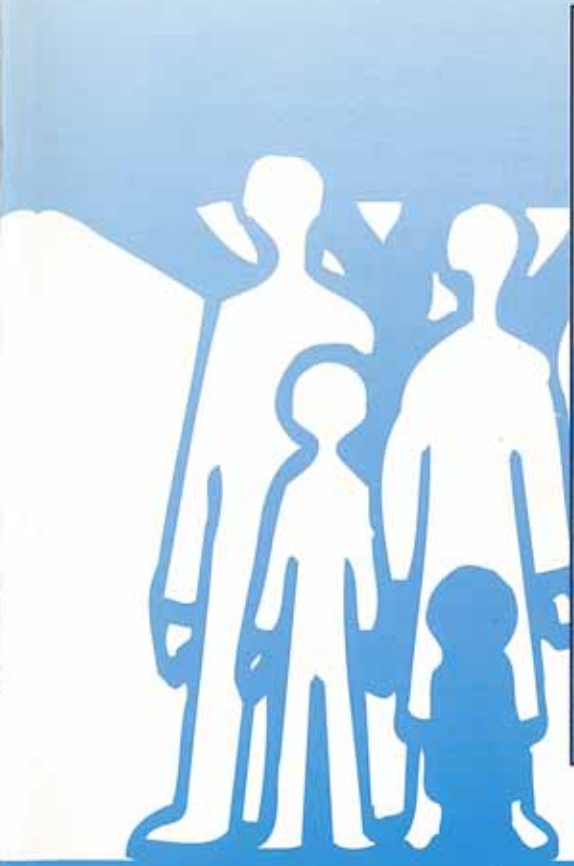
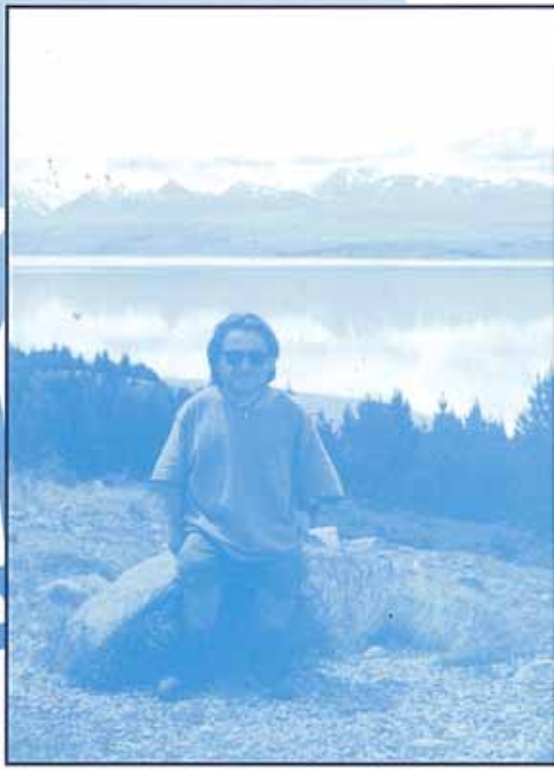
What is

SED?





Restricted Growth Association

Working to benefit people with restricted growth and their families

RGA

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

Dr Will Christian is a paediatrician who 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.

RGA would like to acknowledge the grants from the Department of Health and the National Lottery Charities Board which have made it possible to produce this booklet, together with the help of the Wellcome Trust Medical Photographic Library which has supplied many of the photographs.

'When you're first told the diagnosis, you don't know what it means and the doctors don't have much to tell you either. But don't be scared. Our son is adorable – bright and alert with a great personality.'

The contents of this booklet are believed to be correct at the time of publication. However, knowledge about spondyloepiphyseal dysplasia 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 SED (*spondyloepiphyseal dysplasia*)?

by Will Christian MBBS, BSc

'Looking back on my child, she has achieved more understanding and compassion than I have ever done. In the beginning this is something that I never thought she would achieve.'

SED is a rare condition causing restricted growth, with both short body size and short arms and legs. As with many causes of short stature, SED 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. SED is a condition that covers a range of heights and abilities. Most people with SED live normal, happy lives although there are, of course, some physical limitations.

'Physical limitations don't matter. It's what you believe inside that counts.'

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

'It was devastating to be told that our son had dwarfism and with all the potential health problems SED can bring, the future seemed very bleak. But people are constantly amazed by Alex's determination to have a go at things. He finds his own level and way of interpreting everyday living.'

'Guid gear comes in small bulk.'

Old Scottish saying

What does SED *mean?*

Spondyloepiphyseal dysplasia (SED) is one of a group of disorders known as the 'epiphyseal dysplasias'. SED literally means 'abnormal formation of the spine and ends of long bones'. There are three commonly described forms of SED, each sharing many similar characteristics:

- SED congenita – this condition is noticeable around the time of birth
- late, X-linked SED – this form of SED does not become fully apparent until a child is 4-6 years old. It occurs most frequently in males.
- SED tarda – this form is inherited in the same way as SED congenita. It does not usually become obvious until mid-childhood.

'My abnormalities did not start to show until I was 3-4 years old. I fell more than most children of my age and I could not lift my arms high.'

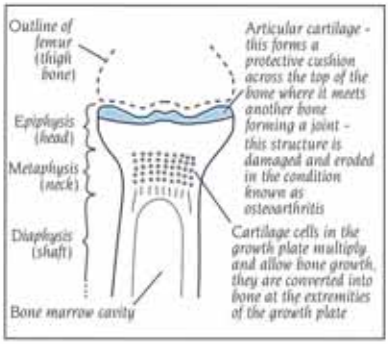
All forms of SED are genetic conditions, beginning at conception and continuing throughout life. They arise because of the faulty production of a protein called *collagen**. Collagen forms an essential part of many of the body's tissues, including cartilage, bone and connective tissue. Connective tissue is a special tissue that supports and binds other tissues and is found in many areas of the body (e.g. the eye, the spine and the joints). There are several different types of collagen in the body. Abnormalities in Type-II collagen are responsible for the features of SED.

* Words in italics are found in the glossary

How does SED *affect growth ?*

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 *chondrocytes*. In people with SED, the growth plate is damaged by the build-up of abnormal materials in these chondrocytes. This build-up stops the cells 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.

Collagen is also involved in bone production. It forms the template for normal cartilage and bone development. Cartilage is produced around a meshwork of collagen that gives the bone strength and flexibility (a bit like the steel rods that run through reinforced concrete). In SED, the presence of an abnormal form of Type-II collagen means that this process is interrupted, resulting in faulty bone formation. The effect of this is most noticeable at the ends of the long bones (the bones of the arms and legs). This area is known as the *epiphysis*. In SED, the epiphyses are underdeveloped and fragmented causing joint problems and leading to osteoarthritis in later life. The backbones (*vertebrae*) are also affected, leading to curvature of the spine and other problems.



Section of long bone – in this case the top of the tibia (shin bone)

The final adult height achieved by both men and women with SED ranges between about 85cm and 1m 30cm (2'9"- 4'3").

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

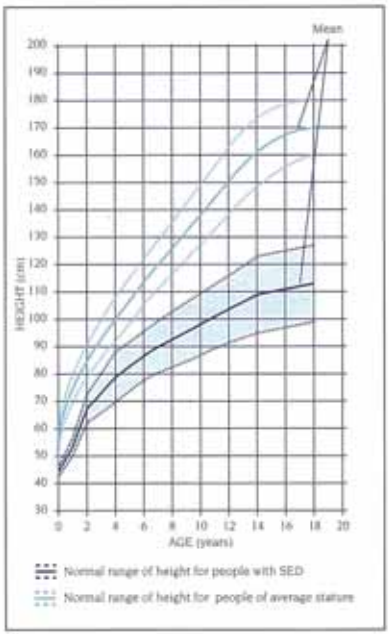


Diagram of height chart for people with SED

What are the other effects of SED?

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

- a short body length which is accompanied by some shortening of the arms and legs: the arms may appear long in relation to the body – can affect people with all forms of SED
- a normal head size with a slight tendency towards widely spaced eyes and mildly 'flattened' features – can affect people with all forms of SED
- an increased inward curvature of the lower spine (*lumbar lordosis*) and outward curvature of the upper spine (*kyphosis*), sometimes accompanied by an S-shaped bend (*scoliosis*) which develops progressively through life – can affect people with all forms of SED

- distortion of the lower leg joints resulting in either knock-knees (your doctor may refer to this as 'genu valgum') or bowing of the lower legs ('genu varum') – can affect people with all forms of SED
- in a baby with SED congenita there may be a *cleft palate* requiring corrective surgery between 12 and 24 months.
  Support with the care and treatment of a cleft lip and/or palate is available from the Cleft Lip and Palate Association (CLAPA). They organise parent-to-parent support and provide information and specialist items to help in feeding babies with clefts.
- in a baby with SED congenita there is an increased tendency towards *club foot* at birth which may require corrective surgery or splinting
- a chest wall which is broad or barrel shaped because of the shape of the breast-bone (*sternum*) – can affect people with all forms of SED
- in a person with SED congenita there may be eye problems, including shortsightedness and retinal detachment – this can cause blindness so must be treated by an eye specialist
- in people with SED congenita some people have progressive hearing loss due to electrical sound messages not getting through from the ear to the brain (sensorineural deafness) – people with this form of the condition should have regular hearing checks, especially in childhood
- difficulties in straightening the arms and legs (limited extension) – can affect people with all forms of SED
- osteoarthritis of the hips, knees and spine – can people with affect all forms of SED
- a short neck with neck bones . which are more likely to dislocate (see below) – can affect people with all forms of SED
- Babies with SED congenita, tend to be more floppy (*hypotonia*) than those without SED and reach their developmental milestones later.

'My hands and feet appear normal although my feet sprain, often spontaneously.'

'I've always suffered from decreased endurance and overwhelming fatigue. Standing still without support for more than a few seconds has always been a problem – my father used to call me lazy.'

It is important to remember that people with SED have normal intelligence, and that although babies with SED develop more slowly than normal, they will usually achieve all their developmental milestones, albeit at a later age.

Specific complications

There are some medical complications that are often associated with SED. 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 SED (which is unlikely), he or she may be equally uncertain.

'When our son was diagnosed as having SED the GP knew no more than we did, which was nothing.'

The back

People with SED are especially prone to back problems. The curvature of the spine is likely to be more exaggerated in the lower back (lumbar *lordosis*). This increases the possibility of injury, especially if using poor lifting techniques when carrying heavy objects. It also means that people with SED are more prone to *sciatica*, a condition caused by compression

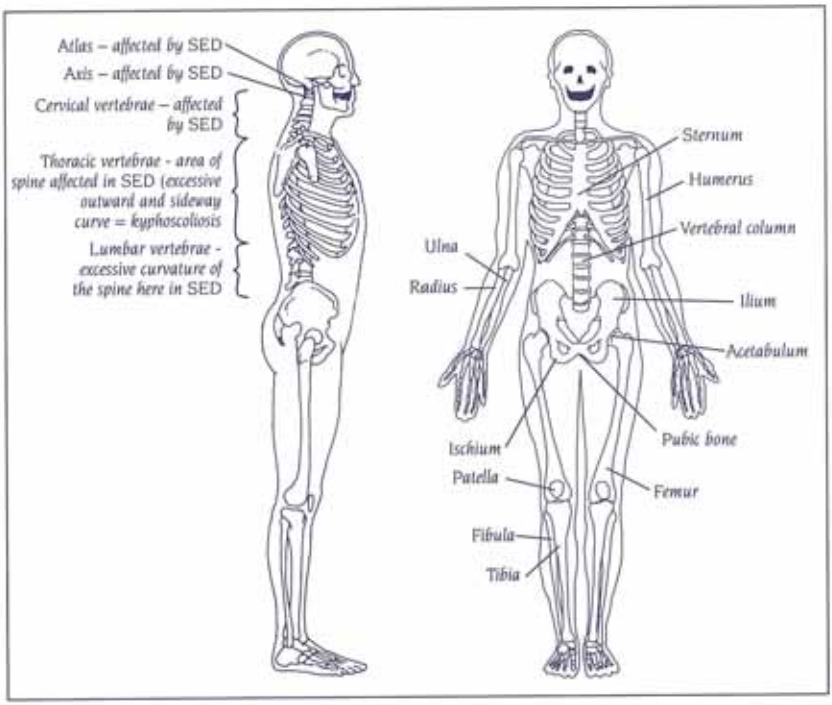


Diagram of skeleton – anterior and lateral views

or squeezing of the nerves that supply the lower legs at the point at which they leave the back bone. It results in an unpleasant tingling/numb sensation in the lower limbs, usually when walking or lifting.

The lordosis in SED congenita is often severe, and is made worse by limited movement at the hips. This causes the back to bend even more to try to compensate and allow walking to occur. Bracing for lordosis is not usually effective and early surgical intervention may be necessary.

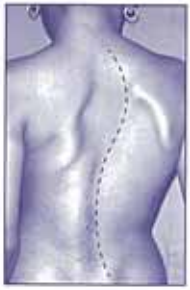
'Having a lumbar lordosis can cause problems in later life with osteoarthritis of the spine. Certain movements can cause the back and legs to lock [due to the pain], causing you to fall where you stand. This can be embarrassing at times. Emotionally it can be draining as well as painful. Care is needed not to do too much – avoid impact exercises and a lot of walking.'

The spine may also curve in other places. Occasionally the upper spine curves outwards (a condition known as *kyphosis*) giving the appearance of a "hunched back"/outward kink at the junction between the thoracic and lumbar vertebrae, though this is usually mild.

Lordosis

Kyphosis

Scoliosis refers to a sideways curve in the back, and may develop as the child grows older. Early detection leads to more effective treatment (for example, by bracing).



Scoliosis

The combination of kyphosis and scoliosis (*kyphoscoliosis*), and an abnormally-shaped chest wall (see above) does make the work of breathing more difficult in some cases. In the long term, this makes people with SED more prone to chronic respiratory problems and can sometimes lead to heart failure in middle age. Again, early treatment of back/chest problems can help reduce the likelihood of this happening.

'Alec's spinal jackets come in a wide range of colours and designs. He's set a new fashion trend amongst his peers. The release of Walt Disney's Hunchback of Notre Dame has made us all aware of what he might face in the future.'

'The spinal curvature caused the bottom of the ribcage to rub on the top of the pelvis so that my left side went numb.'

The neck

Most importantly, there may also be problems in the neck region. Underdevelopment of one of the bones of the neck, the 'axis' or second cervical vertebra, which is responsible for neck stability, 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 damage to the spinal cord may be permanent or even fatal. To be alert to its possibility, a neck X-ray should be taken as soon as the diagnosis of SED is made to detect any structural problems. Call a doctor immediately if anyone with SED experiences the following symptoms of spinal cord compression:

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

People with SED should receive regular neurological check-ups (with x-rays as required) to assess neck stability and spinal cord function, and to identify problems at an early stage.

Prevention

Back problems can often be lessened by these simple measures:

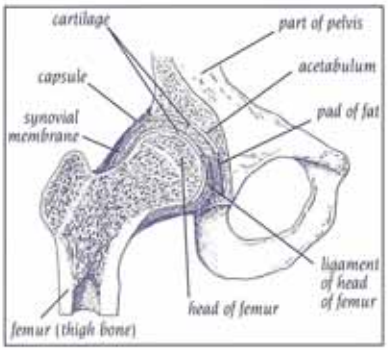
- following a well-balanced diet which will prevent overweight (in fact, many problems can be avoided or lessened by getting

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/poor lifting technique and high-impact sports such as jogging.

In young children, it is important to make sure the 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, avoid using baby walkers, jumpers or backpacks or similar items that do not have adequate back support.

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 previous page).

Limbs, joints and hips



The majority of people with SED are affected by hip problems. The shape of the hip may cause the foot to turn inwards (coxa vara). The pelvis and ends of the femurs (thighbones) that make up the hip joint are underdeveloped, causing restricted hip movement and a characteristic 'waddle'. Curvature of the spine aggravates the problem as it causes the pelvis to be tilted and further reduces free movement of the hip joint. These abnormal stresses on hips, knees and ankles, together with abnormal joint cartilage, lead to osteoarthritis later in life, sometimes requiring surgery.

'When I was about ten, I had three operations, two hip and one leg, to try to improve my walking ability, but to no avail. Fortunately I could ride a motorcycle and drive a car, but the restriction of movement has always to be considered.'

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 you feel happy with. The RGA will be able to help you contact specialists with the appropriate experience.

Breathing

Young babies with SED congenita may experience breathing problems in the first few weeks after birth if their rib cage is particularly small. Breathing may be eased by the clearing of secretions from the lungs. These difficulties should pass as the baby grows. Breathing problems may persist if they are related to the flattening of the face and/or a cleft palate repair that restricts the upper airways. Removing tonsils and adenoids or other nasal surgery may improve the situation.

The eyes

Over 40% of people with SED congenita have varying degrees of shortsightedness (*myopia*). These people may also be more prone to retinal detachment – a condition where the thin, light-receptive membrane across the back of the eye which allows us to see, becomes removed from the back of the eye. This is a potential cause of blindness. People with SED should receive regular eye-checks to ensure that any problems are treated early.

'I've had severe eye problems since I was about 40: cataracts, detachments, glaucoma. Moorfields Eye Hospital made no connection between these problems and SED.'

Childbirth

Women with SED are just as capable of having babies as women without SED, but need to be closely monitored throughout pregnancy. Narrowing of the pelvic bones means that a planned caesarean section will probably be necessary. Ideally this should be done under an epidural to avoid the serious risks of a general anaesthetic (see below). You will find more information in the **Lifestyles** booklet entitled *Having a Baby*.

Anaesthetic complications

People with SED who face an operation that requires a general anaesthetic should make the anaesthetist aware of their condition well in advance of the actual operation. The fact that people with SED are prone to kyphoscoliosis (which can make breathing difficult and lead to long-term lung problems), unstable necks and narrowing of the vocal cords and windpipe (laryngotracheal stenosis), means that a general anaesthetic is a high-risk procedure. Alternatives (such as epidural anaesthesia) should be considered.

Living with SED

There are many ways of overcoming some of the difficulties associated with having shorter arms and legs, and back problems, that may restrict mobility. Contact the RGA office for information on a wide range of gadgets: reaching aids and adaptions (refer to the **Lifestyles** series).

Take the lead

One of the greatest problems for people with SED 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 to 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, especially *Going to School* and *The Teenage Years*.

'We had been told by our GP that "babies come in all shapes and sizes so take him home and care for him as normal". We did exactly that, giving him the same love, care and attention as our daughter. Luckily for us, our family life was not damaged by the stress and strain and my husband was very good, understanding and strong. His love for our son gave our family the best possible reasons to carry on as normal despite Richard not growing properly.'

'My depression lasted for 18 months after Alex was born. Going to RGA conventions brought mixed emotions. This is how he will look as an adult. But now, with so much love and support, there is light at the end of the tunnel, and hope for the future.'

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 – SED 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, chest physician, 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 quotations would have summed up the experience of many parents of a baby newly diagnosed with SED.

'I didn't see my child for three days. No explanation was given. I was just fobbed off. Finally, after prolonging the agony, there was shock, horror, total numbness. A lot of tears shed. Fear. Denial. "Oh God, this isn't happening to us." Questions constantly agonising us.'

'One of the most frustrating aspects of the diagnosis was the lack of information. I was desperate to find out what SED actually meant to our family.'

Now, however, there are other possibilities.

'Being able to talk to another mother whose child has the condition helped enormously, as she understood completely my fears and problems. I could relax and talk about my son knowing we had a great deal in common.'

'Socialising with others with similar problems is a great way of gaining useful, practical information and also is an invaluable support emotionally.'

What causes SED?

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 and tendencies to certain diseases and conditions.

Sometimes a genetic problem arises which affects an aspect of the person controlled by a particular gene.

The gene responsible for SED congenita and SED tarda is known as COL2A1 and was identified in 1989. It was found to lie within chromosome 12q, and it is part of the code for an extremely important protein, Type-II collagen. The function of Type-II collagen and its role in SED is described earlier in this booklet. The gene for late X-linked SED lies on the X chromosome (Xp22).

How is SED *inherited* ?

No-one knows how many people in the United Kingdom have SED since the condition does not have to be registered. Some estimates suggest about one person in a million has the disorder.

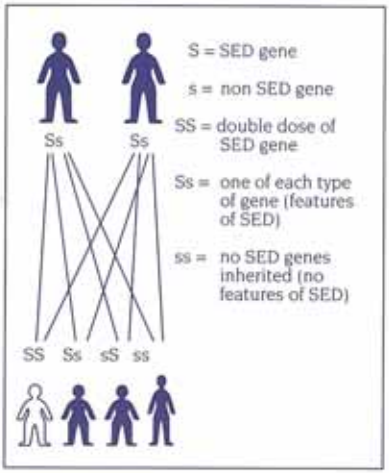
SED *congenita and* SED *tarda*

Most children with these types of SED are born to parents of average stature. In these cases a spontaneous change (mutation) of the COL2A1 gene has occurred in either the mother's (egg) or father's (sperm) genetic contribution to their child some time prior to conception.

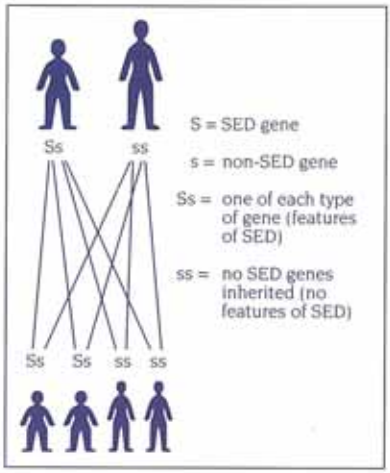
Once the genetic change has occurred, a person with SED can pass the condition on to his or her children. However, every individual with SED carries two copies of the gene responsible: one normal and one causing SED. So if one parent is already affected by SED (and the other parent is not), their children have a 50% chance of being affected. This is because SED congenita and SED tarda are examples of *dominant* conditions where the effects of the SED 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 SED 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 SED gene from either parent and a one in four (25%) chance that the baby would inherit the SED gene from both parents. It is not known what would happen if this occurred in SED. A number of different genetic changes in the COL2A1 gene all produce the same features of SED. If the child of two people with SED inherits two copies of the abnormal COL2A1 gene, the copy inherited from the father is likely to contain a different mistake from that inherited from the mother. This will result in varying degrees of severity in the type of SED that manifests.

This is best illustrated in the following diagrams :



Both parents with SED



One parent with SED

Some combinations of conditions (such as achondroplasia and SED) can give rise to severe, often fatal, deformities in the offspring. If two people of restricted growth are planning to start a family, they should seek *genetic counselling*.

'Having SED helps me as a parent bringing up Annabelle, by knowing what lies ahead for her, especially when I am dealing with doctors.'

Samantha, Annabelle and Warwick

Late X-linked SED

X-linked inheritance is a special way of transferring genetic information from one generation to the next. There are two types of sex chromosomes: X and Y, which determine whether you are male (XY) or female (XX). Several genetic conditions are caused by genes carried on the X-chromosome. In people with late X-linked SED, affected males have one affected X-chromosome and one unaffected Y-chromosome and therefore show the condition. Females who carry the affected X-chromosome will always have one unaffected X-chromosome and therefore will not usually display features of the condition, but can pass it on to their male children. Sometimes females do show symptoms of the condition due to a process known as 'Lyonisation', whereby one of the two X-chromosomes in each cell is randomly switched off. If the abnormal X-chromosome is switched on in more cells than the unaffected X-chromosome, then the woman will show signs of SED.

Diagnosis and treatment

Genetic Testing

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

How is SED *Diagnosed?*

The diagnosis of SED, like all other conditions, is based on a combination of clinical suspicion combined with various investigations. Many of the features of SED congenita are, by definition, present at birth and some indications may be picked up before birth on ultrasound screening. Late X-linked SED and SED tarda are not usually diagnosed until mid-childhood.

Doctors will look for a number of different characteristic x-ray features which can be used to confirm the diagnosis:

- generalised delay in early bone development (at ossification centres)
- small, irregular, fragmented epiphyses (the ends of the long bones) that progressively worsen with time
- the spinal vertebrae appear oval in childhood – later they appear flattened (*platyspondyly*). The second cervical vertebra (the second neck bone or axis), which forms a stable base for neck rotation, may be underdeveloped. This can lead to problems with neck instability (see earlier in this booklet).
- hips and legs: lack of bone development in the pubic bone; the heads (epiphyses) of the *tibia* (shin bone) and *femur* (thigh bone) are small and underdeveloped in both shape and bone formation. The

combination of delayed bone development, the high-riding position of the femur and limited hip movement often gives the false impression of a dislocated hip

- the long bones have irregular flaring of the *metaphyses* (neck of the bone) and underdeveloped epiphyses (bone endings) – this is especially noticeable in the femur or thighbone (see earlier diagrams). The bones become more 'normal' towards the ankles and wrists.

Treatment

At the moment, there is no single treatment that will result in a child with SED attaining a 'normal' appearance by the time they are fully-grown. There is no magic bullet that targets the abnormal COL2A1 gene in SED congenita and SED tarda or the Xp22 gene in late X-linked SED, and the effect of growth hormone on people with SED is not fully known.

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

Surgery has been used to lengthen the legs of people with SED who feel that their short stature is a disadvantage. However, it is not

without complications and the process can often be long and painful. It remains a controversial issue amongst affected people and the severity of hip and spinal problems may have a bearing on the medical advisability of this procedure. 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. This is one of the reasons why the RGA encourages its young members to socialise together and develop positive attitudes. Unfortunately, this surgical procedure can sometimes be 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 the treatment of SED.

In this country the largest organisation for parents, their children and other individuals with SED 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 specific to SED. Their 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: 01132 703791

CLAPA (Cleft Lip and Palate Association)
138 Buckingham Palace Road
London SW1 9SA
Tel: 0171 824 8110

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.

McKeand J., Rotta J. and Hecht J.T. (1996) 'Natural History Study of Pseudoachondroplasia'. *Am. J. Med. Genetics* 63: 406-410.

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.

Glossary of terms

Autosomal Dominant/Recessive – these terms refer to two 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 chromosomes: 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 of 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.

Cleft palate – fissure (or break) in the roof of the mouth resulting from a failure of the two sides to fuse in the developing embryo

Clubfoot (known medically as *talipes*) – a twisting or turning of the foot making it impossible to stand with the sole of the foot flat on the ground.

Collagen – a protein contained in cartilage, bone and connective tisue

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.

Femur – the long bone of the thigh

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 – the process by which knowledge and advice concerning inherited disorders and the possibilities of passing on particular conditions from one generation to the next, together with possible options regarding diagnosis and management, are given to affected people and their families.

Hypotonia – excessive 'floppiness' caused by insufficient muscle tone.

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

Kyphoscoliosis – a combination of an outward kink and a sideways curve of the spine.

Lordosis – inwards curvature of the lower spine.

Metaphysis – neck of a long bone.

Myopia – shortsightedness.

Platyspondyly – flattening of the spinal vertebrae.

Sciatica – pain or tingling in the legs caused by compression (or squeezing) of the nerves at the base of the spine.

Scoliosis – a sideways curve of the spine.

Sternum – the central breast bone.

Tibia – the larger of the two bones in the lower leg.

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).

List of publications available from the Restricted Growth Association

What is . . .?

Achondroplasia

Hypochondroplasia

Pseudoachondroplasia

SED

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

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