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Chapter 15

The Chromosomal Basis of Inheritance

PowerPoint® Lecture Presentations for

Biology

Eighth Edition

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Lectures by Chris Romero, updated by Erin Barley with contributions from Joan Sharp

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What we've learned since Mendel: Chromosomes

- Mendel's laws described in 1860
- Mitosis and meiosis described in the late 1800's
- The chromosome theory of inheritance formed in the early 1900's:
  - Mendelian genes have specific loci on chromosomes
  - Chromosomes undergo segregation and independent assortment

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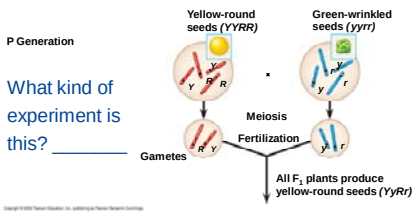
Fig. 15-2a



What kind of experiment is this? _____

(Peas have 7 pairs of chromosomes.)

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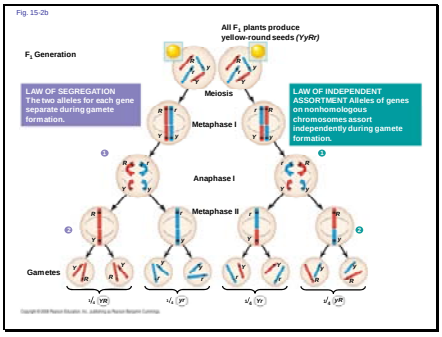

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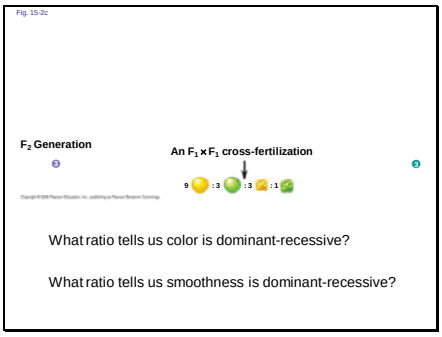

What ratio tells us color is dominant-recessive?

What ratio tells us smoothness is dominant-recessive?

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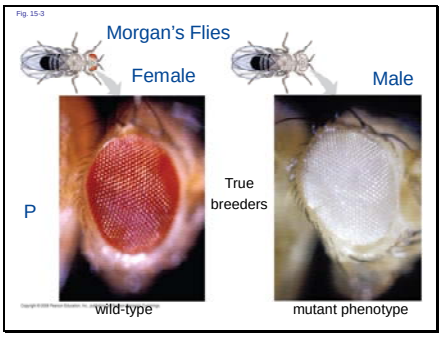

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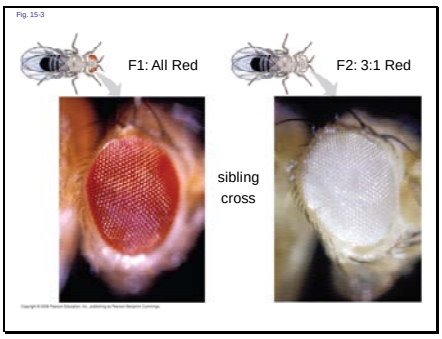

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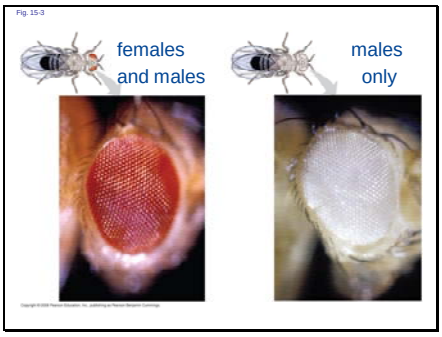

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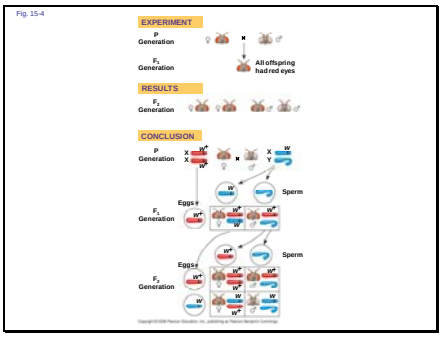

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The Chromosomal Basis of Sex

- In humans and some other animals, there is a chromosomal basis of sex determination
- In humans and other mammals, there are two varieties of sex chromosomes: a larger X chromosome and a smaller Y chromosome
- Only the ends of Y have regions that are homologous with X
- The *SRY* gene on Y codes for the development of testes

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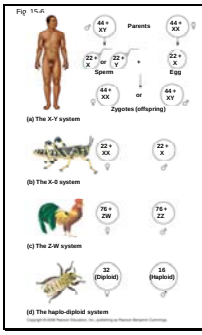

- Females are XX, and males are XY
- Each ovum contains an X chromosome, while a sperm may contain either an X or a Y chromosome
- Other animals have different methods of sex determination

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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**
- In humans, sex-linked usually refers to a gene on the larger X chromosome
- Sex-linked genes follow specific patterns of inheritance

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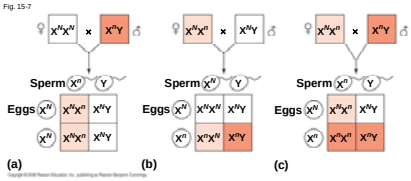

Example - Colorblindness: N = normal color vision; n = colorblind
White = unaffected; light orange = carriers; dark orange = colorblind

- For a recessive sex-linked trait to be expressed
  - A female needs _____ copy/copies of the allele
  - A male needs _____ copy/copies of the allele
- Sex-linked recessive disorders are much more common in _____.

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Some disorders caused by recessive alleles on the X chromosome in humans:

- Color blindness
- **Duchenne muscular dystrophy**
- **Hemophilia**

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X Inactivation in Female Mammals

- In mammalian females, one of the two X chromosomes in each cell is randomly inactivated during embryonic development
- The inactive X condenses into a **Barr body**
- What modification to DNA causes the condensation of one X chromosome?
- If a female is heterozygous for a particular gene located on the X chromosome, she will be a mosaic for that character

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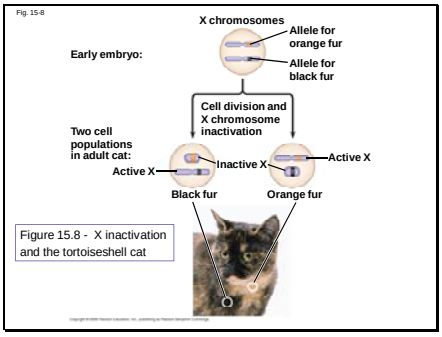

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Concept 15.3: Linked genes tend to be inherited together because they are located near each other on the same chromosome

- Each chromosome has hundreds or thousands of genes
- Genes located on the same chromosome that tend to be inherited together are called **linked genes**

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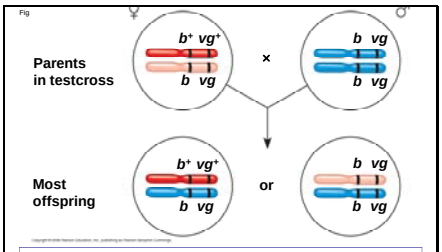

- Morgan found that Drosophila body color and wing size are usually inherited together in specific combinations (parental phenotypes)
- He noted that these genes do not assort independently, and reasoned that they were on the same chromosome

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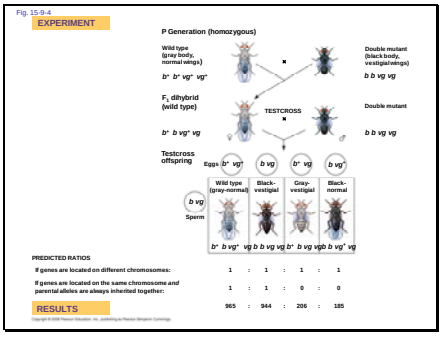

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- However, nonparental phenotypes were also produced from Morgan's Drosophila crosses.
- Understanding this result involves exploring **genetic recombination**, the production of offspring with combinations of traits differing from either parent
- The genetic findings of Mendel and Morgan relate to the chromosomal basis of recombination

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Recombination of Linked Genes: Crossing Over

- Morgan discovered that genes can be linked, but the linkage was incomplete, as evident from recombinant phenotypes
- Morgan proposed that some process must sometimes break the physical connection between genes on the same chromosome
- That mechanism was the **crossing over** of homologous chromosomes

http://www.sinauer.com/cooper/4e/animations0602.html - animation

PLAY Animation: Crossing Over

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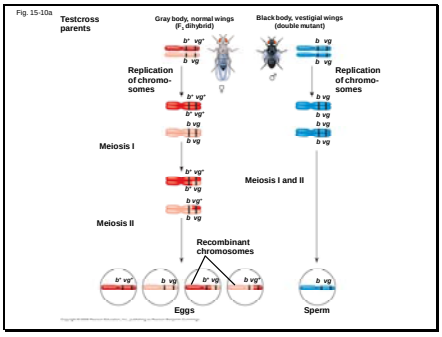

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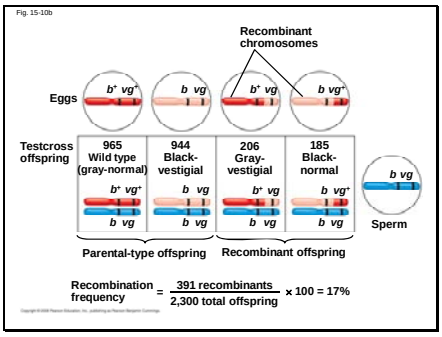

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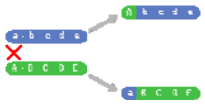
Mapping the Distance Between Genes Using Recombination Data: *Scientific Inquiry*

- Alfred Sturtevant, one of Morgan's students, constructed a **genetic map**, an ordered list of the genetic loci along a particular chromosome
- Sturtevant predicted that *the farther apart two genes are, the higher the probability that a crossover will occur between them and therefore the higher the recombination frequency*



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- A **linkage map** is a genetic map of a chromosome based on recombination frequencies
- Distances between genes can be expressed as **map units**; one map unit, or centimorgan, represents a 1% recombination frequency
- Map units indicate relative distance and order, not precise locations of genes

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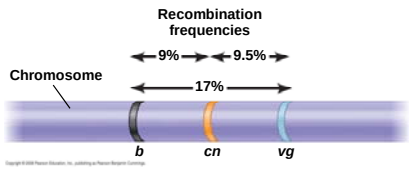

• Genes that are far apart on the same chromosome can have a recombination frequency near 50%

• Such genes are physically linked, but genetically unlinked, and behave as if found on different chromosomes

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Concept 15.4: Alterations of chromosome number or structure cause some genetic disorders

- Large-scale chromosomal alterations often lead to spontaneous abortions (miscarriages) or cause a variety of developmental disorders
- In **nondisjunction**, pairs of homologous chromosomes do not separate normally during meiosis
- As a result, one gamete receives two of the same type of chromosome, and another gamete receives no copy

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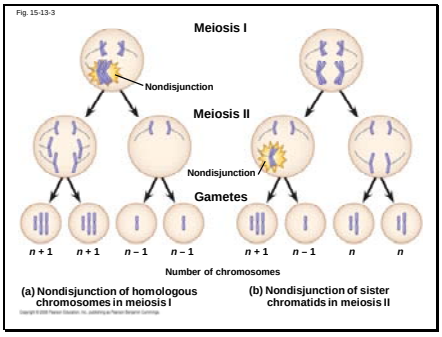

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- **Aneuploidy** results from the fertilization of gametes in which nondisjunction occurred
- Offspring with this condition have an abnormal number of a particular chromosome
- A **monosomic** zygote has only one copy of a particular chromosome
- A **trisomic** zygote has three copies of a particular chromosome

Down Syndrome
(Trisomy 21)

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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
- Monosomy X, called *Turner syndrome*, produces X0 females, who are sterile; it is the only known viable monosomy in humans

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- **Polyploidy** is a condition in which an organism has more than two complete sets of chromosomes
  - Triploidy (3*n*) is three sets of chromosomes
  - Tetraploidy (4*n*) is four sets of chromosomes
- Polyploidy is common in plants, but not animals
- Polyploids are more normal in appearance than aneuploids

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Alterations of Chromosome Structure

- Breakage of a chromosome can lead to four types of changes in chromosome structure:
  - **Duplication** repeats a segment
  - **Translocation** moves a segment from one chromosome to another
  - **Inversion** reverses a segment within a chromosome
  - **Deletion** removes a chromosomal segment

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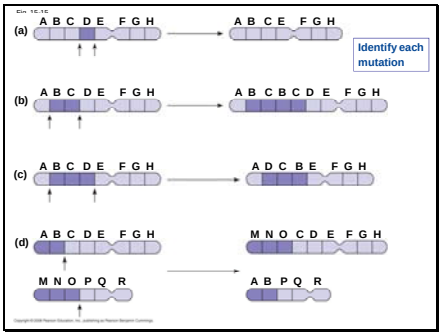

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Disorders Caused by Structurally Altered Chromosomes

- The syndrome *cri du chat* ("cry of the cat"), results from a specific deletion in chromosome 5

http://learn.genetics.utah.edu

- A child born with this syndrome is mentally retarded and has a catlike cry; individuals usually die in infancy or early childhood
- Certain cancers, including *chronic myelogenous leukemia* (CML), are caused by translocations of chromosomes

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Concept 15.5: Some inheritance patterns are exceptions to the standard chromosome theory

- There are two normal exceptions to Mendelian genetics
- One exception involves genes located in the nucleus, and the other exception involves genes located outside the nucleus

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Genomic Imprinting

- For a few mammalian traits, the phenotype depends on which parent passed along the alleles for those traits
- Such variation in phenotype is called **genomic imprinting**
- Genomic imprinting involves the silencing of certain genes that are"stamped" (epigenetically marked) with an imprint during gamete production (in the egg or sperm)

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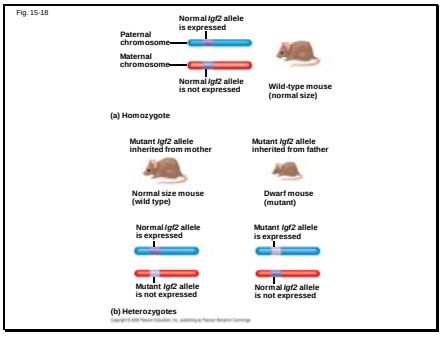

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- It appears that imprinting is the result of the methylation (addition of $–CH_3$) of DNA
- Genomic imprinting is thought to affect only a small fraction of mammalian genes
- Most imprinted genes are critical for embryonic development
- Diseases?

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Inheritance of Organelle Genes

- *Extranuclear genes* (or *cytoplasmic genes*) are genes found in organelles in the cytoplasm
- Mitochondria, chloroplasts, and other plant plastids carry small circular DNA molecules
- Extranuclear genes are inherited maternally because the zygote's cytoplasm comes from the egg

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