Heredity Outline

29.1 Genes are the vocabulary of genetics (pp. 1107–1108; Fig. 29.1)

A. Introduction (p. 1107; Fig. 29.1)

1. The nuclei of all human cells except gametes contain the diploid number of chromosomes (46), consisting of 23 pairs of homologous chromosomes.
   a. Homologous chromosomes are pairs of chromosomes, one paternal and one maternal, which carry the same genes, but do not necessarily express the trait in the same way.
2. Two of the 46 chromosomes are the sex chromosomes (X and Y), and the remaining 44 are the 22 pairs of autosomes that guide the expression of most other traits.
3. The karyotype is the diploid chromosomal complement displayed in homologous pairs; the genome represents the entire genetic makeup of both egg and sperm.

B. Chromosomes are paired, with one coming from each parent, and the genes on those chromosomes are also paired (pp. 1107)

1. Alleles are any two matched genes at the same locus (location) on homologous chromosomes.
   a. If the two alleles controlling a trait are the same, the genotype is homozygous.
   b. If the two alleles controlling a trait are different, the genotype is heterozygous.
2. A dominant allele is one that will mask or suppress the expression of the other allele, and is indicated by a capital letter (for example, X).
3. A recessive allele is the allele that is suppressed by the dominant allele and will only be expressed in the homozygous condition; it is indicated by a lowercase letter (for example, x).

C. A person's genetic makeup is called his or her genotype, but the way the genotype is expressed in the body is that individual's phenotype. (p. 1108)

29.2 Genetic variation results from independent assortment, crossover of homologues, and random fertilization (pp. 1108–1110; Figs. 29.2–29.3)

A. Chromosome Segregation and Independent Assortment (pp. 1108–1109; Fig. 29.2)

1. During meiosis I, the alignment of the tetrads along the center of the cell is completely random, allowing for the random distribution of maternal and paternal chromosomes into the daughter nuclei.
2. In meiosis I, the two alleles determining each trait are segregated, or distributed to different gametes.
3. Alleles on different pairs of homologous chromosomes are distributed independently of each other, resulting in each gamete having one of the four possible parental alleles for each trait.
4. Independent assortment of homologues during meiosis I can be calculated as 2^n, where n is the number of homologous pairs.

B. During meiosis I, homologous chromosomes may exchange gene segments, a process called crossing over, which gives rise to recombinant chromosomes that have contributions from each parent. (p. 1109; Fig. 29.3)
C. Random fertilization occurs because a single human egg is fertilized by a single human sperm on a completely haphazard basis. (pp. 1109)

29.3 Several patterns of inheritance have long been known (pp. 1110–1112; Figs. 29.4–29.6; Tables 29.1–29.2)
A. Dominant-Recessive Inheritance (pp. 1110–1112; Fig. 29.4; Tables 29.1–29.2)
1. A Punnett square is used to determine the possible gene combinations resulting from the mating of parents of known genotypes and the probability of each combination.
2. Dominant traits include widow's peaks, dimples, and freckles.
3. Genetic disorders caused by dominant genes are uncommon because lethal dominant genes are always expressed and result in the death of the embryo, fetus, or child.
4. Recessive traits include some desirable conditions such as normal vision, but they also include most genetic disorders.
a. Recessive genetic disorders are more frequent than those caused by dominant genes, because individuals who are heterozygous for the trait do not express the trait, and can survive to reproduce, passing the trait to some of their offspring.
5. Some traits exhibit incomplete dominance, wherein the heterozygote has a phenotype intermediate between those of the homozygous dominant and the homozygous recessive.
6. In multiple-allele inheritance, while we inherit only two alleles for each gene, some genes exhibit more than two allele forms: These alleles may be codominant, in which case, both phenotypes are expressed.
7. Sex-linked traits are determined by genes on the sex chromosomes: These differences in expression occur because the Y chromosome is much smaller than the X chromosome and lacks many of the genes present on the X that code for nonsexual characteristics.
8. The Y chromosome is much smaller than the X chromosome and lacks many of the genes present on the X that code for nonsexual characteristics, such as red-green color blindness.
a. Genes found only on the X chromosome are said to be X-linked; those found only on the Y chromosome, such as those that code for maleness, are said to be Y-linked.
B. Polygene inheritance results in continuous or quantitative phenotypic variation between two extremes and depends on several gene pairs at different locations acting in tandem (an example of this is skin color). (p. 11012; Fig. 29.6)

29.4 Environmental factors may influence or override gene expression (p. 1112–1113)
A. Maternal factors, such as drugs or pathogens, may influence gene expression during development. (p. 1112)
B. Phenocopies are environmentally produced phenotypes that mimic conditions that may be caused by genetic mutations. (p. 1113)

C. Environmental factors may also influence genetic expression after birth, such as the effect of poor infant nutrition on brain growth, body development, and height. (p. 1113)

29.5 Factors other than nuclear DNA sequence can determine inheritance (pp. 1113–1114)

A. Nontraditional inheritance is the result of control mechanisms outside the coding portion of DNA and of the chromosome entirely. (p. 1113)

B. Beyond DNA: Regulation of Gene Expression (pp. 1113–1114)

1. RNA-only genes are found throughout the non-protein coding DNA and code for RNAs that regulate gene expression.
2. MicroRNAs (miRNAs) and short interfering RNAs (siRNAs) can help control transposons by disabling or hyperactivating them.
3. Much of the individual complexity seen is the result of small RNAs that control gene expression.
4. Epigenetic marks, proteins, and chemical groups that bind to and around DNA determine which areas in the DNA are ready for transcription or are silenced.
5. Genomic imprinting somehow tags genes during gametogenesis as either paternal or maternal and confers important functional differences in the resulting embryo.

C. Extranuclear (mitochondrial) inheritance is based on mitochondrial genes that are transmitted to the offspring almost exclusively by the mother. (p. 1114)

29.6 Genetic screening is used to determine or predict genetic disorders (pp. 1114–1116; Figs. 29.7–29.8)

A. There are two main ways to identify carriers of detrimental genes: pedigrees, which trace a trait through generations, or blood tests. (p. 1114; Fig. 29.7)

B. Fetal testing is used when there is a known risk of a genetic disorder. (pp. 1115–1116; Fig. 29.8)

1. The most common type of fetal testing is amniocentesis, in which a needle is inserted into the amniotic sac to withdraw amniotic fluid for testing.
2. Chorionic villi sampling (CVS) suctions off bits of the chorionic villi from the placenta for examination.

C. Human gene therapy and genetic engineering have the potential to alleviate or even cure diseases, especially those traced to one defective gene. (p. 1116)

1. Genes may be inserted into viruses and used to infect body cells, or corrected DNA can be inserted directly into cells.