

pregnant women can avoid having a child with one of the fetal alcohol spectrum disorders by not ingesting alcohol during pregnancy.

Genetic Disorders

Genetic disorders can be caused by chromosome abnormalities as seen in Turner syndrome, Down syndrome, or velocardiofacial syndrome (VCFS); single-gene mutations as seen in sickle cell anemia, neurofibromatosis, or Duchenne muscular dystrophy; a combination of genetic and environmental factors as seen in NTDs or maturity-onset diabetes in the young; and mitochondrial deoxyribonucleic acid (mtDNA) mutations as seen in nonsyndromic deafness susceptibility caused by aminoglycoside sensitivity.

Both numeric and large structural abnormalities of **autosomes** (all chromosomes except the X and Y chromosomes) account for a variety of syndromes usually characterized by cognitive deficiencies. Nurses often note dysmorphic facial features, behavioral characteristics such as an unusual cry and poor feeding behavior, and other neurologic manifestations such as hypotonia or abnormal reflex responses, which may alert them to these and other chromosome abnormalities.

Somatic cells contain 44 autosomes (the 22 pairs of chromosomes that do not greatly influence sex determination at conception) and two sex chromosomes, XX in females and XY in males. For the purpose of cytogenetic studies, chromosomes are usually displayed in a karyotype, the laboratory-made arrangement of specially prepared chromosomes according to their size, centromere position, and band pattern. Numeric chromosome abnormalities occur whenever entire chromosomes are added or deleted. Down syndrome is an example of a condition caused by having an extra autosome, chromosome 21. Turner syndrome is the only example of a condition compatible with life that is caused by the absence of a chromosome. Children with Turner syndrome have one X chromosome. Chromosomes are subject to structural alterations resulting from breakage and rearrangement. A chromosome deletion occurs when chromosome breakage results in loss of the broken fragment at a chromosome's terminal end or within the chromosome. Some structural chromosome abnormalities are too