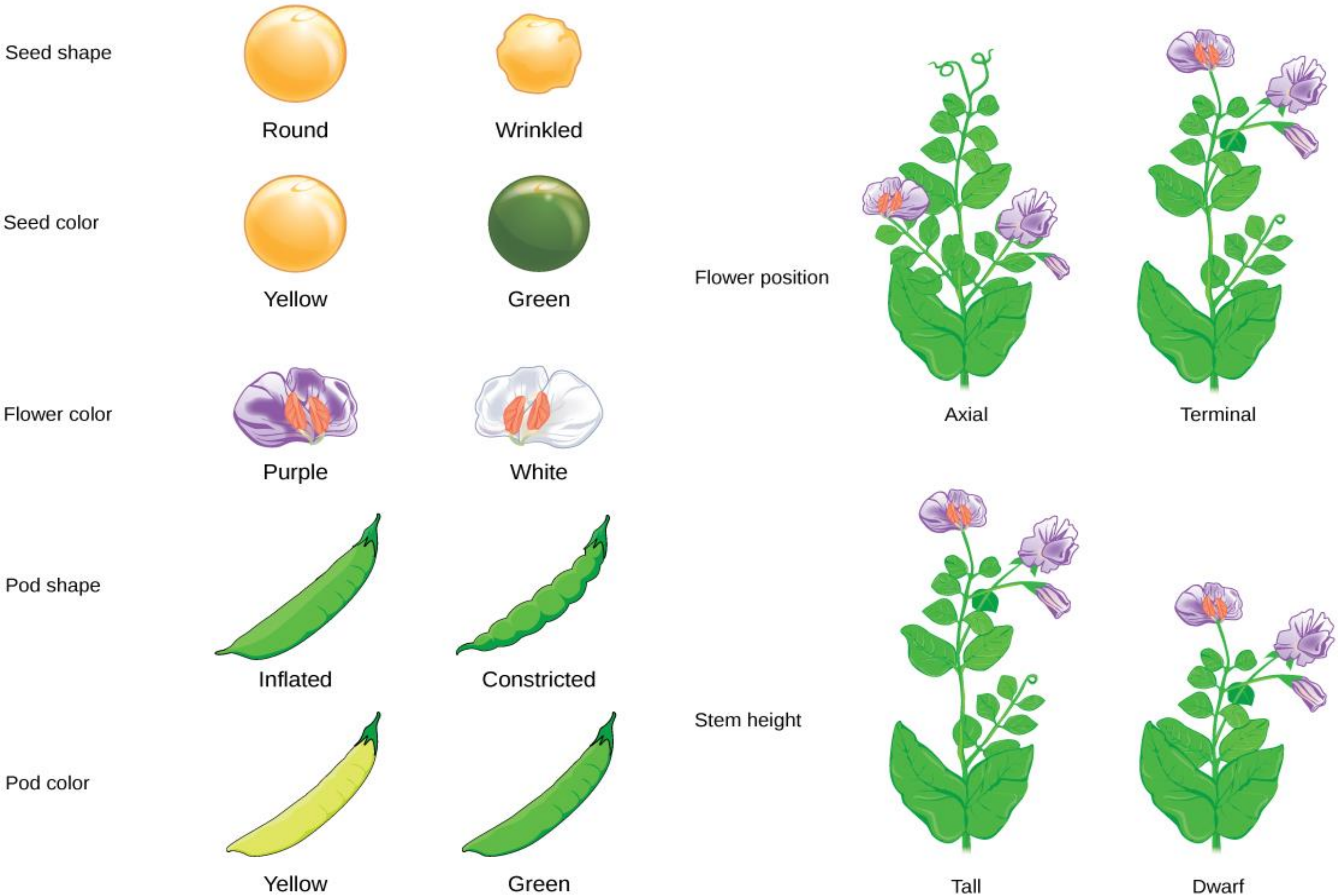
Mendelian concept of inheritance

Gregor Johann Mendel (1822–1884)
[Father of Genetics]

Gregor Johann Mendel (1822-1884)

- Breeding/crossing experiments-Pea plants
- Rediscovered after his death- 1900
- Referred to "Father of Genetics"- contribution to an understanding – some of the basic principles of hereditary - His principles -" Mendelian Genetics"

7 Characteristics in Peas



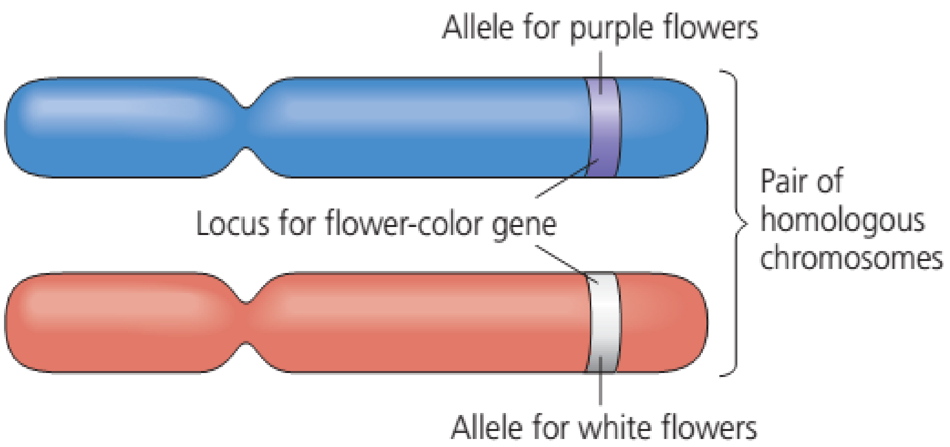
Gregor Johann Mendel (1822-1884)


Gregor Mendel

- Green pea
- Available in many varieties
- Short generation time
- Self pollinating- strictly control the mating

Genetics terms you need to know

- **Gene** – a unit of heredity; a section of DNA sequence encoding a single protein



- **Alleles** – Alternate version of a gene that occupy the same position on homologous chromosomes.

- **Locus** – a fixed location on a strand of DNA where one of its alleles is located.

- **Homozygous** (**True breeding**) – having identical alleles for a particular characteristic.

- **Heterozygous** – having two different alleles for a particular characteristic.

- **Dominant** – the allele of a gene that masks or suppresses the expression of an alternate allele

- **Recessive** – an allele that is masked by a dominant allele

- <u>**Genotype**</u> – the genetic makeup of an organisms

- <u>**Phenotype**</u> – the physical appearance of an organism

- **Character-** A heritable feature that varies among individuals- Flower color

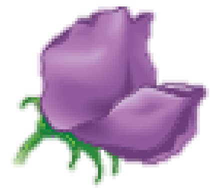
- **Trait-** Each variant of the character- purple or white color for flower

Monohybrid cross: a genetic cross involving parents possessing a pair of contrasting characters

Hybridization- mating or crossing of two true breeding varieties

Monohybrid cross for Flower color

P = parents
<u>true breeding,</u>
<u>homozygous plants</u>

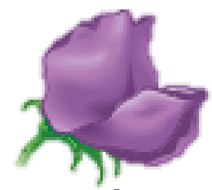


RR × *rr*

(Purple) (White)

R = allele for Purple
r = allele for White

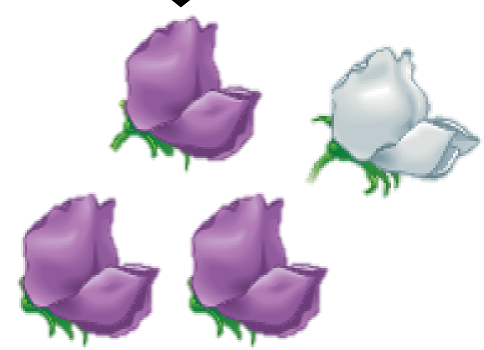
F_1 generation
is heterozygous



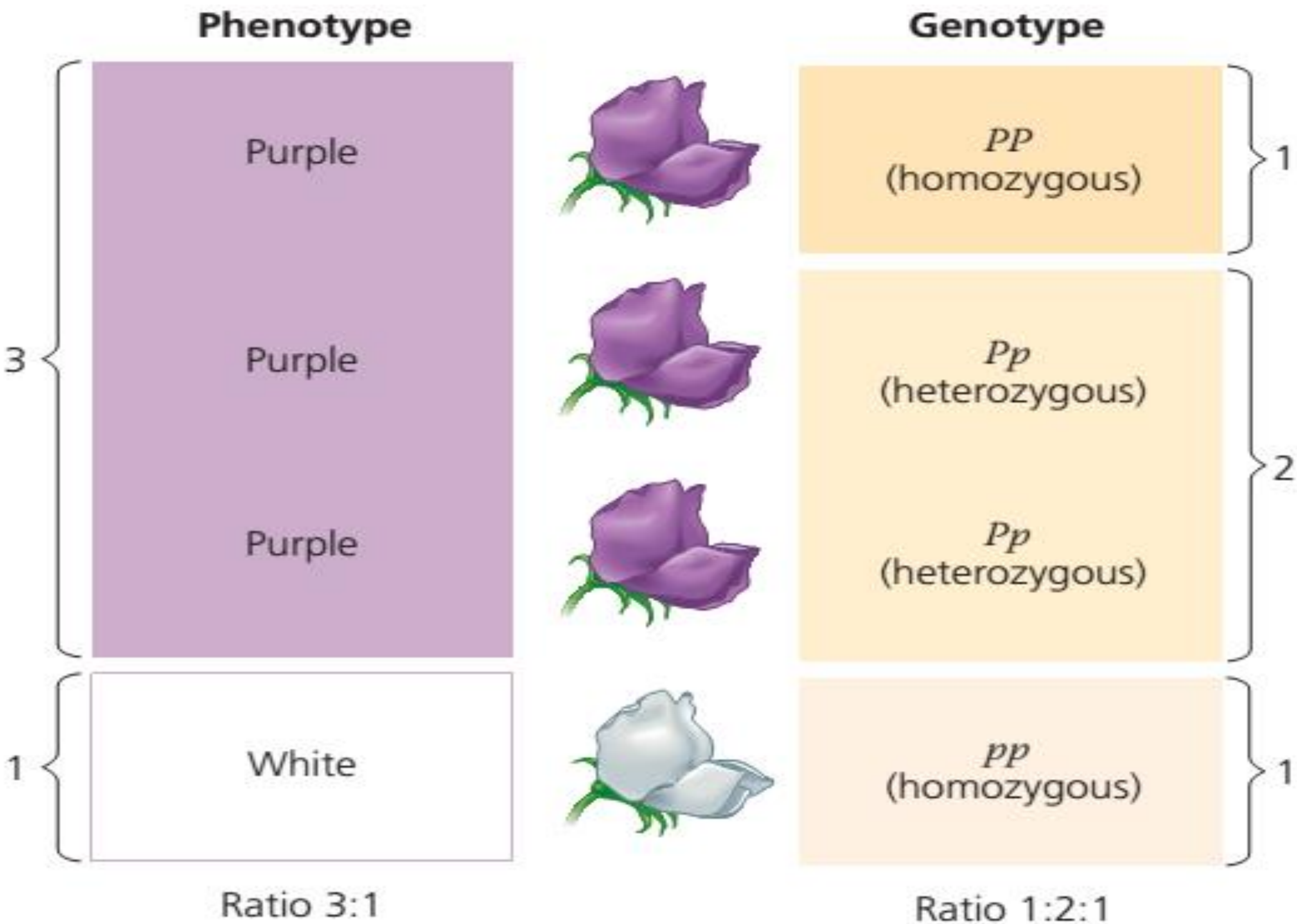
Rr
(all puple)

Self pollinate

F_2 generation



Purple : white is 3:1



Phenotype
Genotype
Purple
PP
(homozygous)
1
3
Purple
Pp
(heterozygous)
2
Purple
Pp
(heterozygous)
1
White
pp
(homozygous)
1
Ratio 3:1
Ratio 1:2:1

Using a Punnett Square

Parent genotypes:

PP and *pp*

Cross

Pp--------F1 Generation

F1 Selfed

Pp× *Pp*

Pp× *Pp*

	P	**p**
P	**PP**	**Pp**
p	**Pp**	**pp**

Genotypes:

1 PP= Purple

2 Pp = Purple

1 *pp* = white

Genotypic ratio= 1:2:1

Phenotype:

3 Purple

1 White

Phenotypic ratio= 3:1

	Parental generation phenotypes: Dominant		Parental generation phenotypes: Recessive		F1 Phenotypes	F_2 generation phenotypes: Dominant	F_2 generation phenotypes: Recessive	Total	Ratio
	Spherical seeds	×	Wrinkled seeds		Spherical	5,474	1,850	7,324	2.96:1
	Yellow seeds	×	Green seeds		Yellow	6,022	2,001	8,023	3.01:1
	Purple flowers	×	White flowers		Purple	705	224	929	3.15:1
	Inflated pods	×	Constricted pods		Inflated	882	299	1,181	2.95:1
	Green pods	×	Yellow pods		Green	428	152	580	2.82:1
	Axial flowers	×	Terminal flowers		Axial	651	207	858	3.14:1
	Tall stems (1 m)	×	Dwarf stems (0.3 m)		Tall	787	277	1,064	2.84:1

Observations of **Monohybrid cross**

(A) Only one trait appeared in the F1 generation

(B) Both traits were appeared in the F2 generation, but not in equal percentage

(C) A trait which disappeared in the F1, reappeared in F2

(D) The results are consistent in all the 7 characters

(E) There is no blending of characters

Observations of Monohybrid cross

- The hybrid offspring - resembled one of the parents, no intermediate flower color.
- The first filial generation F1 plants all had **Dominant Characters.**
- **Trait expressed in the F1 plants –Dominant**; Alternative trait - not expressed in the F1- **Recessive**.
- 3/4th of the F2 individuals exhibited the dominant trait and 1/4th displayed the recessive trait. The ratio of dominant to recessive among the F2 plants was always 3:1

Mendel’s model

1. Alternative versions of a gene- **“Alleles”**

2. For each character an organism inherits two copies of a gene, one come from each parent

3. **“dominant”** allele determines the organism’s appearance, the other **“recessive”** allele has no noticeable effect on the organisms appearance

Law of Segregation/Law of purity of gametes

- **Principle of Segregation**:

It states that "whenever a pair of factors for character brought together in a hybrid, they segregate during the formation of gametes and each gamete is pure with reference to this character"

Back cross

Cross involving **F1 individuals with either of two parents**- Back cross

Cross with a F1 hybrid and dominant parental type- Dominant individuals.

Cross with a F1 hybrid and recessive parental type- both the phenotype appear in the progeny 50: 50 %

Test cross

Test cross: Cross with **F1 and a homozygous recessive individual**.

Test cross helps- **test individual homozygous or heterozygous**

Test cross

For example, a plant with **purple** flowers can either be **PP** or **P*p***… therefore, to know the genotype - cross the plant with a *pp* (white flowers, homozygous recessive)

 P *?* × *pp* 

- If you get all 100% purple flowers, then the unknown parent was PP…

- If you get 50% white, 50% purple flowers, then the unknown parent was P*p*…

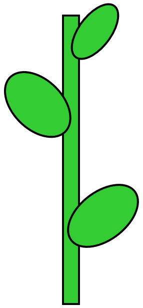
Dihybrid cross: a genetic cross involving **parents differ by a Two characters**. **Stem Height** and **Flower color**

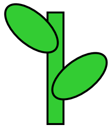
For example, Flower color:

P = purple (dominant) 

p = white (recessive) 

and Stem length:

T = tall 

t = short 

Dihybrid cross: flower color and stem length (shortcut)

TT PP × *tt pp*

(tall, purple) (short, white)

Possible Gametes

T P *t p*

	T P
t p	T*t* P*p*

F1 Generation: All tall, purple flowers (T*t* P*p*)

Dihybrid cross F_2

If F_1 generation is allowed to self pollinate, Mendel observed 4 phenotypes:

T*t*P*p* × **T*t*P*p***

(tall, purple) (tall, purple)

Possible gametes: **TP** **T*p*** ***t*P** ***tp***

	TP	T*p*	*t*P	*tp*
TP	TTPP	TTP*p*	T*t*PP	T*t*P*p*
T*p*	TTP*p*	TT*pp*	T*t*P*p*	T*tpp*
*t*P	T*t*PP	T*t*P*p*	*tt*PP	*tt*P*p*
tp	T*t*P*p*	T*tpp*	*tt*P*p*	*ttpp*

Four phenotypes observed

Tall, purple (9); Tall, white (3); Short, purple (3); Short white (1)

Law of Independent Assortment

- The factors for two or more pairs of contrasting character, each pair of alleles segregates independently of each other pair of the alleles during gamete formation

Patterns of inheritance

- Mendel explained inheritance in terms of discrete "**factors**"

- Passing of information through "factors"- generation to generations- **rules of probability**

Patterns of inheritance

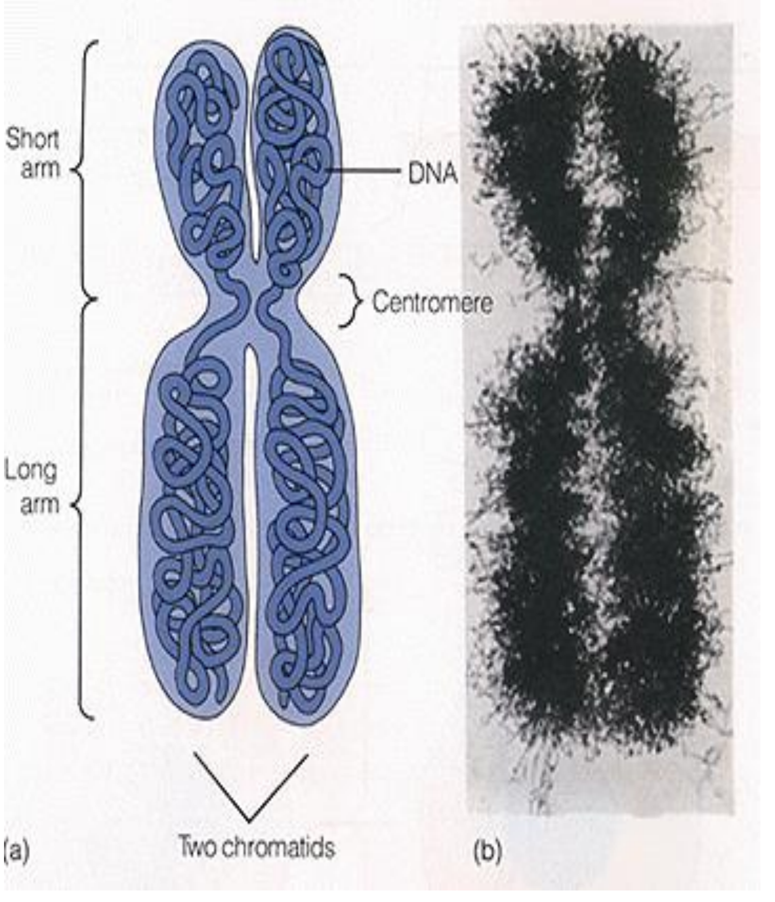
- Mendel's law stop short of explaining- pattern of genetic inheritance in <u>Sexually reproducing organisms – pattern of inheritance – often more complex</u>

- Offspring of Mendel's pea cross- one of the two parental varieties- dominant- <u>**"Complete dominance"**</u>

What Exactly is a chromosome?

- Chromosomes- rod shaped, present in the nucleus

- They are the carriers of the gene or unit of heredity.



Chromosome number

- Constant - throughout the life of an individual

- Gametes normally contain only one set of chromosome – this number is called **Haploid (n)**

- Somatic cells usually contain two sets of chromosome - **Diploid (2n)**

Morgan's Experimental Evidence

- The first solid evidence associating a specific gene with a specific chromosome came from Thomas Hunt Morgan, an embryologist

- Morgan's experiments with fruit flies provided convincing evidence that chromosomes are the location of Mendel's heritable factors

Drosophila melanogaster (Fruit fly)

Morgan’s Choice of Experimental Organism

- Several characteristics make fruit flies a convenient organism for genetic studies
  - They produce many offspring
  - A generation can be bred every two weeks
  - They have only four pairs of chromosomes
  - 3 pairs –autosome, 1 pair- sex chromosomes

How Linkage Affects Inheritance

- Morgan did experiments with fruit flies to see inheritance of two characters

- Morgan crossed flies that differed in traits of **body color and wing size**

- **GRAY BODY & NORMAL WINGS (Dominant)**
- **Black body & Vestigial wings (Recessive)**

12.18 Some Alleles Do Not Assort Independently

Morgan's studies showed that the genes for body color and wing size in *Drosophila* are linked, so that their alleles do not assort independently.

HYPOTHESIS Alleles for different characteristics always assort independently.

METHOD

Parent (P)

BbVgvg
Wild type
(gray body,
normal
wings)
♀

bbvgvg
(black body,
vestigial
wings)
♂

RESULTS

These are the results expected from Mendel's second law (independent assortment)...

F_1				
Genotypes	*BbVgvg* Wild type	*bbvgvg* Black vestigial	*Bbvgvg* Gray vestigial	*bbVgvg* Black normal
Expected phenotypes	575	575	575	575
Observed phenotypes (number of individuals)	965	944	206	185
	Parental phenotypes		Recombinant phenotypes	

...but the actual results were inconsistent with the law.

CONCLUSION The hypothesis is rejected. These two genes do not assort independently, but are linked (on the same chromosome).

PREDICTED RATIOS

If genes are located on different chromosomes:	**1**	**:**	**1**	**:**	**1**	**:**	**1**
If genes are located on the same chromosome *and* parental alleles are always inherited together:	**1**	**:**	**1**	**:**	**0**	**:**	**0**
RESULTS	**965**	**:**	**944**	**:**	**206**	**:**	**185**

What Morgan expected is a 1:1:1:1 ratio (Recollect Mendel's dihybrid test cross ratio)

Total individuals = 965+944+206+185 = 2300

Parental types = 965 + 944 = 1909

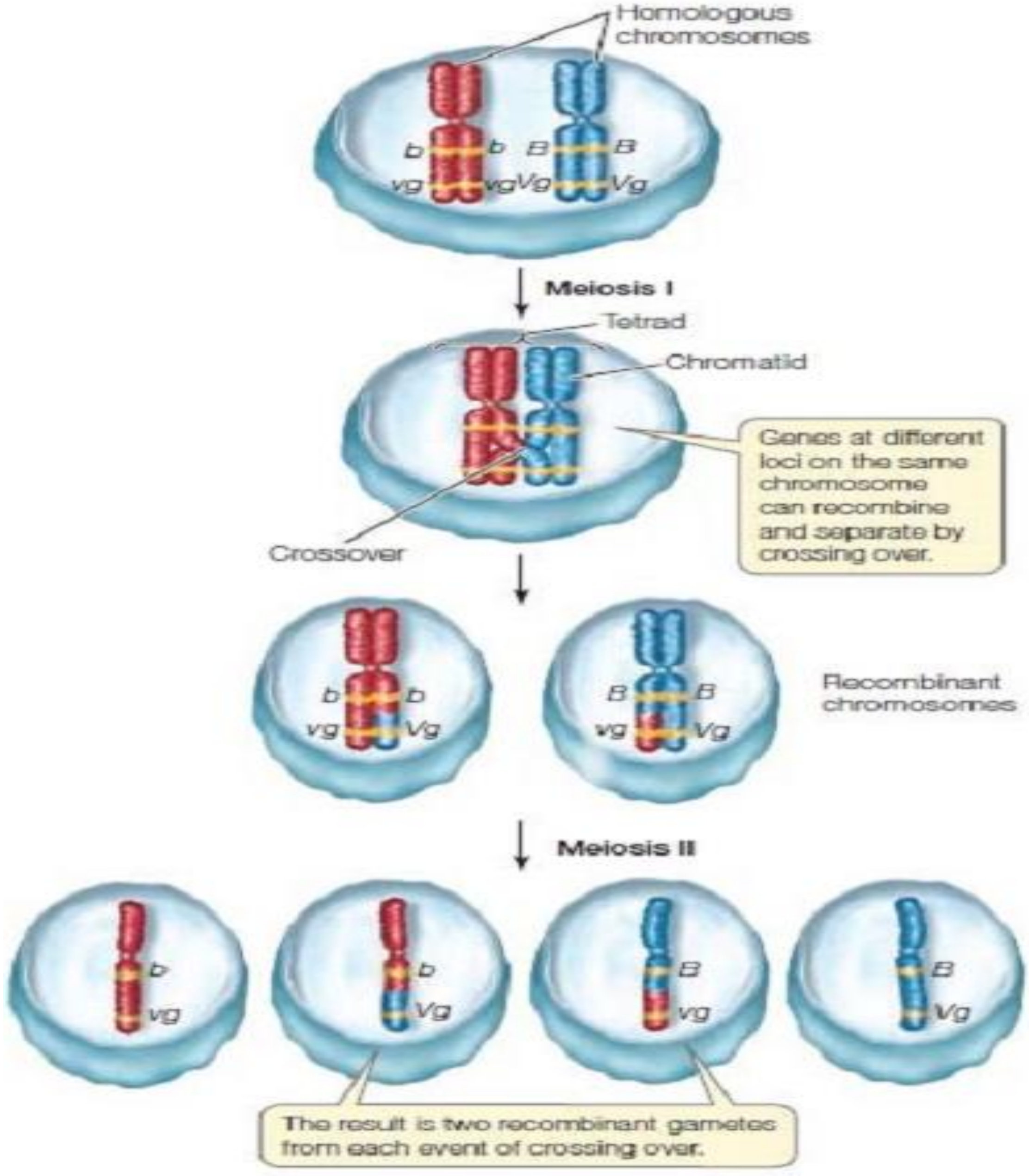
Non parental types or recombinant types = 391

- Morgan found genes for **body color** and **wing size** do not assort independently, and reasoned that they were on the same chromosome

- However, non-parental phenotypes were also produced

- Understanding this result involves exploring **genetic recombination**, the production of offspring with combinations of traits differing from either parent

The new phenotypes appear because of exchange of genes between homologous chromosomes that occurs during meiosis (swapping). This event is known as crossing over. Look at the following illustration to understand the process.



Linked genes

- Genes located on the same chromosome that tend to be inherited together are called **linked genes**

- Linked genes don't sort independently, unless crossing over occurs

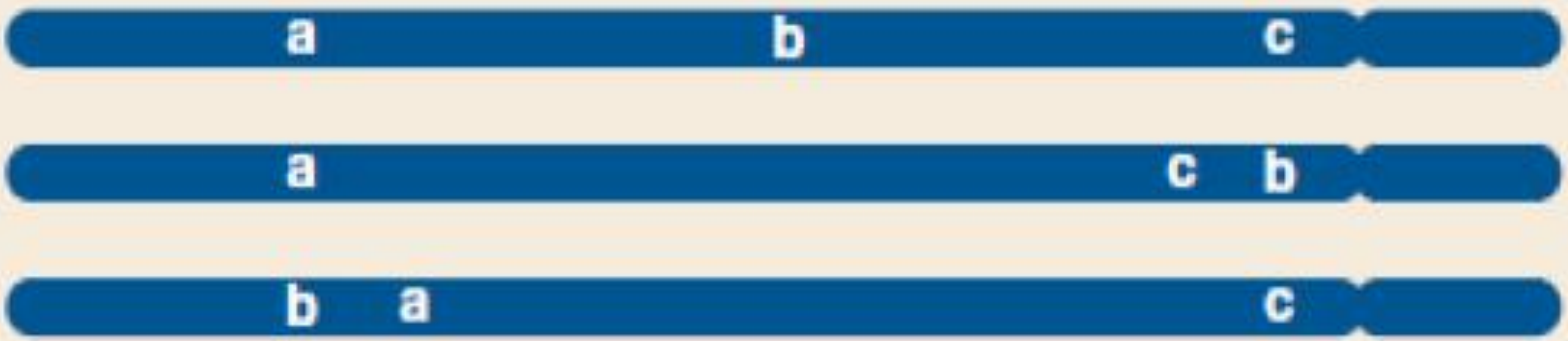
- Linked genes are predicted to always be transmitted together during gamete formation

Genetic Map

- A genetic map tells the distance between two genes

- Probability of recombination between two loci increases with distance

1 At the outset, we have no idea of the individual distances between the genes, and there are several possible sequences (*a-b-c*, *a-c-b*, *b-a-c*).



We make a cross *AABB* × *aabb*, and obtain an F_1 generation with a genotype *AaBb*. We test cross these *AaBb* individuals with *aabb*. Here are the genotypes of the first 1,000 progeny:

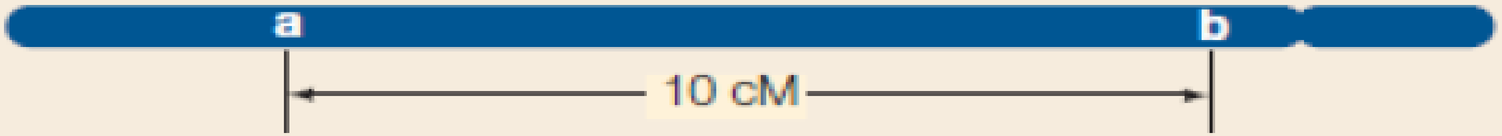
450 *AaBb*, 450 *aabb*, 50 *Aabb*, and 50 *aaBb*.
(parental types) (recombinant types)

2 How far apart are the *a* and *b* genes?

What is the recombinant frequency? Which are the recombinant types, and which are the parental types?

Recombinant frequency (*a* to *b*) = (50 + 50)/1,000 = 0.1
So the map distance is

$$\text{Map distance} = 100 \times \text{recombinant frequency} = 100 \times 0.1 = 10 \text{ cM}$$



3 How far apart are the a and c genes?

Now we make a cross *AACC* × *aacc*, obtain an F_1 generation, and test cross it, obtaining

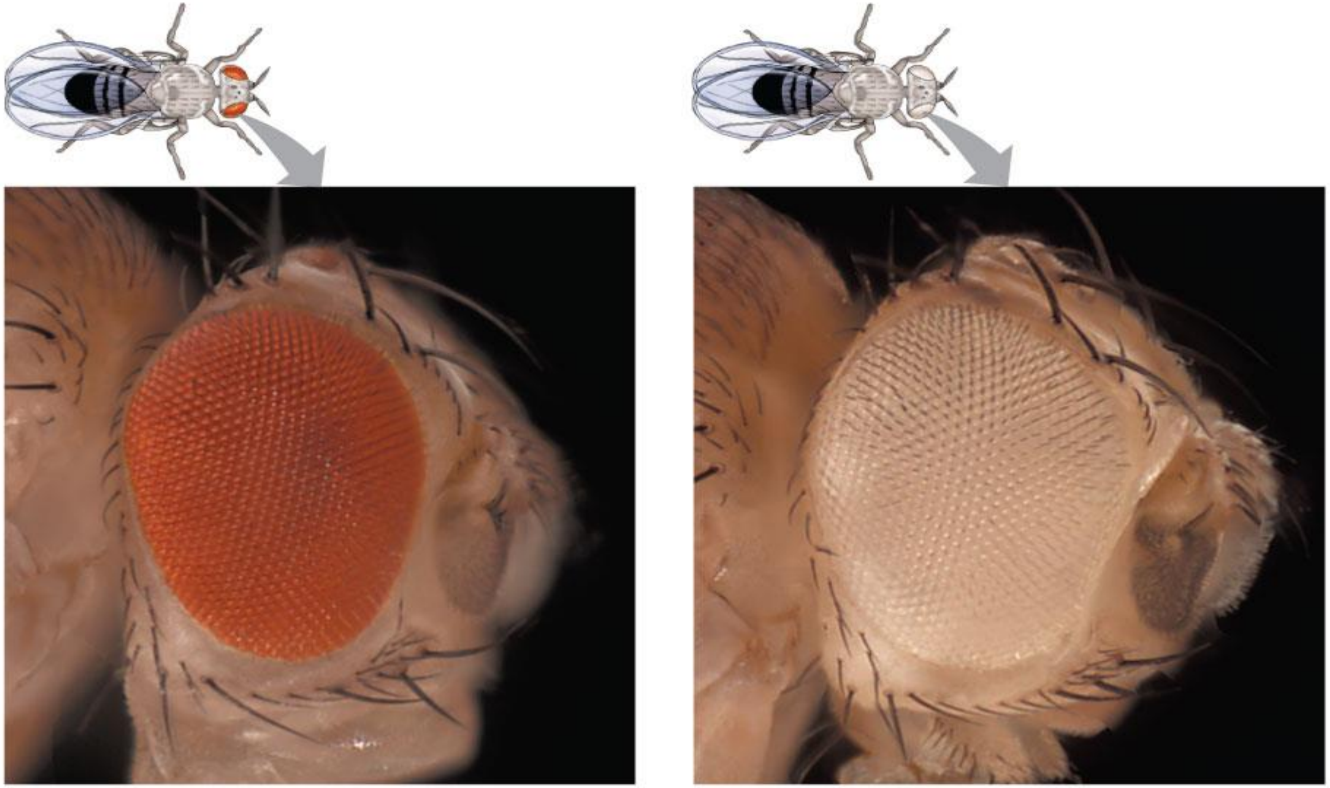
460 *AaCc*, 460 *aacc*, 40 *Aacc*, and 40 *aaCc*

Recombinant frequency (*a* to *c*) = (40 + 40)/1,000 = 0.08

$$\text{Map distance} = 100 \times \text{recombinant frequency} = 100 \times 0.08 = 8 \text{ cM}$$

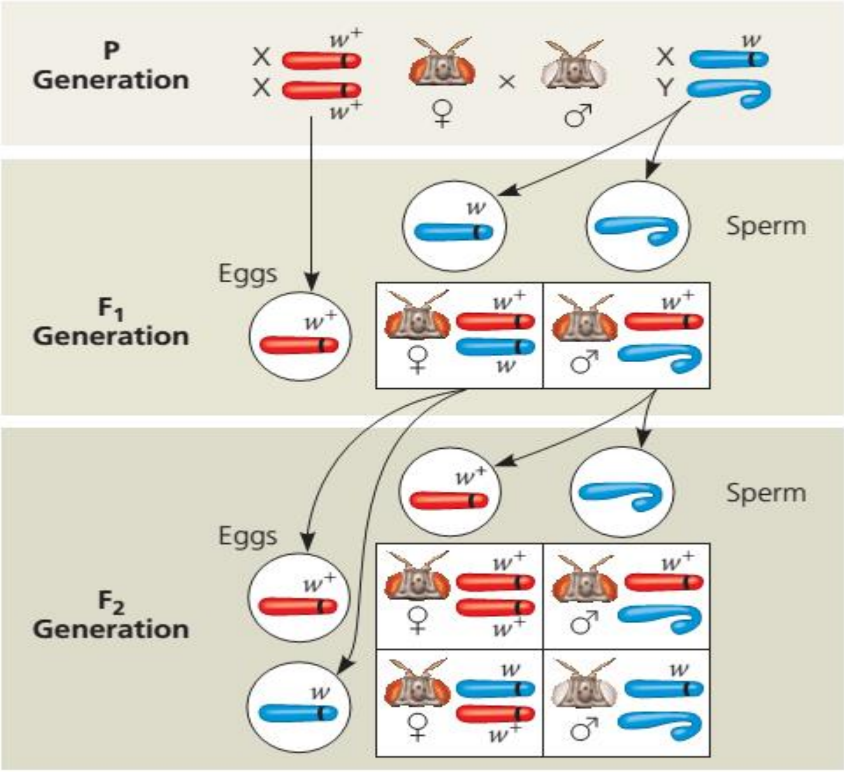
Sex linked inheritance

- Morgan noted **wild type**, or normal, phenotypes that were **common in the fly populations**
- Traits alternative to the wild type are called **mutant phenotypes**



Correlating Behavior of a Gene's Alleles with Behavior of a Chromosome Pair

- Morgan mated male flies with white eyes (mutant) with female flies with red eyes (wild type)- Got Red eyed flies (F1).F_2 generation 3:1 red : white eye ratio, **but only males had white eyes**
- Morgan determined that the **<u>genes for eye color must be located on the X chromosome</u>**



- A gene that is located on either sex chromosome is called a **sex-linked gene**

- Genes on the Y chromosome are called Y-linked genes;

- Genes on the X chromosome are called **X-linked genes**

X Linked Recessive

- <u>Males get their X from their mother</u>

- <u>Fathers pass their X to daughters only</u>

- **<u>Females express it only if they get a copy from both parents.</u>**

Y-Linked Inheritance

- Traits on the Y chromosome are only found in males, never in females.

- **The father's traits are passed to all sons.**

- Dominance is irrelevant: there is only 1 copy of each Y-linked gene (hemizygous).

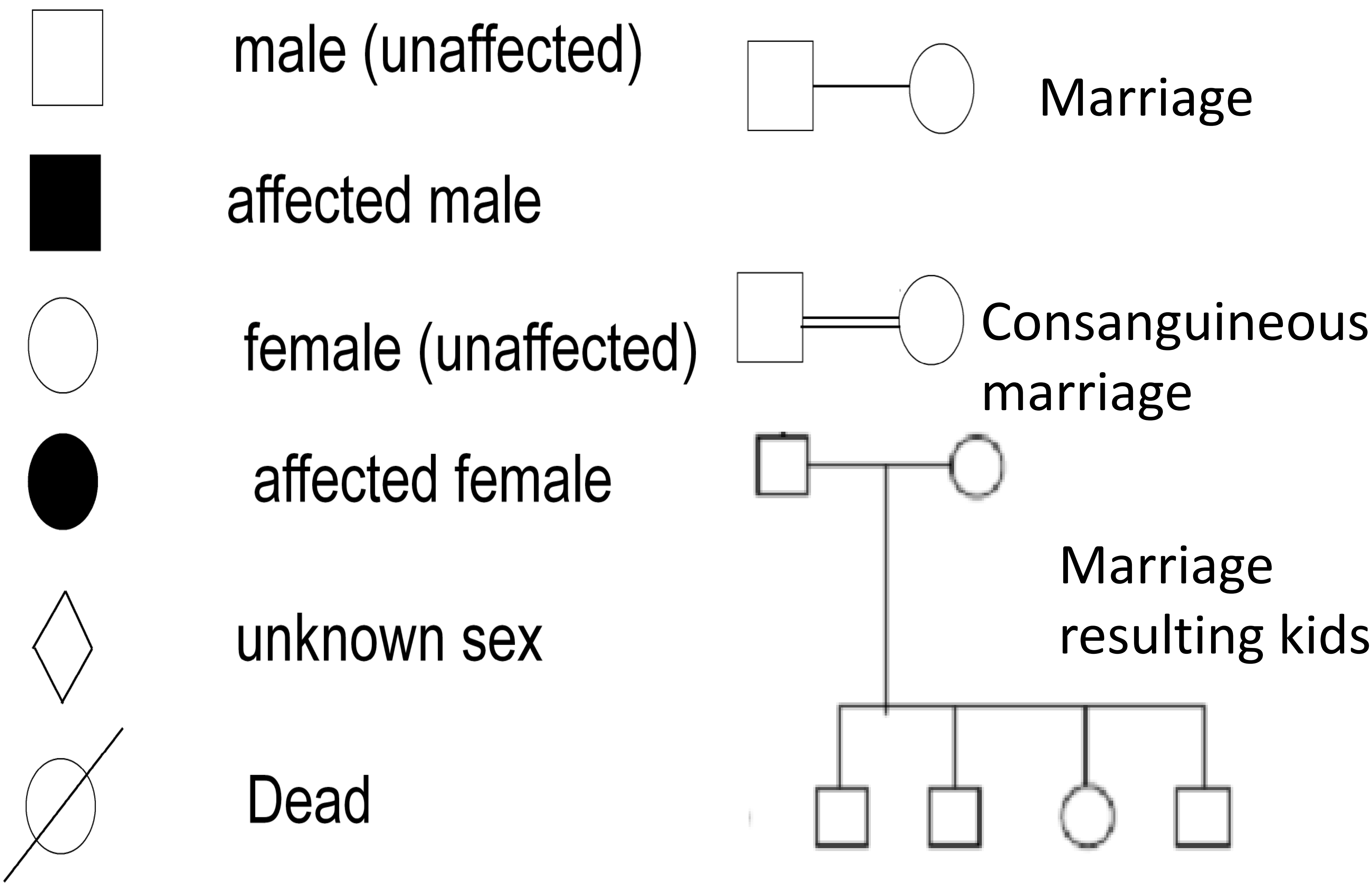
Pedigree Analysis

Goals of Pedigree Analysis

- Determine the mode of inheritance: dominant, recessive, sex-linked inheritance, autosomal etc.

- Determine the probability of an affected offspring for a given cross.

Basic Symbols in Pedigree Analysis



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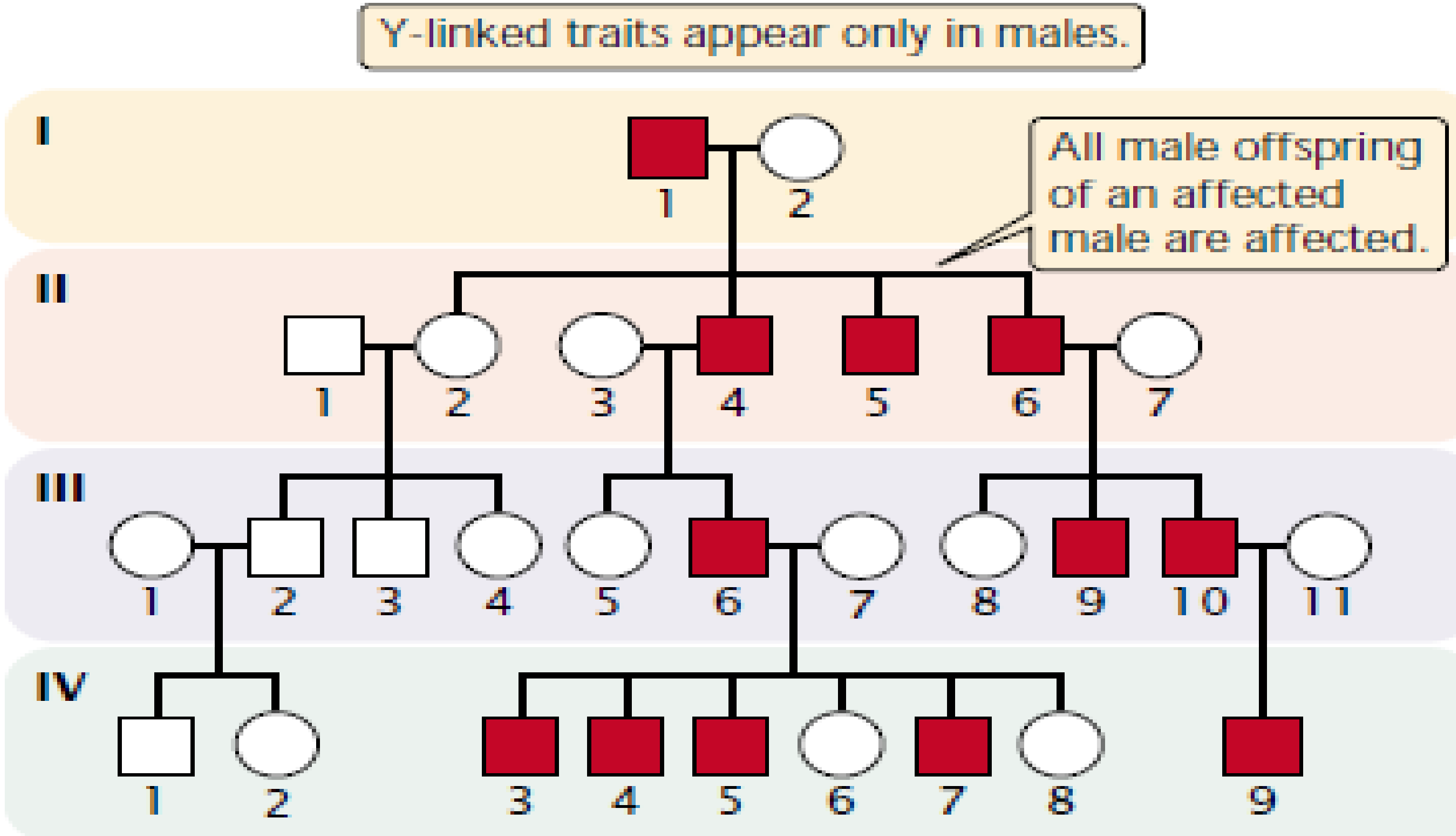
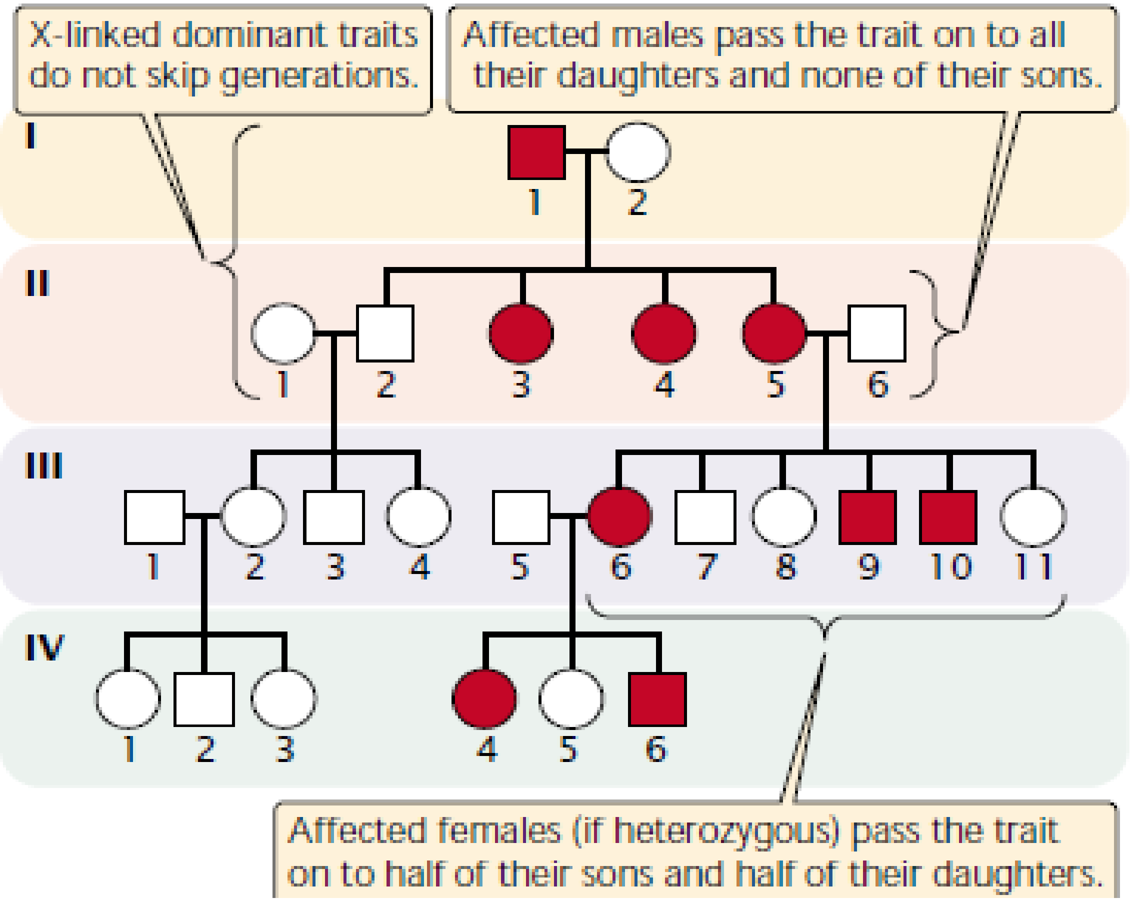


X-Linked Dominant

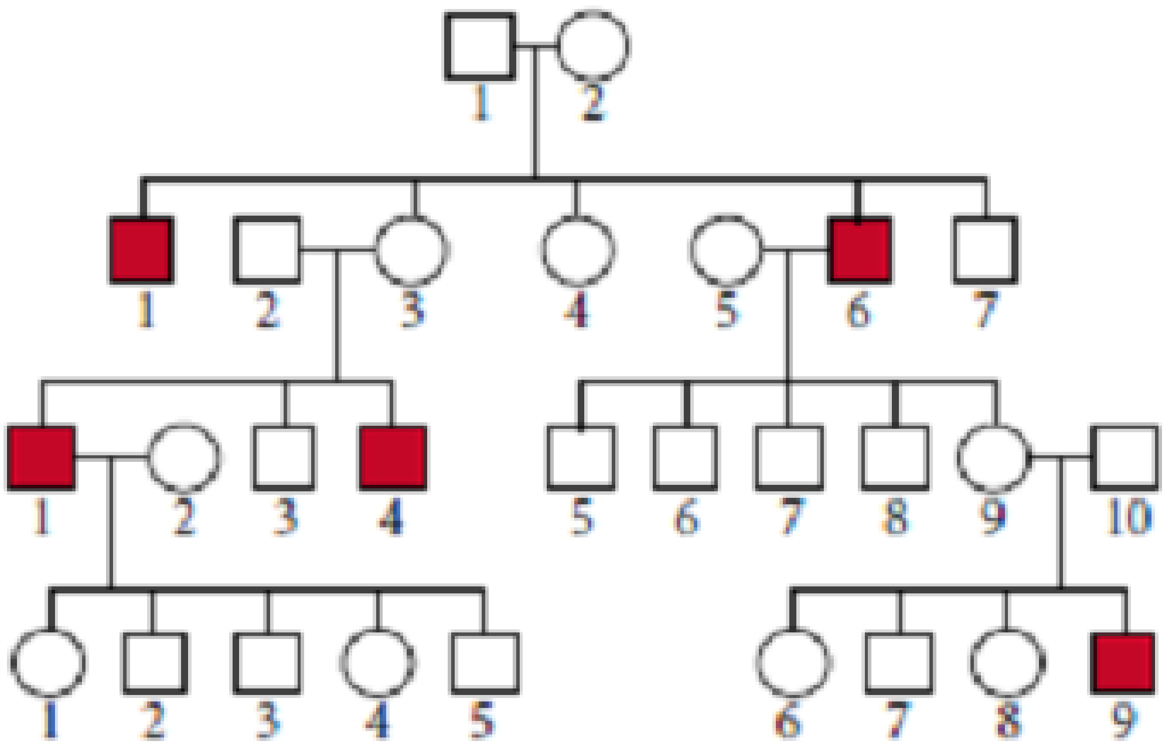
- Affected father will pass the trait to all the daughter
- Both males and females are affected- More females affected
- Doesn't skip the generation. Affected son/daughter will either one of the parent affected

- **X^D = dominant mutant allele**
- **X^d = recessive normal allele**



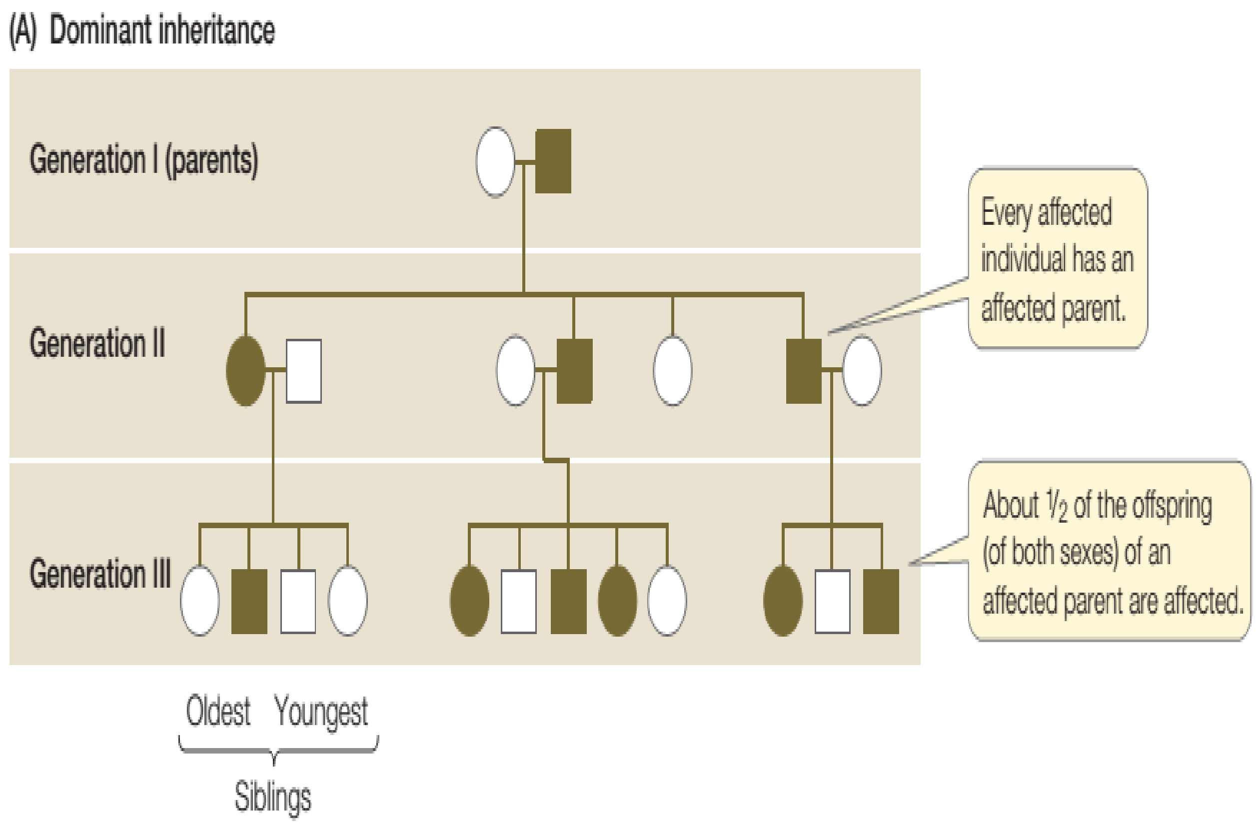
X Linked Recessive

- Females express (affected), only if they get a copy of the allele from both parents.
- More males than females are affected. Affected son born to unaffected mother
- All the daughter of the affected father are carriers
- Traits tend to skip the generation



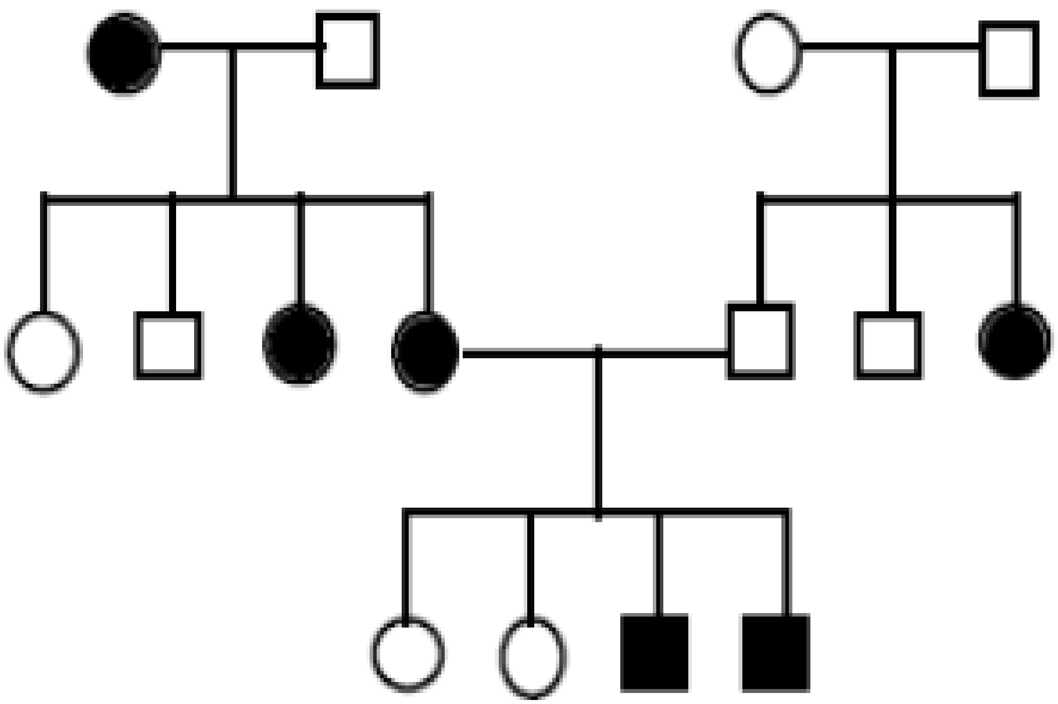
Autosomal Dominant

- Appears in the both the sexes
- Both the sexes transmits their traits to offspring
- <u>Doesn't skips the generation</u>
- Affected offspring must have a affected parent



Autosomal Recessive

- Appears in the both the sexes with equal frequency
- <u>Traits tend to skip the generation</u>
- Affected offspring are usually born to un affected parent



Autosomal recessive trait

1. Appears in both sexes with equal frequency.
2. Trait tends to skip generations.
3. Affected offspring are usually born to unaffected parents.

Autosomal dominant trait

1. Appears in both sexes with equal frequency.
2. Both sexes transmit the trait to their offspring.
3. Does not skip generations.
4. Affected offspring must have an affected parent, unless they possess a new mutation.
5. When one parent is affected (heterozygous) and the other parent is unaffected, approximately 1/2 of the offspring will be affected.
6. Unaffected parents do not transmit the trait.

X-linked recessive trait

1. More males than females are affected.
2. Affected sons are usually born to unaffected mothers; thus, the trait skips generations.

4. Is never passed from father to son.
5. All daughters of affected fathers are carriers.

X-linked dominant trait

1. Both males and females are affected; often more females than males are affected.
2. Does not skip generations. Affected sons must have an affected mother; affected daughters must have either an affected mother or an affected father.
3. Affected fathers will pass the trait on to all their daughters.

Y-linked trait

1. Only males are affected.
2. Is passed from father to all sons.
3. Does not skip generations.