Mader/*Biology*, 11/e – Chapter Outline

Chapter 10

10.6 Change in Chromosome Number and Structure

A. Aneuploidy

1. **Chromosomal mutations** are **c**hanges in chromosome number or structure.
2. Mutations, along with crossing-over, recombination of chromosomes during meiosis, and gamete fusion during fertilization, increase the amount of variation among offspring.
3. The correct number of chromosomes in a species is called **euploidy;** changes in chromosome number resulting from nondisjunction during meiosis is called **aneuploidy.**
4. **Monosomy** (2n – 1) occurs when an individual has only *one* of a particular type of chromosome.
5. **Trisomy** (2n + 1) occurs when an individual has *three* of a particular type of chromosome.
6. **Nondisjunction** is the failure of chromosomes to separate at meiosis—both members of the homologous pair go into the same gamete.
    a. *Primary nondisjunction* occurs during meiosis I when both members of a homologous pair go into the same daughter cell
    b. *Secondary nondisjunction* occurs during meiosis II when the sister chromatids fail to separate and both daughter chromosomes go into the same gamete.
7. Monosomy and trisomy occur in plants and animals; in autosomes of animals, it is generally lethal.
8. **Trisomy 21** is the most common autosomal trisomy.
    a. Trisomy 21 (also called *Down syndrome*) occurs when three copies of chromosome 21 are present.
    b. Usually two copies of chromosome 21 are contributed by the egg; in 23% of the cases, the sperm had the extra chromosome 21.
    c. A Down syndrome child has many characteristic signs and symptoms, including a tendency for leukemia, cataracts, faster aging, mental retardation, and an increased chance of developing Alzheimer disease later in life.
    d. Chances of a woman having a Down syndrome child increase with age.
    e. A **karyotype,** a visual display of the chromosomes arranged by shape, size, and banding pattern, may be performed to identify babies with Down syndrome and other aneuploid conditions.
9. Nondisjunction during oogenesis can result in too few or too many X chromosomes; nondisjunction during spermatogenesis can result in missing or too many Y chromosomes.
10. **Turner syndrome** females have only one sex chromosome, an X; thus, they are XO, with O signifying the absence of a second sex chromosome.
    a. Turner females are short, have a broad chest and folds of skin on back of neck.
    b. Ovaries of Turner females never become functional; therefore, females do not undergo puberty.
    c. They usually have normal intelligence and can lead fairly normal lives with hormone supplements.
11. **Klinefelter syndrome** males have one Y chromosome and two or more X chromosomes (e.g., XXY).
    a. Affected individuals are sterile males; the testes and prostate are underdeveloped.
    b. Individuals have large hands and feet, long arms and legs, and lack facial hair.
    c. Presence of the Y chromosome drives male formation but more than two X chromosomes may result in mental retardation.
    d. A *Barr body*, usually only seen in the nuclei of a female's cells, is seen in this syndrome due to the two X chromosomes.
12. **Poly-X** females (or *superfemale*) have three or more X chromosomes and therefore extra Barr

bodies in the nucleus.
    a. There is no increased femininity; most lack any physical abnormalities.
    b. XXX individuals are not mentally retarded but may have delayed motor and language development; XXXX females are usually tall and severely mentally retarded.
    c. Some experience menstrual irregularities but many menstruate regularly and are fertile.
13. **Jacobs syndrome** (XYY) are males with two Y chromosomes instead of one.
    a. This results from nondisjunction during spermatogenesis.
    b. Males are usually taller than average, suffer from persistent acne, and tend to have speech and reading problems.
    c. Earlier claims that XYY individuals were likely to be aggressive were not correct.

B. Changes in Chromosome Structure
1. Environmental factors including radiation, chemicals, and viruses, can cause chromosomes to break; if the broken ends do not rejoin in the same pattern, this causes a change in chromosomal structure.
2. **Deletion:** a type of mutation in which an end of a chromosome breaks off or when two simultaneous breaks lead to the loss of a segment.
3. **Translocation:** a chromosomal segment is removed from one chromosome and inserted into another nonhomologous chromosome; in Down syndrome, 5% of cases are due to a translocation between chromosome 21 and 14, a situation that runs in the family of the father or mother.
4. **Duplication:** the presence of a chromosomal segment more than once on the same chromosome.
    a. A broken segment from one chromosome can simply attach to its homologue or unequal crossing-over may occur.
    b. A duplication may also involve an **inversion** where a segment that has become separated from the chromosome is reinserted at the same place but in reverse; the position and sequence of genes are altered.

C. Living with Klinefelter Syndrome (*Evolution* reading)
1. Stefan Schwartz is a man who has Klinefelter syndrome.
2. As a child, some of his symptoms included being shy, trouble making friends, and emotional instability.
3. Teacher and doctors thought he had “learning disabilities,” and was lazy.
4. Eventually a doctor tested his blood and discovered he had Klinefelter sydrome, a genetic disorder having sex chromosomes XXY.
5. Physical treatment can include testosterone injection. Emotional treatment can include support groups.

D. Human Syndromes
1. Deletion Syndromes
    a. *Williams syndrome* occurs when chromosome 7 loses an end piece: children look like pixies, have poor academic skills but good verbal and musical skills; lack of elastin causes cardiovascular problems and skin aging.
    b. *Cri du chat syndrome* (“cry of the cat”) is a deletion in which an individual has a small head, is mentally retarded, has facial abnormalities, and an abnormal glottis and larynx resulting in a cry resembling that of a cat.
2. Translocation Syndromes
    a. If a translocation results in the normal amount of genetic material, the person will remain healthy; if a person inherits only one of the translocated chromosomes, that person may have only one allele or three alleles rather than the normal two.
    b. In *Alagille syndrome*, chromosomes 2 and 20 exchange segments, causing a small deletion on chromosome 20 that may produce some abnormalities.