

Chromosomal aberrations are disruptions in the normal chromosomal content of a cell, and are a major cause of genetic disease in humans, such as Down syndrome. Some chromosome abnormalities do not cause disease in carriers, such as translocations, or chromosomal inversions, although they may lead to a higher chance of having a child with a chromosome disorder. Abnormal numbers of chromosomes or chromosome sets, aneuploidy, may be lethal or give rise to genetic disorders.

The gain or loss of chromosome material can lead to a variety of [genetic disorders](#). Human examples include:

- [Cri du chat](#), which is caused by the [deletion](#) of part of the short arm of chromosome 5. "Cri du chat" means "cry of the cat" in French, and the condition was so-named because affected babies make high-pitched cries that sound like a cat. Affected individuals have wide-set eyes, a small head and jaw and are moderately to severely mentally retarded and very short.
- [Wolf-Hirschhorn syndrome](#), which is caused by partial deletion of the short arm of chromosome 4. It is characterized by severe growth retardation and severe to profound mental retardation.
- [Jacobsen syndrome](#), also called the terminal 11q deletion disorder.^[1] This is a very rare disorder. Those affected have normal intelligence or mild mental retardation, with poor expressive language skills. Most have a bleeding disorder called [Paris-Trousseau syndrome](#).
- [Down's syndrome](#), usually is caused by an extra copy of chromosome 21 ([trisomy 21](#)). Characteristics include decreased muscle tone, asymmetrical skull, slanting eyes and mild to moderate mental retardation.
- [Edward's syndrome](#), which is the second most common trisomy after Down syndrome. It is a trisomy of chromosome 18. Symptoms include mental and motor retardation and numerous congenital anomalies causing serious health problems. Ninety percent die in infancy; however, those who live past their first birthday usually are quite healthy thereafter. They have a characteristic hand appearance with clenched hands and overlapping fingers.
- [Patau Syndrome](#), also called D-Syndrome or trisomy-13. Symptoms are somewhat similar to those of trisomy-18, but they do not have the characteristic hand shape.
- [Klinefelter's syndrome](#) (XXY). Men with Klinefelter syndrome are usually sterile, and tend to have longer arms and legs and to be taller than their peers. Boys with the syndrome are often shy and quiet, and have a higher incidence of speech delay and [dyslexia](#). During puberty, without testosterone treatment, some of them may develop [gynecomastia](#).
- [Turner syndrome](#) (X instead of XX or XY). In Turner syndrome, female sexual characteristics are present but underdeveloped. People with Turner syndrome often have a short stature, low hairline, abnormal eye features and bone development and a "caved-in" appearance to the chest.
- [XYY syndrome](#). XYY boys are usually taller than their siblings. Like XXY boys and XXX girls, they are somewhat more likely to have learning difficulties.
- [Triple-X syndrome](#) (XXX). XXX girls tend to be tall and thin and are often shy. They have a higher incidence of dyslexia.