chromosomes, whereas the other receives only one, yielding, for example, a 45,X/47,XXX mosaic. All descendent cells derived from each of these precursors thus have either a 47,XXX complement or a 45,X complement. Such a patient is a mosaic variant of Turner syndrome, with the extent of phenotypic expression dependent on the number and distribution of the 45,X cells.

Autosomal mosaicism seems to be much less common than that involving the sex chromosomes. An error in an early mitotic division affecting the autosomes usually leads to a nonviable mosaic due to autosomal monosomy. Rarely, the nonviable cell population is lost during embryogenesis, yielding a viable mosaic (e.g., 46,XY/47,XY,+21). Such a patient is a trisomy 21 mosaic with variable expression of Down syndrome, depending on the proportion of cells containing the trisomy.

A second category of chromosomal aberrations is associated with changes in the structure of chromosomes. To be visible by routine banding techniques, a fairly large amount of DNA (approximately 2 to 4 million base pairs), containing many genes, must be involved. The resolution is much higher with fluorescence in situ hybridization (FISH), which can detect changes as small as kilobases. Structural changes in chromosomes usually result from chromosome breakage followed by loss or rearrangement of material. In the next section the more common forms of alterations in chromosome structure and the notations used to signify them are reviewed.

*Deletion* refers to loss of a portion of a chromosome (Fig. 5-18). Most deletions are interstitial, but rarely terminal deletions may occur. Interstitial deletions occur when there are two breaks within a chromosome arm, followed by loss of the chromosomal material between the breaks and fusion of the broken ends. One can specify in which regions and at what bands the breaks have occurred. For example, *46,XY,del(16)(p11.2p13.1)* describes breakpoints in the short arm of chromosome 16 at 16p11.2 and 16p13.1 with loss of material between breaks. Terminal deletions result from a single break in a chromosome arm, producing a fragment with no centromere, which is then lost at the next cell division, and a chromosome bearing a deletion. The end of the chromosome is protected by acquiring telomeric sequences.

A *ring chromosome* is a special form of deletion. It is produced when a break occurs at both ends of a chromosome with fusion of the damaged ends (Fig. 5-18). If significant genetic material is lost, phenotypic abnormalities result. This might be expressed as *46,XY,r(14)*. Ring chromosomes do not behave normally in meiosis or mitosis and usually result in serious consequences.

*Inversion* refers to a rearrangement that involves two breaks within a single chromosome with reincorporation of the inverted, intervening segment (Fig. 5-18). An inversion involving only one arm of the chromosome is known as *paracentric.* If the breaks are on opposite sides of the centromere, it is known as *pericentric.* Inversions are often fully compatible with normal development.

*Isochromosome* formation results when one arm of a chromosome is lost and the remaining arm is duplicated, resulting in a chromosome consisting of two short arms only or of two long arms (Fig. 5-18). An isochromosome has morphologically identical genetic information in both arms. The most common isochromosome present in live births involves the long arm of the X and is designated *i(X)(q10)*. The Xq isochromosome is associated with monosomy for genes on the short arm of X and with trisomy for genes on the long arm of X.

In a *translocation*, a segment of one chromosome is transferred to another (Fig. 5-18). In one form, called *balanced*

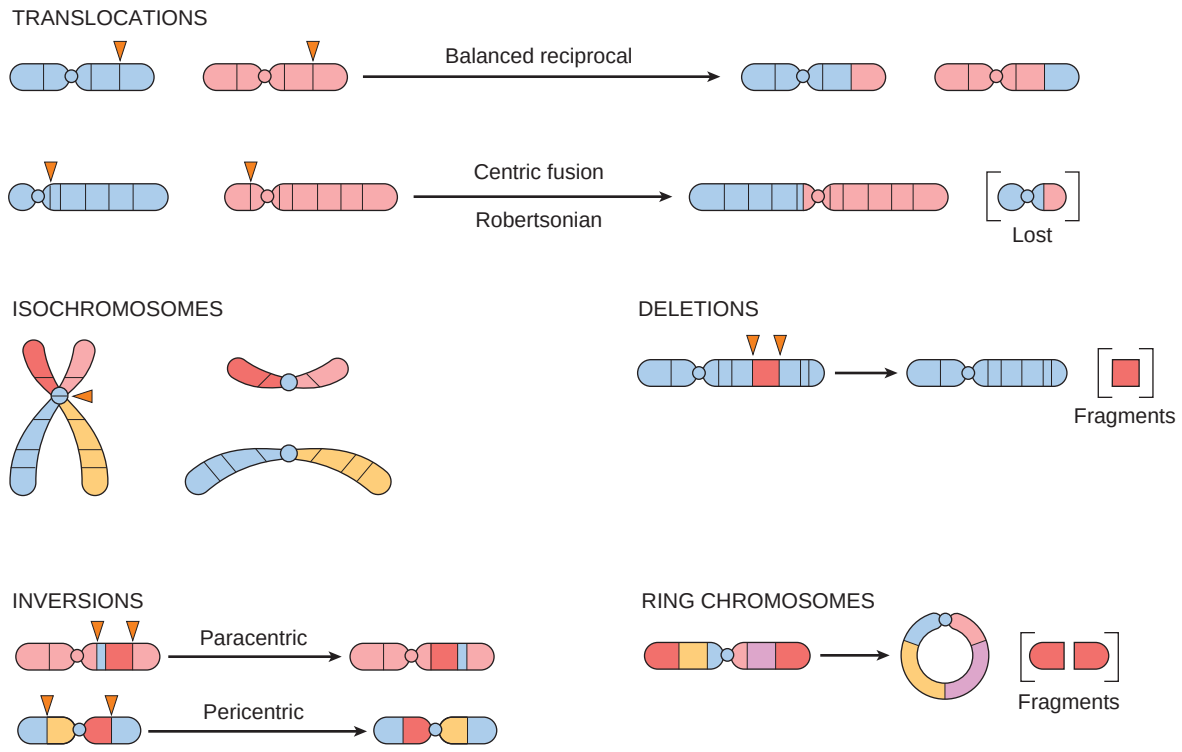


**Figure 5-18** Types of chromosomal rearrangements.