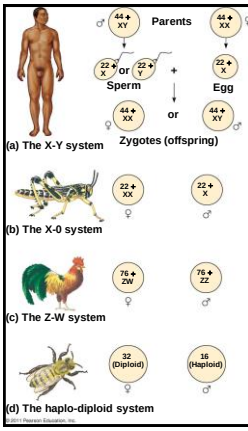
Chapter 15:
The Chromosomal Basis of Inheritance

1. Sex Linkage
2. Linked Genes
3. Chromosome Abnormalities

1. Sex Linkage

Chapter Reading – pp. 294-298

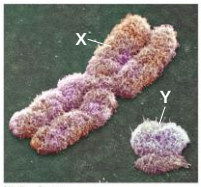


Sex Determination

Sex determination in mammals involves the X and Y chromosomes:

XX = female

XY = male



- **other members of the animal kingdom use slightly different systems**

Sex Linkage in Mammals

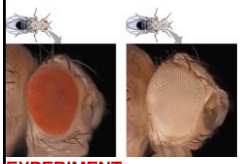
Sex linkage refers to inherited traits for which the inheritance pattern is different for males vs females.

Sex linkage involves genes on the X and Y chromosomes:

- **X-linked genes are located on the X chromosome**
- **Y-linked genes are located on the Y chromosome**

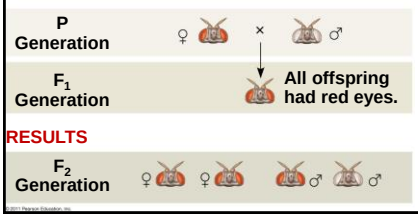
The vast majority of sex linked inheritance is due to X-linked genes since there are many more genes on the X chromosome (~1100) than the Y chromosome (~78)

Evidence for X-linked Genes



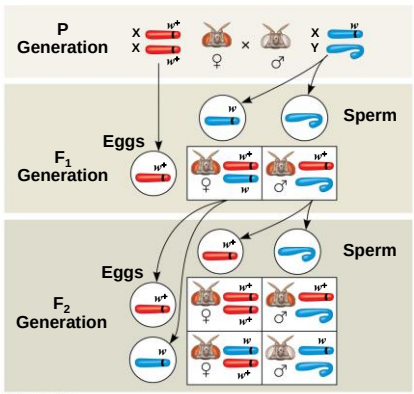
In 1910, Thomas Hunt Morgan discovered a Drosophila mutant phenotype that appears only in males.

EXPERIMENT

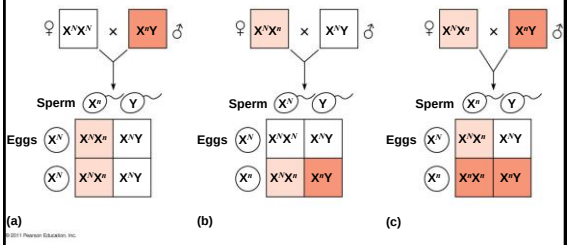


- **this mutation revealed both the chromosomal basis of inheritance and sex-linked inheritance involving the X chromosome**

CONCLUSION



Transmission of X-linked Traits



- females have 2 copies of each allele on the X chromosome and are homozygous or heterozygous
- males have 1 copy of each allele on the X chromosome and are hemizygous

An X-linked Human Trait

Genes on the X chromosome have a unique inheritance pattern in males:

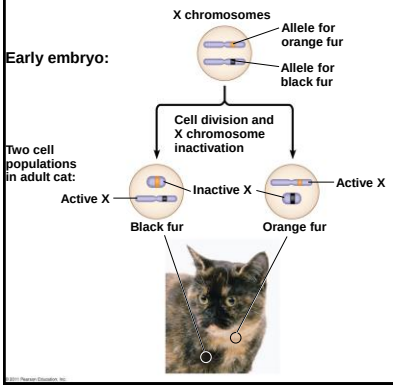
- only 1 allele, so no masking of recessive alleles
- X-linked alleles are always inherited from the mother (carrier)

e.g., Hemophilia is caused by a recessive X-linked allele (h):

♀ X^HX^h x ♂ X^HY

	X^H	Y
X^H	X^HX^H ♀	X^HY ♂
X^h	X^HX^h ♀	X^hY ♂

X Inactivation in Mammals

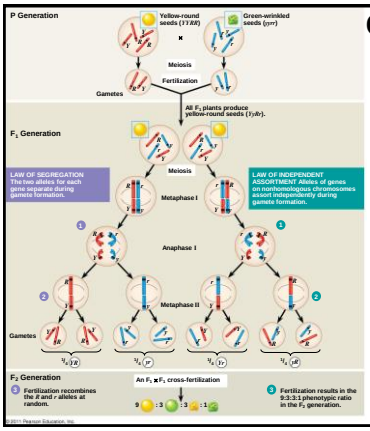


In females, the genes on only 1 X chromosome are expressed per cell, the other X chromosome is an inactive Barr body.

- during embryonic development, 1 X chromosome per cell is randomly inactivated
- all daughter cells inherit the same inactive chromosome

2. Linked Genes

Chapter Reading – pp. 292-294, 299-304



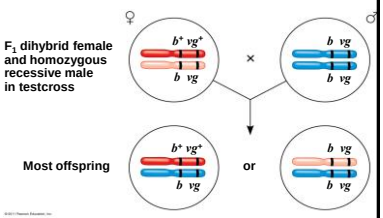
Chromosomal Basis of Mendel's Laws

- as we shall see, Mendel's "Law of Independent Assortment" does not apply to genes on the same chromosome

Linked Genes

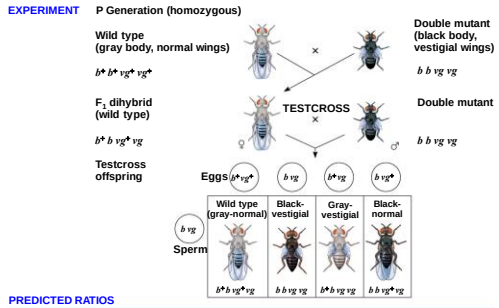
Genes on the same chromosome are said to be linked.

- alleles for linked genes will be inherited together unless crossing over during Prophase I occurs between them

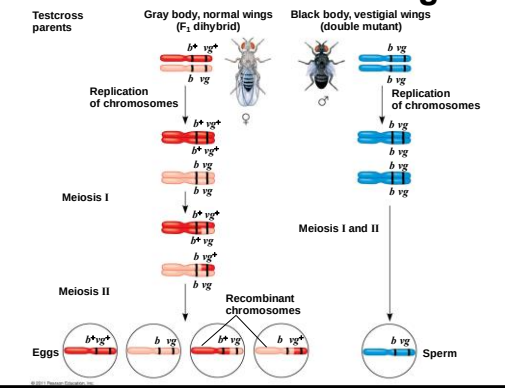


Gene linkage is indicated when actual ratios of phenotypes deviate from expected ratios, and revealed by doing a test cross…

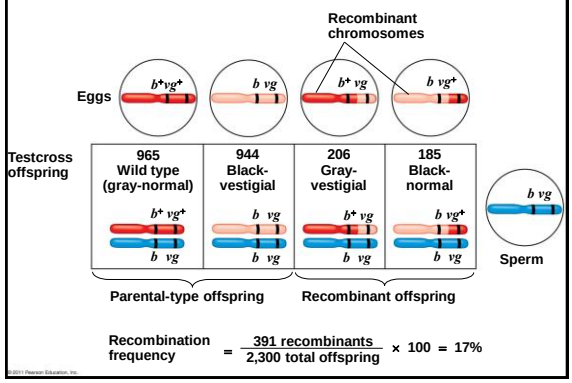
Example of Linked Genes



Test Cross reveals Linkage…



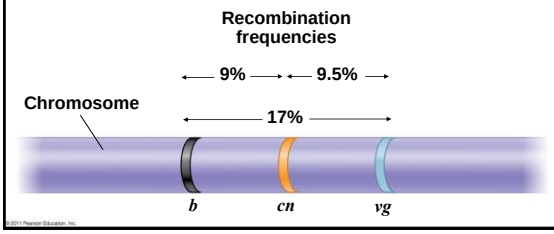
…results of Test Cross



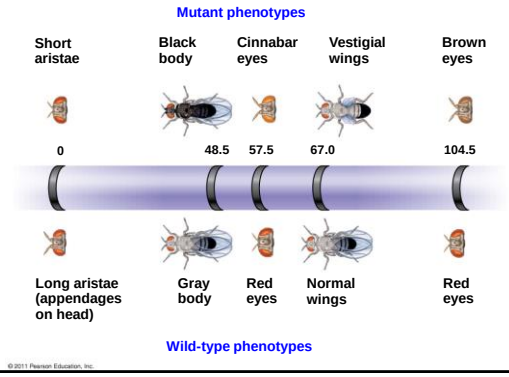
Using Recombination Frequencies to Map Genes

Frequency of recombinant chromosomes reflects the relative location of genes.

RESULTS



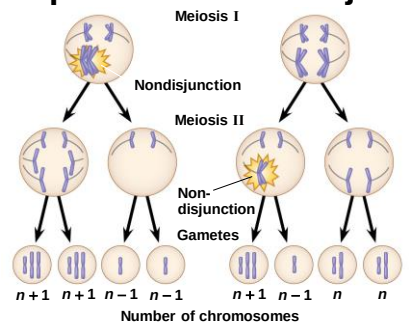
A Linkage Map in Drosophila



3. Chromosome Abnormalities

Chapter Reading – pp. 304-309

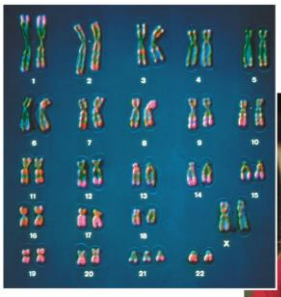
Consequences of Nondisjunction



(a) Nondisjunction of homologous chromosomes in meiosis I

(b) Nondisjunction of sister chromatids in meiosis II

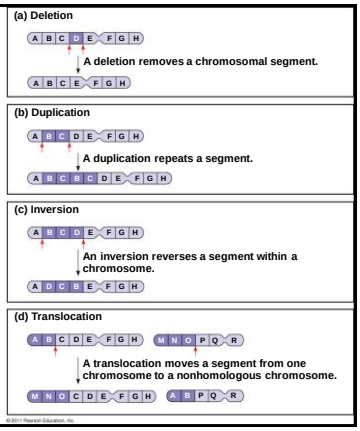
Down Syndrome



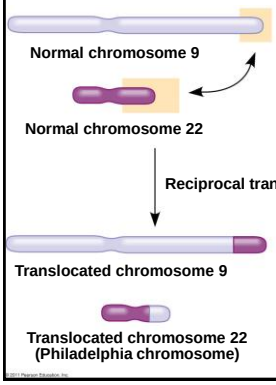
Trisomy 21

- example of aneuploidy
- due to nondisjunction

Other Chromosomal Aberrations



Chromosome Translocation



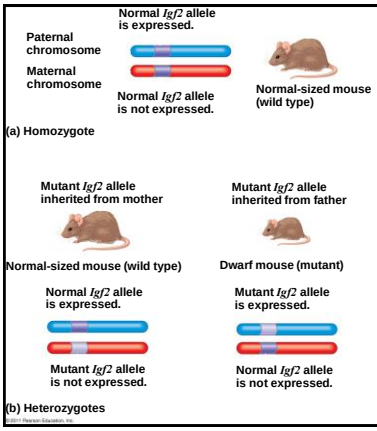
Translocation is due to the exchange of chromosomal fragments between non-homologous chromosomes

- **when the fusion occurs within a gene involved in cell cycle regulation, the result can be an increased likelihood of cancer**

Genomic Imprinting

Genomic imprinting is when only one allele is expressed for certain autosomal genes while the other is inactive.

- **occurs in <1% of mammalian genes**
- **inactive allele consistently comes from males or females, depending on the gene**
- **inactivation involves methylation of DNA in gene, modification of histones during gamete formation**



Example of Genomic Imprinting

In this example, the paternal allele will always be expressed whereas the maternal will be inactive

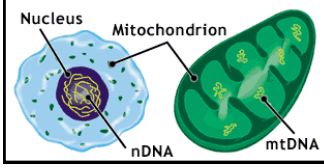
- **mutant phenotype will only appear when inherited from the father**

Mitochondrial Inheritance

Mitochondria contain their own small circular chromosome containing genes involving in mitochondrial gene expression, energy metabolism.

- **human mitochondrial genome is only ~16,000 bp**

Mitochondria are inherited almost exclusively from the mother.



- **mutations in mitochondrial genes thus follow maternal inheritance and can affect energy metabolism**

Key Terms for Chapter 15

- **sex-linkage, X-linked, Y-linked, hemizygous**
- **X-inactivation, Barr body**
- **linked genes, linkage map**
- **nondisjunction, aneuploidy, polyploidy**
- **deletion, duplication, inversion, translocation**
- **genomic imprinting**

Relevant Chapter Questions
1-9, 12