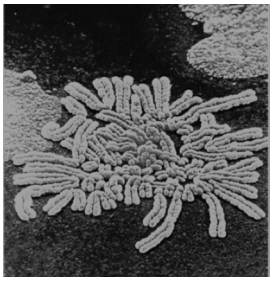
Chapter 15
Chromosomes, Mapping, and the Meiosis - Inheritance Connection



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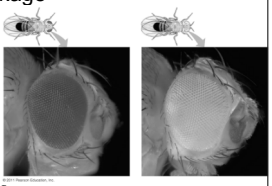
EXTENSIONS (not really EXCEPTIONS)
Sex Linkage



Drosophila melanogaster
fruit fly species
eats fungi on fruit

generation time 2 weeks

Fruit flies -three pairs of autosomes
and a pair of sex chromosomes
(XX in females, XY in males).

Thomas Hunt Morgan discovered a mutant male fly with white eyes
Instead of the usual red eyes (wild type).

Could this new trait be inherited in a Mendelian fashion?

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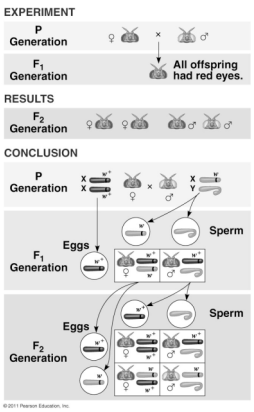
Results :

F1 progeny = red dominant

F2 progeny = actually >3:1 overall

all white eyed F2 flies were males!

all females and ½ males are wild type



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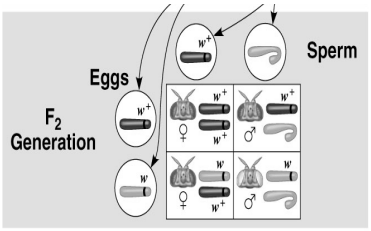
Possible explanation for F2 results = white eyed female flies don't exist? Not viable?



Test cross shows white eyed females can exist

Need alternative explanation – eye color LINKED TO SEX

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X-linked

- Gene with the white-eyed mutation is on the X chromosome alone
  - Males with mutation have white eyes because have only 1 X chromosome
  - For females to have white eyes, must have a mutation on both chromosomes.
- Why not on Y chromosome?
  - Y chromosome in flies carries almost no functional genes

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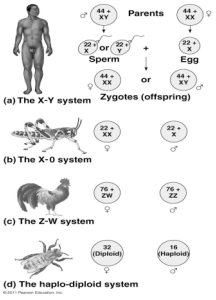
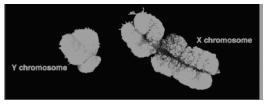
Sex Chromosomes and Sex Determination

Structure and # sex chromosomes vary in different species



autosomes- = all other chromosomes besides sex chromosomes

In humans, Y chromosome determines "maleness"
-very condensed



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- Default = female?
  - SRY gene on Y Chromosome responsible for "maleness"

EXCEPTIONS

-movement of a part of the Y chromosome to the X chromosome = male

-Androgen insensitivity syndrome – XY individuals develop as female because body fails to respond to androgens (female hormones)

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Human Genetic
Disorders & Sex Linkage

Affects males more than females

- Red-green color blindness—ability to absorb the different-colored wavelengths in light.
- Hemophilia—blood-clotting protein (Factor VIII)
- Allele for hemophilia introduced into European royal families by Queen Victoria of England
  - in 5 generations after Victoria, 10 males decedents had hemophilia

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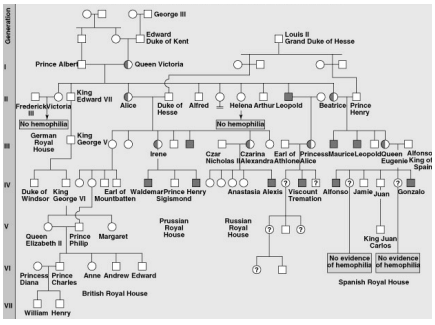

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Human Chromosomes

- Human somatic cells normally have 23 pairs of chromosomes.
  - divided into seven groups characterized by size and shape
  - 22 pairs of autosomes
  - 1 pair of sex chromosomes
    - XX = Female
    - XY = Male

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- **Value of Having Two X Chromosomes**
  - X chromosome that male gets from his mother determines phenotype
  - Diseases caused by X-linked recessive alleles more common in males than females
  - Women have two X chromosomes, so can be heterozygous but still have normal color vision

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Dosage compensation- one of X chromosomes in females inactivated early in development

Ensures equal level of expression from sex chromosomes

X inactivation Random

Inactivated chromosome is highly condensed and is visible as darkly staining Barr body attached to nuclear membrane

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RANDOM X inactivation

- female mammals inherit two X chromosomes, BUT only ONE is active
  - one X chromosome in each cell condenses into a **Barr body**
  - its genes are inactivated
- The condensed Barr body chromosome is reactivated in ovarian cells that produce ova

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- Mary Lyon- Barr body occurs randomly and independently in embryonic cells at the time of X inactivation
- females are mosaics -some active paternal X, others active maternal X
  - After Barr body formation, all descendent cells have the same inactive X
  - If female is heterozygous for sex-linked trait, approximately half her cells will express one allele and other half will express other allele

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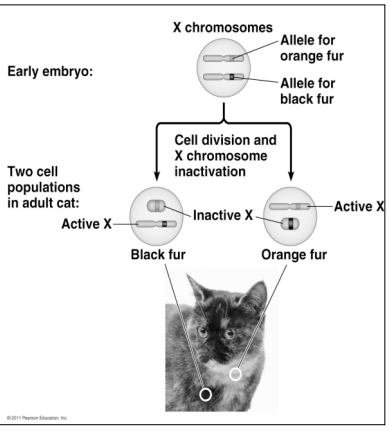
X inactivation = genetic mosaics

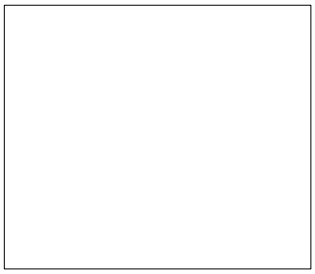
cells express different alleles, depending on which chromosome inactivated

also epistasis!

Expressed gene is epistasic suppressed is hypostatic)



Genetic Maps from Human Genome



- Anonymous markers- molecular techniques
  - no detectable phenotype
- Single Nucleotide Polymorphims (SNPs)
  - polymorphisms – any differences b/w individuals in a population
  - (2 million!!!)
- SNPs- affect single base of a gene loci

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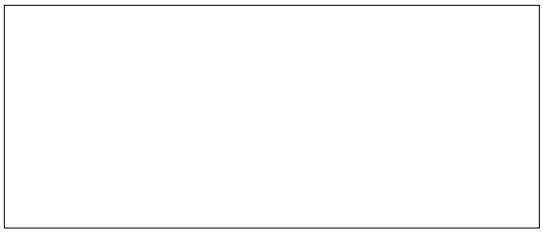

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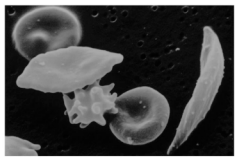
Gene Disorders Due to Protein Alteration

- Sickle-cell anemia recessive
- defective hemoglobin, unable to properly transport oxygen
- Homozygotes have sickle-cell.
  - Heterozygotes usually appear normal, but are resistant to malaria.



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Sickle cell disease

- 1 in 400 African Americans
- Single amino acid substitution!
- Hemoglobin deforms
- Carriers healthy – except under stress
- Carriers – alleles ar COdominant
- Benefit? Malaria resistance
- Homozygous normal – die of malaria
- Homozygous recessive die of sickle cell anemia
- Carriers - OK

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Nondisjunction of Chromosomes

Failure of homologues (sister chromatids) to separate during meiosis = nondisjunction

Leads to gain or loss chromosome - aneuploidy

humans who've lost a copy of an autosome - monosomics

gained - trisomics

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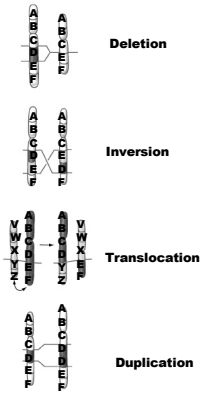
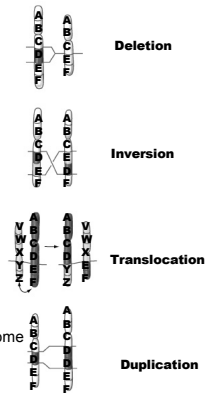


Chromosome 5
Cri du Chat

Chromosome 16
Form of leukemia

Chromosome 9
Normal, common

Chromosome 17
Charcot Marie Tooth syndrome
Muscles, touch sensation



Exceptions to Chromosomal Theory of Inheritance

- MITOCHONDRIA from mom
  - Organelles usually come from mom; ALL mitochondria come from egg
  - Maternal Inheritance
- Chloroplast genes
  - Usually maternal, depends on species

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