CRI-DU-CHAT SYNDROME

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2nd Edition

In association with

Cri Du Chat

Syndrome
Support Group

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Foreward

The past decade has seen tremendous advances in our knowledge of cri-du-chat syndrome. At the *genetic* level, we can now pinpoint the critical region that is responsible for cri-du-chat to a small part within the short arm of chromosome 5 (5p 15.2-5p 15.3). At the *developmental* level, there is strong evidence that toddlers and children with the syndrome, although displaying developmental delay, do go on to reach major milestones. And, at the *behavioural* level, many children and young adults live at home, interact socially and develop some language and communication skills. This is not to say that individuals with cri-du-chat do not experience difficulties in many aspects of their lives and these will be described in greater detail in the sections below. What this recent research has highlighted that has been absent in earlier studies is the potential of children and young adults with this syndrome to develop and maintain important life skills. It is hoped that the information presented in this second edition of the handbook will not only provide a more realistic and optimistic portrayal of the syndrome but that it will enable parents and professionals to deal more effectively with the implications of a diagnosis of cri-du-chat syndrome in their child.

The new information reported in the handbook was made possible by a PPP Foundation research project grant awarded to Professor Kim Cornish and Drs Penny Standen, Margaret Collins and Professor Helene McNulty. This research represents the world's largest study to date of children and adolescents with cri-du-chat syndrome with over 75 families representing all corners of the UK and Southern Ireland. The study was conducted from 2000 to 2002 and information was gathered by a team of talented researchers including Esme Ferguson, Lindsay Pearce, Megan Fisher, Natalie Stoles and Richard Cant.

I would like this opportunity to say thank you to all the families who participated in this important research and to the many professionals who gave their time and expertise.

Kim Cornish
May 2003

The 2nd edition of this Handbook is dedicated in memory of Dr Margaret Collins, a clinical psychologist who worked tirelessly to improve the quality of life for children and adults with cri-du-chat syndrome, who was also a friend to many families and a wonderful colleague to work with. Her vision and energy have transformed the lives of many individuals and her work will continue to inform and contribute to knowledge for years to come.

What is cri-du-chat syndrome and how is it diagnosed?

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 cri-du-chat syndrome. 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.

It is also important to note that a growing number of studies have described individuals with 5p deletions outside the critical region. Most often these individuals present with the cat-like cry from which the syndrome derives its name but not severe learning disability or significant developmental delay. These studies highlight the importance of careful and accurate differentiation between deletions that result in the typical CDCS profile and those that result in an atypical, milder profile.

At birth, the main clinical diagnostic feature of the syndrome is a high pitched, monochromatic 'cat-like' cry that is always present in the new-born but may disappear with age. Other features include a round, full face ("moon face"), wide-ly spread eyes (hypertelorism), an extra fold of skin at the inner corners of the eyes (epicanthal folds), a flattened and widened nasal bridge and ears that are positioned low on the head. Most children with CDCS will have feeding problems from birth including failure to thrive, poor sucking and slow weight gain. They may also be some medical complications but these will not affect every child and are not frequent.

"Having our daughter is like having a little person with all the seriousness taken out. Everything is fun for her"

Physical and Medical Features

Growth and Feeding Problems

At birth most CDCS babies are small. Studies have reported two-thirds below the 50th percentile for length after adjusting for gestational age and almost 90% below the 50th percentile for weight. More than 50% have a head circumference below the 10th percentile.

Through childhood and adolescence CDCS children tend to be shorter and lighter than the majority of children the same age in the general population. It is important to monitor growth but weight for height is a more useful comparison for this group than weight for age or height for age. A recent study of children and young people with CDCS in the British Isles found that most were small and lean but as most were at or above the 9th percentile for Body Mass Index (Body Mass Index is weight (kg.) divided by the square of height (m)) they were not disconcertingly thin for their height. At the present time, it is unclear how much of the short stature is due directly to genetics or indirectly due to suboptimal nutrition. In a recent series of studies, Dr Margaret Collins outlines some of the main findings regarding growth curves in children with CDCS. These articles can be assessed directly from the CDCS Support Group at the address at end of booklet

Feeding concerns in infancy

Several recent studies have reported that feeding difficulties in infancy are very common in CDCS children. In one study, 63% of parents reported that their child had had feeding difficulties in the neonatal period, associated with poor sucking (47%), reflux vomiting (42%) and failure to thrive (47%). Some parents have also reported that as a new-born their child had been unable to suck and breathe simultaneously. One approach that may help breathing during feeding is to clear the mouth and nose of mucus with a syringe before attempting to feed.

Poor muscle tone (hypertonia) can also cause a poor sucking response. The baby will have a weak suck and milk will leak from the mouth making it very difficult for the baby to take in enough milk to meet energy requirements. A poor sucking response in infancy is likely to improve as the child gains experience in sucking and also matures. Tube feeding may be necessary to ensure that the child does not fail to thrive while the sucking response is poor. Tips which have proved helpful with babies with a poor suck include making sure that the infant is wide awake before attempting feeding, extra support for the infant during feeding, and supporting the infant's chin during feeding to help steady the jaw. Parents have reported very varied responses by the medical profession to these feeding problems in infancy. It is important that parents ask for help if it is not offered. Although medical staff may have little or no experience with infants with CDCS they will have extensive experience in dealing with hypotonic and low birth weight babies with feeding

problems. Hospital and community paediatric teams may include a paediatric speech and language therapist who will advise about improving the suck (and later, chewing), an occupational therapist who can advise about utensils and equipment, a dietician who can advise on all aspects of nutrition and a clinical psychologist who can advise about behaviour. Parents have also commented that they have found Health Visitors supportive and helpful.

Feeding concerns in childhood

Children with CDCS need vitamins, minerals, protein and carbohydrate just like other children. In a recent UK study over 60% of children and adolescents with CDCS (2-18 years) always consumed the usual family diet while the remaining 40% consumed the usual family diet either frequently or occasionally. Unfortunately, only 21% were always able to cope with the normal consistency of the diet and 37% could only cope with pureed food. Difficulties in sucking, swallowing and chewing often lead to a prolonged use of pureed foods and delayed introduction to solids. However, as with other children with feeding problems, the initial cause of the problem can be followed by persistent, difficult to correct behaviour patterns. It is therefore crucial that parents and carers can be advised to be alert for the signs that their child is developmentally ready for the introduction of soft, mashed food, then finger feeding of large pieces of food which are soluble in the mouth to prevent choking, next finger feeding of smaller pieces of food and on then to more textured food from the family's meals.

Any parent or carer anxious about a feeding problem should seek professional advice. There may be a simple solution to the problem, but if not then an interdisciplinary assessment of the developmental, nutritional and behavioural aspects of the problem will aid the drawing up of a treatment programme.

Encouragingly, in our recent survey of parents we asked whether their child eats a normal family diet (71% responded yes) and whether the consistency of food was normal (56% yes), mashed (30% yes) and pureed/liquidized (3% yes). This finding is strongly age-related and suggests a positive developmental progression from a reliance on pureed and mashed foods to eating textured food. We have also found that even though many parents experience feeding problems with their infant or child leading to concerns about compromised nutritional status, the blood levels of a whole range of important nutrients appear to more than adequate. Importantly, we found no evidence of compromised vitamin status in children even when food was taken in a liquidized form. As long as the liquidized meals provided are nutrient-dense, there is no reason to suppose that micronutrient status will be compromised by this approach, and it may in fact be improved. With these findings, we hope that parents will be reassured to know all your efforts in trying to ensure the provision of adequate nutrition to your children are not in vain. You are to be congratulated on achieving what appears to be a very well balanced diet to your children.

Dribbling

Most CDCS children will have problems with excessive dribbling which, in many cases, requires frequent changes of clothing throughout the day. Dribbling is due to weak muscle control and can be helped by early speech therapy, which aids swallowing. Another successful way parents have found to alleviate dribbling altogether is by surgery, an operation called Submandibular duct Translocation. In most cases, your child will not need an over night stay. They will arrive at the hospital on the day of surgery and should be home by the evening. The procedure involves relocating the salivary ducts underneath the tongue to the back of the throat so that the saliva trickles down the throat and is swallowed. The operation is fairly painless but does involve a general anaesthetic. Most parents have reported a high success rate and stated how much it has added to their child's quality of life. In addition to this technique, other procedures are available and these should be discussed in consultation with your GP.

Constipation

Constipation, not diarrhoea, is a problem that appears to be present in over 70% of children in infancy and remains a problem for the majority of children in later childhood and adolescence. An increase of fibre in the diet by encouraging the child to take fruits, vegetables and whole grain cereals can be helpful, and many parents swear by the use of prunes and prune juice which contains a natural laxative. There are specific medical treatments for constipation but advice should be sought from your GP or the specialist most involved in the care of your child.

Other medical problems

Congenital scoliosis (curvature of the spine), gastrointestinal and cardiovascular problems can occur in some but not all children and adults with CDCS. Alongside these major health problems, there is a proneness to develop recurrent upper respiratory tract infections and an increased risk of dental problems. However, at the moment there is no research that can inform as to the trajectory of any medical problems and we do not know if there is a change in the severity of any problems with increasing age.

"If I had to describe my child, I would say 'A joy with a lovely nature'"

Early Developmental Milestones

Most children with CDCS will reach their developmental milestones later than typically developing children but with the help of early therapeutic interventions, most children will eventually reach major milestones including learning to sit up, crawl, walk, and eat independently. In the section below we describe some of the main difficulties you might encounter with your child as they begin to reach towards their milestones. What is important to remember is that every CDCS child is unique and not all children will perform at the same level at exactly the same time.

Ian aged 5

Debbie aged 4

Motor difficulties

The majority of CDCS children will experience some problems in motor control particularly in skills that require the use of motor co-ordination (e.g. walking, dressing and feeding self) and fine motor skill (e.g. holding a pencil). However, contrary to early advice that children with CDCS would never walk and have only limited mobility, there is now growing evidence to indicate that the majority of children will acquire some degree of mobility and dexterity. For example, many children are able to sit independently and to feed themselves to varying degrees. Walking is also achieved by many CDCS children, although almost all will experience co-ordination problems which make them appear overly clumsy.

Washing, dressing and undressing

Less well developed are skills that require fine motor control and planning such as those involved in washing, dressing and undressing. Many children with CDCS find it difficult to master these skills and often require considerable help from parents. However, these skills do appear to be sensitive to age and many young adults, although not able to perform them totally independently, need only minimal help. You will probably find your child is more than willing to help with washing and this should be encouraged from an early age, even if it is just letting them place their hands into a bowl of water and splashing. Washing hair and using soap are skills that may take much longer to develop. In terms of dressing, encourage your child to put items of clothing on him/herself, even if it's just very basic items, and again start doing it from an early age. The fine motor skills needed for zips and buttons, however, are going to be beyond most children's ability so don't expect too much! Also, allow extra time for dressing and undressing so your child doesn't feel rushed and you then don't feel tempted to simply do it yourself to save time! For these skills, Occupational Therapists can be very helpful. They can assess and provide exercises for your child in the areas of co-ordination, balance and gross and fine motor skills, and can advise on building up skills in dressing, washing, eating with a knife and fork etc. Where appropriate, your GP may be able to refer you to an Occupational Therapist.

Toilet Training

The majority of CDCS children will experience problems with wetting and soiling and few will have achieved bladder and bowel control by early childhood. You will find that some CDCS children are toilet trained by the age of four or five years while for many children it takes much longer. So don't feel discouraged if your child is still in nappies at seven or more years old, they are not going to be the only one. Toileting starts with the task of getting your child to associate the lavatory with urination and defecation. This is best achieved by sitting the child comfortably on the toilet for a short period (maximum of a minute or two) after every meal of the day. This is the time when food in the stomach naturally triggers a reflex in the lower bowel to contract and defecate. Should this happen then praise and rewards will help to reinforce the habit. However, praise and rewards should be given initially for simply sitting on the toilet. If your child has mobility problems the lavatory may have to be physically adapted to allow comfortable sitting. Even a footstool or box will help most children to use a normal lavatory satisfactorily.

If your child has constipation problems they may benefit from high fibre diet, brief courses of laxatives and, very occasionally, enemas. You might also find that your child is "dry" at school but not at home. If this is the case, then find out the routine or procedure your school uses and follow it at home. District nurses, specialist health visitors and occupational therapists provide invaluable support and advice

with these problems. More complex problems may require the help of specialist continence nurses, paediatricians, psychologists or psychiatrists.

"She is everything you could wish for in a child. Loving, affectionate and loves life. She has learning difficulties and has to learn everything from sitting up to talking. It takes a long time to learn skills but she does manage to master. She is a special and important member of our family"

Sleeping Difficulties

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 parents 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. Furthermore, although the sleep problems were commonly longstanding being usually present from early infancy, few parents reported actually seeking any help. Few professionals and particularly GPs and Health Visitors receive any training in this area despite the fact that children's (and adults') sleep problems are commonplace. This is a serious situation because chronically disturbed sleep for children can result in major daytime behavioural problems such as aggressive behaviour and overactivity and can significantly interfere with learning. For parents, 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. Indeed, in studies examining the burden of care faced by parents of handicapped children, chronic sleep problems always figure prominently and, most seriously, are the most important reason for parents not coping with their children and requesting help from Social Services (respite care, foster care and even adoption!).

Well, what can be done about this? Of course, most babies, with or without CDCS, do indeed cry regularly at night but it may be more beneficial to allow some crying, in the hope that this will be a natural prelude to sleep, before rushing to comfort them. Parents could help a young infant to get into a good sleep pattern by quickly establishing a regular settling routine in which the baby or infant is put to sleep on its back in a quiet darkened room (or part of a room) which is safe, warm and comfortable. By doing this, infants should quickly settle and learn to fall asleep without any fuss by 3 to 6 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".

S.L.E.E.P.

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!

No negative effects have been reported following the use of this somewhat dramatic technique and this approach can work very quickly (a few nights). Furthermore, it commonly results in reduced levels of stress for parents and conspicuous improvements in children's daytime behaviour and capacity to learn.

However, if you are not prepared to ignore your child at night, there is an alternative. You can try to remove your presence from him or her in *gradual stages:* this involves initially lying on the bed until they fall asleep for a few nights, then by the bed, then near the bedroom door etc. This can be highly effective but usually takes much longer than the technique described above. Ask a trusted friend or Health Visitor to help you with this approach.

A small minority of children will not respond to the techniques described previously for a variety of reasons. In these circumstances it is best to stop, reflect upon why the approach did not work (for example, a parent who eventually "gives in" to their child's insistent nocturnal demands!) and then to try again when you feel it would be the best time to do so. Some children, and particularly those with visual impairments or other problems like autism, may benefit from taking capsules (2 to 12 milligrams) of a sleep-inducing agent called MELATONIN half an hour before their usual bedtime. This is a natural hormone that promotes sleep in humans and most other animals. It is effective in about two thirds of patients and is to be preferred to traditional "sleeping pills" such as "Vallergan" and "Phenergan", that can cause many unwanted problems through side-effects. Melatonin should only be prescribed by doctors who are experienced in its use, such as community paediatricians and child psychiatrists, and only for a fixed period. Melatonin levels in the body can also be boosted naturally by exposure of the child to daylight in the mornings and also by them eating foods rich in melatonin, or the substance that the body makes it from, shortly before bedtime. These foods include oats, sweet corn, bananas and milk. A banana "smoothie" or a bowl of porridge or corn flakes may just do the trick!

A Child Psychiatrist or Psychologist may also be able to help you with your child's sleep problems: your GP or Health Visitor can arrange this.

"We tried this method on Carol and within a week she was sleepimg much better and her behaviour has improved too. It is much easier to deal with your child when you have had a good nights rest. I wish someone had told me about this method years ago"

Hearing and Vision

Visual problems are not frequent in CDCS children but when they do occur they can include near sightedness (myopia), dancing eyes (nystagmus) and optic nerve abnormalities. Hearing problems are also rare but there is a high incidence (between 70-80%) of a condition known as 'hyperacusis' (hypersensitivity to noise) in children with CDCS. This can manifest itself in a number of ways but the most frequent is for severe distress or agitation at a wide range of sounds, including sudden noises made from aeroplanes, lawn mowers, a balloon bursting, thunder, loud clapping etc. It is difficult to know whether these noises may be physically painful to the ears, or whether they may simply startle the child. As yet, we don't know if hyperacusis gets better with age or continues into adulthood. It may very well depend upon the individual child. What we do know, however, is that this condition can be extremely distressing for both the child, and those around him or her (parents, siblings, teacher).

To help reduce the distress, you can *comfort* and try to explain in a way your child can understand about the source of the sound. It might also help to allow your child some *control* over certain loud noises. For example, encourage them to turn on the vacuum cleaner or shut the car door. Even learning to pop balloons themselves or clap other people's hands may also help reduce the distress. Gentle but repeated exposure to some of the noises that most frequently distress your child may encourage *familiarity* and thus reduce its impact (i.e. make tape recordings of sounds that you and your child, in play, could switch on and off. You could also encourage your child to gradually turn up the volume on the recording so that the noise level gradually increases).

Language and Communication

CDCS children are particularly slow to develop language abilities and speech delay is one of the main characteristics of the syndrome. However, previous and current findings confirm that CDCS children do have far better developed comprehension skills compared to their expressive skills (speech). What is especially encouraging about our findings is that a lack of speech does not seem to impede communication. Using data from children in the UK and in the US over two-thirds of children were able to communicate their needs using non-verbal methods (sign language, symbol

cards). The consistency of these findings should encourage parents, teachers and professionals to facilitate effective communication with CDCS children. You should be optimistic about your child's capacity to understand more complex verbal commands than their speech would suggest. It should be possible for your child to see a Speech Therapist who could develop programmes that focus upon extending even further their relatively good comprehension and vocabulary skills. Early stimulation and introduction to sign language (i.e. Makaton) might also prove to be a very effective tool in developing communication skills in your child.

Makaton is a very simple and effective sign language technique and was devised for children and adults with a variety of communication and learning disabilities. Makaton is used extensively throughout the UK and has been adapted for use in over 40 other countries. It incorporates both basic sign and speech and can be used by parents as well as children. As parents you will need to go on a course and to register you will need to contact either your Health Visitor or Speech and Language Therapist. Makaton have now also produced a range of resources for parents and teachers and they can be contacted on their website at: www.makaton.org/

"She seems to understand everything and uses less gestures to make us understand what she wants. She is hard work but all of it is worth it, just for the hugs!"

Common Behavioural Difficulties

Several surveys have discovered an excess of behavioural problems in CDCS children. These include hyperactivity, aggressive and oppositional behaviour as well as sleep problems (see above/below). Why this should be is a complex question; however, the degree of learning disability, communication difficulties and aspects of temperament such as high levels of irritability are important factors. 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. For example, a regular tantrum at the checkout of the supermarket may result in the child being given a sweet "to shut him up", thus paradoxically "rewarding" the behaviour.

Tantrums can be alarming especially in larger and stronger children. They are upsetting for everyone involved. Here are a few tips to handle them:

- nip them in the bud by distracting the child if they are "brewing up" by an activity that they usually enjoy

- avoid situations where they are more likely to occur (e.g. supermarket checkouts)
- don't respond: full-blown tantrums thrive with an audience
- don't attempt to "talk them down": this rarely works and becomes stressful when parents "lose their cool" and make a bad situation worse
- walk away or, without any fuss, put the child into a safe unstimulating place until they calm down
- expect an apology if the child can understand cause and effect

It is also important that children know what is expected of them in terms of acceptable behaviour: they will not magically "discover" this. Communicate clearly, concisely, and with conviction using words or symbols that the child readily understands. Parents must agree to back one another up in matters of discipline: inconsistency can paradoxically increase bad behaviour. Parents should remember that their own behaviour is the most important example for their children: well-behaved adults encourage the same in children. Finally, rather than focussing on bad behaviour, promote 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. Some CDCS children have developmental problems such as ADHD that may need to be treated before behavioural measures can work effectively. Your GP will be able to refer your child to specialist paediatric or mental health services for this sort of help.

Some common behavioural problems are cited below:

(i) *Concentration difficulties and hyperactivity.*

About 90% of CDCS children will experience some problems with poor concentration, impulsiveness and overactivity. It might be that some CDCS children will be diagnosed with Attention Deficit Hyperactivity Disorder (ADHD). The extent to which these behaviours develop into adulthood is not yet known, although overactivity may become less of a problem in late adolescence and adulthood but they may still remain very distractible and have poor attention spans. Before we consider some straightforward means which may promote attentional skills in children it is important to highlight some other reasons why children may be overactive and demonstrating poor attention: Firstly, the importance of children to get a regularly refreshing night's sleep needs to be emphasised. All children who are sleepy are likely to be both inattentive and overactive. By promoting healthy sleep habits in your child in the manner previously described this may well have a beneficial effect. Secondly, children may show attention difficulties if they are not

provided with the necessary limits and boundaries for their behaviour by their parents. A firm but fair parenting approach usually reaps rewards in these circumstances. Lastly, children who are generally either under or over stimulated through either neglect or overindulgence may show these features.

A prerequisite for promoting optimal attention is effective communication with your child. Both verbal and non-verbal (body language and expression) methods need to be employed in a manner which your child understands and demonstrably responds to. Generally speaking, in order for a child with a short attention span to understand what is expected of them communication has to be clear and concise. Strong eye contact is also important. Once one is sure that they are receiving the message loud and clear the tasks required should be quite simple and well within your child's general ability to perform. As soon as it is performed satisfactorily, praise should be given. Simple tasks requiring brief attention and concentration are very slowly over weeks and months replaced by progressively more complex and time consuming tasks. Certain constructional toys and educational video games may also promote attentional skills. One word of warning, expecting your child with a mental age of two years to learn to pay attention for an hour is unrealistic. The normal (non-novelty) attention span increases developmentally by a minute or two for every year of mental age. This means that a child with a mental age of two is unlikely to be able to be trained to pay attention for longer than three or four minutes. While your child is being trained it is important that extraneous sources of noise and other distractions (especially televisions and music centres) are avoided.

In addition to the therapeutic techniques described above, there is now increasing evidence to suggest that some forms of medication may also prove beneficial in reducing attention problems in some children.

Medication.

For children who are pervasively and chronically overactive and demonstrate very poor attention and impulse control which handicaps them in addition to their other problems the diagnosis of clinical hyperactivity or attention deficit hyperactivity disorder (ADHD) is made. When this condition presents to a severe degree, which often appears to be the case in CDCS, interventions based around behaviour modification, parenting skills and special educational provision may not help these core symptoms which cause tremendous problems for affected children's parents and teachers. In such circumstances medication may prove beneficial. In the past, tranquillisers would have been used but we now know that these produced no immediate or lasting gain. Indeed, sedatives have been shown to make some children much worse! It may seem strange but a class of drug called the stimulants have proved beneficial in this condition. Most of the clinical evidence for these

drugs' efficacy has been obtained from work with non-learning disabled ADHD children. However, children with learning disabilities are now benefiting from this treatment. Child psychiatrists and paediatricians are the specialists who diagnose this condition and prescribe the medication.

(ii) *Self-injurious and compulsive behaviour:*

In recent years we have attempted to understand the nature of the self-injurious behaviour in CDCS children. From parental reports, we know that this behaviour causes demonstrable damage to the body and is particularly distressing for parents to witness. In our recent survey of the UK families, we found the main problems to be related to self-biting, hitting head with hand, and hitting head against objects. Indeed, 92% of parents surveyed reported some incidence of self-injurious behaviour with a third reporting that it occurred on a regular basis either weekly or daily. In a more detailed study we observed CDCS children's behaviour during a typical school day and looked at both the "intervals/frequency" of a specific behaviour and the "duration" of an episode of the behaviour. We also interviewed their teachers to ascertain the range of self-injurious behaviours displayed within the classroom. We found that *hand biting* and *skin picking* were the two most common behaviours and were performed more frequently in children with CDCS compared to a comparison group of children who did not have CDCS.

"Kate is predictably unpredictable. You know she is going to do something but not quite when."

Why do children with CDCS display these specific self-injurious behaviours?

Self injurious behaviour occurs in approximately 4 to 10 per cent of people who have intellectual disability. The most common forms of SIB are picking and scratching, biting and head banging and hitting. There are some individual characteristics that are related to self-injurious behaviour such as degree of intellectual disability (self-injury is approximately four times more common in those who have severe or profound intellectual disability) some genetic syndromes, such as Lesch-Nyhan and Prader-Willi syndromes, autism and poor expressive communication. Generally, the prevalence of self injurious behaviour rises with age up until the mid-twenties.

There are three main theories that have been proposed to account for self injurious behaviour. The first focuses on neurotransmitters. These are chemicals in the nervous system that form links between the ends of different nerves. There are different types of chemicals and each has a different function. It has been

suggested that a disorder of any of three neurotransmitters might be involved in self injurious behaviour. These are dopamine, serotonin and the endorphins. However, the evidence to support this theory is not very strong and at present there are no specific medications that have been shown to be effective. The second theory focuses on self injurious behaviour in response to pain or discomfort. Whilst there is little research literature to support this theory this is most likely due to the difficulty in establishing this association. This is because pain and discomfort can be rather transient and for people who are non-verbal it is hard to confirm the presence of pain and discomfort. The third theory has the strongest evidence and there is a research literature spanning over 40 years demonstrating various aspects of the theory and interventions that can be successful. This theory views self injurious behaviour as a learned behaviour that can occur because of the rewards that follow an act of self-injury. These rewards may be invisible to an observer as they comprise stimulation that follows the self injurious act. The best example of this is eye pressing which results in bright stimulatory flashes in the eye. Other rewards are more observable and are socially mediated. The most commonly cited are attention from carers and escape from difficult task demands. The evidence in the research literature suggests that for approximately 70 per cent of people with intellectual disability who show self injurious behaviour this theory is the most applicable.

When beginning the process of intervention for self injurious behaviour the importance of thorough assessment cannot be emphasised enough. The causes of self injurious behaviour will differ between individuals and there is no single intervention that can be applied to everyone. Generally the strategy that can be adopted is to first rule out any medical causes that can be giving rise to pain and discomfort. There is little to guide carers on how to approach this apart from their knowledge of the person they care for and how that person responds when in discomfort. It is important to be aware of common conditions that can give rise to local pain and discomfort such as tooth infection, middle ear infection and to note whether the self injurious behaviour is targeted at those parts of the body where pain and discomfort might be localised. In addition, when self injurious behaviour is associated to pain and discomfort it will be unlikely that the behaviour will vary with environmental events. It is critical that a careful examination of the association between self injurious behaviour and environmental events is conducted in order to see whether this is the case. Clinical experience suggests that addressing pain and discomfort as a potential cause of self injurious behaviour should be pursued vigorously in the first instance before psychological interventions are considered.

There is considerable evidence that self injurious behaviour can occur because it is rewarded either by social contact or other more tangible rewards, such as food, drinks and activities, or it is rewarded by escape from unpleasant tasks or other difficult situations. In order to evaluate whether this is the case it is essential that an assessment of the behaviour and the response of others to the behaviour is

conducted. Once it is established that rewards might be influential then interventions based on behavioural principles can be constructed that comprise behaviour management strategies in response to the behaviour and increasing behaviours that can replace the function of self injurious behaviour. This often means increasing the functional communication of individuals who shows self-injurious behaviour and there is a strong emerging literature that this is an effective strategy. Finally, when self injurious behaviour is rewarded by the sensory stimulation that follows the self-injury the general strategy is to try to increase alternative forms of stimulation and minimise the stimulation that results from the behaviour.

This brief summary of the potential causes and interventions for self-injury gives an idea of the sorts of strategies that can be adopted in response to his behaviour. It must be emphasised that assessment is the key to successful intervention and that at present there is no evidence that these sorts of intervention that are effective for all people with intellectual disabilities will not be effective for individuals who have CDCS. Help in putting together effective assessments and interventions can come from clinical psychologists and professionals who had been trained in applied behaviour analysis and it is important to seek professional advice on the assessment and treatment of self injurious behaviour.

Finally, there are three important things to remember when confronting the problem of self injurious behaviour. First, self injurious behaviour very rarely occurs at a stable rate all the time and in every situation. Because this is the case there is a cause. This is why assessment is critical. Second, whatever intervention strategy is adopted it is important that a careful record of the rate or frequency of self injurious behaviour is kept in order to evaluate whether the intervention is working. Sometimes, we do not know what is causing self injurious behaviour and the theories described above may not appear applicable. However, that does not mean that a trial-and-error process cannot be adopted and it is the records that will indicate when the intervention is working. Third, it is important to persist with any intervention over a reasonable period of time to see if the intervention is working. Self-injurious behaviour can take some time to decrease and it is important that any intervention is given a good trial.

(iii) *Stereotyped behaviour:*

Sterotypies are repeated body movements or postures that are not part of a goal direction act, such as hand flapping, finger twirling and rocking. These can typically occur when a child is agitated or upset and even when they are bored or when totally engrossed in a particular activity. The intensity of stereotyped behaviour does appear to decrease with age although it does not completely disappear. Instead, it may take on a more subtle appearance and only become very intense during peri-

ods of stress and excitement. You may find that your child responds to gentle encouragement to stop their hand flapping or rocking when you ask them to. Providing some token of a reward might also be effective and may well speed up the desired effect!

With all these behaviours a Child Psychiatrist or Community Paediatrician might be able to provide some useful advice. In severe cases medication can sometimes be beneficial but this always has to be prescribed by and under the supervision of a specialist.

Respite Care

For most families respite care is a very welcome and much needed service. It involves a child or adult staying somewhere outside the family home to give the family a break and enable them to recharge their batteries. The type of respite care varies according to the area you are living in. Typically the child or adult stays with either another family (often foster families) or in small group type homes. The length of stay varies according to the need of the family and can be from a couple of hours to a week or two. Respite is organised by the Social Services Department and is closely monitored to make sure that the child or adult is happy. Respite care can start from any age.

"Respite really helps the whole family. We get a chance to let the other children have our undivided attention for a change and know that our son is having a good time too staying with his friends."

"Everything she achieves makes me very proud because I know how hard some activities are to master."

Socialisation and Daily Living Skills

With few exceptions, CDCS children are very friendly and outgoing. They will try and make friends with all ages, including their peers and often demonstrate wonderful sensitivity and concern for others. Because of their nature, CDCS children can integrate very successfully into many social activities such as dance classes, gym clubs, Brownies, Cubs, Boys and Girls Brigade etc. Your child's school may also be able to inform you about local social clubs and sports clubs which have a special needs integration scheme. Within your area there may also be afterschool clubs and play schemes which welcome participation from children with learning disabilities. It is vital for their social development that they are allowed to integrate and enjoy the company of 'typically developing children'. Don't feel that you should isolate them from the outside world.

While socialisation skills are highly developed in CDCS children, *community* skills are under developed. These skills include safety awareness and understanding when something or someone may be dangerous. The impulsive nature of the CDCS child makes it very difficult for them to hold back when they encounter potential danger. For example, when crossing a road they rarely wait for traffic to stop, appearing unable to appreciate the consequences of stepping out into a busy road full of traffic. There is also minimal understanding of the concept of "hot" and many children have received burns by placing their hands in boiling water, or touching hot irons or fires. Because of this lack of awareness, it is important that CDCS children are supervised when out walking along a street, on public transport, helping with cooking, washing up etc. in the home.

Debbie aged 13

Simon aged 11

"We found that taking Gary horse riding, swimming and trampolining has helped his physical side and also helped his confidence."

"I believe that helping your child to integrate with all sections of society not only benefits your child, but those who are then privileged to know him or her."

School-Related Difficulties

In terms of schooling, each child's needs will be different. Some children may begin school life in a school for moderate learning disabilities (MLD) and may move later on into a school for severe learning difficulties (SLD) because they are not coping with the greater demands and pressures of an MLD school. Equally, other children may settle very well at one school (MLD or SLD) appropriate to their needs and level of abilities. It is very rare for a CDCS child to be in mainstream schooling although many special needs schools now have a policy of greater integration. You need to remember that there are many advantages to placing your child within a special needs school. There is, for example, a higher staff: child ratio which will allow for more individualised planning appropriate to your child's level of ability. There will also be a wider variety of communication systems available (i.e. Makaton (see above), symbols/photo timetables) that can greatly assist learning.

Indeed, it is vital that teachers allow communication skills to develop in CDCS children. Primarily because it allows the child to be 'included' in the day to day routine of school life, and also because communication, by whatever means, reduces the level of frustration and aggression that can accompany limited speech production. It is also important that CDCS children are integrated in classrooms with children their own age. Because the CDCS child often look much younger than their actual age you need to ensure that they are learning within an environment that has similar aged children performing age appropriate tasks.

The school should also have access to the services of speech therapists, occupational therapists, and educational psychologists.

On Leaving School: What Next?

The question of what does a CDCS child do when they leave the educational system at 19 years or younger is a major source of worry for most parents. On leaving school, all children will be given a Disabled Persons Assessment (DPA) which is a statement of their needs and requirements which will go with them. You should be encouraged to look at the different options available to you (the range of options will, of course, depend upon the local authority in which you live) well before your child leaves school. You may also find it useful to contact Social Services or a special needs careers advisor on what is available. It is important that you feel comfortable about the final decision and you may want to visit a number of different centres (day or residential) before choosing the most appropriate for your child. Looking first at day centres, these can offer a range of specialist training to help develop communication skills, work skills, and self-help skills. Many centres will also have programmes that involve developing community skills such as shopping, dealing with money, preparing for paid employment and may also offer college courses specifically designed for children with learning disability. Some centres even offer specialist therapies such as music therapy which may work well with a CDCS young adult.

A residential centre may also be an option. It has many of the advantages that a day centre can offer and its grounds may also be extensive enough to provide an on-site workshop or farm. Parents can decide how often their child will visit home although many CDCS young adults will find it difficult emotionally to return back to the centre after each home visit and you may need to devise a system whereby your child knows how long it is before they visit home again.

As within the school environment, it is essential that the young CDCS adult mixes well with his or her own age group and continues to develop age-appropriate skills. You may also need to push to ensure that adequate one to one support is provided and that Makaton is available and used within the centre.

Debbie aged 26

Simon aged 32

The impact upon the Family

Having a child with CDCS is a chronic stressor for most families and one that requires continuous adaptation of the parents, grandparents and siblings over time. Not only do families have to cope and adjust emotionally but they also have to cope with providing more intensive care for their infant and child. For example, meeting additional physical needs such as problems in feeding and weight gain in the early years, alongside knowledge that their child will require special needs education throughout their school lives can create ever increasing levels of stress and feelings of helplessness. Our research highlighted that parents of CDCS children displayed stress levels that are in the top 70% when compared to the stress levels of a normal population of parents but were at a similar level to parents who also had a child with a learning disability but without CDCS. This suggests that increased stress levels are likely to reflect the increased demands of coping with the needs of a child with a severe learning disability rather than specific to being a parent of a CDCS child.

A very worrying concern is that nearly a quarter of parents surveyed, 24% presented with clinical symptoms of depression and 43% experienced clinical symptoms of anxiety. Parents reported that strained relationships with their child and difficulty in managing problem behaviours were a main factor in increasing stress and anxiety levels. When we looked at mothers and fathers separately, mothers experienced greater levels of parental distress, anxiety and depression than fathers. This was particularly pronounced for anxiety with 60% of mothers compared to 20% of fathers experiencing some clinical symptoms, this increasing as the child aged and as problem behaviours (e.g. hyperactivity, self-injury, stereotypy) increased in intensity. What is especially concerning is that few parents reported seeking external help in learning appropriate coping strategies and even fewer reported using medication or alternative therapies to alleviate symptoms of anxiety and depression. However, parents need to recognise that they are not on their own in having to cope with a child with a severe learning disability. Family doctors should be able to put parents in touch with a counsellor and a local self help group. Local branches of Mencap are also a good source of information and will know of any parent groups or activities which you may find helpful.

We also recommend that families (including parents, grandparents and siblings) attend the annual conference of the CDCS Parents Support Group. It provides a once a year opportunity to meet up with other families caring for a Cri du chat child in a fairly informal setting allowing for an exchange of experiences and information. Professional speakers also provide an insight into the Syndrome and development areas. **Contact either ray@criduchat.co.uk (tel-01268 411666) or angie@criduchat.co.uk (tel-01455 841680)**

What about siblings? If parents experience stress through having a child with CDCS then it is highly likely that other children in the family will experience some

difficult emotions too. Sources of difficulty for siblings are not only the effects of their parents' stress but also their feelings that their parents give more attention to their disabled brother or sister, increased responsibilities within the family and fewer social or recreational opportunities. It is therefore important for parents not to overlook their other children's needs even though they have their hands full with their child with CDCS and to appreciate the help given by siblings.

There can be positive effects of having a disabled brother or sister. Several studies have found that siblings often show increased compassion, tolerance, sensitivity, maturity and responsibility. However, siblings in our study did express concern about their own future and parents should be aware that this is a topic that both they and their children need to explore. Siblings too may benefit from some of the self help contacts and information provided for parents and they should be given this option to explore them.

"Our child has taught us tolerance, has given us the ability to not judge quickly and to be aware of others. I wouldn't have been any of those things if it were not for her"

"Life is never dull in our house"

USEFUL DEFINITIONS

Clinical Geneticist: This could well be the person who diagnoses your child. Using very sophisticated techniques, Geneticists look at the blood and are able in most cases are able give you detailed information about the problem. If a specific problem such as CDCS has been diagnosed, the geneticist will usually do studies in the parents to see if the syndrome is likely to occur again. This is called Genetic Counselling.

Genetic Counsellor: This is a specialist whose role is to estimate the probability of recurrence of a genetic disorder, and to assist in deciding what the appropriate action would be. However, the final decision will always be left to the family

Genetic Nurse: This is a nurse with specialist knowledge in genetics and is typically based within a hospital Genetic department. One of their main roles is to talk and discuss with families about the syndrome.

Health Visitor: There are two types of health visitor: (1) The Health Visitor (HV) who is involved in all children who are born. They have the responsibility of checking the child's progress from birth. This includes immunisation and pre-school checks as well as advising on feeding and problems associated with the new-born. (2) The Specialist health Advisor (SHA) who tends to look after the welfare of children with special needs only. Her remit is to monitor the child's progress throughout childhood which includes attending meetings and reviews about the child. They will also look after the welfare of the parents, make sure that they are getting the right benefits, introduce them to a special playgroup, provide free nappies when they are eligible etc. The SHV tends to be more involved than the usual HV.

Occupational Therapist: This is a specialist therapist who works with people of any age to promote their independence. In hospitals they are an important part of the rehabilitation team. In the community, they work on trying to resolve practical problems. They can also provide essential equipment such as Buggies, special high chairs, potty chairs and helmets for children who fit or fall easily.

Paediatrician: This is a medically qualified doctor who specialises in babies and children. If your child has any ongoing problems they are usually kept an eye on by the paediatrician. At school age the child will have yearly checks with the Community Paediatrician.

Psychiatrist (child): This is a medically qualified doctor who specialise in diagnosis and treatment of various behavioural and psychological problems in children.

Psychologist (education): This is a specialist psychologist who assesses the educational needs and development of children with special needs.

School Nurse: This nurse is based in wither a particular school or covers a number of schools. The nurse deals with any medication or specific needs that are required. She will also assist in school examinations.

Speech and Language Therapist: This specialist therapist has a varied role. They will assist in helping the child/adult to be able to communicate. This can take various forms i.e. speech, signing, symbols and electronic machines. They can also offer advice on feeding techniques.

USEFUL ADDRESSES

Some of these contacts will have offices in other areas of the UK and Ireland.

ACE (Aids to Communications in Education)
ACE Centre, 92 Windmill Road, Headington,
Oxford OX3 7DR

Tel: 01865 759800
Fax: 01865 759810

Action for Sick Children
Argyle House
29-31 Euston Road
London NW1 2SD

Tel: 0207 833 1041

Assistive Communication Aids Centre
(other UK and Ireland centres from
http://www.laryngectomees.inuk.com/addresses.htm)
Speech and Language Department
Frenchay Hospital
Frenchey Park Road
Bristol
Avon BS16 1LE

Tel: 0117 970 1212 ext. 2141/2151

Carers National Association
20-25 Glasshouse Yard
London EClA 4JT

Tel: 020 7490 8818
Fax: 020 7490 8824

Contact A Family
209 – 211 City Road
London EC1V 1JN

Tel: 0808 808 3555 (10.00am – 4.00pm Monday to Friday)

ENABLE
6th Floor
7 Buchanan Street
Glasgow G1 3HL

Tel: 0141 226 4541
E-mail enable@enable.org.uk

Family Fund
PO BOX 50
York YO1 9ZX

Tel: 0845 130 45 42 or 01904 658085 (Textphone)
Fax: 01904 652625
e-mail: info@familyfund.org.uk URL: http://www.familyfund.org.uk

Makaton Vocabulary Development Project
31Firwood Drive
Camberley
Surrey GU15 3QD

Tel: 01276 61390
Fax: 01276 681368

MENCAP
123 Golders Lane
London EC1Y ORT

Tel: 020 7454 0454
Fax: 020 7696 5540
Email: information@mencap.org.uk

Motability
Goodman House
Station Approach
Harlow
Essex CM20 2ET

Tel: 01279 635666
Fax: 01279 632000

The SINGALONG Group
Communication and Language Centre
Magpie Hall Road
Chatham
Kent ME4 5NG

Tel: 01634 819915

Useful Books

A Practical Guide for Disabled People
(where to find information, services and equipment)
For a free copy write to:
Department of Health
PO BOX 410
Wetherby LS23 7LN

Code of Practice (on Identification and Assessment of Special Needs)
To obtain a free copy ring 0181 533 2000

THE UK CRI-DU-CHAT SUPPORT GROUP

The Cri-du-Chat Syndrome Support group was first formed in the late 80's by Anne and Stewart Wilson after a request from Ann Worthington of In Touch. They took over the original list of contacts and existed as a phone/letter contact group until 1991 when Gill Watts organised the first Get Together at the Cabury Club near Bristol. From that first meeting, which for many parents was the first time they had seen other children with CDCS, a national steering group was formed and the group has grown from strength to strength. There has been an annual meeting each year since and the membership has continued to grow at a fast rate.

The aims of the support group are to provide support and friendship to families and carers through a network of area families. It also seeks to raise awareness of the syndrome amongst the medical profession, parents, carers and the public.

The group organises a family weekend together each year which provides an ideal opportunity to meet other families and to talk to professionals. Research into the syndrome is also actively encouraged and supported.

The support group also produces two newsletters a year and is in contact with families world-wide through its Web site address: WWW.CRIDCHAT.U-NET.COM. To join the Group or receive more information about us, you can contact the National Co-ordinator, Angie Stokes, at the following address:

7 Penny Lane, Barwell, Leicestershire LE9 8HJ. Tel/Fax: 01455 841680

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CONTRIBUTING AUTHORS

Professor Kim Cornish, PhD, is a developmental neuropsychologist who has worked in collaboration with the UK Cri-du-Chat Syndrome Support Group for the past decade. Until August 2002, Kim was based in the Department of Developmental Psychiatry at the University of Nottingham. More recently, she has moved to Canada to take up the appointment as Canada Research Chair in Neuropsychology and Education at McGill University in Montreal.

Professor Chris Oliver, PhD, is a clinical psychologist with a world reputation in the field of learning disability research. His contribution lies in unravelling reasons why children with severe learning disabilities develop self-injurious behaviour. He and his team have developed innovative technology that can help observe early signs of self-injury in different environments (e.g. home, school). Chris is based in the Department of Clinical Psychology at the University of Birmingham.

Dr David Bramble, MD, is a child psychiatrist who has worked with Professor Cornish on research related to children with CDCS. His special interest is in children with learning disabilities and he works as a consultant for the Telford and Wrekin Primary Care Trust, Harlescott, Shrewsbury.

Dr Penny Standen, PhD, is a Reader in Health Psychology and her main research focus is on addressing issues that can improve the quality of life of individuals with severe learning disabilities. Penny is based in the Section of Learning Disabilities at the University of Nottingham.

Dr Margaret Collins, DClinPsych, was a consultant clinical psychologist with an international reputation in the field of learning disabilities. Her main research focus was on assessing the nutritional and dietary needs of children with CDCS and other developmental disorders. Margaret was based in the Department of Clinical Psychology, Muckamore Abbey Hospital, Antrim, Northern Ireland.