13
GENES, CHROMOSOMES, AND HUMAN GENETICS

Chapter Outline

13.1 GENETIC LINKAGE AND RECOMBINATION
   The principles of linkage and recombination were determined with *Drosophila*
   Recombination frequency can be used to map chromosomes
   Widely separated linked genes assort independently

13.2 SEX-LINKED GENES
   Females are XX and males are XY in both humans and fruit flies
   Human sex determination depends on the Y chromosome
   Sex-linked genes were first discovered in *Drosophila*
   Sex-linked genes in humans are inherited as they are in *Drosophila*
   Inactivation of one X-chromosome evens out gene effects in mammalian females

13.3 CHROMOSOMAL ALTERATIONS THAT AFFECT INHERITANCE
   Deletions, duplications, translocations, and inversions are the most common chromosomal alterations
   The number of entire chromosomes may also change

13.4 HUMAN GENETICS AND GENETIC COUNSELING
   In autosomal recessive inheritance, heterozygotes are carriers and homozygous recessives are affected by the trait.
   In autosomal dominant inheritance, only homozygous recessives are unaffected
   Males are more likely to be affected by X-linked recessive traits
   Human genetic disorders can be predicted, and many can be treated

13.5 NONTRADITIONAL PATTERNS OF INHERITANCE
   Cytoplasmic inheritance follows the pattern of inheritance of mitochondria or chloroplasts
   In genomic imprinting, the allele inherited from one of the parents is expressed while the other allele is silent

Learning Objectives

After reading the chapter, you should be able to:

1. Explain genetic linkage and the causes for two genes to be linked or unlinked.
2. Understand genetic patterns of inheritance based on sex chromosomes.
3. Understand the importance of X-chromosome inactivation.
4. Discuss large scale chromosomal changes, and how they.
5. Discuss how the environment contributes to variations in gene expression.
6. Provide all the factors contributing to an individual’s phenotypic expression.
Key Terms

linked genes
linkage
parental phenotype
recombinant phenotype
genetic recombination
recombination frequency
linkage map
map unit
centimorgan
sex-linked genes
autosomes
autosomes
X chromosome

Y chromosome
pedigree
carrier
Barr body
deletion
duplcaton
translocation
inversion
nondisjunction
aneuploids
euploids
polyploids
autosomal recessive inheritance
autosomal dominant inheritance
X-linked recessive inheritance
genetic counseling
parental diagnosis
amniocentesis
chorionic villus sampling
genetic screening
ctoplasmic inheritance
genomic imprinting
loss of imprinting
dihybrid
dihybrid cross
independent assortment
Principle of Independent Assortment
chromosome theory of inheritance
locus
incomplete dominance
codominance
multiple alleles
epistasis
polygenic inheritance
pleiotropy

Lecture Outline

13.1 Genetic Linkage and Recombination
A. Gregor Mendal used garden peas to study seven different genes, and he concluded that they assorted independently.
B. Genes on the same chromosome may not assort independently and are called linked genes.
C. Linkage principle and recombination were determined with Drosophila.
   1. Thomas Hunt Morgan used the fruit fly as a model organism and performed test crosses to analyze genetics (Figure 13.2, step 1).
   2. Symbols developed are still being used (Figure 13.2, step 2).
   3. Some had parental phenotypes, and others developed recombinant or mixed phenotypes caused by genetic crossover (Figure 11.6).
   4. These rules apply to purple-vestigial crosses (Figure 13.3).
   5. Dihybrid crosses produce four types of gametes (Figure 13.3, step 1).
D. Recombination frequency can be used to map the location of genes on a chromosome.
   1. The percentage of test-cross progeny that are recombinants is indicative of the distance between the two genes in question (Figure 13.3).
   2. A linkage map is created using percent frequencies, but they are not always exact (Figure 13.4).
E. Widely separated linked genes assort independently due to the increased likelihood of recombination somewhere between the two genes (Figure 13.5).

Focus on Research: Model Research Organisms: The Marvelous Fruit Fly, Drosophila melanogaster
A. Reasons that fruit flies are good models for genetics include: easy to cultivate, sex easily determined, rapid life cycle, large chromosomes, wide range of mutants, and easy to collect virgins.
B. This research has led to discoveries of sex-linked genes and an understanding of many birth defects.

13.2 Sex-Linked Genes
A. Sex-linked genes are located on the sex chromosomes and are, therefore, linked to the sex of the individual.
B. Females are XX and males are XY in both humans and fruit flies (Figure 13.6).
C. Human sex determination depends on the Y chromosome.
   1. Sexual traits, such as distribution of body fat and nonsexual color perception, are found on the sex chromosome.
   2. SRY is found on the Y chromosome and switches on maleness during development.
D. Sex-linked genes were first found in the fruit fly (Figure 13.7, 13.8a, and 13.8b).
E. Sex-linked genes in humans are inherited as they are in *Drosophila*.
   1. Color blindness (red-green) is more common in males because females would have to receive two copies of the recessive gene.
   2. Hemophilia is a genetic disorder found in royalty (Figure 13.9).

F. Inactivation of one X chromosome evens out gene effects in mammalian females.
   1. Dosage compensation inactivates one of the two X chromosomes in most body cells.
   2. Inactivated X chromosomes show up as small dark Barr bodies in the cells.
   3. Inactivation is random and can result in patches of activity easily recognized in calico cats (Figure 13.10).

13.3 Chromosomal Alterations That Affect Inheritance
A. Chromosome alterations that affect inheritance result from breaks in the DNA.
B. Deletions, duplications, translocations, and inversions are the most common chromosomal alterations (Figure 13.11).
C. The number of entire chromosomes may also change (Figure 13.12).
   1. Nondisjunction is the failure of homologous parts of chromosomes to separate during meiotic division.
   2. Aneuploids have extra or missing chromosomes and euploids retain a normal complement number.
   3. Triploids have three copies for each chromosome, and tetraploids have four.
   4. An extra copy of chromosome 21 causes Down syndrome (Figure 13.13a and b).
   5. Sex chromosomes can be aneuploid, and typically if a Y chromosome is present the individual will be male (Figure 13.14 and Table 13.1).
   6. Polyploids are often fatal in animals but in plants result in hardier plants.

13.4 Human Genetics and Genetic Counseling
A. Autosomal inheritance makes heterozygotes carriers, and homozygotes are affected by the trait (Table 13.2).
B. Sickle-cell anemia affects 10–15% of African Americans.
C. Autosomal dominants have only the homozygous recessives unaffected.
D. Males are more likely to be affected by X-linked recessive traits.
E. Human genetic disorders can be predicted and many treated.
   1. Amniocentesis and genetic testing can predict disorders.
   2. Treatment usually involves enzyme or hormone replacement.

*Insights from the Molecular Revolution:* Achondroplastic Dwarfing by a Single Amino Acid Change
A. Achondroplasia, a type of dwarfing, is caused by a dominant allele on chromosome 4.
B. One amino acid difference in a receptor for fibroblast growth factor causes this disorder.

13.5 Nontraditional Patterns of Inheritance
A. Cytoplasmic inheritance follows the pattern of inheritance of mitochondria or chloroplasts.
   1. Mendelian segregation is not found because genes in these organelles do not use meiosis.
   2. Mitochondria generally are always inherited from the maternal parent.
   3. In humans, several inherited diseases are mitochondrial gene mutations (Table 13.3).
B. Genomic imprinting results in one allele being silent.
   1. Angleman syndrome (AS) results when paternally derived chromosome 15 and a mutated maternally derived one are inherited.
   2. Prader-Willi syndrome (PWS) results from a normal maternal chromosome 15 and a mutant maternal chromosome.
   3. Loss of imprinting is the cause of some types of cancer.