

CRI DU CHAT FAMILY ROUTE MAP

INTRODUCTION

This leaflet is designed to provide signposts to sources of current information and appropriate services for patients, their families, carers, together with healthcare professionals. The issues and concerns raised during a series of patient focus groups in 2006 organised by the Genetic Interest Group (GIG) now the Genetic Alliance UK (GAUK) identified seven themes which have been used as the basis for developing this resource with the help of patient's families and carers and clinical staff with experience and expertise of this condition.

CRI DU CHAT SYNDROME

Cri du Chat syndrome (CdCS) is a relatively rare chromosome disorder affecting approximately 1 in 37,000-50,000 live births. The exact sex ratio is not known although reports indicate that females outnumber males by 2 to 1. The syndrome is known to result from a deletion from the short arm of chromosome 5 and represents one of most common deletion syndromes in humans. A deletion occurs when there is a loss of material from one chromosome because of either one break (a terminal deletion) or two breaks (an interstitial deletion). Recent molecular research has further highlighted a 'critical region' on chromosome 5 (5p15.2) that appears to be specifically involved in displaying the classical features of CdCS. If the deletion breakpoint includes this 'critical region' the characteristic features diagnostic of the syndrome will be present. Usually the loss from the short arm of chromosome 5 is purely accidental and thus the risk of recurrence is very low, no greater than the original risk of 1 in 37,000-50,000. In 80-95% of cases, the genetic material is lost from the end of chromosome 5 (terminal deletion). When the parental chromosomes of the children with terminal deletions are found to be normal, the deletions are referred to as 'sporadic'. However, in 10-15% of cases this deleted chromosome is inherited from a parent. When this occurs the risk of having another affected child is much greater than when the syndrome results from a sporadic deletion. A Genetic Counsellor or GP is the most appropriate person to provide professional advice on risk in future pregnancies. The following web-link provides details and locations of NHS genetic services in the UK: <http://www.geneticalliance.org.uk/services.htm>

SUPPORT GROUP

Cri du Chat Syndrome Support Group (CdCSSG)

VISION

Our vision is a society that values and nurtures those with Cri du Chat syndrome enabling them to live full, happy and healthier lives.

MISSION STATEMENT

We aim to provide support to people with Cri du Chat syndrome by engaging with them and supporting their families, their carers and professionals. The CdCSSG will also strive to improve

knowledge of CdCS by facilitating appropriate research into the syndrome with the hope of creating a better quality of life for all members of a Cri du Chat family.

OUR PURPOSE

To make a positive difference in the lives of those with Cri du Chat syndrome

OUR BUSINESS

To support family members and professionals who care for those with Cri du Chat syndrome and encourage dedicated research that has the aim of providing a better quality of life to all affected by CdCS.

OUR VALUES

Those with Cri du Chat syndrome and their families can have complete, satisfied and rewarding lives. CdCS does not in any way hinder families from achieving the opportunities open to other members of society.

INFORMATION FOR HEALTH CARE PROFESSIONALS

Cri du Chat syndrome is an autosomal deletion syndrome caused by a partial deletion of chromosome 5p and is characterized by a distinctive, high-pitched, catlike cry in infancy with growth failure, microcephaly, facial abnormalities, and profound intellectual disability throughout life.

Information and articles about CdCS are available from the support's group website:

www.criduchat.org.uk.

More general information can be found on the following websites:

- Medscape Reference: <http://emedicine.medscape.com/article/942897-overview>
- Patient.co.uk: <http://www.patient.co.uk/doctor/Cri-Du-Chat-Syndrome.htm>

INFORMATION AND SIGNPOSTS

Information and Signposts have been divided into the following categories and some issues that may be relevant for you have also been explored:

DIAGNOSIS

Diagnosis is usually performed within the first few days of birth. For some babies this may be longer because the syndrome may not be initially evident. The syndrome is determined by a combination of symptoms, physical examination by a specialist and genetic testing indicating missing part of chromosome 5.

Symptoms include:

- Cry that is high-pitched and sounds like a cat
- Downward slant to the eyes
- Low birth weight and slow growth
- Low-set or abnormally shaped ears

- Intellectual disability
- Partial webbing or fusing of fingers or toes
- Single line in the palm of the hand
- Skin tags just in front of the ear
- Slow or incomplete development of motor skills
- Small head (microcephaly)
- Small jaw (micrognathia)
- Wide-set eyes

Physical examination may identify the following:

- Inguinal hernia
- Diastasis recti (separation of the muscles in the belly area)
- Low muscle tone
- Epicanthal folds, an extra fold of skin over the inner corner of the eye
- Problems with the folding of the outer ears

Fully genetic testing (from a small blood sample) is recommended as it is the only method of obtaining an accurate diagnosis. Parents may also wish to be tested to see if they have the 10-15% chance of being carriers.

Family advice

If it is a new case then there is no risk to the immediate family and close relations. If a parent is found to be a carrier then further investigations may be warranted of their siblings. Leaflets for counselling, advice for appointments and siblings for these question can be sought from; http://www.geneticalliance.org.uk/publications_patients.htm

TREATMENT

The syndrome cannot be cured, however many patients will be referred to and require a variety of different therapies and specialties as they develop. The following sections will provide some information regarding each therapy. More detailed information is available on the website.

Contacting your GP

It would make your visits easier if you made your position clear to your primary care provider i.e. GP surgery. It would be sensible if you could be assigned a regular GP who would know the CdCs case. Providing them with this route map would remind them. It is also recommended that the 'carers' make themselves known to the practice as this has added considerations to their needs.

CHILDREN UP TO THE AGE OF TWO

Children with CdCS may not have all the problems listed below. However these are common problems encountered:

- Delayed milestones and failure to thrive
- Muscle weakness or low-tone
- Poor mobility
- Gastric reflux and vomiting

- Constipation
- Difficulty in feeding
- Heart Problems

CHILDREN OVER THE AGE OF TWO

As children grow older the following additional problems may become more apparent:

- Speech problems
- Hearing problems
- Vision problems
- Behavioural problems
- Orthopaedic problems
- Emotional difficulties

COMMON PROBLEMS & TREATMENTS

GASTRIC REFLUX

Gastric reflux is common in children with CdCS. There are many treatment options ranging from daily medication to surgery. The following are common treatments after consultation with a paediatrician:

- Thickened feeds
- Elimination of cow's milk and products
- Alginate (Gaviscon)
- Histamine-2 receptor antagonists
- Proton pump inhibitors
- Metoclopramide or domperidone
- Fundoplication (Surgery to tighten the top of the stomach)

DRIBBLING

Many children with disabilities experience excessive dribbling or drooling. Treatment options include medication, patches and surgery:

- Scopolamine (Hyoscine) patches
- Botox injections
- Translocation of salivary glands (Surgery to move glands to back of throat)

SLEEPLESSNESS

Sleep problems in CdCS children are very common indeed, occurring far more frequently than in the general population of children. Difficulties settling at night, waking too early in the morning and, especially waking repeatedly through the night, represent a severe cause for concern for almost 50% of the parents who participated in our survey. A quarter of the children had settling problems, but nearly twice as many also woke repeatedly through the night. Sleeplessness can lead to many problems for both child and carer. This includes behavioural problems such as aggression and over activity and can significantly interfere with learning. For carers, the effects can also be severe

resulting in stress, depression, relationship difficulties and also not coping with the many needs and demands of their disabled and other children.

There are many methods that can be employed to combat sleeplessness. Like all children a good sleep pattern established by a regular settling routine should be employed. For many children this will be sufficient and they sleep without fuss by three to six months of age. However, if sleep problems persist into childhood and are characterised by an ingrained pattern of not settling, waking repeatedly through the night or waking up too early, then try a routine known as: "SLEEP".

- Settle your child as quickly as possible at a set time into a safe and secure bedroom and wake him or her up at a set time in the morning (use an alarm clock)
- Leave your child after settling unless you suspect physical illness or danger
- Even if he or she cries out for attention
- Even if he or she screams or does anything else to demand your attention
- Persist and don't give in: this will teach your child quickly to get into a healthy sleep pattern and you will notice the benefits for everyone!

If problems persist do seek advice, this may be from a child Psychiatrist or Psychologist and could include medication, your GP or Health Visitor can arrange this.

BEHAVIOURAL

Several surveys have discovered an excess of behavioural problems in CdCS children. These include hyperactivity, aggressive and oppositional behaviour as well as sleep problems. What can you do if your child is behaving badly? Well, the first thing to do is to keep a record of what's happening in the form of a diary. To do this systematically, it helps to have 4 columns entitled "what happened", "what led up to it"; "what happened as a result" and, usually most importantly "what response to the behaviour did I make". Surprisingly, by doing this you may easily detect what is happening and by altering your response to the particular behaviour, either stop it or change it for the better.

Clear communication that is concise and said with conviction using words or symbols that the child readily understands can help and focussing on good behaviour with lots of praise; furthermore, actively encourage children to do praiseworthy things such as tidying up their toys, playing nicely with their siblings or friends, etc. An ounce of praise is worth a ton of punishment in these circumstances. Special Needs Teachers, Health Visitors and Child Psychologists are also good sources of sensible advice for families with badly behaved children.

THERAPIES

All children with CdCS will require a range of therapies as they develop. We will explore the most common therapies that your child may require. The list is not exhaustive and all children's individual needs will differ. Your family doctor (GP) or community paediatrician will usually be the main point of focus and coordinator of therapies. Advice should be sourced from these contacts when first discussing your child's requirements.

PHYSIOTHERAPY

Many children will experience physical difficulties during their development. Physiotherapy can help your child's physical development including helping them to sit, stand and improve their fine motor

skills. There are specialised physiotherapists who can supply orthotics to help with joint and spinal disorders known as Orthotists.

OCCUPATIONAL THERAPY

Occupational therapists work in-conjunction with physiotherapy to develop children's fine motor movements and activities of daily living. They also will be involved in ensuring that your home has the right adaptations for you and your child. In the unfortunate eventuality that your child requires a wheelchair, OTs will assist you in ensuring that your child has the appropriate chair.

SPEECH THERAPY

A child with CdCS may experience difficulty in swallowing and speaking. Speech therapists may work with your child from an early age to aid swallowing by providing advice on what is the best method of feeding your child. As your child grows older they may experience problems with their speech, speech therapists will provide guidance on how you can assist your child to communicate. In addition, dribbling is also a common problem for children with CdCS; speech therapists will also provide advice regarding how this can be managed.

DIETICIAN

Due to the problems of thriving many children with CdCS experience and their difficulty in swallowing a dietician will provide advice on special dietary requirements, for example high calorie drinks.

PSYCHOLOGIST

Due to the prevalence of behavioural problems in CdCS children many will require psychologically input and advice for their parents. In addition educational psychologists may be involved to assess the best educational placement for your child.

AUDIOLOGY

Glue ear is common in CdCS children and therefore children will often undertake tests to assess the quality of their hearing. Many children end up having hearing aids.

OPHTHALMOLOGY

Ophthalmology will assess your children's eyesight as problems are more prevalent in children with CdCS.

OTHER INFORMATION

INSURANCE

It is common to have difficulties finding adequate and affordable insurance policies once you have a pre-existing condition. The following company offers travel insurance for people with pre-existing conditions. There may be others available and this should not be taken as a recommendation:

The insurance group Banner – see www.bannergroup.com have agreed to look at and consider insuring anyone, however they cannot guarantee that they will cover everyone for all conditions, it will have to be on an individual basis.

FINANCIAL HELP

CAB (Citizens Advice Bureau) can help with advice locally about benefits and can be found through your phone directory.

FAMILY FUND

The Family Fund helps families with severely disabled children to have choices and the opportunity to enjoy ordinary life – giving grants for things that make life easier for the disabled child and their family, such as washing machines, driving lessons, hospital visiting costs, computers and holidays. They will consider funding anything connected to bringing up a disabled child.

DISABILITY LIVING ALLOWANCE

This is a benefit provided to those under the age of 65 with a long term health problem which affects your everyday activities. It is not means tested and not affected by any other benefits. It is split into mobility and personal care and each is assessed separately with a different rate of allowance for each. There is a lengthy form to complete and you may require assistance to complete. The amount awarded can be up to several hundred pounds paid four weekly. It is tax free.

MOTABILITY SCHEME

This is a means of purchasing a brand new car without the worry of servicing, tax, insurance etc. Applications can be processed through accredited dealerships and basically once approved; the car is paid for using your Disabled Living Allowance (higher rate of mobility care). You can make additional contributions but must receive the higher rate of DLA. Motability can be contacted at:

www.motability.co.uk or 01279 635666

Adaptations;

Mostly adaptations are not required though, due to poor muscular control, supporting seats may be required. Advice can be sought from your Occupational Therapist.

DIRECT PAYMENTS

Direct payments are local council payments for people who have been assessed as needing help from social services, and who would like to arrange and pay for their own care and support services instead of receiving them directly from the local council. A person must be able to give their consent to receiving direct payments and be able to manage them even if they need help to do this on a day-

to-day basis. The website www.direct.gov.uk provides further information.

EDUCATION

A leaflet entitled 'Statutory assessment—Education' (leaflet code ES26) from *Early Support* explains the process of requesting an assessment for your child and provides examples of letters to help you. It also describes what a **Statement of Special Educational Needs** is. This can be obtained from the *Early Support* website www.earlysupport.org.uk or by phoning 0845 602 2260.

Mostly preschools and nursery can accommodate CdCs with key worker help, then special primary schools have been found to benefit almost all; very few have been able to go mainstream. For your rights see; <http://www.direct.gov.uk/en/DisabledPeople/index.htm>

SPECIAL EDUCATIONAL NEEDS - IDENTIFICATION AND ASSESSMENT

Early identification, assessment and provision for any child who may have Special Educational Needs (SEN) is crucial. The SEN Code of Practice, to which schools, early education settings, (Local Educational Authority) LEA's and others must by law have regard, promotes a common approach to identifying, assessing and providing for all children's special educational needs.

School Transport

Transport to schools will normally be provided by your local authority and they will do a risk assessment for their seating requirements if necessary.

The National Parent Partnership

The National Parent Partnership Service offers impartial advice, information and support to parents/carers of children and young people with special educational needs. They are very helpful and can often save you hours of worry by simply knowing who you need to contact for what. They will also liaise on your behalf with schools/LEA. Simply log onto www.parentpartnership.org.uk to find your local contact.

SOCIAL CARE INFORMATION

ASSESSMENTS FOR CARERS

If you provide a regular and substantial amount of care for someone aged 18 or over, you can ask the social services department in your local council for a carer's assessment.

PREPARING FOR A CARER'S ASSESSMENT

There is no definition of 'regular and substantial care'. A carer's assessment means social services will look at your situation and see if you are entitled to any services that could make caring easier for you. Further information can be found on www.direct.gov.uk

RESPITE: YOUR RIGHTS AND BREAKS FROM CARING

Breaks or time off from caring are vital. Whether it's an hour every day, a couple of hours a week or a two-week holiday, we all need some time to ourselves, and carers are no exception. Breaks for carers are meant to give you time off from your caring role as well as enable you to do something that you want to do for yourself. The Carers Equal Opportunities Act (2004) was introduced to

ensure that you have the support you need to achieve a better balance in your life. The Act gives you the right to ask the local authority of the person you look after for a carer's assessment of your needs. During a carer's assessment, the local authority will ask you about your work, learning and leisure requirements.

TRANSITION

As the CdCs age their needs change and transition occurs from primary to secondary schools also from childhood and adulthood. So do the rules change and allowances from child to adult services. Keeping a record of all their needs will help with the transitions as will services like ;

<http://www.direct.gov.uk/en/DisabledPeople/EducationAndTraining/Schools/index.htm>

'Connexions' there are local services in each county/city council that help.

Inevitably a time will come when the care will change from parents to siblings or other carers due to natural aging, accident or health reasons. Looking at providers and services is advisable sooner rather than later so that the transition can be gradually introduced.

Wills and Trusts

There are services for making wills and trusts and as such do not recommend any particular group. Mencap will have a list of 'specialised' advisors on making wills that are special needs specific to your needs.

<http://www.mencap.org.uk/what-we-do/our-services/wills-and-trusts>

TRAVEL

Train

National Rail

National Rail needs advance warning if you need any assistance from staff - ideally more than 24 hours' notice. This is particularly important if your journey includes getting on or off a train at an unstaffed station.

Telephone: 0845 7484 950

Textphone: 0845 6050 600

Mainline trains

On mainline (intercity, suburban and cross-country) trains there is a space designed for wheelchair users to travel in safety and comfort. You must always use this space and should apply your brakes when the vehicle is moving. If you use a powered wheelchair, you should make sure that the power is switched off when travelling.

Local and regional services

Most trains can accommodate wheelchair users and new trains also have facilities to assist sensory impaired people, for example public information systems that are both visual and audible.

To arrange a train journey in the UK, contact National Rail Enquiries.

Telephone: 0845 7484 950

Textphone: 0845 6050 600

London Underground

There is information about the accessibility of stations on Transport for London's website.

BUS

Free bus travel in England for disabled people

Please see www.direct.gov.uk for more information.

BLUE BADGE SCHEME

Please see www.direct.gov.uk for more information. Most CdCs qualify.

HOME AND HOUSING OPTIONS

If you have impairment or a health condition that makes living alone difficult, making some adaptations to your home or getting extra support may help you to live in your own home.

Living in your own home

You have the right for your needs to be assessed by the social services department of your local council. Following a health and social care assessment, an occupational therapist may recommend types of equipment and ideas about adapting your home. You may be entitled to financial help, such as a Disabled Facilities Grant, to pay for adaptations or improvements to your home. Please see www.direct.gov.uk for more information.

Independent Living

There are difficult choices to be made as whether the CdCs stay with their family's or be a 'looked after person' and then go into independent living or sheltered accommodation; at each stage a multi-agency team [health, social services, educational] will assess the needs of all and come to a decision as what is the best compromise for CdCs, families and carers.

WHEELCHAIRS

One of the ways to get a wheelchair is through the National Health Service (NHS) Wheelchair Service. This includes an assessment to make sure your needs are met. Wheelchairs can be manual or powered; there are four different types of powered wheelchair.

NHS WHEELCHAIR SERVICE

NHS Wheelchair Services are run by local health authorities. They are responsible for allocating funds to the Wheelchair Service and Primary Care Trusts who are responsible for providing the service itself. This sometimes includes contracting the running of the service to an outside company.