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ABOUT US

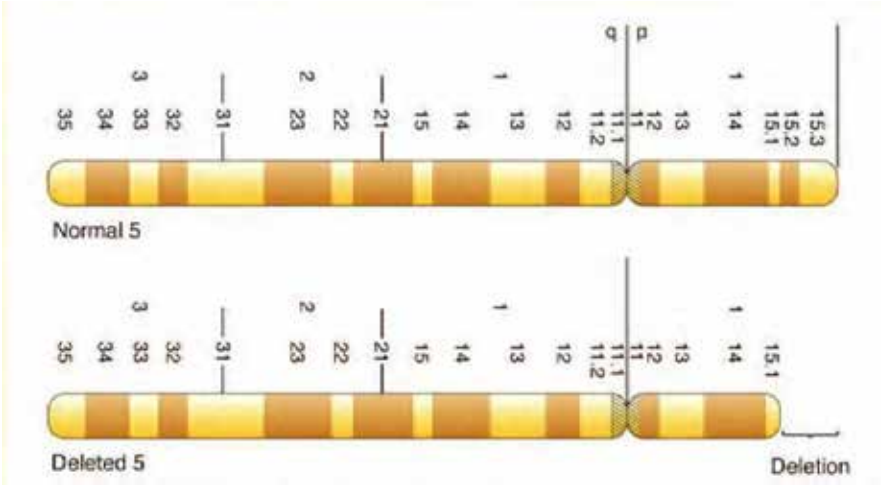
Chromosome Disorder Outreach provides support and information to anyone diagnosed with a rare chromosome change, rearrangement or disorder. CDO actively promotes research and a positive community understanding of all chromosome disorders.

CDO is a 501c3 organization founded in 1992.

5p deletion

5p deletion

Besides the cat-like cry, patients often also show other clinical features, including, but not limited to, microcephaly (i.e. significantly smaller than average-sized heads), broad nasal bridges, epicanthic folds (i.e. skin-folds of the upper eyelid), preauricular tags or pits, undersized jaws, and psychomotor and mental retardation. At least 20% of affected patients have different types of congenital heart disease. Defects of other systems may be found in several patients. Because individuals with this syndrome vary so greatly in their genotype (that is, how much and which portions of the short arm of chromosome five are deleted) their phenotypes (or observable characteristics) vary drastically, meaning that patients can show any range of the aforementioned features, as well as a wide-range of unlisted characteristics
(1). In some rare cases, even the cat-like cry is not observed (2). Researchers have mapped the typical Cri du Chat clinical symptoms and the typical cat-like cry to 'critical regions' of chromosome five named 15.2 and 15.3, respectively (1, 3). Further, they have found that the size of the deletion correlates with the severity of mental retardation (4)



Cri du Chat Syndrome (CdCS) was first described in 1963 by Jerome Lejeune, who named the syndrome after it most conspicuous clinical feature, a high-pitched, cat-like cry observed in infants with the disorder. Cri du chat, which translates to 'cry of cat' in English, is also known as 5p- syndrome, which refers to the syndrome's genotype as opposed to its phenotype--that is, it refers to the genetic constitution of the syndrome instead of its observable characteristics. All patients of the syndrome are missing (denoted by '-') some portion of the short arm (denoted by 'p') of their fifth chromosome.

Although it is known that severe mental and psychomotor retardation are common in those with CdCS, researchers are still working to characterize their exact behavioral profile. The research that does exist is sometimes contradictory. Some common behaviors may include hyperactivity, loss of attention, aggression, and self-injury, although there are conflicting reports on the topic hyperactivity (5). Patients with CdCS also often have difficulty with language development, to the degree that some are not able to develop spoken language; patients' comprehension, however, is oftentimes much better than their ability to speak (6, 7).

Most premature deaths due to this syndrome occur during the first months of life and nearly all occur within the first year. The most frequent causes of death are pneumonia, aspiration, congenital cardiac defects, and respiratory distress syndrome. After the child's first year, however, life expectancy is high, and there have been reports of patients with the syndrome living into their 50s (8).

While there is no specific treatment for CdCS, it can be managed, especially if intervention takes place early enough. Parents or guardians might consider seeking speech therapy and/or physical therapy, depending on the patient's symptoms.

Approximately 15% of deletions are caused by parental balanced rearrangements (mainly translocations or inversions). Parents concerned with the chance of having affected children should seek genetic counseling.