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# Overview of Sex Chromosome Abnormalities

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**Sex chromosome abnormalities may be caused by full or partial deletions or duplications of sex chromosomes.**

Chromosomes are structures within cells that contain DNA and many genes. A gene is a segment of deoxyribonucleic acid (DNA) and contains the code for a specific protein that functions in one or more types of cells in the body (see Genes and Chromosomes for a discussion about genetics). Genes contain instructions that determine how the body is supposed to function. Except for certain cells (for example, sperm and egg cells or red blood cells), every human cell contains 23 pairs of chromosomes, for a total of 46 chromosomes. There are 22 pairs of chromosomes that are not sex chromosomes (called nonsex chromosomes, numbered chromosomes, or autosomal chromosomes) and one pair of sex chromosomes. The sex chromosomes determine whether a fetus becomes male or female. A pair of X and Y chromosomes (XY) results in a male, and a pair of X and X chromosomes (XX) results in a female.

Sex chromosome abnormalities occur when a person is missing a whole sex chromosome (called monosomy) or has an extra sex chromosome (one extra is trisomy). Abnormalities can also occur when a person is missing *part* of a sex chromosome (called a deletion).

Sex chromosome abnormalities are common and cause syndromes that are associated with a range of physical and developmental problems. Many of these syndromes are not noticed while the mother is pregnant but may be discovered if prenatal testing is done for other reasons, such as older age in the mother. The abnormalities are often hard to recognize at birth and may not be diagnosed until puberty.

Syndromes that are caused by a sex chromosome abnormality are less severe than those caused by a nonsex chromosome abnormality. For example, girls who have an extra sex chromosome (an extra X) often appear normal physically and mentally and are fertile. In contrast, children who have extra numbered (1 to 22 ) chromosomes typically have severe abnormalities such as Down syndrome, which commonly results from a person having an extra chromosome 21. An extra chromosome 1 can be fatal to a fetus. Similarly, girls who are *missing* a sex chromosome have a specific syndrome (Turner syndrome), whereas fetuses who are missing a nonsex chromosome do not survive.