# What is Pseudoachondroplasia?





Restricted Growth Association

axong to benefit people with restricted growth and their familia





This booklet has been written to provide information and support for parents of children with pseudoaochondroplasia, 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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Thank you for sending me the draft copy of 'What is Pseudoachondroplasia?'. At last 1 feel that this is my real diagnosis and feel really relieved that 1 now belong to something, even if it is a condition that has pinched some other condition's name.'

The contents of this booklet are believed to be correct at the time of publication. However, knowledge about pseudoachondroplasia 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.

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# What is Pseudoachondroplasia? by Will Christian MBBS, BSc

Our daughter is remarkable. Shes done much more than we ever expected.

I think I'm tougher because of it. If I were of average height, there would be no reason to be strong.

seudoachondroplasia is a rare condition and is one of the types of restricted growth with disproportionate stature (in this case the term refers to people with an average size body but shorter limbs). As with many causes of short stature, pseudoachondroplasia 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.
Pseudoachondroplasia is a condition that covers a wide range of heights and abilities. Most

people with pseudoachondroplasia 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, cars and bicycles can be adapted (see the RGA Lifestyles series), and there is no reason why someone with pseudoachondroplasia should not participate in most activities. All children (and adults!), including those with pseudoachondroplasia, 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.



# What does pseudoachondroplasia mean?

Pseudoachondroplasia is a misleading term. It literally means 'false achondroplasia' and arose because some of the clinical features bear a resemblance to another restricted growth condition called achondroplasia. This is deceptive however. Achondroplasia is an unrelated condition caused by changes in a completely different gene (see below) and has many features that are not shared by people with pseudoachondroplasia. For example, children with pseudoachondroplasia have normal head growth and facial appearance. This is not so in achondroplasia.

Indeed, pseudoachondroplasia is commonly not diagnosed until a child is 18 to 36 months old. This is because affected children often appear normal at birth, with normal length and birthweight, and it is not until growth starts to slow (from about 2 years onwards) that the condition becomes apparent.

As a child of a mother with pseudoachondroplasia, Chiloe had regular X-rays and check-ups from birth. But she was not diagnosed until she was three years and three months old, when she had an x-ray of her colon which showed up abnormalities of the spine. Chioe had features which to me, her mother, indicated pseudoachondroplasia, i.e. wide thumbs, small poday hands, small wide feet, but nothing showed on the x-ray and the doctors were emphatic that she did not have the condition.

When I was born, I looked just like my brother, but when I started to walk I had some difficulties and also developed convaisions. When the doctors looked into it, they first diagnosed achiendroplasia.

<sup>\*</sup> Words in italics are found in the glossary



# How does pseudoachondroplasia arise?

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 by special cells called chondrocutes. In people with pseudoachondroplasia, the growth plate is damaged by the build-up of an abnormal form of a material called 'cartilage oligomeric matrix protein' (COMP for short) in these chondrocytes (see below). This build-up stops the cell working properly and 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, however, are not affected by the growth plate and continue to grow. This is why people with pseudoachondroplasia sometimes have much bulkier legs and arms.



Section of long bone - in this case the top of the tibia (skin bone)

The average height for both men and women with pseudoachondroplasia is between Im 8cm and Im 32cm (3ft 7in and 4ft 4in). However, the variation amongst individuals is great, and some may be shorter or taller than this.

Paediatricians and GPs should use growth charts designed specifically for children with pseudoachondroplasia to predict their growth rates and expected heights. These charts can be obtained from the RGA (address at the end of this book).



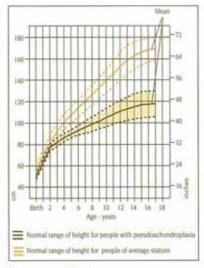


Diagram of height chart

### What are the other effects of pseudoachondroplasia?

People with pseudoachondroplasia are as varied in appearance as people without pseudoachondroplasia. However, they may share certain physical characteristics, and may be more prone to certain medical problems. These include:

- a near-normal body length with short arms and legs
- a normal head size with normal facial features

- an increased curvature of the lower spine (lumbar lordosis)
- lax ligaments (loose joints)
   affecting all joints which may be
   very severe

'Naomi often has pains due to her hips, knees and ankles dislocating on a regular hasis.'

- the lower legs may be bowed (genu varum) or knock-kneed (genu valgum)
- bowing/curving of the forearms
- short fingers of similar length (this is often described as 'Telescopic' due to the fact that they can be extended in a telescopic fashion)
- angling of the hand at the wrist so that it is not quite in line with the arm (ulnar deviation)
- · short, broad, flat feet and hands
- limited extension at the hips and elbows
- premature osteoarthritis of the joints, particularly the hips and knees
- neck problems (see diagram of skeleton).



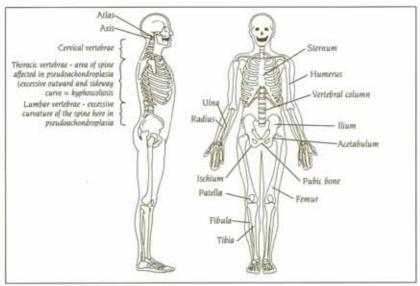


Diagram of skeleton - anterior and lateral views

It is important to remember that people with pseudoachondroplasia have normal intelligence and children with pseudoachondroplasia will usually reach their developmental milestones at the appropriate age, although walking may be delayed due to loose joints. This may in turn affect when the child begins to talk. The first indication of the condition may be related to mobility.

#### Possible complications

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



#### The back

People with pseudoachondroplasia are especially prone to back problems. The curvature of the spine is often more exaggerated in the lower back (lumbar lordosis). This increases the likelihood of injury to the lower back, especially if poor lifting techniques are used when carrying heavy objects. It also means that people with pseudoachondroplasia are more prone to Sciatica, a condition caused by compression or squeezing of the nerves that supply the lower legs, at the point at which they leave the back bone. It can result in an unpleasant tingling/numb sensation in the lower limbs, usually when walking or lifting.

The spine may also curve in other places. In the condition known as kyphosis, there is an outward kink at the junction between the thoracic and lumbar vertebrae, giving the appearance of a 'hunched back'. Treatment, either through bracing or surgery, may be necessary.





Lordosis

Kuphosis



Scolinsis

Scoliosis refers to a sideways curve in the back. This is less common in pseudoachondroplasia, but it is best to be alert to its possibility, as early detection leads to more effective treatment. Again, bracing may be necessary.



My ribs rest on my pelvis on the right side — this has been exacerbated by sitting unsupported for 60-odd years.

#### The neck

Most importantly, there may occasionally be problems in the neck region. A dangerous combination of underdeveloped neck bones (the cervical vertebrae) and looseness of the ligaments responsible for supporting the neck, can lead to a rare but extremely serious situation. In these exceptional cases the neck repeatedly subluxates (a term referring to partial dislocation of the neck), damaging the spinal cord (the main bundle of nerves that travels in the backbone from the brain to the body). This is a very grave condition. To be alert to its possibility, an x-ray should be taken of your child's neck as soon as the diagnosis of pseudoachondroplasia is made, to

detect any structural problems with the neck. In addition, a doctor should be called immediately if anyone with pseudoachondroplasia experiences the following features of spinal cord compression:

- neck or back pain.
- numbness or pain in the arms, hands or legs
- loss of arm / leg control.





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 from an early age)
- regular gentle and supported exercise, such as swimming
- avoiding high-risk tasks, such as lifting heavy objects and poor lifting technique.

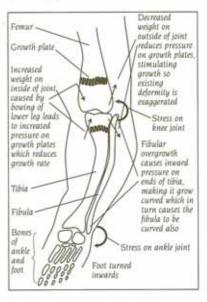
In young children, it is important to make sure their back is always well supported. It is therefore best to avoid unsupported sitting and/or any activity that involves being in a curved position for any length of time. In addition, baby walkers/jumpers/backpacks or similar items that do not have adequate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

Because of the rare but serious problem of neck instability, children should be advised to avoid doing things that may place a strain on the neck, such as forward rolls, trampolining, rugby, etc. It is

important to see your doctor urgently if any of the features of spinal cord compression become apparent (see above).

#### Limbs, joints and hips

Many people with pseudoachondroplasia 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.



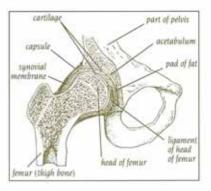
The development of bowing in the lower legs



The lower legs may be bowed as a result of a combination of loose ligaments (causing joint distortion) and 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 this is severe, orthopaedic surgery, either using fixators or metal plates, can correct bowing. If you are considering leg surgery, please refer to the **Lifestyles** booklet on this topic.

The majority of people with pseudoachondroplasia are affected by a severe form of premature osteoarthritis involving most of the joints in the body, particularly those that are weight-bearing, such as the hips and knees. This is one of the most frequent and serious complications of pseudoachondroplasia and means that many affected people will require joint-replacement surgery in early adult life (most frequently, the hips).

The had two replacements and I'm now able to get around more easily.



Medical opinion on the treatment of all 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 with whom you feel happy. The RGA will be able to help you contact specialists with the appropriate experience.

#### Childbirth

Women with pseudoachondroplasia are just as capable of having babies as women without



pseudoachondroplasia, but need to be closely monitored throughout pregnancy. Delivery by planned caesarean section will usually be necessary due to hip joint immobility. You will find more information in the **Lifestyles** booklet entitled Having a Baby.



Boy on right has pseudoachondroplasia

# Living with pseudoachondroplasia

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 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. (See the Lifestyles booklet Going to School for more practical ideas on this subject, and contact the RGA office for information on a wide range of gadgets.)

#### Take the lead

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



looks 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 they 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. However, 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.

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 – pseudoachondroplasia is a rare condition and many doctors have not encountered it before. Parents may need to ask for referral to an appropriate specialist such as an orthopaedic surgeon, rheumatologist, physiotherapist or other specialist clinics. RGA is assembling the names of specialists with appropriate experience, but in the meantime parents need to know what 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 pseudoachondroplasia:

'I used to be awake worrying about Sue and when I finally fell asleep, wake up thinking I had had a nightmare that Sue was small — only to realise that it was true.

Now, however, there are other possibilities: If only I had been able to go to something like an RGA social when Sue was diagnosed, I would not have had one sleepless night. It was one of the happiest days of my life, seeing other people like Sue, all enjoying life.

# What causes pseudoachondroplasia?

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 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 an individual controlled by a particular gene.

Pseudoachondroplasia is caused by a change in one very specific gene



which was discovered in 1995. It was found to lie within chromosome 19, and it is the code for an extremely important protein, the cartilage oligomeric matrix protein (COMP). COMP is normally present in special cells known as chondrocytes (see the glossary at the end of this booklet) and in tendon tissue. In pseudoachondroplasia, there is a build-up of abnormal COMP within these areas, leading to altered bone growth and loose ligaments.

Initially, pseudoachondroplasia was divided into four separate disorders (pseudoachondroplasia types I-IV). However, as work on the genetic basis of pseudoachondroplasia continues, it is becoming apparent that these are all forms of the same disorder, with varying degrees of severity.

### How is pseudoachondroplasia inherited?

When a child with pseudoachondroplasia is diagnosed, the fact that he or she has pseudoachondroplasia does not have to be registered. Consequently no one knows exactly how many people have the condition, though some estimates suggest that it affects about 1 in every 50,000 people.

Most children with pseudoachondroplasia are born to parents of average stature. In these cases a spontaneous mutation of the COMP gene has occurred in either the mother's (egg) or father's (sperm) genetic contribution to their child sometime prior to conception

Once the change has occurred, a person with pseudoachondroplasia can pass the condition on to his or her children. However, every individual with pseudoachondroplasia carries two



copies of the gene responsible, one normal and one pseudoachondroplastic. So if one parent is already affected by pseudoachondroplasia (and the other parent is not), their children have a 50% chance of being affected. This is because pseudoachondroplasia is an example of a dominant condition where the effects of the pseudoachondroplastic gene override the effects of the normal gene inherited from the other parent.

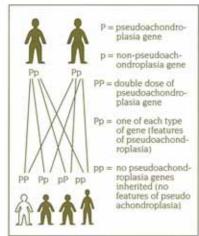
If both parents are affected (i.e. each has one copy of the pseudoachondroplastic gene), the chances of passing on the condition to their children is increased. Such a couple has a one in four (25%) chance of having a child of average stature. There is a one in two chance (50%) that a child would inherit one copy of the pseudoachondroplasia gene from either parent and a one in four (25%) chance that the baby would

inherit the pseudoachondroplasia gene from both parents. It is not known what would happen if this occurred in pseudoachondroplasia. A number of different genetic changes (or spelling mistakes) in the COMP gene all produce the same features of pseudoachondroplasia. Therefore, if the offspring of two people with pseudoachondroplasia inherit two copies of the abnormal COMP gene, the copy inherited from the father is likely to contain a different mistake to that inherited from the mother. This will result in varying degrees of severity in the type of pseudoachondroplasia that results.

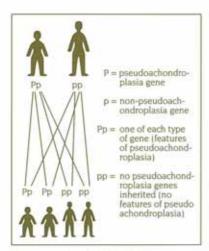
This is best illustrated in the diagrams on the next page.

Occasionally, parents of average stature may have more than one affected child. Normally the odds of this happening are extremely low. However, due to a phenomenon known as mosaicism, this can happen more frequently than would be expected. In these cases, two





Both parents with pseudoachondroplasia



separate populations of sperm cells or egg cells arise in the testes of the father/ovaries of the mother. One population carries the pseudoachondroplasia gene and the other carries the normal gene. If a sperm/egg from the pseudoachondroplasic gene pool is passed on to the offspring, they will show the characteristic features of pseudoachondroplasia. If the normal gene is passed on then the child will be unaffected. This point should be discussed in more detail with your hospital's clinical geneticist.

If two people with different restricted growth conditions plan to start a family, they can seek genetic counselling to find out about the risk and consequences of their two conditions combining in their offspring.

One parent with pseudoachondroplasia



#### Diagnosis and treatment Genetic testing

The fact that the gene for pseudoachondroplasia has now been discovered means that a test is now theoretically possible to diagnose the condition before birth. The test is not used in routine screening because the incidence of pseudoachondroplasia is so low. Indeed, many would argue that the introduction of a screening program for pseudoachondroplasia where neither parent has the condition demeans individuals with the condition and ignores the fact that the majority of people with pseudoachondroplasia live active, healthy fulfilling lives.

"When Paul was born, the medical staff described him as a "floppy" baby. They asked me if I had noticed anything different about him but since he was my first child. I didn't have anyone to compare him with. Paul only weighed 5lbs 10oz at birth but I don't think his length was particularly short at that stage."

## How is pseudoachondroplasia diagnosed?

The diagnosis of pseudoachondroplasia, like all other conditions, is based on a combination of doctors' suspicions along with various investigations.

I thought there was something wrong with my daughter when she was about three months old. I was particularly aware that her limbs did not move as they should, due to poor muscle tone, but she looked just like any other baby and her birth weight, length, etc., were within the normal range. I had suspected that there was something wrong during my pregnancy, but when she was born, everuthing appeared to be all right.

By the time doctors suspect the diagnosis (usually at the age of 2-4 years, though this may be earlier where one or both of the parents has pseudoachondroplasia), a number of different characteristic X-ray features can be used to confirm it:

 Limbs: The limbs are short in relation to trunk length



- Spine: The vertebrae appear
  flattened (platyspondyly) with a
  characteristic oval shape in
  early childhood. A bony shelf on
  the front of the vertebrae gives
  them a 'beaked' appearance.
  The second cervical vertebra
  (the second neck bone), the
  axis, which forms a stable base
  for neck rotation, may be
  underdeveloped. This can lead
  to problems with neck
  instability (see above)
- Hip joint (see previous diagram): The femoral ephiphysis (head of the thighbone) is small and underdeveloped; the acetabulum (socket into which the head or ball of the thighbone fits) is poorly formed
- Pelvis: The ischium and pubis at the front of the pelvis are underdeveloped
- Hands: The fingers are short and of equal length, with flared irregular metaphyses and small irregular epiphyses
- Long bones: Irregular flaring of the metaphyses occurs, with under-

developed epiphyses (especially noticeable in the femur).

Paul was slower than average to do things like lift his head from the mattress or sit up. He was also very reluctant to use his leas for anything. When he was a year old, he didn't move around at all. If I sat him on the floor with a pile of tous, he would stay there until I moved him again. He never crawled at all but did eventually get around by rolling everywhere. Still he showed no signs of wanting to put any weight on his legs and I took him for physiotherapy twice a week to encourage him to use his legs. He was two years old before he started to walk.

#### **Treatment**

At the moment, there is no single treatment that will result in a baby with pseudoachondroplasia attaining a 'normal' physique/ appearance by the time they are fully-grown. There is no 'magic bullet' that targets the abnormal COMP gene. Trials looking at the affect of growth hormone on other



conditions with restricted growth are ongoing. Early data has indicated some success in improving initial growth rate, but the increase in final height may not be significant. At present, the effect of growth hormone on people with pseudoachondroplasia is unknown.

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

"As I grew older, my limbs became a lot thicker as the muscles were growing, but the bones were not, so it became much more difficult to walk. My leas also became bowed and my joints became painful. I thought long and hard and decided that having some lea lengthening would correct the bowing and enable me to walk better."

Surgery can be used to lengthen the arms and legs of people with pseudoachondroplasia who feel that their short stature is a disadvantage. However, this remains a controversial issue amongst affected people and because of the frequent complications of severe osteoarthritis and joint degeneration in pseudoachondroplasia, it is not commonly recommended. It 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.

#### Sources of information

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

In this country the largest organisation for parents, their children and other individuals with pseudoachondroplasia is the Restricted Growth Association. The RGA is a self-help organisation concerned with the welfare of people of restricted growth. Members maintain contact through a magazine, 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 pseudoachondroplasia. Their address is:

Restricted Growth Association PO Box 8, Countesthorpe, Leicestershire LE8 52S 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

Co 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





#### Glossary of terms

#### Autosomal dominant/recessive -

these terms refer to the ways in which individual genes can be inherited. Autosomal means that the gene lies on a non-sex chromosome (there are two types of sex chromosome: x and y, which determine whether you are male or female). Dominant refers to the fact that only one copy of the gene is necessary for the condition to be present. Recessive means that two copies of the gene are required.

Cartilage – a dense white/grey material which has several roles in the body. It is part of a group of structural materials known as connective tissue. In bones it can act as a protective buffer at the ends of joints, protecting them from damage. It is also involved in new bone formation. Cartilage produced at the end of long bones is converted into bone by special cells known as osteoblasts.

Chondroblast/Chondrocytes – a special type of cell that is involved in the production cartilage

Chromosomes – One of 46 structures present in most cells in the body that are composed of long coiled strands of DNA and carry genetic information in the form of genes. Germ cells, i.e. sperm and eggs, only carry 23 chromosomes.

Epiphysis – the end of the long bone which is normally separated from the shaft by the growth plate (the area of cartilage where bone growth occurs). It becomes fused to the bone shaft during puberty to form a complete bone.

Gene – a unit of genetic material which carries instructions for growth, development, the regulation of the bodies internal workings, etc. Genes are grouped together on chromosomes.

Genetic counselling – process by which knowledge and advice concerning inherited disorders and the possibilities of passing on particular conditions from one generation to the next, along with options regarding diagnosis and management, are given to patients, parents and families.

Hypotonia – excessive 'floppiness' caused by insufficient muscle tone

Kyphosis – outwards curve of the back, resulting in a hunched back / outward kink at the junction between the thoracic and lumbar vertebrae.

Lordosis – inwards curvature of the lower spine.



Metaphysis – part of the bone lying between the epiphysis (end of the bone) and the diaphysis (the shaft of the bone).

Scollosis – a sideways curve of spine.

Vertebrae— the individual bones that make up the back-bone, or spinal column. They are divided into four types: Cervical (in the neck region), Thoracic (in the upper and middle back), Lumbar (in the lower back) and Sacral (at the base of the spine).

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

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#### 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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