



Cri du Chat
Support Group
of Australia Inc.

Cri du Chat Syndrome

Information for Professionals



Overview – Cri du Chat Syndrome

Cri du Chat Syndrome (CDCS) is a rare genetic condition that incorporates distinctive physical features, intellectual disability and behavioural challenges. It was first described by French paediatrician and geneticist Dr Jerome Lejeune in 1963 and is caused by a deletion on the short arm of chromosome 5. The Syndrome exists on a spectrum, resulting in a wide degree of presentation variation in individuals with CDCS. As a result of this, some individuals may have been misdiagnosed, diagnosed later in life or may remain undiagnosed. This information pack aims to provide up to date, accurate information about CDCS to health professionals, with the recognition that some aspects may be revised in the future as more information becomes available.

Main features:

- High pitched soft cry.
- Microcephaly.
- Micrognathia.
- Round face.
- Epicanthal folds.
- Low set ears.
- Low birth weight.
- Delayed growth.
- Intellectual disability and developmental delay – moderate to severe.

Diagnosis

- CDCS can be detected prenatally (on ultrasound or prenatal testing), at birth or in mild cases, later in life.
- Clinically, the diagnosis is based on physical symptoms and a high pitched cry.
- When CDCS is suspected genetic testing such as microarray and FISH can confirm the initial diagnosis. A referral to a genetics service should be made to ensure appropriate and up to date information about the required genetic testing is provided.
- As with many genetic conditions, receiving a diagnosis can be a distressing time for parents. It is important that health professionals recognise this and provide support and understanding during this time. Parents need to be made aware of the challenges their children may face but it is also important to mention the strengths of children with CDCS.

Genetics

- CDCS results from a deletion of some, or all, of the short arm of one of the copies of chromosome 5 (5p). The wide spectrum of features is due to the deletion occurring at different breakpoints on the chromosome, and differing amounts of missing chromatin.
- Deletion of region 5p15.31 is associated with a high pitched cry.
- Deletion of region 5p15.33–5p15.32 is associated with speech delay.
- Mosaicism is possible (condition severity depends on how many cells are affected as well as what type of cells are affected).
- The size of the deletion, and whether it is inherited from the mother or father, may be linked to the severity of disability.

Inheritance pattern

- CDCS is an autosomal dominant genetic Syndrome. This means that if a deletion is present on one of the copies of 5p, the individual will have CDCS. Deletion in both copies of 5p is considered embryonically lethal.
- Genetic analysis of the child with CDCS and their parents will determine if the genetic mutation occurred spontaneously or was inherited.
- 85–90% of people who have CDCS have a spontaneously occurring mutation (sporadic or de novo).
- 10–15% of cases are inherited, with most being due to parental translocations.
- Children with break points in the middle of the CTNND2 gene resulting in a partial deletion and duplication of the gene may have less severe disabilities. This is an example of atypical CDCS.



Prevalence

- 1:15,000 to 1:50,000 live born infants.
- Females are affected more than males but the exact ratio is unknown.
- CDCS prevalence is not associated with ethnicity or environmental factors.

Preconception testing

- When CDCS occurs through a spontaneous mutation, carrier testing of parents will not be informative.
- Genetic analysis can determine the carrier status of those with a family history of CDCS. It is recommended that those seeking preconception testing obtain genetic counselling to discuss their options.

Prenatal testing

- Screening CDCS during pregnancy can be done by non-invasive prenatal testing (NIPT). This test gives you a low or increased risk result however is not a diagnostic test.
- Chorionic villus sampling (CVS) or amniocentesis is recommended to confirm a prenatal diagnosis of CDCS prenatally.
- Generally it is not possible to predict the type or severity of clinical features – however with expanded testing there is limited ability to infer related phenotypes.
- Parents have two options when a result of CDCS is obtained during pregnancy; to continue or terminate the pregnancy. A referral to a prenatal genetic counselling service is recommended to enable parents to have an appropriate discussion about their options.
- If parents want to speak with families with children with CDCS, they can be referred to the CDCS support group.

Strengths

It is well recognised that obtaining a diagnosis can be a difficult time for parents and individuals. During the early stages of diagnosis, it is important to recognise the strengths of individuals with CDCS, and the variability in features depending on deletion size, location, and type.

Children with CDCS have better receptive language skills (understanding speech) than expressive language skills (using speech). This means they are better able to understand the world around them, but may not be able to communicate this understanding to others. Some children use nonverbal communication, basic sign language or an augmented device to express themselves.

Most children with CDCS possess a happy and loving disposition, demonstrate an interest in others and enjoy social interaction.

Clinical features

All clinical features may not be present in all patients and may vary in severity:

Developmental delay

- Mild to profound intellectual disability.
- Language/speech delay – some are unable to develop spoken language.
- Communication issues, particularly expressive language.
- Sensory processing difficulties.
- Motor activity delay.
- Motor co-ordination difficulties (clumsiness).
- Milestone delay.

Behavioural or emotional problems

- Features of attention deficit hyperactivity disorder (70%). These features are predominantly in relation to impulsivity and response inhibition.
- Hypersensitivity to sounds.
- Self-injuring behaviour; hitting head against objects, hitting the head against body parts and self-biting (30–50%).
- Repetitive behaviours.
- Echolalia and compulsive attachments to objects.
- Obsessive-compulsive disorder.
- Aggressiveness.
- Stubbornness.
- Sleeping difficulties (30–50%).
- Snoring/sleep apnoea.
- Poor oral hygiene.
- Autistic traits – some children may meet criteria for Autism spectrum disorder.
- Challenging behaviours, self-injury and aggression can escalate due to pain from gastro-oesophageal reflux and constipation. Self-injury is twice as high in children with gastro-oesophageal reflux problems (50%).

Medical conditions & issues requiring management

- Feeding difficulties (44%).
 - Dysphagia.
- Gastrointestinal issues (50%).
 - Gastro-oesophageal reflux.
 - Chronic constipation.
 - Protein Energy Malnutrition (PEM) (47%). Close nutritional monitoring, and support required along with swallowing education from Speech Therapist.
- Hyperacusia (70–80%).
- Recurrent respiratory infections (52%).
- Otitis media.
- Strabismus.
- Chronic periodontitis/dental decay.
- Scoliosis (43%).
- Cardiac anomalies (15–20%) (Ventral/atrial septal defects, patent ductus arteriosus, etc).
- Renal, metabolic or immune anomalies.
- Hernias.
- Hearing difficulties.
- Vision abnormalities (46%).
- Epilepsy (not common).
- Peripheral sensory neuropathy.

Physical characteristics

- High pitched cry.
- Abnormal dermatoglyphics (92%).
- Variable malocclusion with anterior open-bite (75%).
- High palate (50%), rarely, with a cleft (5%).
- Prominent supra-orbital arches (31%).
- Microcephaly.
- Hypotonia and hyperlaxitude which, with age, then becomes hypertonia.
- Low birth weight.
- Delayed growth.
- Cryptorchidism in youth and hypogonadism in adulthood.
- Narrow auditory ducts.
- Enamel hypoplasia.
- Single transverse palmar creases.
- Premature grey hair.





Facial characteristics

- Epicanthal folds (90.2%).
- Broad nasal bridge (87.2%).
- Rounded face (83.5%).
- Down-turned corners of the mouth (81%).
- Low-set ears (69.8%).
- Antimongoloid palpebral fissures (56.9%).
- Hypertelorism.
- Micrognathia.
- Preauricular tags.
- Strabismus.

Change of facial characteristics with age

- Convex facial profile mandibular microretrognathia (96.7%).
- Short philtrum (87.8%).
- Palpebral fissures tend to become horizontal (70.2%).
- Macrostomia.
- Dymorphic facial features may become less apparent with age.

Life expectancy for individuals with CDCS is difficult to predict, however in the absence of a serious medical condition, most individuals will live well into adulthood.

Management guidelines for Paediatricians & General Practitioners

A multidisciplinary approach to the clinical management of individuals with CDCS is invaluable. Co-ordination and communication between specialists across all areas will ensure the most effective care of the patient. There are some commonalities found in this phenotype and an excellent overview of CDCS is available at the University of Birmingham's Further Inform Neurogenic Disorders (FIND) site.

See: <http://www.findresources.co.uk/the-Syndromes/cri-du-chat>

However, due also to the large variability of physical, learning, and behavioural difficulties that result from CDCS, clinical management should be tailored to the needs of the individual.

A paediatrician or general practitioner will play an important and integral role in the co-ordination of care. The following points may be of use:

- Motor skills and life skills will take longer to develop in children with CDCS, however with assistance, patience, and encouragement these skills will improve. Encouraging children to do as many things as possible independently will assist the continual development of new skills.
- Feeding difficulties are common in children with CDCS. This is often associated with a poor sucking response, reflux vomiting, and failure to thrive.
 - Breastfeeding counsellors can be accessed through the Australian Breastfeeding Association.
<https://www.breastfeeding.asn.au/breastfeeding-helpline>
 - If appropriate, swallowing ability can be assessed by a barium swallow test. It is important to be aware that aspiration pneumonia can occur and treatment is possible.
 - An enteral tube may be used to overcome feeding difficulties and subsequent failure to thrive.
- Growth charts specific for CDCS
 - <http://criduchat.org.uk/downloads/growth-charts.pdf>
- Occupational therapy and physiotherapy can be beneficial in assisting the development of motor co-ordination, fine motor skills, and life skills. Occupational therapy is also useful for implementing intervention strategies to reduce distress for those with hyperacusis.
- Expressive speech difficulties may be improved through speech and language therapy. Augmentative and alternative communication devices or sign language development may assist some individuals. Consultation with a speech therapist is recommended.
- Audiological evaluations are important for surveillance of hearing and hearing aids may be beneficial. Hearing loss may present as behavioural issues, difficulty with language and speech and immature social interactions.
- Self-injurious and aggressive behaviour is common in children with CDCS and can be distressing for family members and carers. With the assistance of paediatricians, some basic steps can be taken to manage these behaviours:
 - It is critical to rule out medical causes that may be causing pain or discomfort, particularly gastro-oesophageal reflux, otitis media and constipation. Assessment of pain is always an important part of assessing challenging, aggressive and self-injurious behaviours. Chronic pain signatures may include facial expression, leg movements, crying and poor consolability. Also may be characterised by bursts of behaviour, which are apparently 'out of the blue' (no obvious trigger) and unrelated to the environment. In some children, pain may also be indicated by erratic and destructive behaviour.
 - The Face, Legs, Activity, Cry, Consolability Scale (The FLACC: A behavioral scale for scoring postoperative pain in young children) can be a useful tool for assessment.
<http://wps.prenhall.com/wps/media/objects/3103/3178396/tools/flacc.pdf>
 - Parent videos of their child taken on smart phones during periods of self-injurious and aggressive can also be very useful assessment tools.

- Spend time trying to decipher the cause of the behaviour, the possible triggers, contingencies and individual and/or environmental factors.
- Devise interventions and evaluate the effectiveness of interventions (consistency, reactive and proactive, long term strategies are recommended).
- Interventions that develop expressive language may reduce the cause of some self-injurious behaviours.
- Sleeping difficulties - Sleeping difficulties for these children often relate to abilities settling or frequent waking. These may be related to anxiety, difficulties settling, sleep association disorders, hyperacusis or other medical issues such as pain from gastro oesophageal reflux. These should be managed pre-emptively. Management should include settling and calming techniques with good pre sleep and settling routines. Occasionally white noise may help to minimise other noise waking the child.
- Learning difficulties are most effectively managed through an individualised education plan. An individual plan, developed through collaboration between professionals and parents can set achievable goals specific and appropriate for the individual.
- Surgical interventions for high-pitched voice and cry are generally not recommended. However surgical intervention may be necessary for other conditions such as heart defects.
- In a small number of cases there can be potential risks related to intubation as a result of underlying larynx and epiglottis malformations.
- Genetic testing can be important to identify specific genes affected by a deletion. This information can then be used to guide clinical care and management, which is personalised to the individual.

Specialist areas required for patient care may include:

- Gastroenterology.
- Orthopaedic.
- Respiratory.
- Cardiac.
- Dental.
- Audiology.
- Ophthalmology.
- Otolaryngology.
- Psychology and Psychiatry.
- Sleep medicine.
- Speech therapy.



Guidelines for Allied Health Professionals

A multidisciplinary team of medical and allied health professionals can co-ordinate the most effective treatment for children with CDCS.

Maternal & Child Health Professionals

- You may observe signs of CDCS in your interactions with undiagnosed infants/children, such as; high-pitched cry, low birth weight, hypotonia, feeding difficulties and delayed motor development.
- Children with CDCS may have sleeping difficulties due to overactivity, anxiety, sleep apnoea or gastro oesophageal reflux disease (GORD).
- Autism Spectrum Disorder is considered to have the same prevalence in individuals with CDCS as those with intellectual disability, although some of the traits of CDCS are considered to be autism-like.
- Immunisations can still be administered and are important in protecting the child's future health.

Gastroenterologists

- Many children with CDCS will experience gastrointestinal issues including gastro oesophageal reflux and constipation. Managing these issues will improve the child's quality of life and often helps to significantly reduce behavioural problems.

Speech Therapists

- Comprehension skills of children with CDCS are better than their communication skills. Delayed speech development can be assisted by early intervention of alternative communication methods such as Makaton sign language, PODD communication book, Dynovox and/ or the use of some iPad apps such as Pictello. Difficulties with language and speech may also be attributable to hearing loss, which can be managed through hearing aids.

Occupational & Physiotherapists

- Delayed motor development means that children with CDCS may require assistance in movement abilities such as sitting, walking and jumping.
- Fine motor skill development can be delayed so children may also require programs to manage personal skills such as feeding, washing and dressing.
- Graduated guidance programs have been shown to be effective in teaching children with CDCS daily life skills.

Dieticians

- A change in diet may be required to meet children's medical needs such as constipation and low mouth and oesophagus muscle tone.
- Some children may require their food to be pureed. It is important to introduce challenging foods to assist with oral function and speech development but this should be assessed on an individual basis as some children with CDCS will require pureed/soft food for longer than others.

Psychologists

Psychologists can provide assistance to the wellbeing of children with CDCS as well as their parents/carers/family who can experience a high level of stress and mental health problems.

Children with CDCS

- Children may demonstrate challenging behaviour such as self-injury, aggression, shouting and screaming. The triggers of this behaviour can be due to internal factors (pain or sensory stimulation) or external factors (positive and negative social reinforcement). Pain related factors should be ruled out prior to instigating behavioural interventions.
- Time and a greater number of sessions needs to be allowed to change response contingencies in behavioural interventions.
- Emotionally, most adults with CDCS report a high level of positive mood although low mood levels can be found in individuals who also have other health problems.

- Sleeping difficulties that are not caused by health issues can be managed by developing a sleep support plan and medication only if good sleep behaviours and routines are not successful. It may be worth consulting a sleep physician as in many cases other causes such as difficulty in calming might be more of an issue than sleep latency.

Parents/carers/family

- Parents/carers may experience grief and loss upon the diagnosis of their child with CDCS as well as at other points during their lifetime. They may also experience social isolation and stigmatisation in their daily life, so accessing parent support services can be useful.
- Children with CDCS can sometimes exhibit challenging behaviour which can be exhausting of a parent/carer/families' coping resources.
- In cases of an inherited form of CDCS, parents may experience guilt of 'contributing' to the Syndrome.
- The birth of a child can cause changes, both positive and negative, within a family and this can be compounded either way when a child also has a genetic condition.
- As with other disabilities, siblings can also experience negative emotions and behaviours in response to a child with CDCS as they will often receive less attention from caregivers.
- Caring for a child with a disability can be tiring. It is important for parents/carers to remember to look after themselves. This can include utilising respite services.

Educators

- Children with CDCS may experience moderate to severe learning difficulties. They often have better receptive language than expressive language skills so may require alternative communication tools to demonstrate that they have understood the lesson.
- School also provides an opportunity for children with CDCS to further develop their communication and life skills, such as stranger awareness, following directions, describing personal feelings/events and responding appropriately to questions.
- Students with CDCS also appear to have similarities to students with non-verbal learning disability, especially higher functioning children with CDCS. These higher functioning children may have normal phonological processing (understanding speech sounds that make up spoken language), but struggle to use grammar correctly, and understand text adequately.
- Students with atypical CDCS appear to have learning difficulties as opposed to learning disability with average or near average cognition.
- Children with CDCS aged 2-7 years have been found to have a low level of object directed behaviour with attention directed to off-task behaviour and this should be considered in providing the individual's education.
- Adult social interaction and attention is a reinforcer and motivator of behaviour in students with CDCS.

Family & individual support

- Strategies should be developed to enable support for the entire family including parents, siblings and grandparents. Support groups can provide valuable peer support, allowing individuals to connect and share experiences with others in a similar situation.
- Individuals and family members should be informed about the use of the internet to gain information about CDCS. They should be warned about the inaccuracies present on some websites and should be encouraged to use reputable websites.



Supports & resources

Genetic support & advocacy

Cri du Chat Support Group of Australia Inc.
Ph: 61 3 9775 9962
info@criduchat.asn.au
www.criduchat.asn.au

Genetic Support Network Victoria
Ph: 03 8341 6315
Email: info@gsnv.org.au
www.gsnv.org.au

Genetic and Rare Disease Network (WA)
www.geneticandrarediseasenetw.org.au

Association of Genetic Support Australasia
www.agsa-geneticsupport.org.au

Support & advocacy

Association for Children with a Disability (VIC)
Advocacy and information service for children with a disability and their family
<http://acd.org.au/>

People with a Disability Australia
<http://www.pwd.org.au>

Best Life Foundation
Peer support and out of home experiences for children and young adults living with disabilities
www.bestlifefoundation.com.au

Support for carers

Carers Australia
Ph: 1800 242 636
www.carersaustralia.com.au

Commonwealth Respite and Carelink Centre
Ph: 1800 052 222
www9.health.gov.au/ccsd/index.cfm

Links to financial support & intervention

Better start
Early intervention funding service for children with a disability for which CDCS is eligible.
Ph: 1800 242 631
www.betterstart.net.au

National Disability Insurance Scheme (NDIS)
Ph: 1800 800 110
<https://myplace.ndis.gov.au/ndisstorefront/index.html>

Centrelink
Ph: 13 27 17
www.humanservices.gov.au

Resources

Cri du Chat Support Group of Australia Inc.
'Welcome Pack' - Contact info@criduchat.asn.au

Facebook: Cri du Chat: Australian and New Zealand Families.
<https://www.facebook.com/groups/193249824069090/>

Cri du Chat factsheet
www.betterhealth.vic.gov.au/bhcv2/bhcarticles.nsf/pages/Cri_du_chat_Syndrome

First Responder Information Card
<https://fivepminus.org/wp-content/uploads/2017/02/First-Responder-Info-Revsed-4-14-2014.pdf>

Personal stories

<http://www.findresources.co.uk/the-Syndromes/cri-du-chat/child-stories>

<http://www.criduchat.asn.au/#!/family-stories/c21kz>

International sites

5 p Minus Society (USA)
<https://fivepminus.org>

Cri du Chat Support Group (UK)
<http://criduchat.org.uk>

Cri du Chat Support (Italy)
<http://www.criduchat.it>
http://www.criduchat.it/ABC_Criduchat_English.htm

Cri du Chat Support (Brazil)
<http://www.portalcriduchat.com.br/novo/>

Cri du Chat Support Group (Germany)
<https://5p-syndrom.de/5p-/index.php>

Cri du Chat Support Group (Spain)
<http://fundacionsindrome5p.org>

Cri du Chat Support Group (Turkey)
<http://criduchatturkiye.com>

Cri du Chat Support Group of New Zealand
<http://www.criduchat.org.nz>

Email contacts for other countries
<http://www.criduchat.org/index.php/contact/>

For Health Professionals

Cri du Chat Support Group of Australia Inc.
<http://www.criduchat.asn.au>

The Cerebra Centre for Neurodevelopmental Disorders, School of Psychology, University of Birmingham
www.findresources.co.uk/the-Syndromes/cri-du-chat

Centre for Developmental Disability Health Victoria
<http://www.cddh.monash.org/>

Genetics Home Reference
ghr.nlm.nih.gov/condition/cri-du-chat-Syndrome

Referral pathways

Clinical Genetics Services
A GP referral is usually required to access clinical genetics services. Genetics consultations often include consultations with both a geneticist and genetic counsellor.

Victoria
www.vcgs.org.au

New South Wales
www.genetics.edu.au

Queensland
www.health.qld.gov.au/ghq

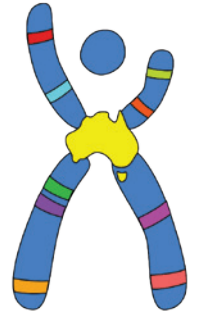
Western Australia
<http://kemh.health.wa.gov.au/services/genetics>

New Zealand
www.genetichealthservice.org.nz

Other Services
Medical specialists and allied health professionals can be accessed through public and private hospitals. Referrals should be forwarded to the appropriate centre.

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Peer reviewed by Dr Susan Green, Psychologist and CDCS parent, Susan Green and Associates, Dr Stephen Withers, Paediatric Clinical Geneticist, Gold Coast Paediatrics and Associate Professor Honey Heussler, Developmental and Behavioural Paediatrician, University of Queensland.



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