CONDITION

Cri du chat syndrome

Cri du chat syndrome is a lifelong genetic condition caused by missing material from chromosome 5.

WHAT IT IS
Lifelong chromosome condition affecting development and health

BODY SYSTEM
Nervous system, growth, movement, and several organs

MAIN SYMPTOMS
Developmental delay, distinctive infant cry, low muscle tone, small head

WHAT CAUSES IT
Missing material from chromosome 5

HOW IT IS DIAGNOSED
Chromosomal microarray, karyotype, or targeted chromosome testing

MAIN TREATMENT
Developmental therapies, supportive care, and complication management

CAN IT BE CURED?
No - the chromosome deletion remains lifelong

IS IT AN EMERGENCY?
Sometimes - with breathing trouble or prolonged seizure

LONG-TERM OUTLOOK
Lifelong disability; many reach adulthood with supportive care

Overview

Cri du chat syndrome is a genetic condition caused by a missing section of chromosome 5. Chromosomes are packages of DNA that contain the instructions for growth and development. In this condition, part of the chromosome's short arm, called 5p, is missing. It is therefore also known as 5p deletion syndrome.

The name means "cry of the cat" in French. Many affected newborns have a high-pitched cry that can sound cat-like. This cry often becomes less noticeable as the child gets older and may disappear. Its absence does not rule out the condition.

Cri du chat syndrome affects development from birth. Most people have some degree of intellectual disability, delayed speech and movement skills, and low muscle tone. Other health needs differ according to the size and location of the missing chromosome section and

whether other genetic changes are present.

The condition is lifelong, but it does not progress in the way a degenerative illness does. Children can continue learning skills throughout childhood and adulthood. Early therapies, educational support, and treatment of associated health problems can help a person communicate, move, participate, and remain as healthy as possible.

Symptoms

Features may be noticeable at birth, while others become clearer as a child develops. Not every person has every feature.

Common early signs include:

Developmental delay affects skills such as sitting, walking, speaking, learning, and self-care. Speech is often more affected than understanding. Some people communicate using signs, pictures, communication devices, or a mixture of methods.

Movement may be delayed by low muscle tone, loose joints, balance difficulties, or later muscle tightness. Some children develop repetitive movements, sleep difficulties, high activity levels, or behaviours linked to frustration and difficulty communicating.

Possible associated health concerns include hearing or vision problems, dental problems, constipation, curvature of the spine, seizures, and frequent respiratory infections. A minority of children have structural differences affecting the heart, kidneys, digestive system, or other organs.

- A high-pitched, cat-like cry during infancy
- Low birth weight or slow growth
- A smaller-than-expected head, called microcephaly
- Low muscle tone, meaning the muscles feel less firm
- Feeding difficulties, weak sucking, or trouble coordinating swallowing
- Distinctive facial features, which may become less obvious with age

Causes and risk factors

Cri du chat syndrome occurs when genetic material is missing from the short arm of chromosome 5. A larger deletion may affect more genes, but deletion size alone cannot predict exactly what one person will be able to do or which health problems they will have.

Most cases begin as a new genetic change when an egg or sperm forms, or very early after conception. This is called a de novo change. It is not caused by anything either parent did before or during pregnancy.

Less often, a child inherits an unbalanced chromosome arrangement from a parent who has a balanced rearrangement. In a balanced rearrangement, chromosome material has changed position, but none is missing or added. The parent is usually healthy because they still have the expected amount of genetic material.

There are no known lifestyle choices that cause the chromosome deletion. A family history of cri du chat syndrome or a known parental chromosome rearrangement can increase the chance of it happening in another pregnancy. A genetics service can assess this chance using the family's actual test results.

Diagnosis

A clinician may suspect cri du chat syndrome because of a newborn's cry, facial appearance, small head size, low muscle tone, feeding problems, or developmental delay. Symptoms alone cannot confirm it.

Diagnosis requires genetic testing. A chromosomal microarray can find missing or extra sections of DNA and usually shows the size of the 5p deletion. A karyotype, which is a picture of the chromosomes, can identify a larger deletion and some chromosome rearrangements. Targeted testing, such as fluorescence in situ hybridisation, may be used to examine a specific chromosome region or clarify family results.

After diagnosis, chromosome testing may be offered to both biological parents. This helps establish whether the deletion was a new event or related to a balanced parental rearrangement. It also gives more accurate information about the chance in a future pregnancy.

Testing during pregnancy can diagnose the condition if chromosome material is examined from chorionic villus sampling or amniocentesis. These are invasive diagnostic procedures. Screening tests may suggest a chromosome difference but do not confirm cri du chat syndrome.

Further assessments may check feeding and swallowing, hearing, vision, heart structure, kidney function, growth, movement, and development. The tests needed are guided by the individual's symptoms and genetic findings.

Treatment and management

There is no treatment that can replace the missing chromosome material. Care focuses on development, communication, independence, comfort, and any associated medical problems.

Infants with feeding difficulties may need support with positioning, feeding techniques, or higher-energy nutrition. A swallowing assessment can look for aspiration, which means food or liquid entering the airway. Some children need temporary or longer-term tube feeding if eating is unsafe or does not provide enough nutrition.

Early developmental support commonly includes:

Communication support should begin early rather than waiting for speech to develop. Many children understand more than they can express, so access to another communication method can reduce frustration.

Medical treatment is matched to the problems present. This may include treatment for reflux, constipation, infections, seizures, hearing loss, dental disease, or sleep difficulties. Structural heart, kidney, eye, or bone problems may require specialist care and, in some cases, surgery.

Regular follow-up often involves a primary care clinician, paediatric or adult disability services, therapists, dentists, and relevant specialists. As a young person approaches adulthood, planning should cover healthcare transfer, decision-making support, education, work or daytime activities, housing, and financial or social services.

- Physiotherapy for strength, posture, balance, and movement
- Occupational therapy for play, daily activities, and adapted equipment
- Speech and language therapy for communication and swallowing
- Sign language, picture systems, or communication devices
- Individual educational and behavioural support

Complications

Feeding and swallowing difficulties can lead to poor growth, dehydration, or aspiration. Aspiration may cause coughing during meals or repeated chest infections. Prompt assessment can make eating safer.

Low muscle tone and reduced mobility may contribute to joint problems, flat feet, or spinal curvature. Physical therapy, suitable seating, and monitoring can help identify changes before they interfere with comfort or movement.

Limited speech can make it difficult to report pain or illness. Changes in sleep, eating,

movement, behaviour, or usual activity may be signs that something is wrong. Dental pain, constipation, reflux, ear infections, and vision problems can otherwise be missed.

Some people develop seizures. Heart or airway differences can also cause serious problems, particularly in infancy, although these do not affect everyone.

Intellectual disability means that many adults need continuing help with healthcare, money, travel, daily routines, or personal care. The amount of support is individual and may change over time.

Prevention

Cri du chat syndrome usually cannot be prevented. Most cases result from a new chromosome deletion that occurs by chance, and there is no known action during pregnancy that causes or prevents it.

If a child or family member has the condition, genetic counselling can explain the test results and the chance of it happening in another pregnancy. When a parent carries a balanced chromosome rearrangement, options may include diagnostic testing during pregnancy or genetic testing of embryos created through in vitro fertilisation. These choices are personal, and testing does not guarantee every possible outcome.

Living with it and what to expect

Cri du chat syndrome remains present throughout life, but the high-pitched infant cry often fades. Facial features may also become less noticeable. Developmental and intellectual disability generally continue, although people can keep gaining skills.

Children often reach milestones later than their peers. Walking, communication, toileting, dressing, and eating independently may take years of teaching and practice. Some people learn spoken words or short sentences, while others rely mainly on signs, pictures, gestures, or electronic devices.

School support is usually needed. A practical learning plan may focus on communication, mobility, self-care, safety, social skills, and academic learning at an appropriate level. Predictable routines and clear communication can help with anxiety or difficult behaviour.

Many people are sociable and take part in family life, school, recreation, supported work, or community activities. Some can complete daily tasks with limited help; others need close supervision and lifelong personal care.

Cri du chat syndrome is not usually considered a degenerative condition, so previously learned abilities are not expected to be lost simply because of the chromosome deletion. Any clear loss of skills should be assessed rather than assumed to be part of the condition.

Life expectancy is influenced by the severity of associated problems, especially major heart defects, breathing or swallowing difficulties, and serious infections. Early childhood can carry greater medical risk for those with complex health needs. Many people without severe organ complications live into adulthood, but long-term information is limited.

When to get medical help

Arrange routine medical review if there are concerns about growth, feeding, hearing, vision, sleep, constipation, dental health, movement, behaviour, or progress at school. New coughing during meals, repeated chest infections, or meals that are consistently long and tiring may need a swallowing assessment.

Seek prompt medical advice for:

Call emergency services for severe breathing difficulty, blue or grey lips, choking that blocks breathing, unresponsiveness, or a seizure lasting five minutes or longer. Emergency help is also needed if repeated seizures occur without recovery between them.

- A first suspected seizure or a change in known seizures
- Noticeably reduced eating or drinking
- Repeated vomiting or signs of dehydration
- New weakness, loss of skills, or reduced alertness
- Persistent pain or an unexplained major behaviour change

Common questions

Is cri du chat syndrome inherited?

Usually not. Most deletions are new genetic changes. In a smaller number of families, one parent carries a balanced chromosome rearrangement that raises the chance of an affected pregnancy. Parental chromosome testing can clarify this.

Can someone with cri du chat syndrome talk?

Some people develop spoken words or sentences, while others have little or no speech. Understanding may be stronger than speaking ability. Signs, pictures, gestures, and

communication devices can provide an effective way to communicate.

Will the cat-like cry last forever?

Usually, it becomes less noticeable or disappears as the child grows. It is most characteristic during infancy. The sound of the cry does not measure the person's level of disability.

Can a person with cri du chat syndrome walk?

Many children eventually learn to walk, often later than usual. Others need braces, walking aids, wheelchairs, or ongoing physical assistance. Muscle tone, balance, joint health, and associated medical problems affect mobility.

What is the life expectancy for cri du chat syndrome?

There is no single life expectancy that applies to everyone. Major heart defects, severe swallowing or breathing problems, and serious infections can shorten life, especially in early childhood. Many people without severe complications survive into adulthood.

Can cri du chat syndrome be detected before birth?

Yes. Diagnostic genetic testing on samples obtained through chorionic villus sampling or amniocentesis can identify the chromosome deletion. Prenatal screening alone cannot provide a definite diagnosis.

Sources

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