Aneuploidy

Scientists call an individual with the appropriate number of chromosomes for their species **euploid**. In humans, euploidy corresponds to 22 pairs of autosomes and one pair of allosomes. An individual with an error in chromosome number is described as **aneuploid**, a term that includes **monosomy**, losing one chromosome, or **trisomy**, gaining an extra chromosome.

Trisomy 21, or Down syndrome, is a condition that occurs when an individual has a third copy of chromosome 21. Down syndrome is characterized by short stature, stunted digits, facial distinctions that include a broad skull and large tongue, and significant developmental delays. The occurrence of Down syndrome can be correlated with parental age. Older parents are more likely to produce fetuses carrying the trisomy 21 genotype (Figure 8.32). Turner syndrome, which is characterized by the presence of only one X allosome, is an example of a monosomy condition. Females that have Turner syndrome are typically sterile and cannot reproduce.

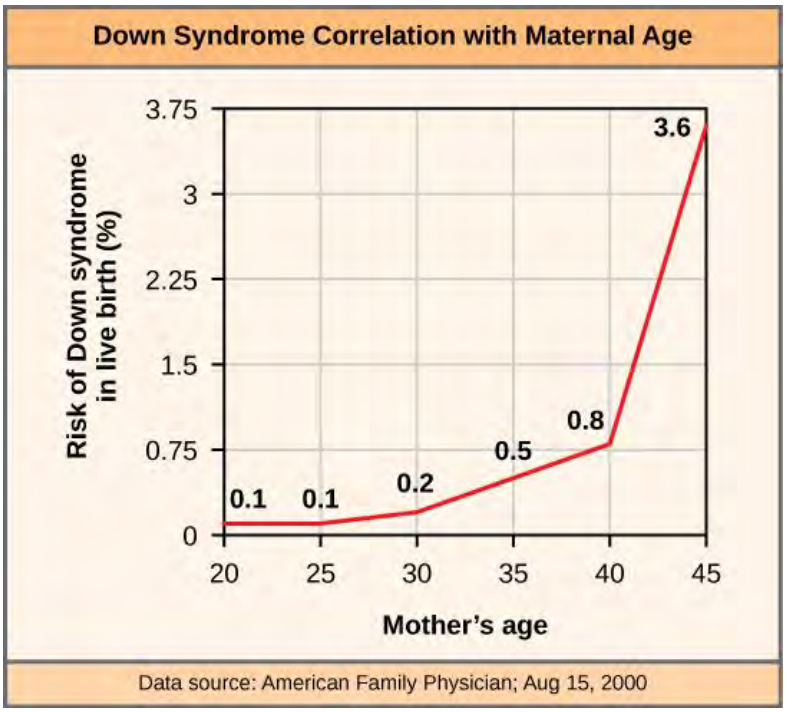

Figure 8.32 The incidence of having a fetus with trisomy 21 increases dramatically with maternal age. (credit: Clark et al. / Biology 2E OpenStax)

CONCEPTS IN ACTION- Visualize the addition of a chromosome that leads to Down syndrome in this video simulation.

Polyploidy

We call an individual with more than the correct number of chromosome pairs a **polyploid**. For instance, fertilizing an abnormal diploid egg with a normal haploid sperm would yield a polyploid. Polyploid animals are extremely rare, with only a few examples including some flatworms, crustaceans, amphibians, fish, and lizards. Polyploid animals are sterile because meiosis cannot occur normally. Rarely, polyploid animals can reproduce asexually when an unfertilized egg divides mitotically to produce offspring. In contrast, polyploidy is very common in plants, and polyploid plants tend to be larger and more robust than the euploids of their species (Figure 8.33).

Figure 8.33 As with many polyploid plants, this triploid orange daylily (*Hemerocallis fulva*) is particularly large and robust and grows flowers with triple the number of petals of its diploid counterparts. (credit: Steve Karg / Biology 2E OpenStax)

Chromosomal Structural Rearrangements

In addition to errors in chromosome number, numerous structural chromosomal rearrangements can occur. These include duplications, deletions, inversions, and translocations.

Duplications and Deletions

In chromosomal **duplications**, a part of a chromosome is duplicated. The duplicated DNA can then either be inserted into a different position on the same chromosome or a completely different chromosome (Figure 8.34). In chromosomal **deletions**, a part of the chromosome is lost or removed (Figure 8.34).

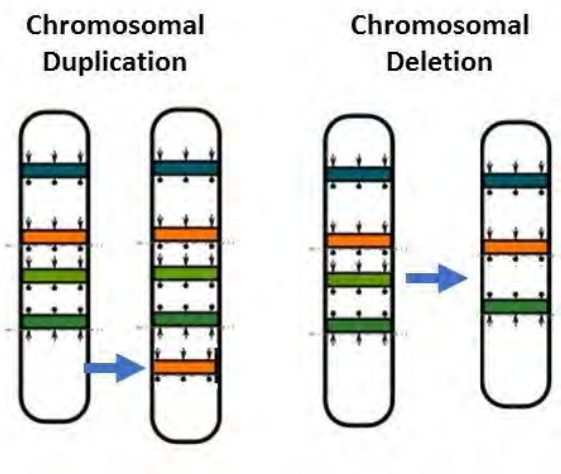

Figure 8.34 Chromosomal arrangements include both duplications and deletions. (credit: Modified by Elizabeth O'Grady original work of Guy Leonard Wikimedia Commons)

Both duplications and deletions often produce offspring that survive but exhibit physical and mental abnormalities. A deletion of a region on chromosome 11 leads to a condition called 11q terminal deletion disorder or Jacobsen syndrome. Jacobsen syndrome involves distinct changes to facial features as well as heart and bleeding defects. A gene duplication on chromosome 17 leads to a condition known as Hereditary motor and sensory neuropathy or Charcot-Marie-Tooth (CMT) disorder. CMT results in individuals that have nervous system issues involving nerves that carry and deliver information to an individual's legs, arms, hands, and feet.

Inversions

A chromosome **inversion** is a detachment, 180° rotation, and reinsertion of part of a chromosome (Figure 8.35). Inversions may occur in nature as a result of damaged or cut DNA or from transposable elements, special DNA sequences capable of rearranging chromosome segments. Unless a gene sequence is disrupted, inversions are likely to have minor effects. However, inversions that disrupt genes can result in abnormally high or low levels of specific proteins.

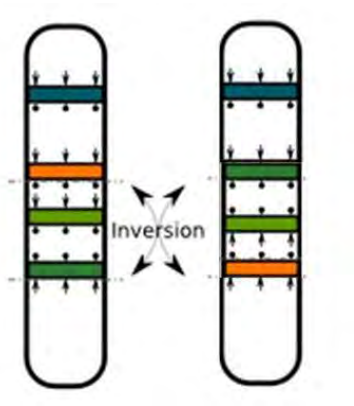

Figure 8.35 An inversion is an example of a chromosomal arrangement. (credit: Modified by Elizabeth O'Grady original work of Guy Leonard Wikimedia Commons)

Translocations

A **translocation** occurs when a segment of genetic material breaks from one chromosome and reattaches to another chromosome or a different part of the same chromosome. Translocations can either have minimal to no impact or have devastating effects depending on how the positions of genes are altered. Notably, specific translocations have occurred with several cancers and with schizophrenia. Reciprocal translocations result from exchanging chromosome segments between two nonhomologous chromosomes such that there is no genetic information gain or loss (Figure 8.36).

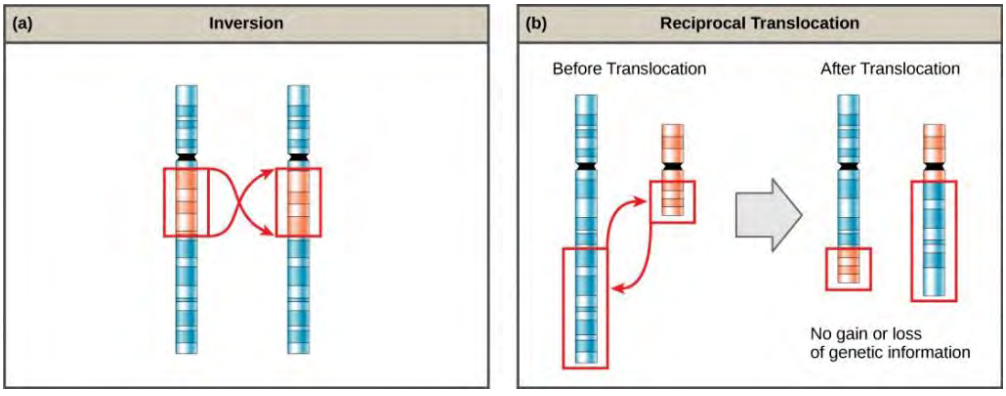

Figure 8.36 (a) chromosomal inversion (b) reciprocal translocation (credit: modification of work by National Human Genome Research Institute / Concepts of Biology OpenStax)