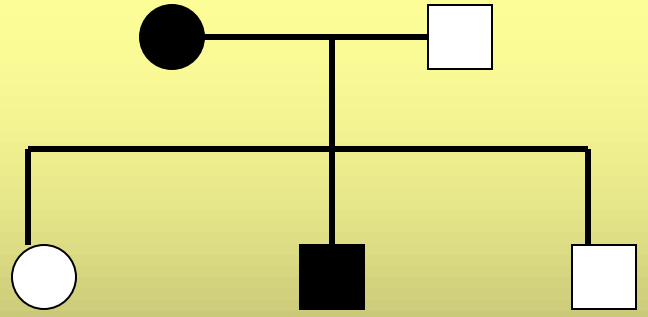
Chapter 15
The Chromosomal Basis of Inheritance

Constructing a Pedigree

- Use the appropriate symbols:
  - Unaffected Male
  - Unaffected Female
  - Affected Male
  - Affected Female
  - Male carrier of trait
  - Mating of Offspring



2. Label each generation down the left hand side of your pedigree.
3. Label each individual in your pedigree with his or her name.
4. It is easiest to construct your pedigree working from the most recent generation backwards.

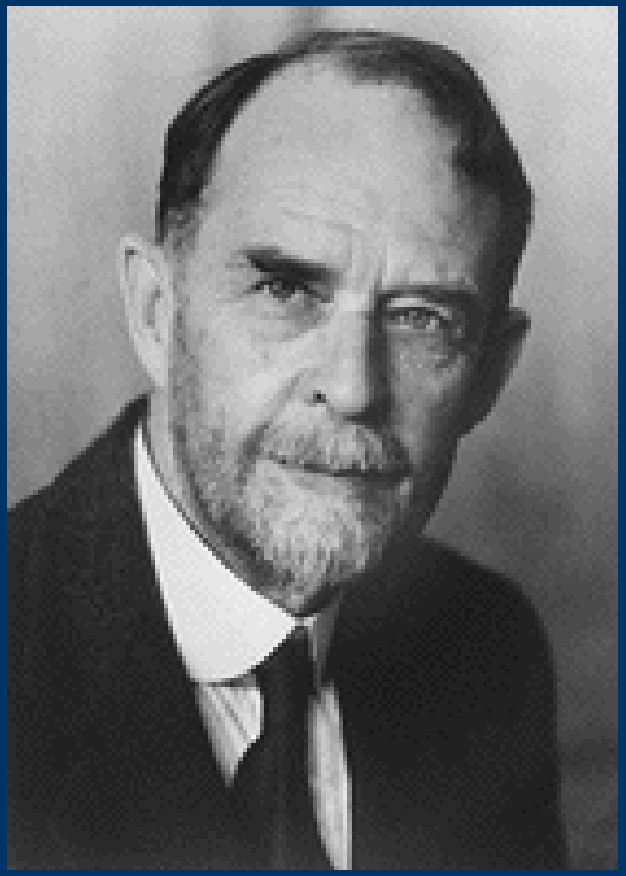
Chromosome theory of Inheritance

* The unifying theory stating that inheritance patterns may be generally explained by assuming that genes are located in specific sites on chromosomes and found in a linear sequence.

* Chromosomes undergo segregation and independent assortment

The Chromosome Theory

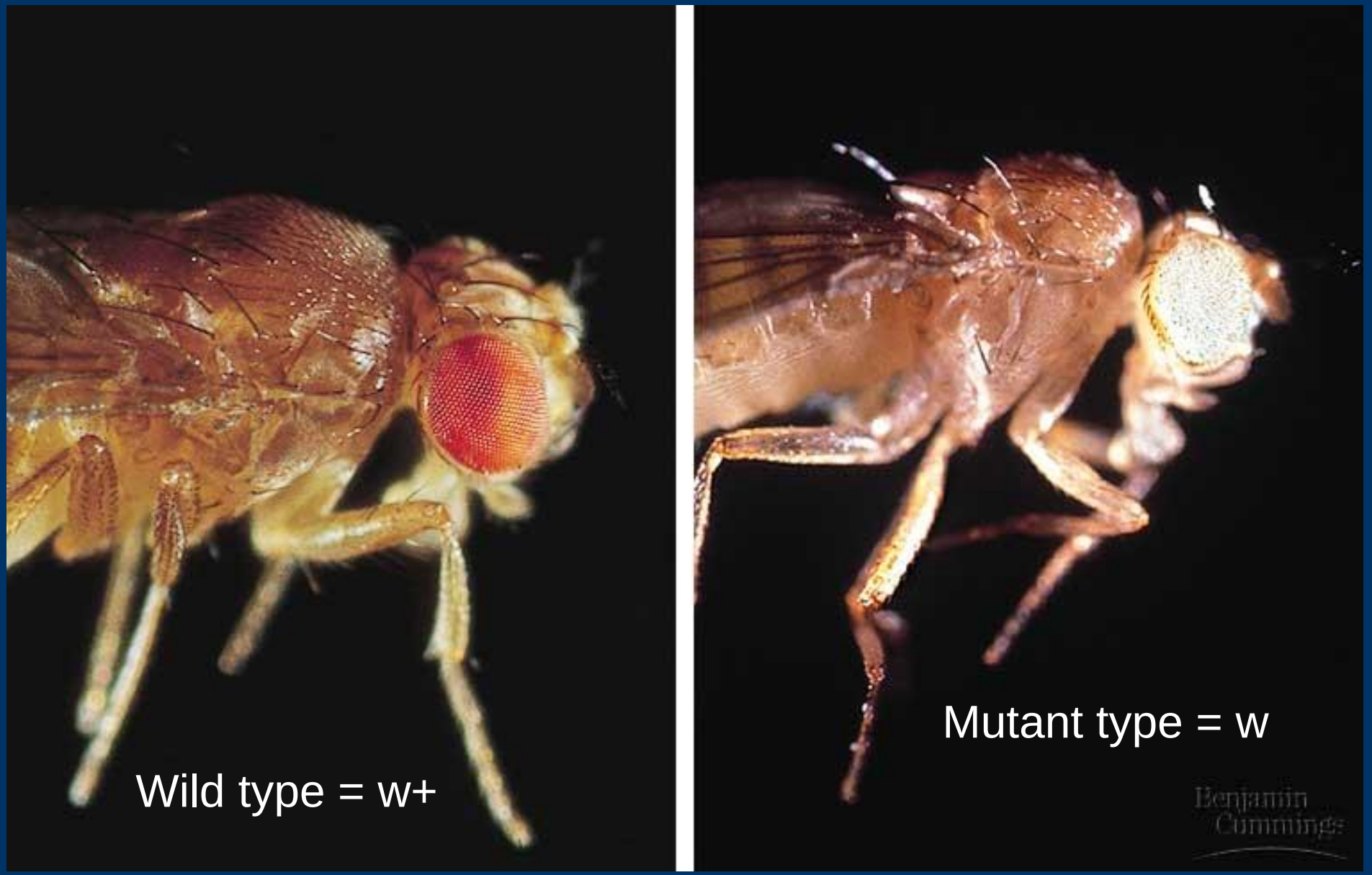
- Thomas Hunt Morgan worked with *Drosophila* (dew loving) and showed a connection between chromosomes and inheritance
- Experimental embryologist that provided evidence that chromosomes are the location of Mendel's heritable factors



Female

Male

Figure 15.2 Morgan's first mutant



Morgan's Experiment

- First took male white eyed fruit fly and mated it with a red eyed female
- F1 generation had all red eyes
- F2 generation had a 3:1 ratio of red to white eyes, but white eyes were only found in males, all females had red eyes, and half the males had red eyes
- Discovered that the gene for eye color in fruit flies was on the sex chromosome, specifically the X chromosome

Sex linked traits occur because…

they are determined by genes located (linked) on either the X or Y chromosome, but not on both.

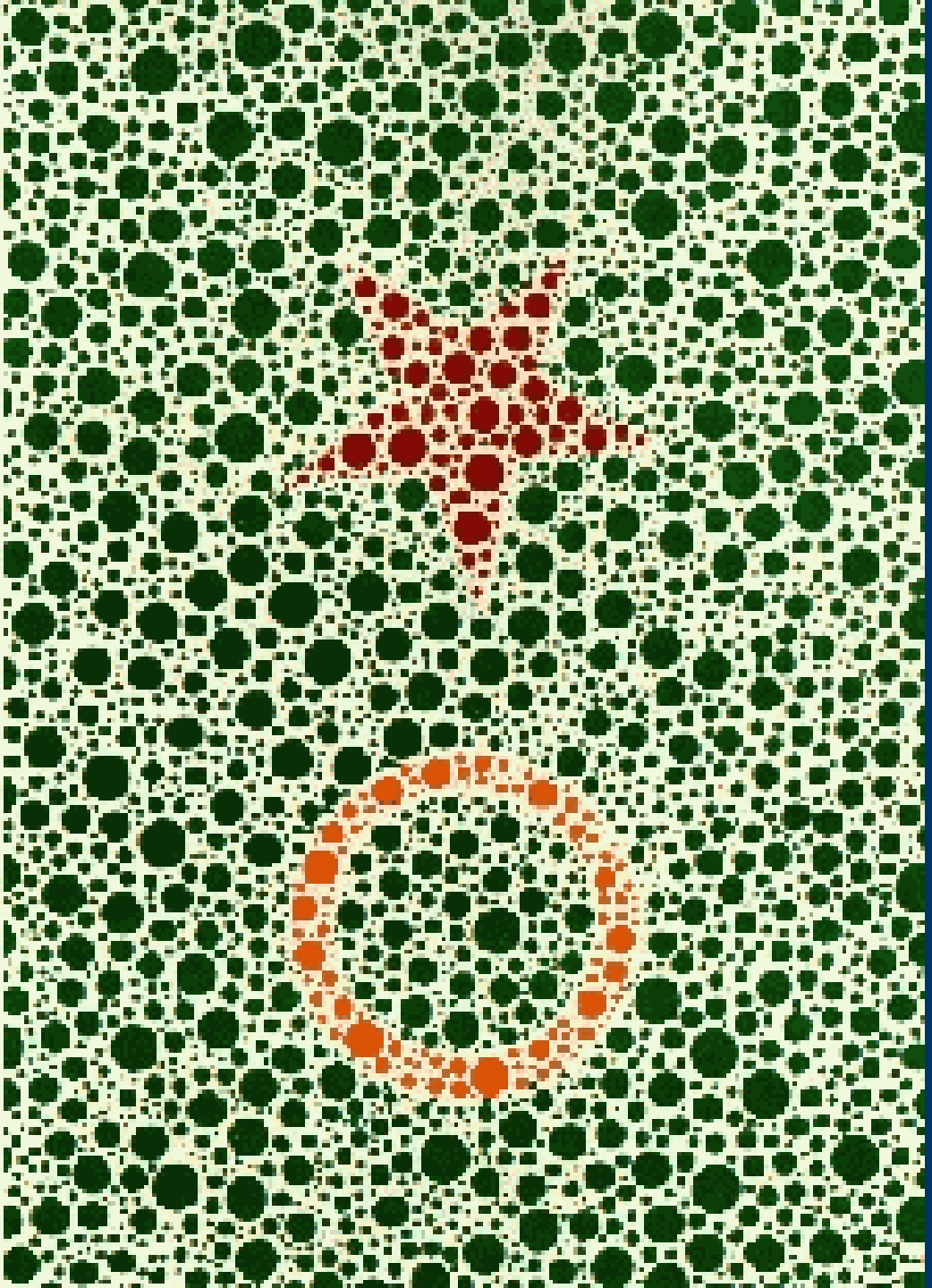

X
Y

Sex linked traits…

- Hemophilia: **an X-linked genetically inherited recessive disease in which one or more of the normal blood clotting factors is not produced.**
  - **Hemophilia most often afflicts males.**

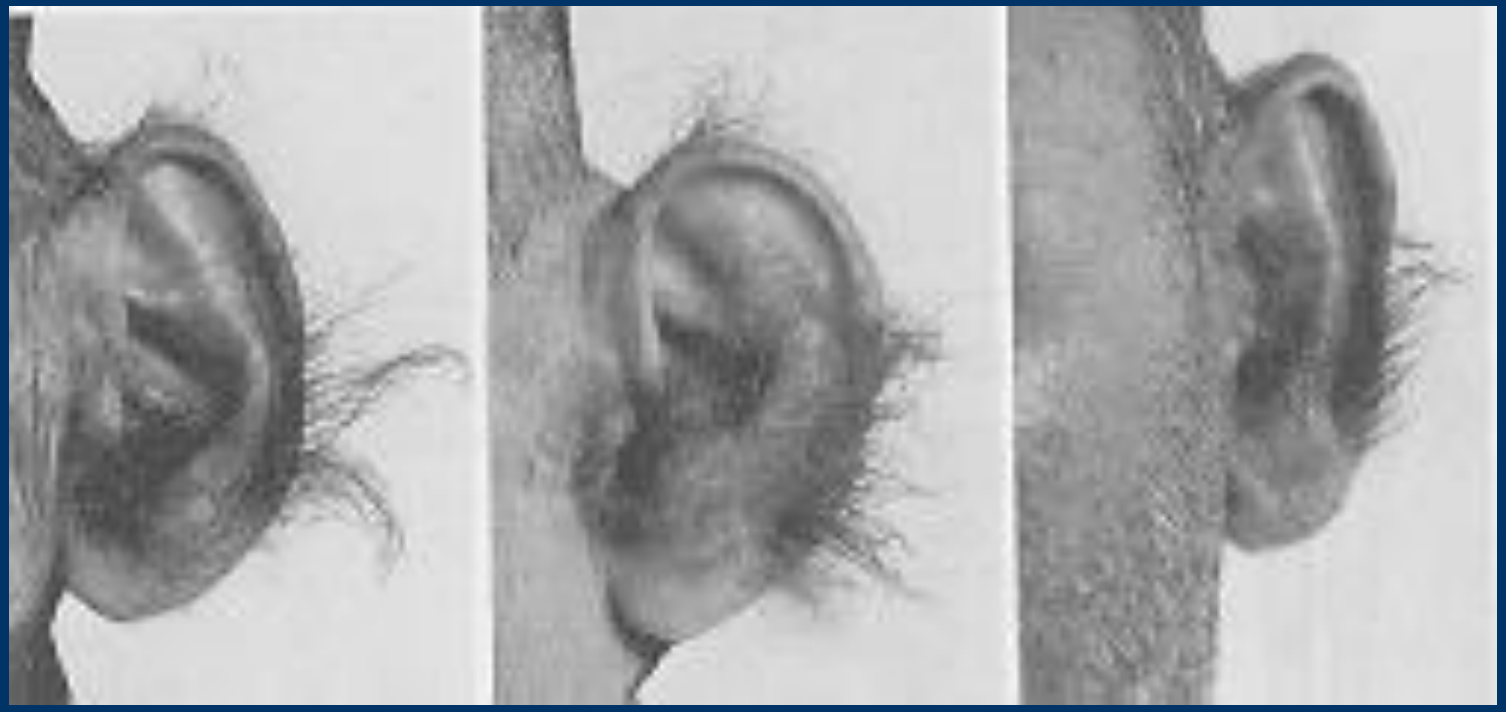
Other X - Linked traits

- Hemophilia
- Red-green color blindness
- Duchene muscular dystrophy
- Fragile X-syndrome

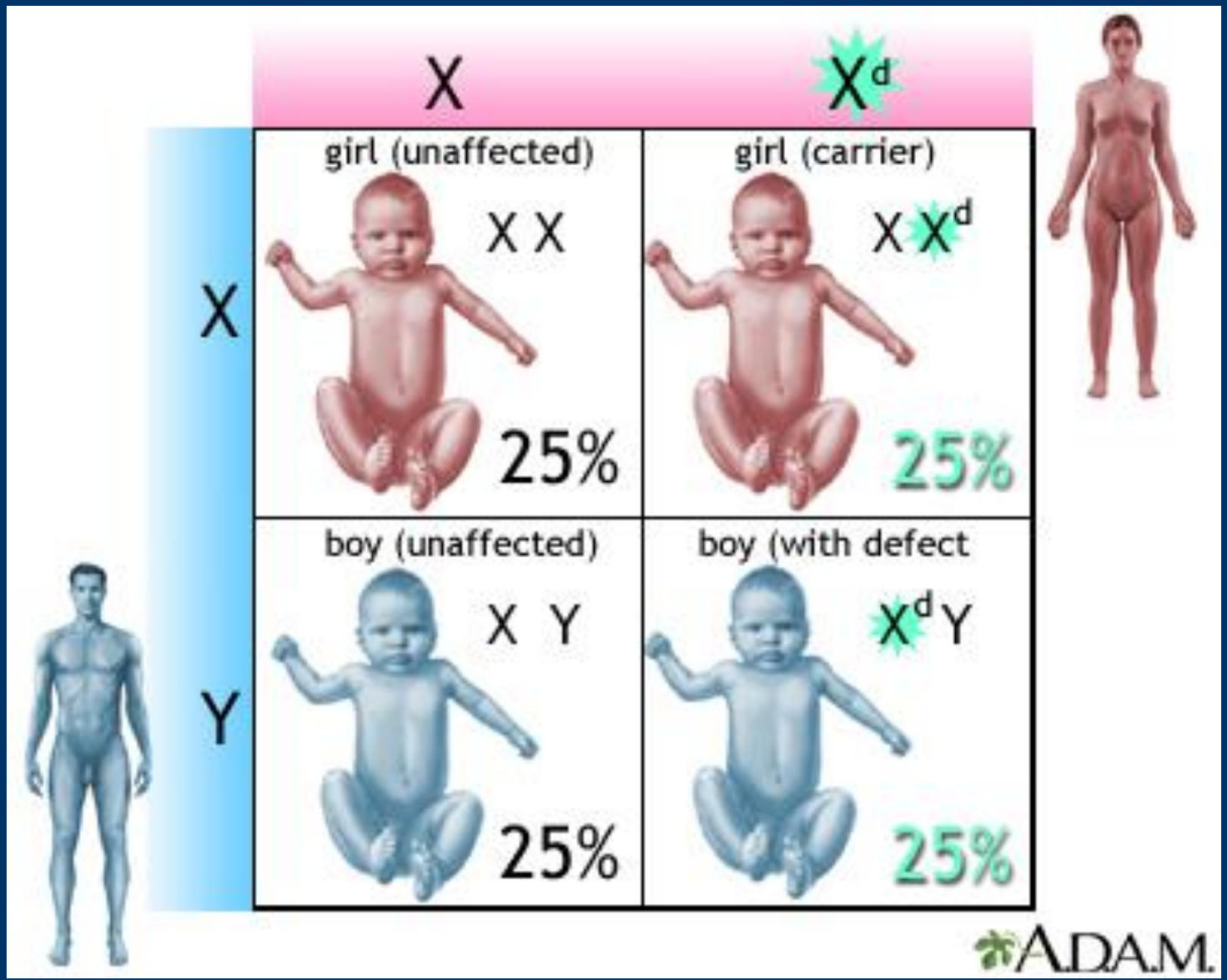


- Colorblind persons can see the circle but not the star

Y - linked trait…



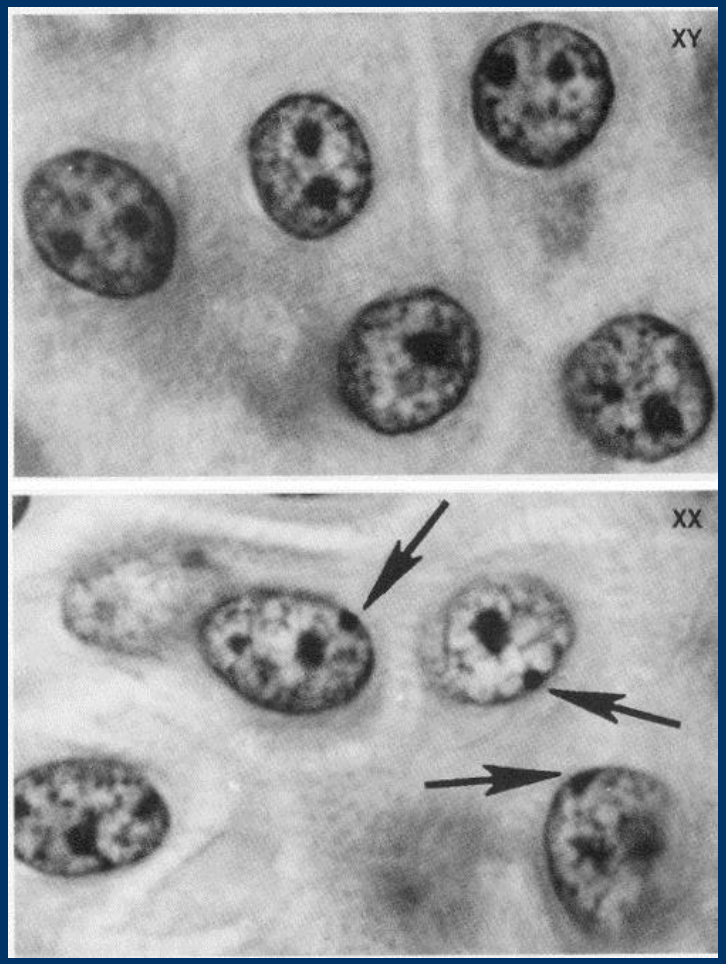
This trait always occurs when the gene is present…but we can’t know if it is dominant. Why not?


X
X^d
girl (unaffected)
X X
25%
girl (carrier)
X X^d
25%
X
boy (unaffected)
X Y
25%
boy (with defect
X^d Y
25%
Y
A.D.A.M.

X Inactivation in Female Mammals

- During fetal development, one female X chromosome becomes inactive and lies along the inside of the nuclear membrane
- Which X chromosome inactivates is random and varies from cell to cell, however during mitotic division the same X chromosome remains inactive
- The inactive X chromosome is called a *Barr body*

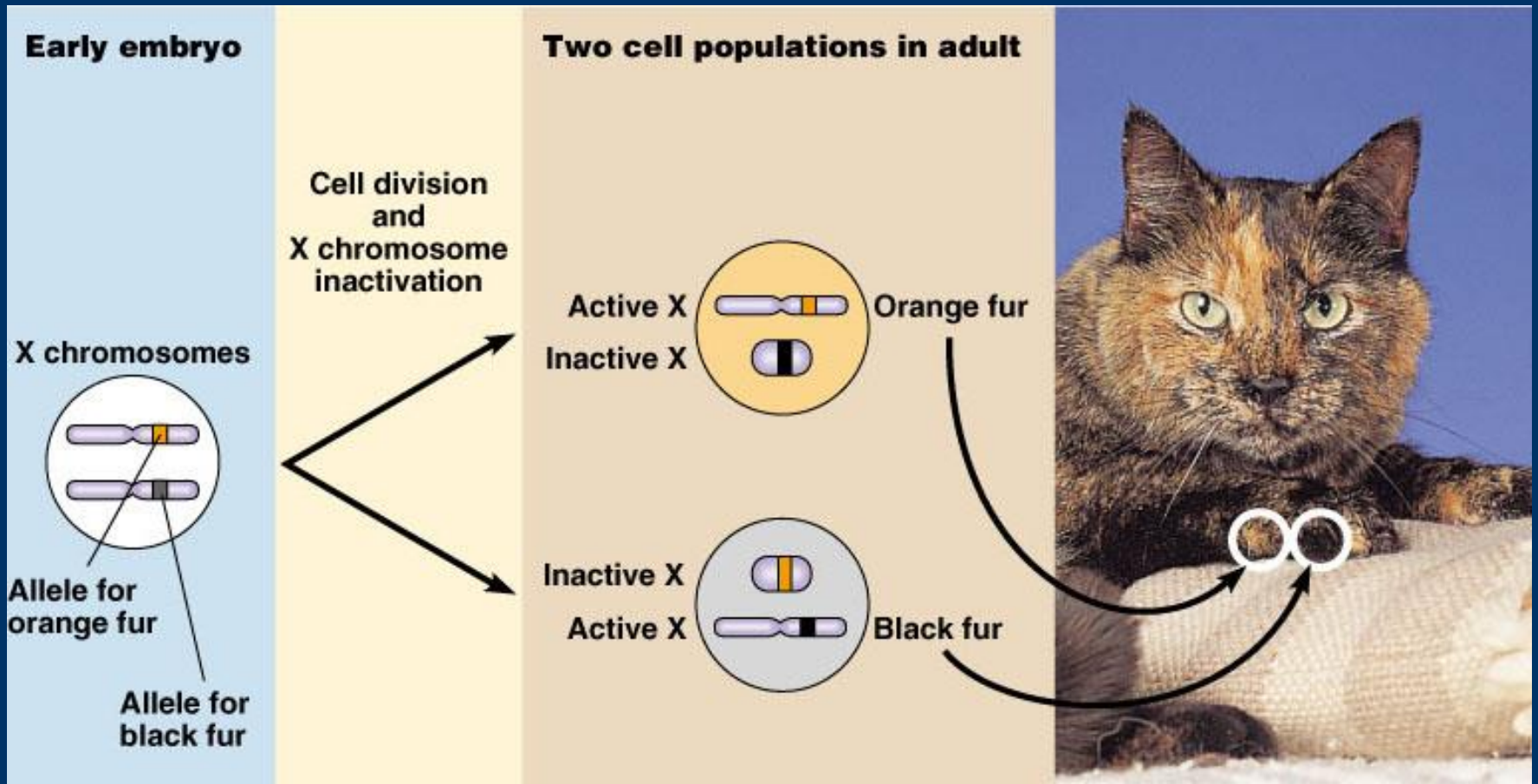
BARR BODIES



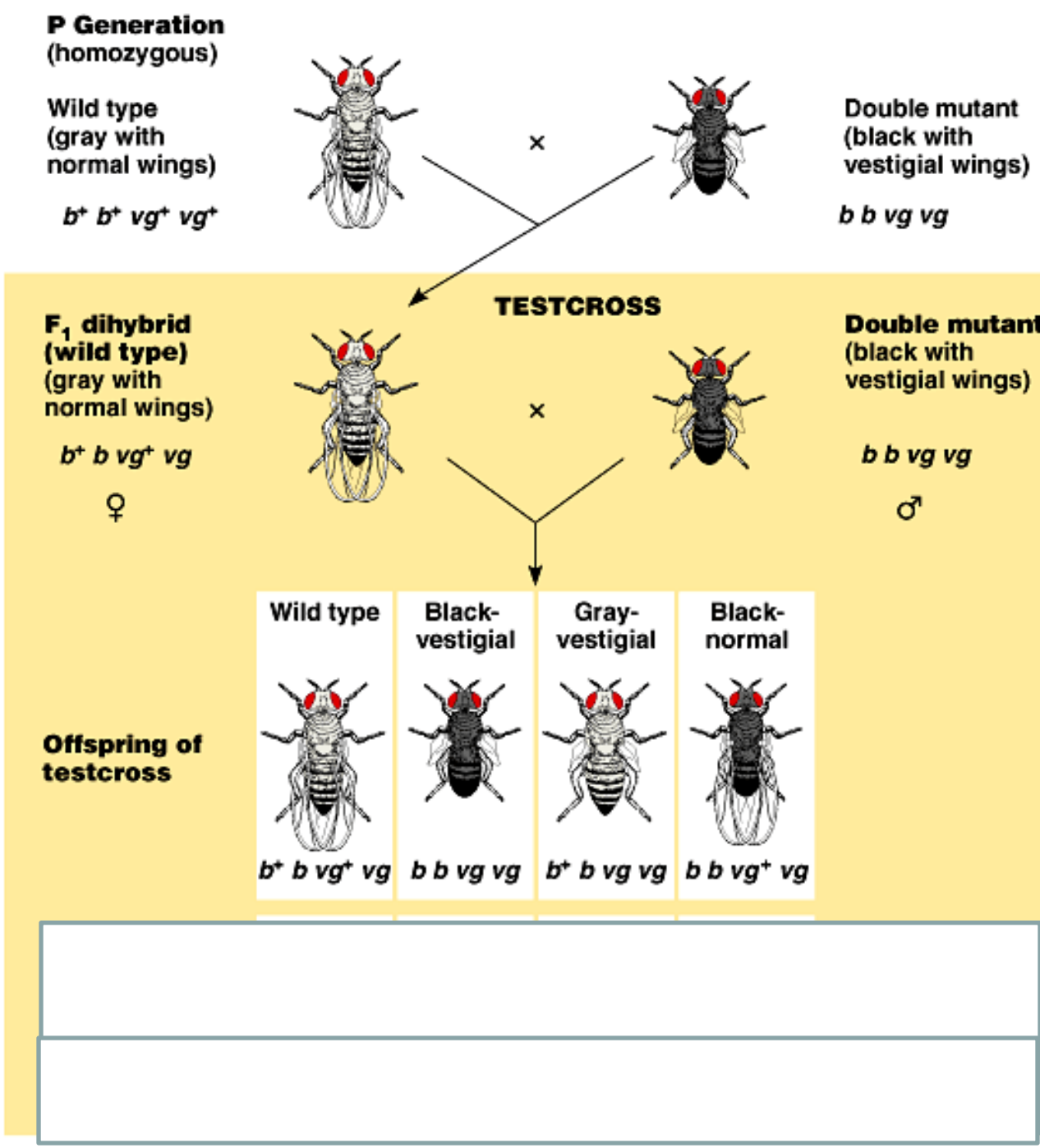
- **Only occurs in female mammals since they have two X chromosomes, unlike men**
- **Females consist of a mosaic of two types of cells; those with active and those with inactive X chromosomes**

X Inactivation in Female Mammals

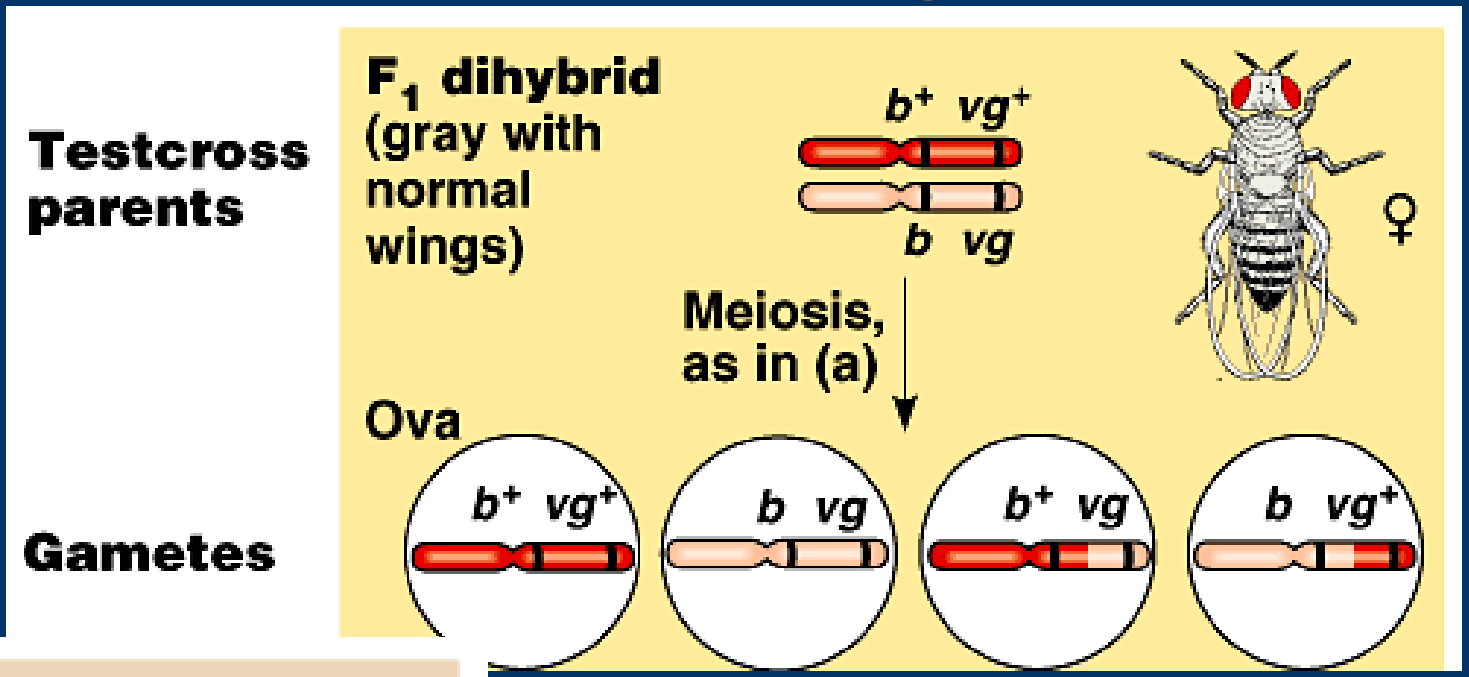
Tortoiseshell cats show mosaicism with patches of orange and black fur. Therefore, all tortoiseshell cats are female.

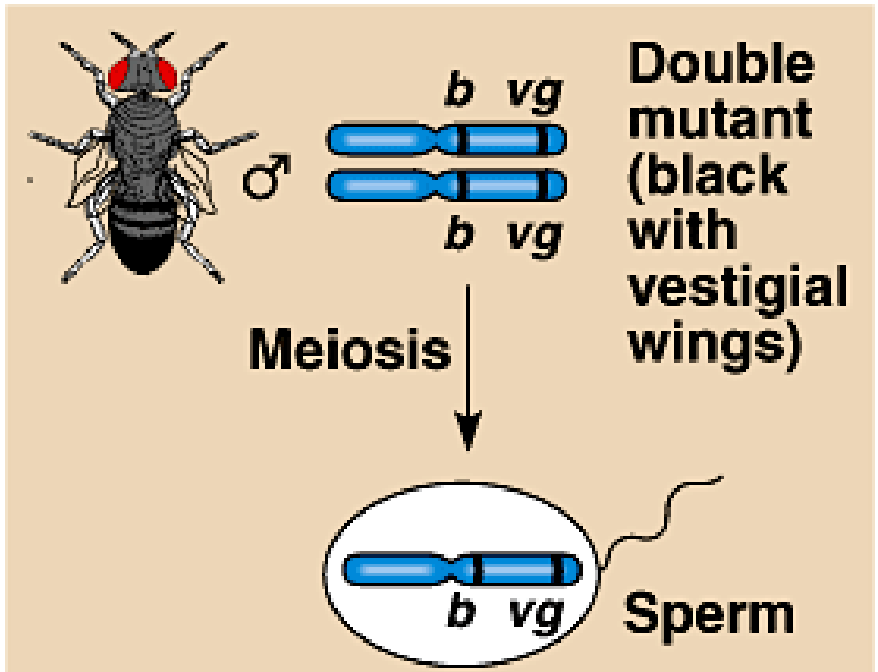


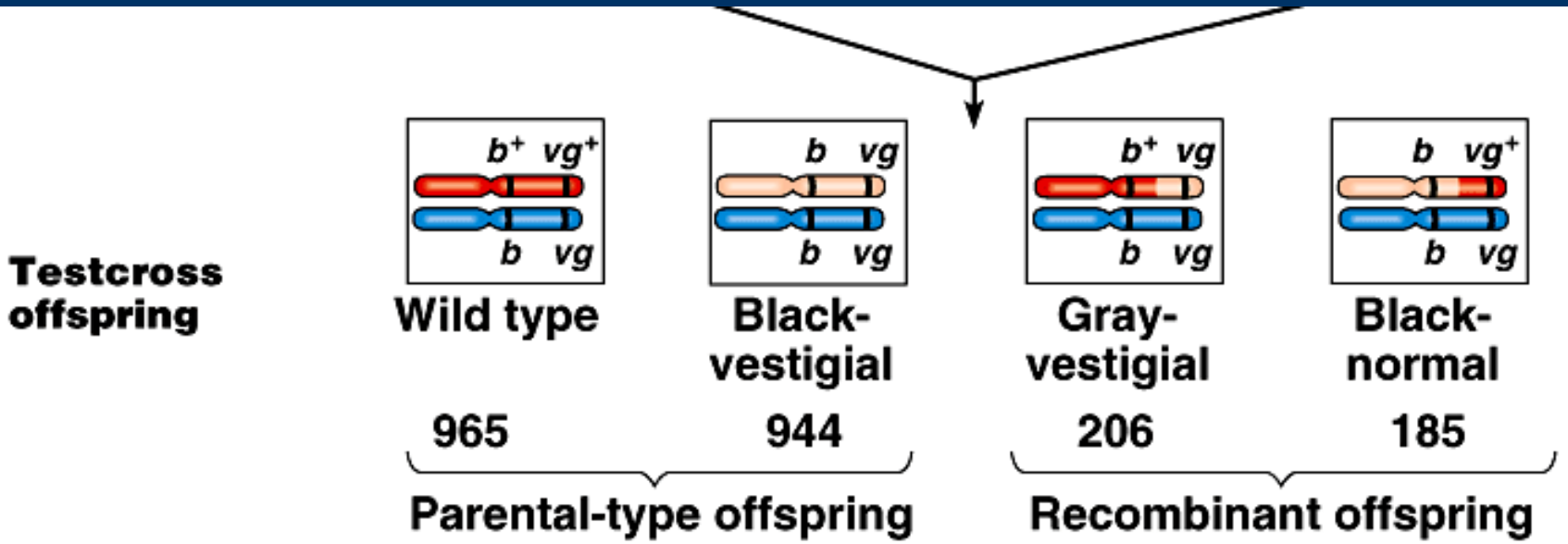
Fruit Fly Inheritance: linked genes



Recombination due to Crossing Over







$$\text{Recombination frequency} = \frac{391 \text{ recombinants}}{2{,}300 \text{ total offspring}} \times 100 = 17\%$$

(b) Production of recombinant offspring

Linked Genes

- Linked genes can occur when genes are located on the same chromosome
  - Don’t assort independently of each other in meiosis
- The further apart the 2 genes are, the more likely that a crossing over events will occur between them
  - The result is recombination

Linked Genes

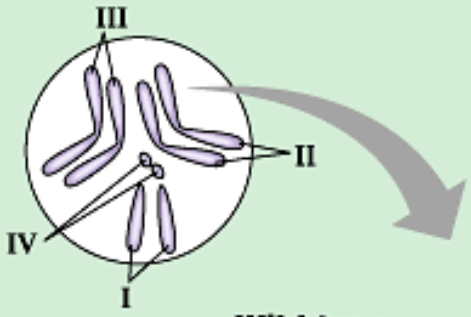
- Map Unit = arbitrary unit of measure used to describe the relative distance between linked genes
  - # of map units between 2 genes or between a gene and the centromere is equal to the percentage of recombinants
- As gene's recombination frequencies gets closer to 50%, it becomes hard to tell if the genes are linked or on separate chromosomes
  - So far apart on same chromosome that it is difficult to distinguish it from unlinked genes

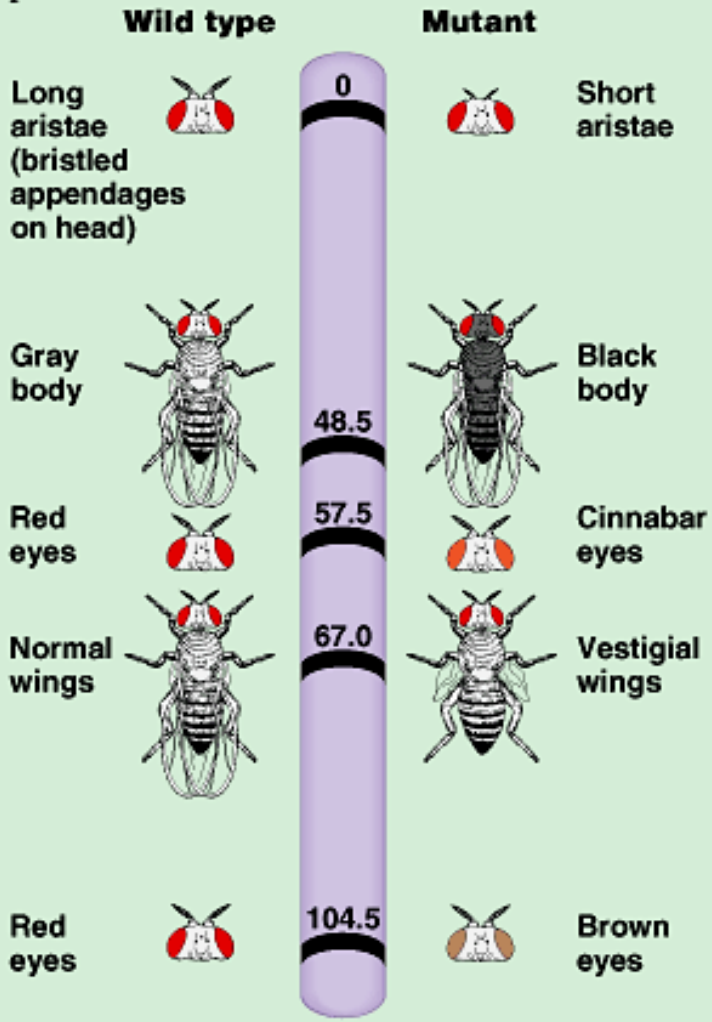
Chromosome Map of *Drosophila* (Fruit Flies)

Gene	b	cn	rb	vg
b	0			
cn	9	0		
rb	3.5	6.5	0	
vg	19	9.0	16	0

b = black body
cn = cinnabar eyes
rb = reduced bristles
vg = vestigial wings

The numbers in the boxes are the recombination frequencies in between the genes (in percent).

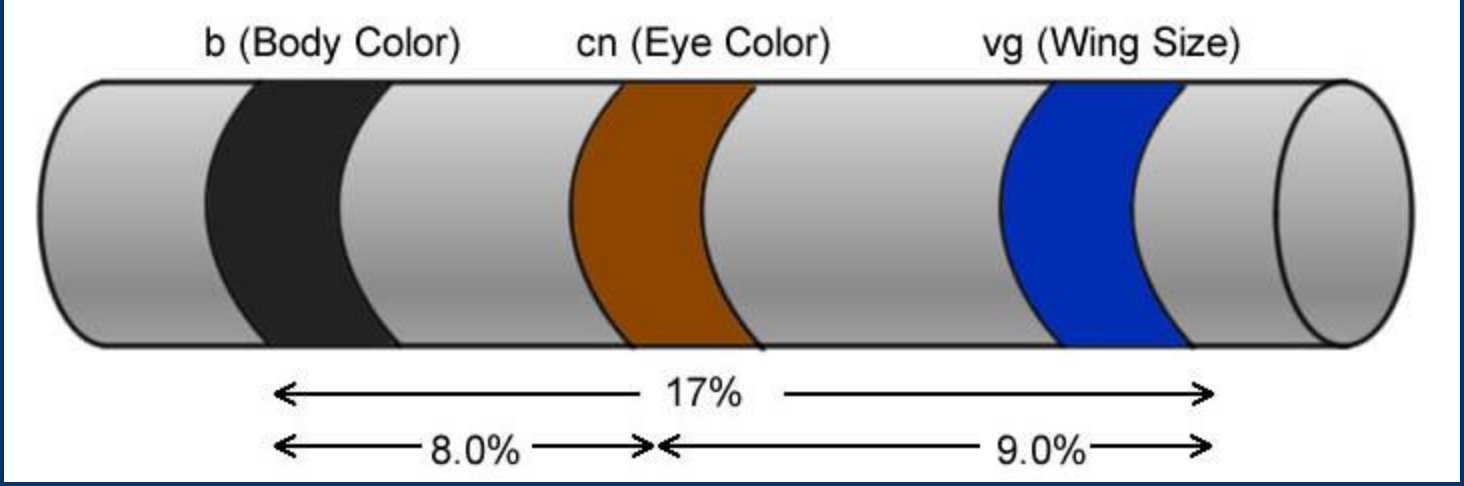




Practice Problems

- Body color (b)
- Eye color (cn)
- Wing size (vg)

- b and cn = 8%
- vg and b = 17%
- cn and vg = 9%



Practice Problems

1. Given the crossover frequencies of each of the gene on the chart, construct a chromosome map.

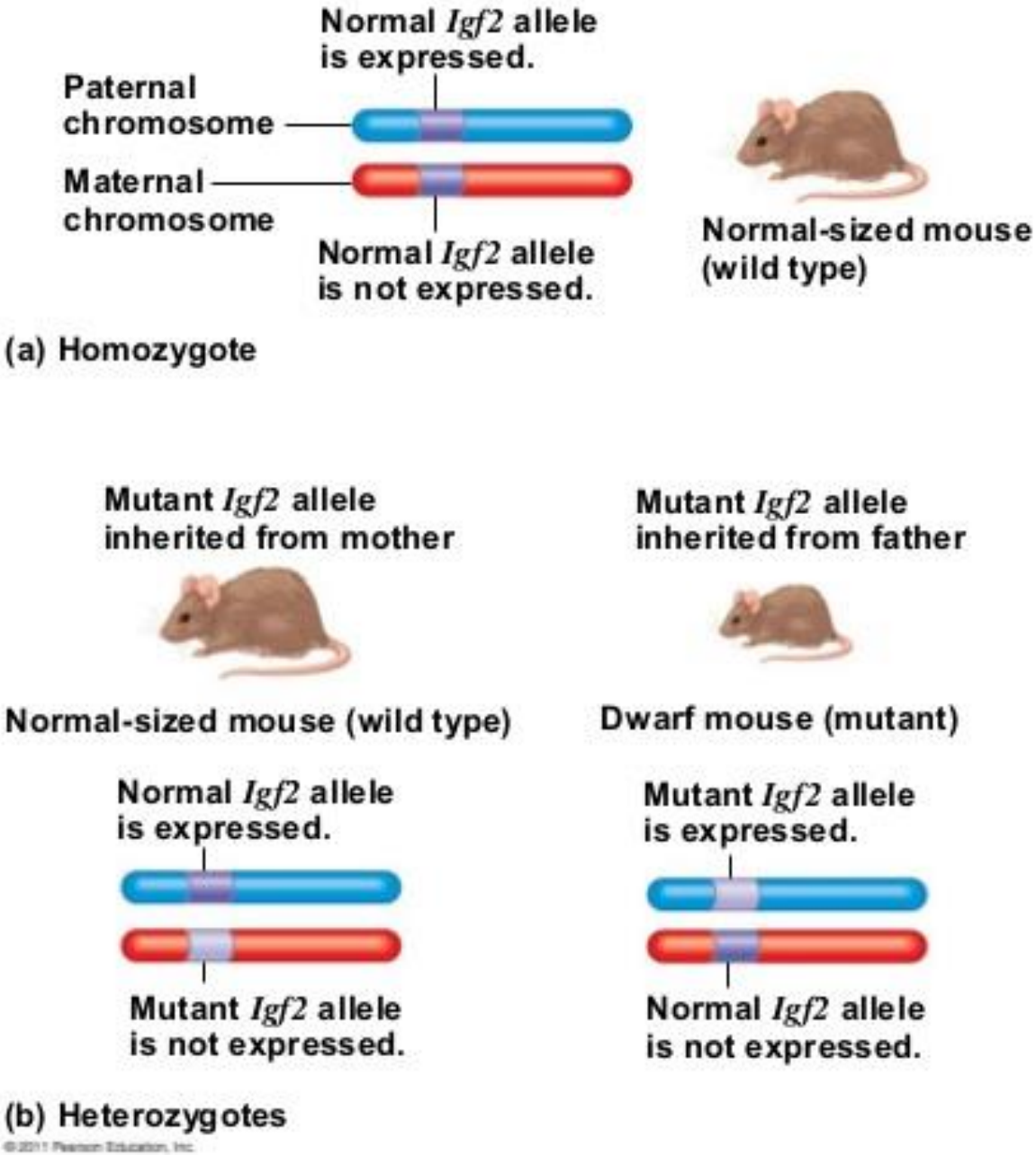
Gene	Frequency of Crossover
A-C	30%
B-C	45%
B-D	40%
A-D	25%

Answer

Exceptions to Mendel's Laws

- So far, we have assumed that any given allele will have the same effect whether inherited from the mother or father
- Genomic imprinting – variation in phenotype depending on whether an allele is inherited from the male or female parent
  - There are about 2-3 dozen genes associated with embryo development that work through genomic imprinting

Figure 15.17



Non-nuclear inheritance

- Mitochondria and chloroplast (plastids) have small circular DNA (extranuclear genes) that code for proteins and RNA
- These gene don’t display Mendelian inheritance because the organelles are randomly assorted to the gametes
- In animals, mitochondrial DNA is transmitted by the egg so these genes are maternally inherited

- Studies of yellow or white patches on leaves of green plants
  - Showed that variation in plastid genes altered pigmentation
  - Plastids came from the cytoplasm in the egg, and the pollen has no control over expression
  - Depending on the ratio of wild-type to mutant plastids, the plant will have a variety of spots on its leaves
- Same is true for mitochondrial genes
  - Mutations in mitochondrial DNA have been linked to Alzheimer's, heart disease, and aging