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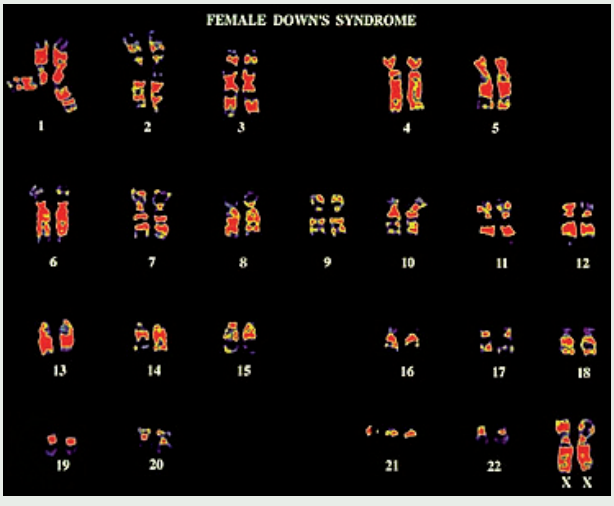
Genetic diseases caused by chromosomal aberrations.

About 1 out of 150 live newborns has a detectable chromosomal abnormality. Yet even this high incidence represents only a small fraction of genetic disease since the vast majority are lethal and result in prenatal death or stillbirth. Indeed, 50 percent of all first-trimester miscarriages and 20 percent of all second-trimester miscarriages are estimated to involve a chromosomally abnormal fetus.

Chromosome disorders can be grouped into three principal categories: (1) those that involve numerical abnormalities of the autosomes, (2) those that involve structural abnormalities of the autosomes, and (3) those that involve the sex chromosomes. Autosomes are the 22 sets of chromosomes found in all normal human cells. They are referred to numerically (chromosome 1, chromosome 2) according to a traditional sort order based on size, shape, and other properties. Sex chromosomes make up the 23rd pair of chromosomes in all normal human cells and come in two forms, termed X and Y. In humans and many other animals, it is the constitution of sex chromosomes that determines the sex of the individual, such that XX results in a female and XY results in a male.

Numerical abnormalities, involving either the autosomes or sex chromosomes, are believed generally to result from meiotic nondisjunction – that is, the unequal division of chromosomes between daughter cells – that can occur during either maternal or paternal gamete formation. Meiotic nondisjunction leads to eggs or sperm with additional or missing chromosomes. Although the biochemical basis of numerical chromosome abnormalities remains unknown, maternal age clearly has an effect, such that older women are at significantly increased risk to conceive and give birth to a chromosomally abnormal child. The risk increases with age in an almost exponential manner, especially after age 35, so that a pregnant woman age 45 or older has between a 1 in 20 and 1 in 50 chance that her child will have trisomy 21 (Down syndrome), while the risk is only 1 in 400 for a 35-year-old woman and less than 1 in 1,000 for a woman under the age of 30. There is no clear effect of paternal age on numerical chromosome abnormalities.



Although Down syndrome is probably the best-known and most commonly observed of the autosomal trisomies, being found in about 1 out of 800 live births, both trisomy 13 and trisomy 18 are also seen in the population, albeit at greatly reduced rates (1 out of 10,000 live births and 1 out of 6,000 live births, respectively). The vast majority of conceptions involving trisomy for any of these three autosomes are nonetheless lost to miscarriage, as are all conceptions involving trisomy for any of the other autosomes. Similarly, monosomy for any of the autosomes is lethal in utero and therefore is not seen in the population. Because numerical chromosomal abnormalities generally result from independent meiotic events, parents who have one pregnancy with a numerical chromosomal abnormality are generally not at markedly increased risk above the general population to repeat the experience.

(www.britannica.com)

a) What is meant by chromosomal abnormality? What is it usually determined by?
b) What are the most common autosomal trisomies occurring in humans? Why are other autosomal abnormalities rare?