

Cat eye syndrome (CES) is a rare chromosomal disorder that may be evident at birth. Individuals with a normal chromosomal make-up have two 22nd chromosomes, both of which have a short arm, known as 22p, and a long arm, known as 22q. However, in individuals with CES, the short arm and a small region of the long arm of chromosome 22 (i.e., 22pter-22q11) are present four times (partial tetrasomy) rather than twice in cells of the body. In a small number of people with CES, the 22q11 region is present in 3 copies (partial trisomy). The name “cat eye syndrome” is derived from a distinctive eye (ocular) abnormality that is present in a little over half affected individuals. This defect, known as a coloboma, usually appears as a cleft or gap in the iris below the pupil, and the elongated pupil therefore resembles the appearance of a cat’s eye. There are, however, many other features associated with CES involving many organs and systems. These symptoms result from abnormal development during embryo and fetal stages. Associated symptoms vary greatly in presence and severity from one person to another, including among members of the same family. CES may be best thought of as a disorder spectrum. While some people may have few or mild manifestations, others may have multiple severe malformations. CES has been recognized for more than a century. More than 100 cases have been described in the medical literature, including apparently sporadic and familial cases. Many more affected individuals exist but have not been described in the medical literature. However, the syndrome is very rare, and currently there are no accurate estimates of the incidence of CES in the population. Schinzel et al (1981) estimated an incidence of approximately one in 50,000 to one in 150,000 individuals in Northeastern Switzerland. Because some affected individuals develop few associated features, however, the disorder may in some people remain unrecognized. There is currently no way to estimate how underdiagnosed this syndrome is.