The Chromosomal Basis Of Inheritance
Chapter 15

• Objectives
  • Explain the chromosomal theory of inheritance and its discovery.
  • Explain why sex-linked diseases are more common in human males than females.
  • Distinguish between sex-linked genes and linked genes.
  • Explain how meiosis accounts for recombinant phenotypes.
  • Explain how linkage maps are constructed.
  • Explain how nondisjunction can lead to aneuploidy.
  • Define trisomy, triploidy, and polyploidy.
  • Distinguish among deletions, duplications, inversions, and translocations.

• Explain genomic imprinting.
• Explain why extranuclear genes are not inherited in a Mendelian fashion.
Locating Genes on Chromosomes

- Genes are located on chromosomes and can be visualized using certain staining techniques

The Behavior of Chromosomes Explains Mendelian Inheritance

- Several researchers proposed in the early 1900s that genes are located on chromosomes
- The behavior of chromosomes during meiosis was said to account for Mendel’s laws of segregation and independent assortment
  - homologous pairs of chromosomes explains the principle of segregation
  - multiple sets of homologous chromosomes explains the principle of independent assortment
Chromosomes and Inheritance

- The chromosome theory of inheritance states that Mendelian genes have specific loci on chromosomes
  - Chromosomes undergo segregation and independent assortment

- Thomas Hunt Morgan provided convincing evidence that chromosomes are the location of Mendel’s heritable factors
- Morgan worked with fruit flies because they breed at a high rate
  - A new generation can be bred every two weeks
  - They also have only four pairs of chromosome
• Morgan first observed and noted wild type, or normal, phenotypes that were common in the fly populations.
  – Traits alternative to the wild type are called mutant phenotypes.
• In one experiment Morgan mated male flies with white eyes (mutant) with female flies with red eyes (wild type)
  – The F₁ generation all had red eyes
  – The F₂ generation showed the 3:1 red:white eye ratio, but only males had white eyes
• Morgan determined that the white-eye mutant allele must be located on the X chromosome

• Morgan’s discovery that transmission of the X chromosome in fruit flies correlates with inheritance of the eye-color trait was the first solid evidence indicating that a specific gene is associated with a specific chromosome
Sex-linked genes exhibit unique patterns of inheritance

- The Chromosomal Basis of Sex
  - An organism’s sex is an inherited phenotypic character determined by the presence or absence of certain chromosomes
- In humans and other mammals there are two varieties of sex chromosomes, X and Y
  - Different systems of sex determination are found in other organisms

Inheritance of Sex-Linked Genes

- The sex chromosomes have genes for many characters unrelated to sex
- A gene located on either sex chromosome is called a sex-linked gene
- Sex-linked genes follow specific patterns of inheritance
Some recessive alleles found on the X chromosome in humans cause certain types of disorders:
- Color blindness
- Duchenne muscular dystrophy
- Hemophilia

X inactivation in Female Mammals

- In mammalian females one of the two X chromosomes in each cell is randomly inactivated during embryonic development.
- If a female is heterozygous for a particular gene located on the X chromosome she will be a mosaic for that character.
Genes on Same Chromosome tend to be Inherited Together

• Each chromosome has hundreds or thousands of genes
  ‒ traits of characters located on the same chromosome tend to be inherited together
  ‒ described as linked traits
• When Morgan followed the inheritance of two characters he observed that some offspring have combinations of traits that do not match either parent in the P generation
Morgan determined that genes that are close together on the same chromosome are linked and do not assort independently. Unlinked genes are either on separate chromosomes or are far apart on the same chromosome and assort independently.

Recombination of Linked Genes: Crossing Over

Morgan proposed that some process must occasionally break the physical connection between genes on the same chromosome. Crossing over of homologous chromosomes was the mechanism.
• Recombinant offspring are those that show new combinations of the parental traits
  – When 50% of all offspring are recombinants geneticists say that there is a 50% frequency of recombination
  – Linked genes exhibit recombination frequencies less than 50%

Linkage Mapping

• A genetic map is an ordered list of the genetic loci along a particular chromosome
  – It can be developed using recombination frequencies
• A linkage map is the actual map of a chromosome based on recombination frequencies
  – The farther apart genes are on a chromosome the more likely they are to be separated during crossing over
• Many fruit fly genes were mapped initially using recombination frequencies
Changes in chromosome number or structure

• Large-scale chromosomal alterations often lead to spontaneous abortions or cause a variety of developmental disorders

Abnormal Chromosome Number

• When nondisjunction occurs pairs of homologous chromosomes do not separate normally during meiosis
  – Gametes contain two copies or no copies of a particular chromosome
• Aneuploidy results from the fertilization of gametes in which nondisjunction occurred
  – It is a condition in which offspring have an abnormal number of a particular chromosome
• If a zygote is trisomic it has three copies of a particular chromosome
• If a zygote is monosomic it has only one copy of a particular chromosome

• Polyploidy is a condition in which there are more than two complete sets of chromosomes in an organism
  – This is common in plants
    • many important agricultural plants are polyploid
  – This is relatively uncommon in animals
    • known to occur among fish and amphibians
    • two species of mammals may be tetraploid
Alterations of Chromosome Structure

- Breakage of a chromosome can lead to four types of changes in chromosome structure:
  - Deletion
  - Duplication
  - Inversion
  - Translocation

Human Disorders Due to Chromosomal Alterations

- Alterations of chromosome number and structure are associated with a number of serious human disorders.
- Down syndrome is usually the result of an extra chromosome 21, trisomy 21.
Aneuploidy of Sex Chromosomes

- Nondisjunction of sex chromosomes produces a variety of aneuploid conditions
- Klinefelter syndrome is the result of an extra chromosome in a male, producing XXY individuals
- Turner syndrome is the result of monosomy X, producing an X0 karyotype

Disorders Caused by Structurally Altered Chromosomes

- Cri du chat is a disorder caused by a deletion in chromosome 5
- Certain cancers are caused by translocations of chromosomes
  - Chronic myelogenous leukemia results from a translocation from chromosome 22 to chromosome 9
Exceptions to Chromosome-based Inheritance

- Some inheritance patterns are exceptions to the standard chromosome theory
- Two normal exceptions to Mendelian genetics include
  - Genes located in the nucleus
  - Genes located outside the nucleus

Genomic Imprinting

- In mammals the phenotypic effects of certain genes depend on which allele is inherited from the mother and which is inherited from the father
- Genomic imprinting involves the silencing of certain genes that are “stamped” with an imprint during gamete production
Inheritance of Organelle Genes

- Extranuclear genes are genes found in organelles in the cytoplasm.
- The inheritance of traits controlled by genes present in the chloroplasts or mitochondria depends solely on the maternal parent because the zygote’s cytoplasm comes from the egg.
  - Some diseases affecting the muscular and nervous systems are caused by defects in mitochondrial genes that prevent cells from making enough ATP.