochondroplasia

Pseudoachondroplasia

SED

MED

Diastrophic Dysplasia

Rarer Syndromes

Lifestyles

Babies and the Early Years

Going to School

Driving a Car

Bikes, Trikes and Ride-on Toys

The Teenage Years

Sporting Opportunities

What Shall I Wear?

Having a Baby

The Later Years

Adoption

Careers, Employment and Education

Limb Surgery

Personal Hygiene

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