What is Diastrophic Dysplasia?

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 diastrophic dysplasia, 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.

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'There were special adaptations at school – things like pencil grips and foot stools and friends were a great help. My biggest challenge came when pursuing my degree in architecture – the drafting table was bigger than me, so thank God for computers!'

The contents of this booklet are believed to be correct at the time of publication. However, knowledge about diastrophic 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 Diastrophic Dysplasia?

by Will Christian MBBS, BSc

"The ravages of diastrophic dysplasia are irrational, but all is not lost. Life can be lived to the full and enjoyed. I have travelled extensively, lectured in many parts of the world, received many awards and held senior executive positions in large companies and organisations. My motto has been "Use what you've got and don't complain about what you haven't."

Dr Charles Pocock, Founder of the RGA.

Diastrophic dysplasia is a rare condition leading to extremely restricted growth with disproportionate stature ('disproportionate' means that the arms and legs are short in comparison to body length).

When the initial shock of the diagnosis has settled, many parents are anxious to know what their child's future will be. As with many causes of short stature, diastrophic dysplasia is mainly a physical condition – people with the condition have a normal range of intelligence, occupy positions in many areas of society and have a range of heights and abilities. Most people live normal, happy lives (with a normal life-span) although there will, of course, be some physical limitations.

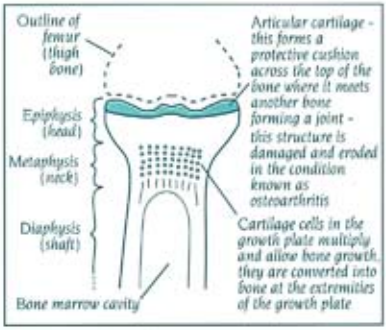
Many practical difficulties can be overcome with a little imagination. Clothes can be altered, and cars/bicycles can be adapted (for more information see the RGA **Lifestyles** series), and there is no reason why someone with diastrophic dysplasia should not participate in most activities. All children (and adults!), including those with diastrophic dysplasia, 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 diastrophic dysplasia mean?

Diastrophic literally means 'twisting' and *dysplasia* means 'abnormal growth'. It is a genetic condition, beginning at conception and continuing throughout life.

Long bones (for example, the arms, legs, fingers and toes) form from *cartilage**. Cartilage is an amazing substance that occurs in many areas of the developing baby and child. It is made up of a complicated chemical gel and often contains special fibres to give it strength and flexibility (a bit like the steel rods in reinforced concrete). As well as forming the basis for bone development, cartilage also acts as a shock absorber between the joints of the spine and the ends of long bones. It is also found in many other areas of the body where it helps to cushion and protect, or give shape to things (e.g. around the ear, keeping it stiff but flexible).

As a baby develops in its mother's womb, cartilage forms the initial template for bone formation. In the older child, its production (by special cells called *chondrocytes*) is restricted to the ends of long bones in an area called the *growth plate* (see diagram). Cartilage is a vital component for normal bone growth. In people with diastrophic dysplasia cartilage, and therefore the growth plate too, is altered in its chemical make-up. Sulphur (a pale yellow, non-metallic chemical) is a vital component of normal cartilage, giving it strength and enabling it to resist compression. Diastrophic dysplasia is thought to be due to a defect in the incorporation of sulphur into cartilage caused by the 'diastrophic dysplasia sulphate transporter' (DTDST), leading to the effects described later.

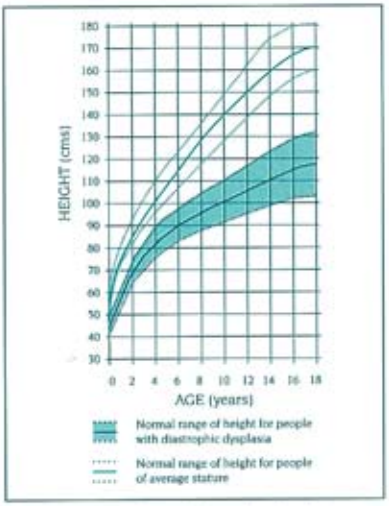


Development of long bones

* Words in italics are found in the glossary

The average height range for both men and women with diastrophic dysplasia is between about 103cm and 132cm (3'4" and 4'4").

Paediatricians and GPs should use growth charts designed specifically for children with diastrophic dysplasia to predict their growth rates and expected heights (see diagram). These charts can be obtained from the RGA (address at the end of this book). Final height is influenced by the progressive effects of the condition on the back, hips, knees and feet.



Height chart for people with diastrophic dysplasia

Other effects of diastrophic dysplasia

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

- extreme short stature
- a near-normal body length at birth with short arms and legs
- normal head size with normal facial features (although people with diastrophic dysplasia all have the same shape nose)
- joint contractures, which may be present from birth and worsen with age – the ligaments that hold certain joints together may get shorter and/or stiffer as time progresses, leading to joint deformity (e.g. spine curvature or foot problems).
- increased curvature of the lower spine (lumbar *lordosis*) and the upper spine (*kyphoscoliosis*) that tends to worsen as the individual grows older

- short fingers with a distinctive appearance of the thumb ('Hitch-hiker thumb' – the thumb tends to stick out from the hand because one of the bones at the base of the thumb is short)

- short, broad, flat feet with rigid deformities that progressively worsen as the child grows, leading to a tendency for the feet to point downwards (as if standing on tip-toe) – known as an *equinovarus deformity*. Unfortunately this can be very difficult to prevent or treat

Adult foot *Baby's foot*

- misshapen or 'cauliflower' ears which may be the result of swelling of the ear at birth due to bleeding from the ear cartilage into the skin of the ear. This condition can occur in anybody. However, because of the abnormalities of cartilage development, it is more frequent in diastrophic dysplasia. The swelling must be drained soon after birth in an attempt to minimise any deformity to the ear that may arise from its presence. Misshaping of the ears can also occur in later childhood due to problems with cartilage development. Surgical removal is possible in adulthood and may also alleviate hearing problems
- cleft palate (a problem with the development of the roof of the mouth leading to a gap in the palate, sometimes associated with cleft lip or hare lip) occurs in about 25% of people with diastrophic dysplasia
- limited movement at the hips

with an increased frequency of hip dislocation, which is worsened by the progressive joint contractures.

- osteoarthritis of the hips and knees
- neck problems (see later).

It is important to remember that overall intelligence is normal and that, although babies with diastrophic dysplasia may reach their developmental milestones at a later age, development will be within the normal range.

Michael is Britain's smallest man according to the Guinness Book of Records

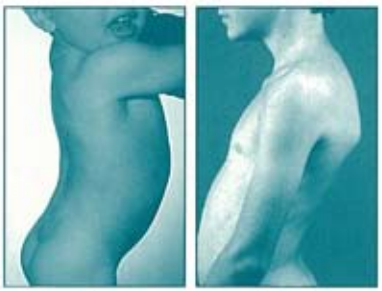
Possible complications and solutions

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

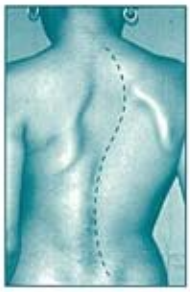
The back

People with diastrophic dysplasia are especially prone to back problems, particularly *kyphoscoliosis* – see next page. 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 diastrophic dysplasia 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 results in an unpleasant tingling/numb sensation in the lower limbs, usually when walking or lifting.



Lordosis *Kyphosis*



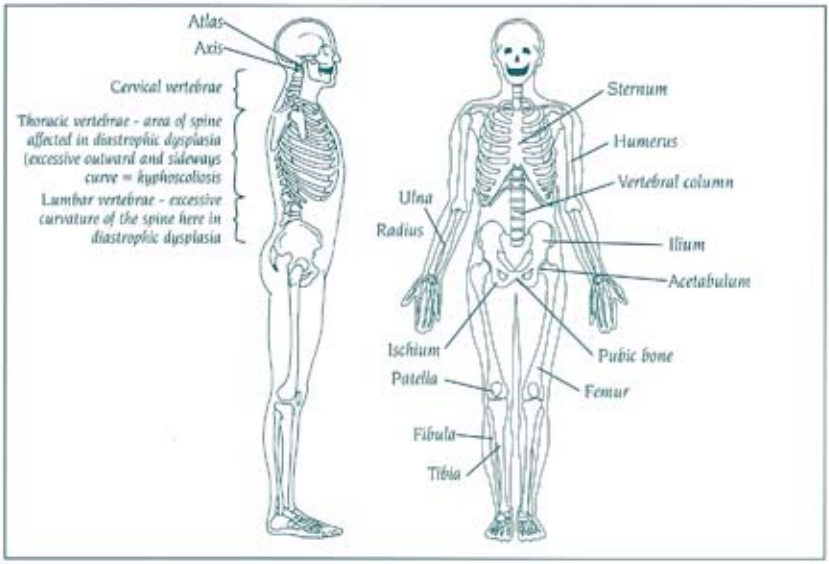
Scoliosis

Kyphosis refers to an outward curving of the spine, giving the appearance of a 'hunched back' or an outward kink at the junction between the *thoracic* and *lumbar* vertebrae. *Scoliosis* refers to a sideways curve to the spine. Both conditions may progressively worsen as the individual gets older.

The use of a back brace while the curvature is still small may help reduce the problem. If bracing does not work, surgery may sometimes be necessary to correct the problem and support the spine. Surgery may also be necessary to alleviate breathing difficulties or heart strain caused by a severely curved back.

The neck

Most importantly, there may be problems in the neck region. A dangerous combination of under-developed 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 diastrophic dysplasia is made, to detect any structural problems with the neck. In addition, a doctor should be called immediately if anyone with diastrophic dysplasia 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.

Reducing back problems

Back problems can sometimes be reduced by:

- dietary measures to prevent obesity (It is important to encourage 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 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, 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 immediately if any of the features of spinal cord compression become apparent (see above).

Limbs, joints and hips

The majority of people with diastrophic dysplasia are affected by hip problems. The hip tends to be rotated so that the leg points inwards ('coxa vara') or outwards ('coxa valga') from the middle of the body. This makes dislocation of the hip more common, especially if the severe and progressive joint contractures that occur in diastrophic dysplasia affect the hip.

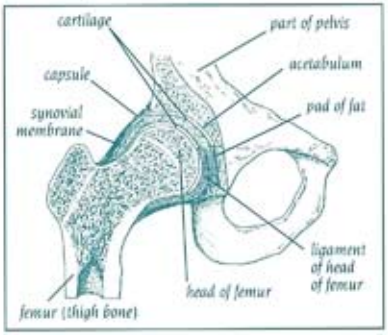


Diagram showing detail of hip joint

A form of arthritis (joint disease) called osteoarthritis, is more common in people with diastrophic dysplasia, and affects most of the weight-bearing joints in the body, starting with the hips. Curvature of the spine aggravates this problem at the hip as it causes the pelvis to be tilted, which in turn reduces free movement of the hip joint. The osteoarthritis is also worsened by joint contractures which pull joints out of line, exerting unnatural stresses upon them.

'Contractures of the hips and knees are already pulling my child into an unnatural standing position. I do physio every day, the school carers are supposed to help but that is an ongoing problem! Jamie's spine seems to be quite good at present but having read your booklet I will ask about the neck problem on my next visit.'

'All deformity occurs during the growing years and is irrational. For example, my right hand is different from my left. Generally, my experience is that where two bones meet, there are problems.'

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 able to help you contact specialists with the appropriate experience.

Childbirth

Women with diastrophic dysplasia are just as capable of having babies as women without it, but they need to be closely monitored throughout pregnancy. Problems with the pelvic bones means that a planned caesarean section will be necessary. There is more information in the **Lifestyles** booklet *Having a Baby*.

Anaesthetic complications

People with diastrophic dysplasia 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 diastrophic dysplasia are prone to kyphoscoliosis (which can make breathing difficult and may lead to long-term lung problems), and unstable necks means that a general anaesthetic is potentially a high-risk procedure. Alternatives should be considered if possible.

Living with diastrophic dysplasia

There will 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. 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.

There is more information in the **Lifestyles** booklet on *Personal Hygiene*, and you are encouraged to contact the RGA office for information on a wide range of gadgets.

Take the lead

One of the greatest problems for many people with diastrophic dysplasia 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 the attributes of good parenting such as listening, educating, loving, setting a good example and exerting 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 earlier) 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 in *Going to School* and *The Teenage Years*.

'J. has always been Mr Happy! His excellent character overcomes most things; we work together and try things out. It is difficult not to be over-protective, but he tells me off! The school are very over-protective, but he does accept that. It doesn't stop him chasing the girls and making them scream or playing with the boys. He is good at swimming. He needs a snorkel though. We haven't managed to keep his head above water yet!'

Parents also need to take the lead in obtaining the most appropriate help and advice for their child. GPs may need to have things explained to them – diastrophic dysplasia is a rare condition and many doctors may not have encountered it before. Parents may need to ask for referral to an appropriate specialist such as an orthopaedic surgeon, rheumatologist, chest physician 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.

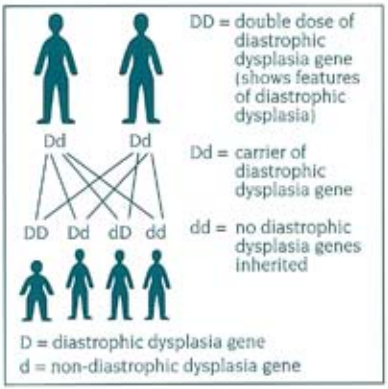
What causes diastrophic dysplasia?

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 cell single cell in the body. *Genes* govern the more obvious things like the colour of the 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. Nearly everyone (including those with diastrophic dysplasia) carries two copies of every gene, one from their mother and one from their father. Specific genetic problems may arise when the aspect of the person controlled by a particular gene is affected.

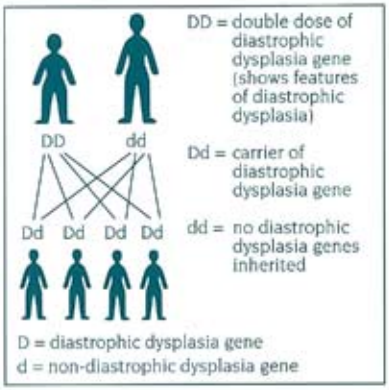
The gene thought to be responsible for diastrophic dysplasia was discovered in 1994. It was found to lie within *chromosome* 5, and it is the code for an extremely important sulphate transporter, responsible for incorporating sulphur into cartilage (see earlier). The gene is called the 'diastrophic dysplasia sulphate transporter' gene (or DTDST for short).

How is diastrophic dysplasia inherited ?

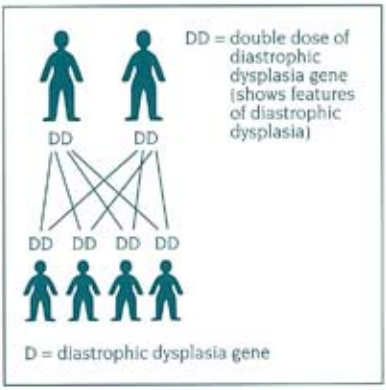
Diastrophic dysplasia is not a notifiable condition. This means



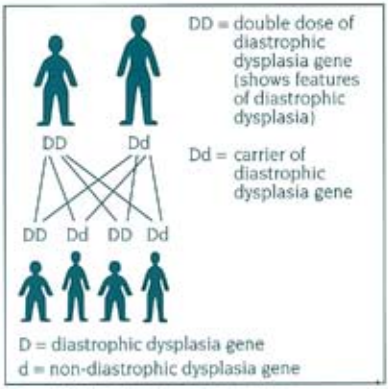
Both parents carriers of diastrophic dysplasia



One parent with diastrophic dysplasia



Both parents with diastrophic dysplasia



One parent with diastrophic dysplasia, one parent carrying one copy of the diastrophic dysplasia gene

that when a child with diastrophic dysplasia is born, the fact that he or she has the condition does not have to be registered. Consequently no one knows exactly how many people have the condition, though some estimate that it affects about one in every one million people.

Most children with diastrophic dysplasia are born to parents of average stature. In these cases, each parent must carry one copy of the affected (DTDST) gene and one normal copy each (see diagram).

Normally, people inherit one copy of every gene from their father and one from their mother. If only one copy of the DTDST gene is inherited (for example, from the father) and the copy received from the other parent is normal, then the child also appears normal ('Dd' in the diagram). If two copies of the DTDST gene are inherited (one from the father and one from the mother), then the child will have diastrophic dysplasia ('DD' in the diagram). This is because diastrophic dysplasia is an example of an *autosomal recessive* condition where two copies of the affected gene are required before the individual can have the condition.

The chances of a second child in the family being affected by diastrophic dysplasia are 25%.

If both parents are affected (i.e. each parent has two copies of the DTDST gene), all of their children will be affected (see diagram).

If only one parent has diastrophic dysplasia, then the odds of having an affected child depend on whether or not the unaffected partner carries the DTDST gene or not. If they do NOT carry it, then the chance of an affected child is zero (as, to be affected, the child needs to inherit one copy of the DTDST gene from each parent) but all of the unaffected children will be carriers of the DTDST gene.

If the average height partner DOES carry the DTDST gene, then the chance of a child being affected is 50% – the rest of the children will be carriers for the gene and will appear normal.

The best advice, especially for couples who are both of restricted growth but have different conditions, is to seek *genetic counselling* from your local genetic

centre if you are planning to start a family. They will help calculate the chances of your child being affected and can give you a lot of practical help and advice.

Diagnosis and treatment

Genetic testing

The fact that the gene for diastrophic dysplasia 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 diastrophic dysplasia is so low. Indeed, many would argue that the introduction of a screening program for diastrophic dysplasia where neither parent has the condition demeans individuals with the condition and ignores the fact that the majority of people with diastrophic dysplasia live active, healthy, fulfilling lives.

How is diastrophic dysplasia diagnosed ?

The diagnosis of diastrophic dysplasia is based on a combination of clinical suspicion along with various investigations. Diastrophic dysplasia 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 the condition, as there are other conditions with a similar appearance.

'I knew I had the diastrophic dysplasia gene, and there were problems at 22 weeks gestation; the scan revealed that the limbs were an abnormal length. My doctor gave me a diagnosis straight away, recognising the "hitch-hiker's" thumb. I thought I was carrying some sort of monster baby and wanted to die. At about 28 weeks I was scanned again. To my delight my baby had beautiful rosebud shaped lips – did that mean no hair lip and no cleft palate? "Maybe", my doctor said. Then I noticed my baby's feet, I thought club foot meant just stumpy leg ends! The second scan probably saved both our lives. I is 7 years old now, and a proper little boy who makes everyone happy!'

After birth the diagnosis can be confirmed on the external features described above, and on X-ray appearance. Diastrophic dysplasia has certain characteristic X-ray features which doctors will look for:

Limbs: Short limbs versus trunk length

Skull: Normal, apart from a possible cleft palate and abnormalities of the external ear.

Spine: Appearance varies greatly between individuals. The vertebrae may appear either normal, flattened (platyspondyly) or unusually tall. A vital part of the second cervical vertebra (the second neck bone or 'axis') called the odontoid process, which forms a stable base for neck rotation, may be underdeveloped. This can lead to problems with neck instability (see earlier). Kyphoscoliosis and lordosis are apparent from an early age.

Hips and femur: Again, appearance is variable. Iliac wings tend to appear flared. Capital femoral *epiphyses* (the head of the thigh bone) are often late to develop but may subsequently be normal. Often however, they are abnormal/fragmented, and the hip is rotated inwards ('coxa vara').

Hands: Irregular shortening of the metacarpals (bones in the hand). The bone at the base of the thumb

(the first metacarpal) is short and underdeveloped, leading to the characteristic 'hitch-hiker' thumb, where the thumb sticks out perpendicularly to the hand.

Long bones: irregular flaring of the *metaphyses* and underdeveloped epiphyses.

Treatment

At the moment, there is no single treatment that will result in a baby with diastrophic dysplasia attaining a 'normal' physique/appearance by the time he or she is fully-grown. There is no 'magic bullet' that targets the abnormal DTDST 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.

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 diastrophic dysplasia as soon as they arise. Most of these complications and their prevention or treatment have been mentioned earlier.

Surgery could theoretically be used to lengthen the arms and legs of people with diastrophic dysplasia who feel that their short stature is a disadvantage, although this remains a controversial issue among affected people. However, because of the frequent complications of joint contractures, severe osteoarthritis and joint degeneration in diastrophic dysplasia, it is not commonly recommended, as this quote from a parent explains:

'I feel leg lengthening is not an option as the joints are unstable and the ligaments lax. I have asked about arm lengthening, that would really help, but it's complicated. We are looking into the problems.'

Sources of information

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

In this country the largest organisation for parents, their children and other individuals with diastrophic dysplasia 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 diastrophic dysplasia. 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: 0113 270391

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 :

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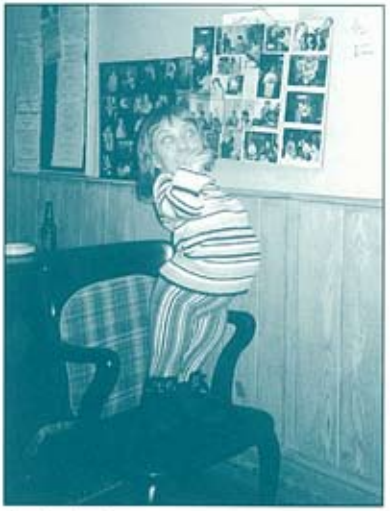

"Playing darts"

Glossary of terms

Autosomal recessive – one way in which conditions can be passed on from one generation to the next. *Autosomal* refers to the fact that the gene responsible for the condition is located on a normal chromosome and not on one of the special chromosomes (X or Y) that determine what sex the individual is. *Recessive* refers to the fact that it requires two copies of the affected gene to produce noticeable effects in the individual concerned.

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 – special types of cells that are 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.

Diaphysis – shaft of a long bone.

Epiphysis – the end of the long bone which is normally separated from the shaft by the growth plate (the area 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 body's 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 patients/parents and their families.

Growth plate – area of the long bone where growth occurs.

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 – neck of the long bone.

Scoliosis – a sideways curve of the 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).

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