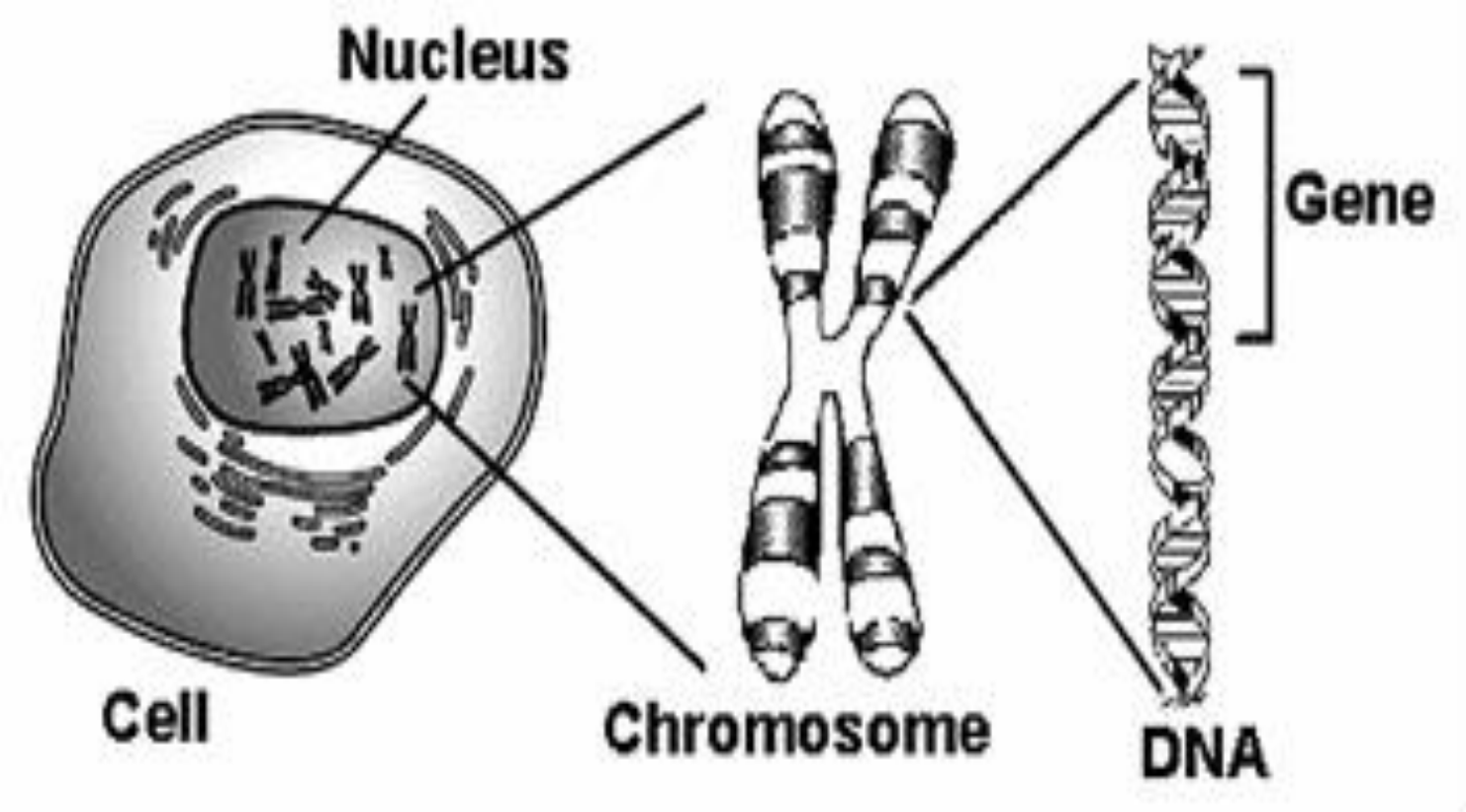
Mendelian Genetics

Biology 3201

Unit 3

Recall:

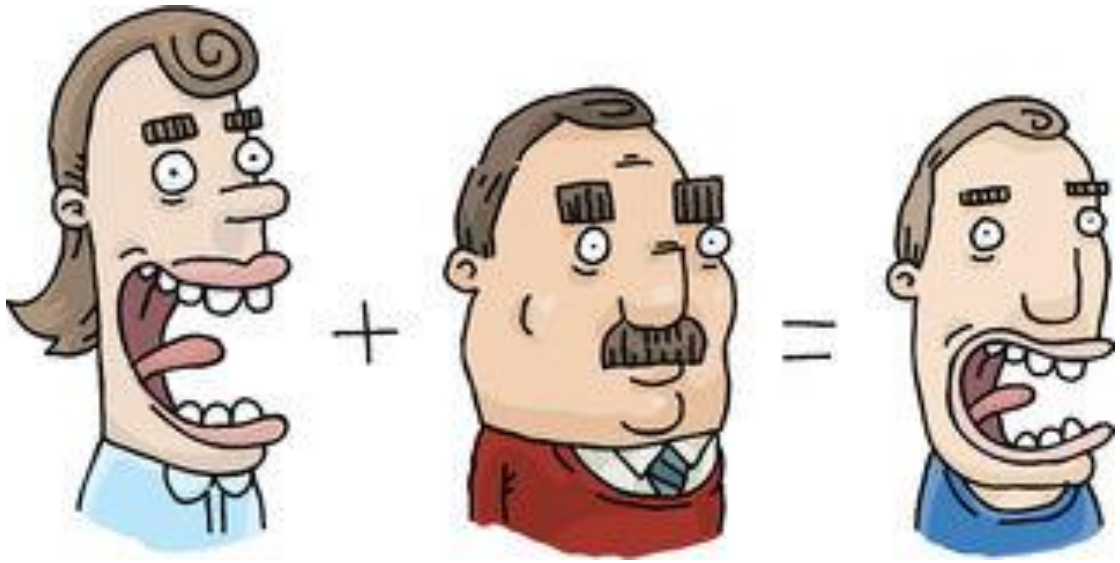


Terms

- **Genetics** is a branch of biology dealing with the principles of variation and inheritance in animals and plants.

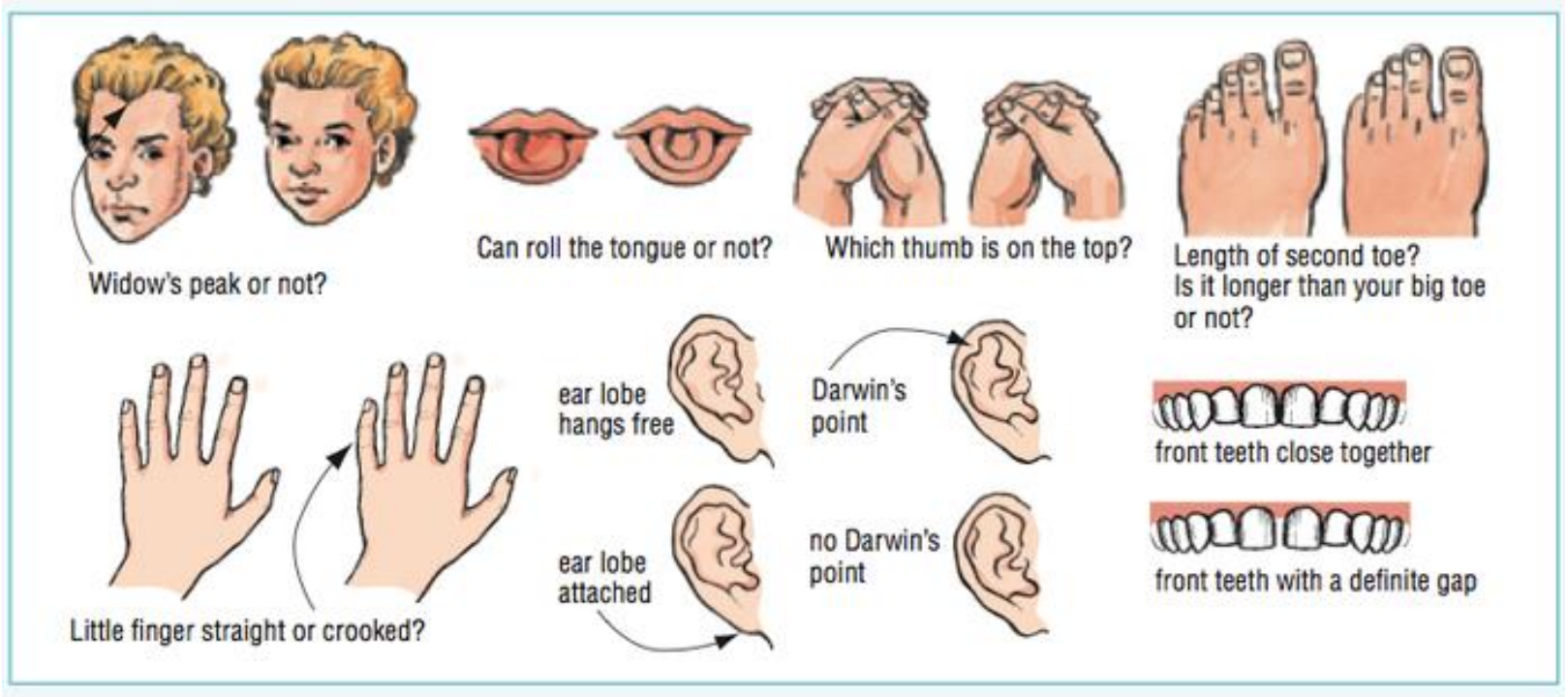
Heredity

- the passing of traits from one generation to another.



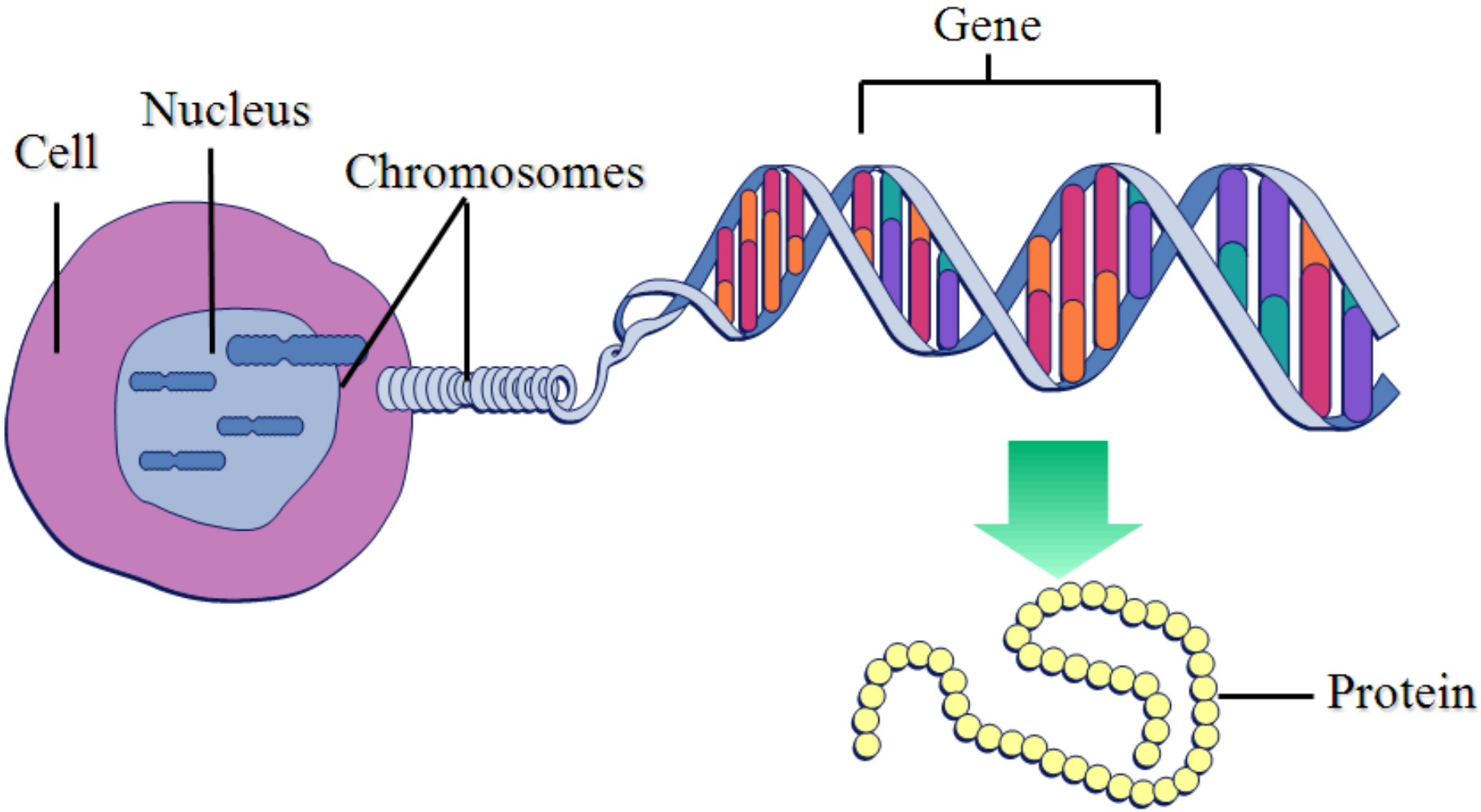
Traits

- sometimes called the characteristics that are inherited. Examples: height, hair colour, eye colour



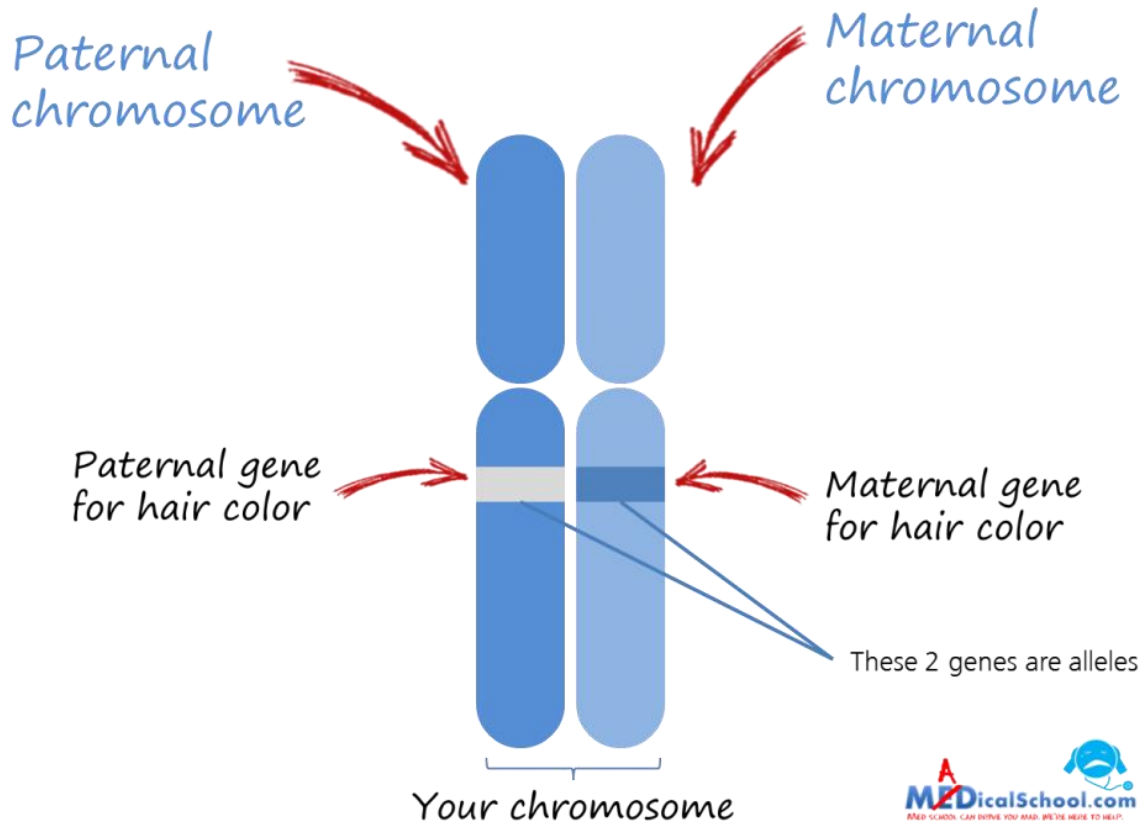
Genes

- A **gene** is the basic physical and functional unit of heredity.
- **Genes**, which are made up of DNA, act as instructions to make molecules called proteins.
- In humans, **genes** vary in size from a few hundred DNA bases to more than 2 million bases.


Gene
Nucleus
Cell
Chromosomes
Protein

Alleles

- An alternate form of a gene. Ex: Blonde, Brown, Black, Red hair



Alleles con’t

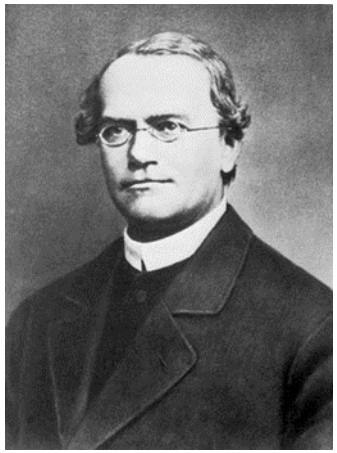
- Every trait can be represented by two alleles. One from each parent. Each allele is written as either a capital letter (dominant form) or lower case letter (recessive).

Example

Trait- stem height

Alleles- Tall (T) or short (t)

Mendel



- Gregory Mendel (1822-1884) The “Father of Genetics”. He was an Austrian monk. He studied garden pea plants as a means to explain inheritance.

Blend Theory

- Pre- Mendel, there was a belief that factors from parents were blended in their offspring. For example, a short parent and tall parent would create a medium height offspring. It did not explain the appearance or disappearance of traits from one generation to the next.

Mendel and the Scientific Process

- Gregor Mendel used the scientific process to lay the foundation for the science of genetics. Mendel used garden peas. They were a good choice because they were

1. **easy to grow**,
2. **matured quickly**
3. **showed distinct traits.**

Mendel's Peas

Trait	Dominant	Recessive
Seed Shape	***Round***	***Wrinkled***
Seed Color	***yellow***	***Green***
Seed Coat Color	***Grayish brown***	***White***
Pod Color	***Green***	***Yellow***
Pod Shape	***Inflated***	***Wrinkled***
Stem Length	***Long***	***Short***
Flower Position	***Lateral***	***Terminal***

	Flower Color	Flower Position	Pea Color	Pea Shape	Pod Color	Pod Shape	Height
Dominant	 purple	 axial	 yellow	 round	 green	 inflated	 tall
Recessive	 white	 terminal	 green	 wrinkled	 yellow	 constricted	 short

Genotype- The combination of alleles for a trait. It is the **written expression** of the trait. Every genotype is **written with two alleles** (one that came from mom's chromosome and one from Dad's)

Ex: BB, Bb, bb

Phenotype- The **physical observable expression** of the trait.

Ex: Blue eyes, Brown eyes

Genotype vs Phenotype

BB, Bb →

bb →

Dominant

- Some alleles are dominant, some are recessive.
- Dominant alleles are ALWAYS expressed in the phenotype. They will mask or hide recessive alleles it is paired with. They are written with CAPITAL LETTERS.

- Ex: Brown eyes (B) are dominant to blue eyes (b)

  BB, Bb

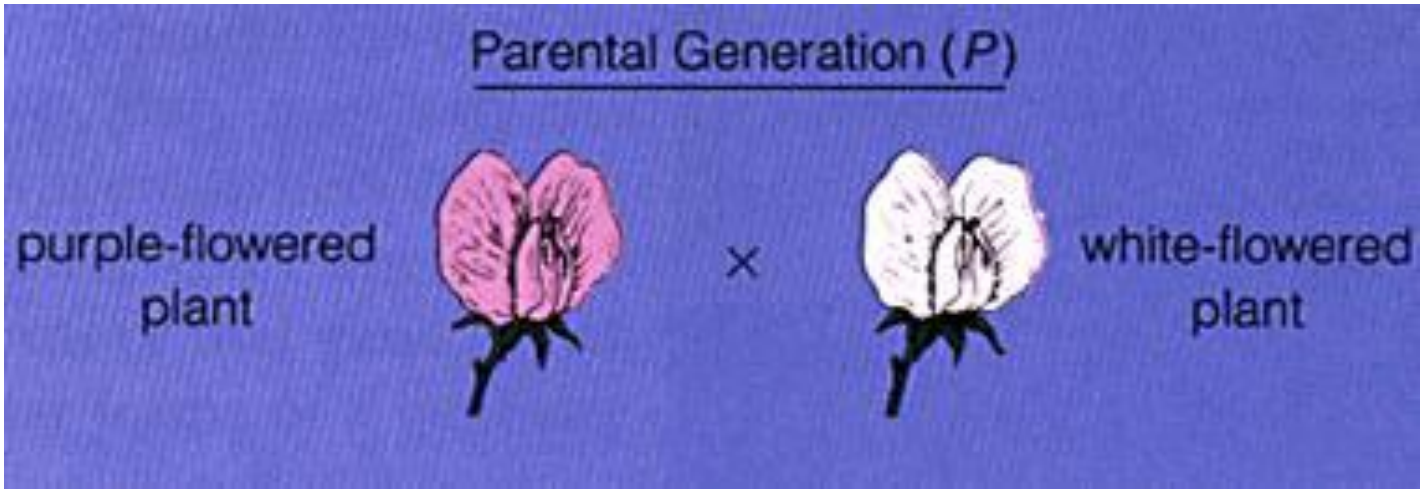
Recessive

Recessive alleles are written with lower case letters. They are NOT expressed unless they are paired with another recessive allele.

Ex: Blue eye colour is recessive to brown. Blue eyes genotype is bb

P Generation

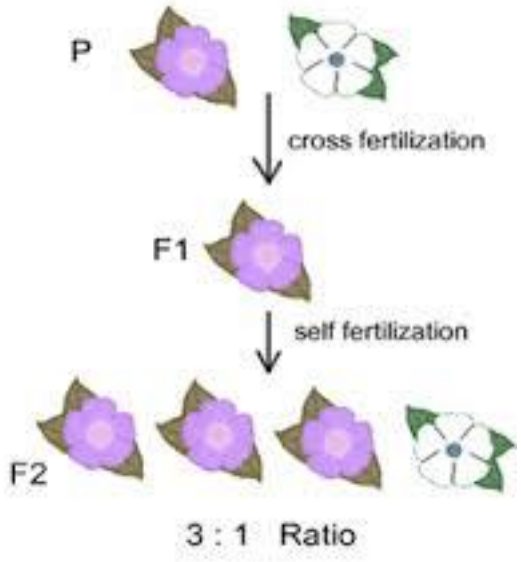
- Parental Generation



F1 and F2 Generation

- 1st Filial Generation is the offspring of the P generation.

- 2nd Filial Generation is the offspring produced by crossing the F1 organisms.



Heterozygous Vs Homozygous

Heterozygous- If a genotype contains both a dominant and recessive allele it is heterozygous.

Ex: Bb, Tt, Ww

Homozygous- If a genotype contains two of the same alleles (dominant or recessive), it is homozygous.

Ex: BB, bb, TT, tt

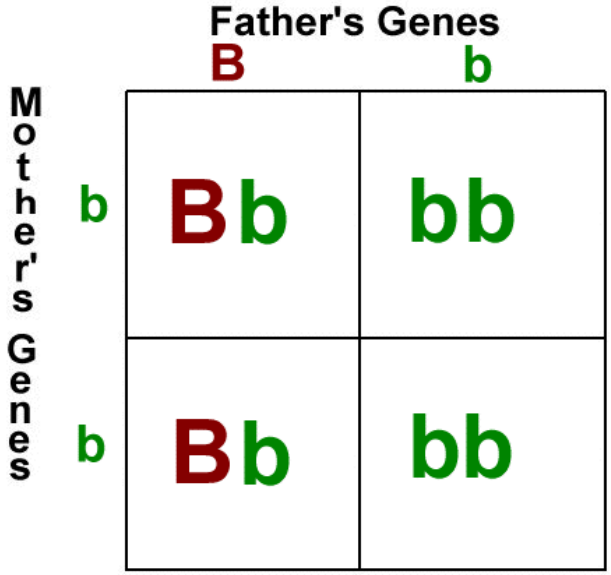
Pure breed- same as homozygous. The genotype contains the same alleles for a trait. Ex: TT or tt

Hybrid- same as heterozygous. The genotype contains two different forms of a trait. Ex: Tt

Punnett Square

- A diagram that shows all the possible combinations of alleles (genotypes) that could be expressed in the offspring of two individuals.

Ex:



Monohybrid Cross

- A mating of two individuals, **involving one trait.**

Ex: Bb (dad) x bb (mom)

Brown eyed father x blue eyed mother

Dihybrid Cross

- The mating of the two individuals **involving two traits.**

Ex: BbDD (dad) x BBdd mom

Brown eyes / Dimples x Brown eyes / no dimples

Product Rule

- To predict a given event will occur repeatedly. We must multiply the probability of each individual event occurring.

Ex: What's the chance that a couple will have 4 girls in a row?

½ x ½ x ½ x ½

Generations

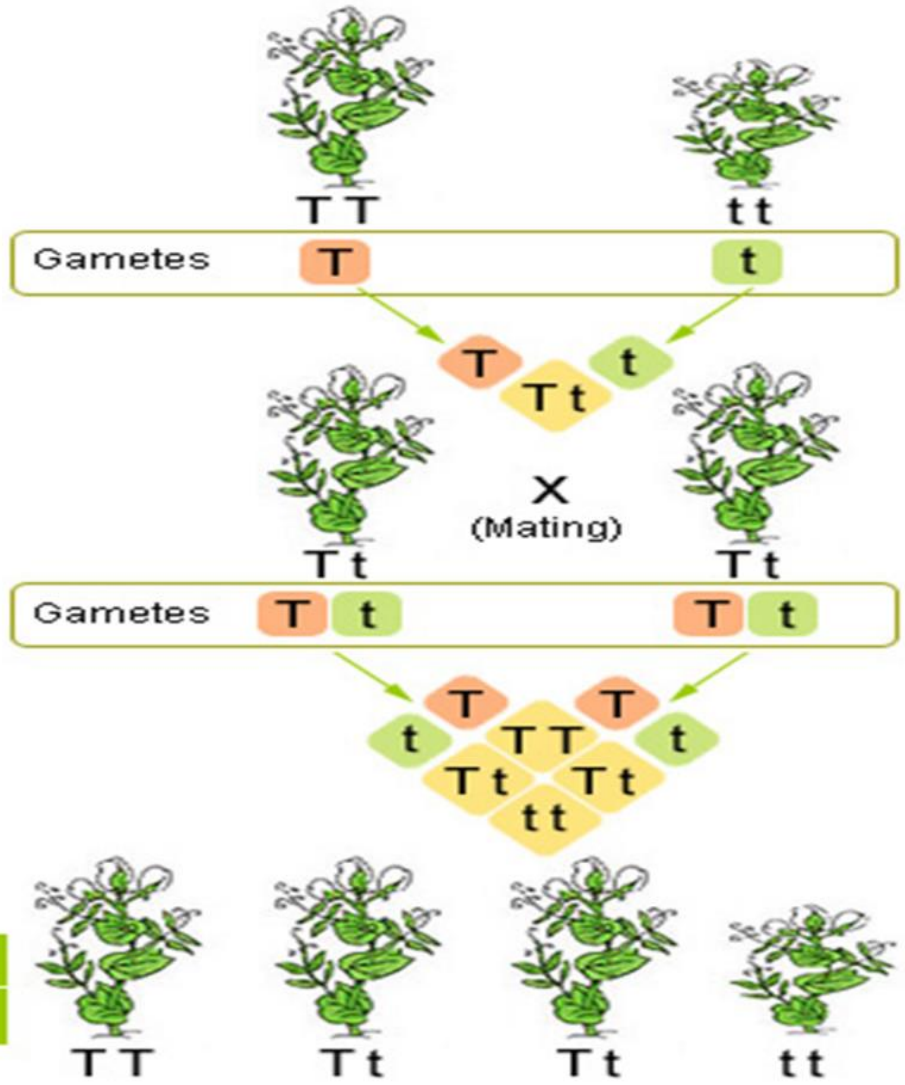
Parental Generation

F1 Generation

Genotype	All are Tt.
Phenotype	All are tall

F2 Generation

Genotype	TT : Tt: : tt = 1 : 2 : 1
Phenotype	Tall:Dwarf 3:1



Single Gene Traits- Humans

Dominant Gene	Recessive Gene
Cleft Chin 	No Cleft 
Widow's Peak 	No Widow's Peak 
Dimples 	No Dimples 
Brown/Black Hair 	Blonde Hair 
Freckles 	No Freckles 
Brown Eyes 	Gray/Blue Eyes 
Free Earlobe 	Attached Earlobe 

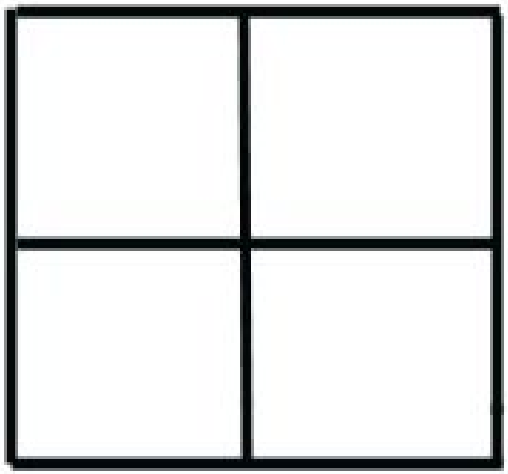
Writing genetics:

For the trait Earlobe attachment, free earlobes (F) are dominant to attached Earlobes (f)

Pure Bred (homozygous)	*Alleles*
Hybrid (heterozygous)	*Alleles*

Mendel's First Cross (P Generation)

- Mendel crossed (P): Pure tall (TT) x pure dwarf (tt)



- The result (F1): All offspring were tall (Tt)

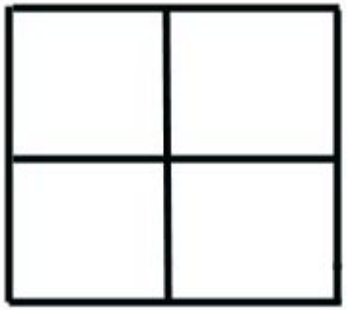
But these were not pure tall, because each parent had a different trait for stem height. Instead, these offspring are referred to as hybrids or hybrid tall.

If there is at least **ONE dominant allele**, they would have the **dominant trait** *(A capital letter is used to represent dominant. Ex: T)*

- In order to show the recessive trait, you must have **TWO recessive alleles**. The dominant allele will mask or prevent the expression of the **recessive allele.** *(A lowercase letter is used to represent recessive. Ex: t)*

Mendel's second cross (F1)

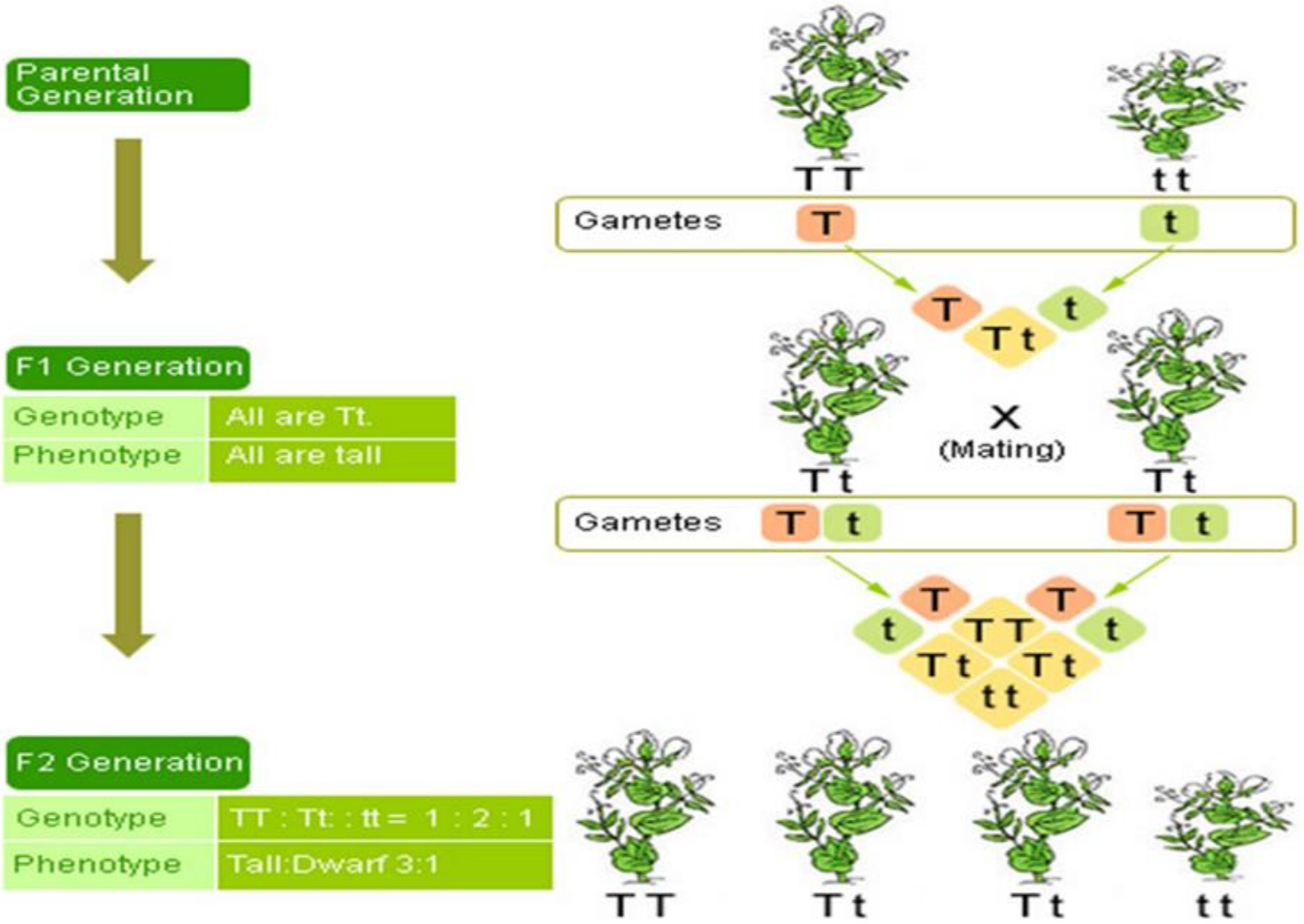
Mendel crossed: Hybrid tall (Tt) x Hybrid tall (Tt)



The result (F2): Some of the offspring were tall and some were short (dwarf).

About ¾ were tall and ¼ were short (3:1 ratio). This showed that the factor that contained the shortness had not disappeared.

Summary


Parental Generation
F1 Generation
Genotype
All are Tt.
Phenotype
All are tall
F2 Generation
Genotype
TT : Tt: : tt = 1 : 2 : 1
Phenotype
Tall:Dwarf 3:1
T T
t t
Gametes
T
t
T t
X
(Mating)
T t
T t
Gametes
T t
T t
T
T
t
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Mendel’s Conclusions

- The inheritance of each trait is determined by "units" or "factors" that are passed on to descendents unchanged. We call these units genes today.

- An individual inherits one such unit from each parent for each trait

- A trait may not show up in an individual but can still be passed on to the next generation.

- If the dominant factor is present, it will be expressed even if the recessive factor is also present. The recessive factor will be expressed if only recessive factors are present

Mendel's Explanations

- **Purebred** plants have two of the same allele.
  - Example: Pure tall = TT or pure dwarf = tt
- **Hybrid** plants have two different alleles.
  - Example: Hybrid tall = Tt
- Mendel concluded that heredity was not just a blending of traits and he developed the **principle of dominance**

Law of Dominance

- **<u>Law of Dominance</u>:** When an organism is heterozygous for a pair of contrasting traits, only the dominant trait can be seen in the hybrid. *Remember: A dominant trait is indicated by an upper case letter (Ex: T) whereas a recessive trait is indicated by a lower case letter (Ex: t)*

Three possible Allele Combinations for plant height	Heterozygous or homozygous?	Dominant or recessive?

Practice....

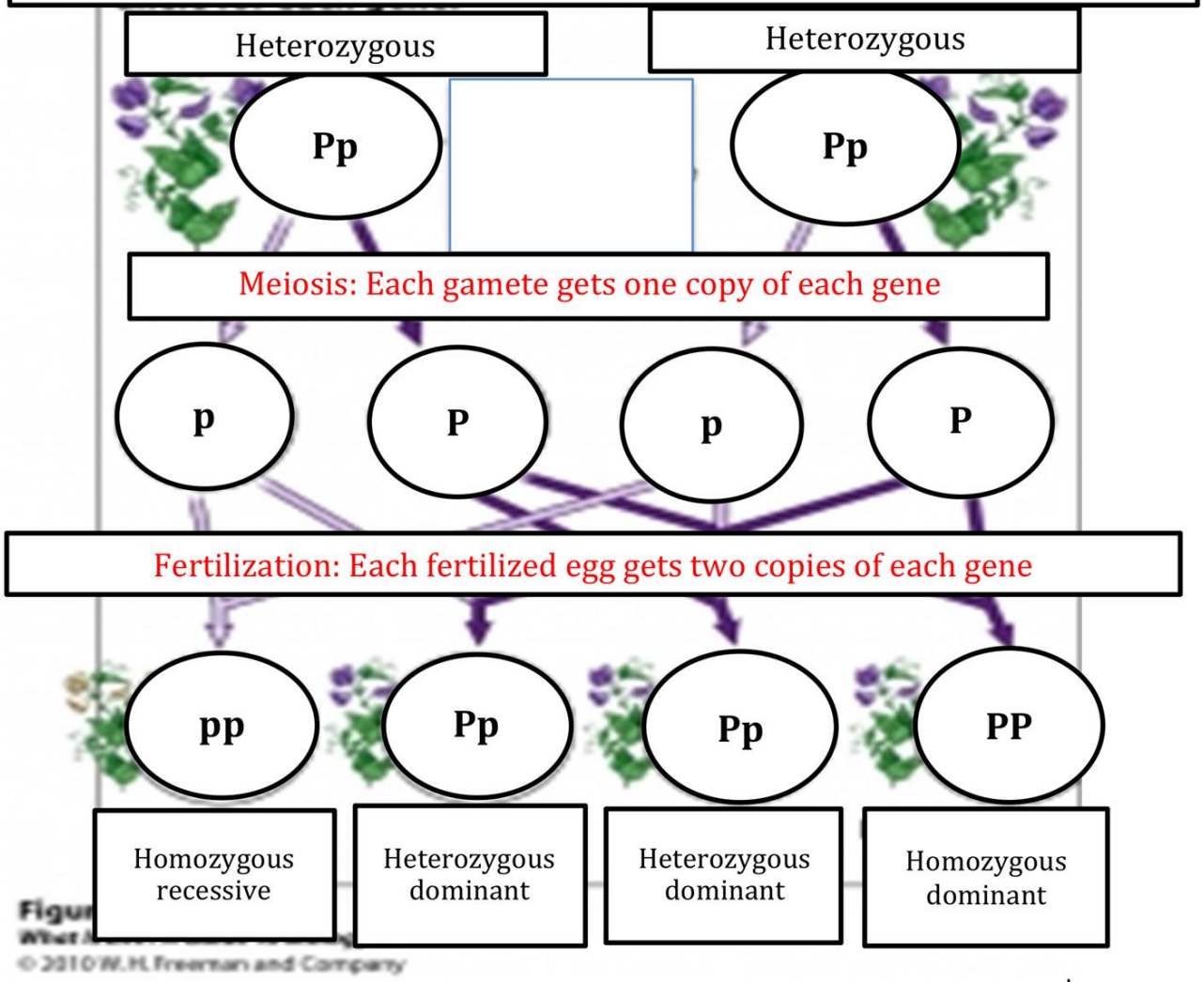
Law of Segregation

- This law states that “factors” (*genes*) occur in pairs (*alleles*) and are separated from each other in gamete formation and recombined in fertilization.

- One of these factors came from the mother and the other factor came from the father during zygote formation

Mendel's Law of Segregation

According to Mendel's law of segregation, only one of the two alleles for a gene is put into a gamete. At fertilization, offspring receive from each parent one allele for each gene



Probability

- **Probability Activity**
- Toss an integer chip 10 times and record how many times you get red and yellow

- Red:
- Yellow:

- **Probability –** the chance, or likelihood, of a particular outcome

- **Product Rule –** the probability that two or more independent events will occur together is the product of their individual probabilities of occurring alone

What is the probability you will get 3 red integer chips in a row? _____

Example 1. What is the probability of having two girls in a row?

- Answer:
- Each time there is ½ chance of having a girl or boy, so using the product rule,
- ½ x ½ = ¼ or 25% chance of having two girls.

Example 2: What is the probability of having a boy, after having two girls in a row?

- Answer:
- Since having two girls in a row has no influence over the sex of the third child, the probability is still ½ or 50%

Example 3:

a. What are the chances of rolling a 2 on a die?

b. What are the chances of rolling a 2 twice in a row?

c. What are the chances of rolling a 2 after having rolled a 5 three times in a row?

Test Cross

- Sometimes you do not know if a parent is homozygous dominant **(TT)** or heterozygous dominant **(Tt).** When this is the case, you can perform a **test cross.**

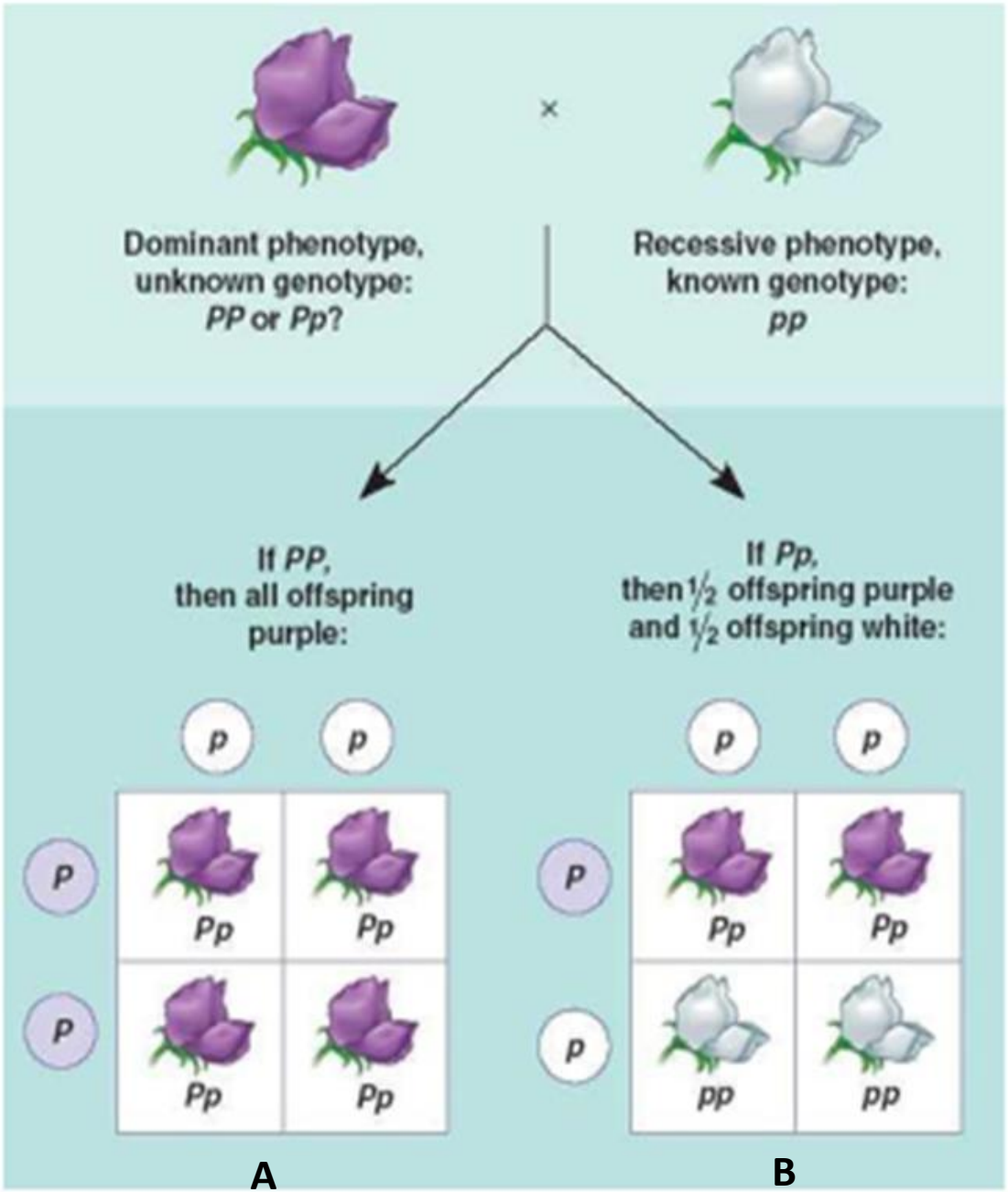
What is a Test Cross?

- A test cross is used to determine an unknown genotype that may either be homozygous dominant or heterozygous for a trait.

- An individual of the unknown genotype is mated with an individual showing the contrasting recessive trait.

Example of a Test Cross:

- Let's say we just bought a purple flower. We want to know; is our purple flower homozygous for purple (PP) or heterozygous (Pp)? To determine this, cross it with a homozygous recessive (white) flower:



- If any of the offspring show the recessive condition then the unknown parent MUST have been heterozygous.

Conclusions:

- Outcome “A”: If the cross resulted in 100% of the offspring being purple, we could assume our Purple flower had the genotype: PP, homozygous dominant.
- Outcome “B”: If the cross resulted in 50% of the offspring being purple and 50% being white, we would know our purple flower had the genotype: Pp, heterozygous dominant.

Example 2: A breeder wishes to know if a pea plant is **TT** or **Tt**. He will cross the plant with a homozygous recessive plant (**tt**).

Practice!

Complex Inheritance

- Mendel wanted to know if the inheritance of one characteristic influenced the inheritance of a different characteristic. (Ex: Does pea shape influence pea colour?).
- Mendel's 2nd experiment looked at how multiple traits are inherited.

- He produced purebred pea plants which were: Round Yellow plants (**RY**) and Wrinkled Green plants (**ry**) and used a Dihybrid Cross to show his results.

What is a Dihybrid Cross?

- This is a chart to show the results of a cross between parents for **two traits**.
- Example: A yellow round seed male is crossed with a green wrinkled seed female.
- Yellow- Round seed Male (**YYRR**)
- Green – Wrinkled seed female (**yyrr**)

What were the results of mendel's 2nd experiment?

- A Dihybrid cross of the F_1 generation produced **nine different genotypes.**
- These results represent a phenotypic ratio of **9:3:3:1**
- This led Mendel to propose the ***Law of Independent Assortment***

♂ gametes

♀ gametes	R Y 1/4	R y 1/4	r y 1/4	r Y 1/4
R Y 1/4	RR YY 1/16 	RR Yy 1/16 	Rr Yy 1/16 	Rr YY 1/16 
R y 1/4	RR Yy 1/16 	RR yy 1/16 	Rr yy 1/16 	Rr Yy 1/16 
r y 1/4	Rr Yy 1/16 	Rr yy 1/16 	rr yy 1/16 	rr Yy 1/16 
r Y 1/4	Rr YY 1/16 	Rr Yy 1/16 	rr Yy 1/16 	rr YY 1/16 

9  : 3  : 3  : 1 

 Round, yellow

 Round, green

 Wrinkled, yellow

 Wrinkled, green

What is Law of Independent Assortment?

- During prophase I of meiosis, the alleles for different traits cross over (tetrads). This mixes chromosomes from your father and mother.

Law of Independent Assortment:

- states that **genes** for different traits are separated and distributed to gametes independently from each other during **crossing over**. The inheritance of alleles for one trait **does not** affect the inheritance of alleles for another trait.
- This means that offspring may have new combinations of alleles that are not present in either parent, making them **genetically unique**.

How To Do A Dihybrid Cross Step By Step!

#1 - Make a legend (assign letters for alleles, i.e. T=tall, t=short)

#2 – Show parents 4 dashes times 4 dashes.

_____ _____ _____ _____ x ___ _____ _____ _____

#3 Perform "FOIL" on each parent to make gametes.

#4 make your Punnett square.

Tip- Keep Same letter together in the boxes and always put the capital letter first (when possible)

Practice…

- 1. A male and a female guinea pig are both **heterozygous** for fur colour and fur texture. Both dark fur (D) and rough fur (R) are dominant traits.

Different Patterns of Inheritance

- Mendel found that inherited traits were either dominant or recessive. The dominant allele in an individual was always expressed, even if the recessive allele was present. However, some organisms show different patterns of inheritance.

What is Incomplete Dominance?

- **Incomplete Dominance** - The blending of two alleles
- There is no definitive dominant or recessive allele.
- Red and white flowers make **pink flowers**

Ex: White and Red snapdragons are homozygous; Pink snapdragons are heterozygous

- Red snapdragon (RR)
- White snapdragon (WW)
- Pink snapdragon (RW)

KEY

R = Red gene

W = White gene

RR = Red

WW = white

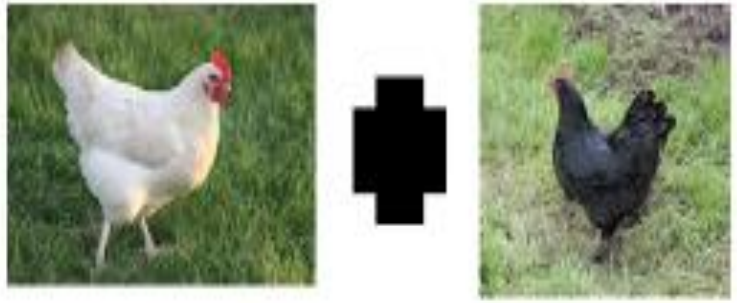
RW = pink

What is Co-dominance?

- A situation where two alleles may be **expressed equally**.
- There is no definitive dominant or recessive allele.
- Ex: Red and white flowers make **red and white spotted flowers**

Another common example…

Another common phenotype in questions about codominance is the black and white feathered checkered chicken. Chickens may be black (= all black feathers), white (= all white feathers), or checkered (= black and white feathers together).

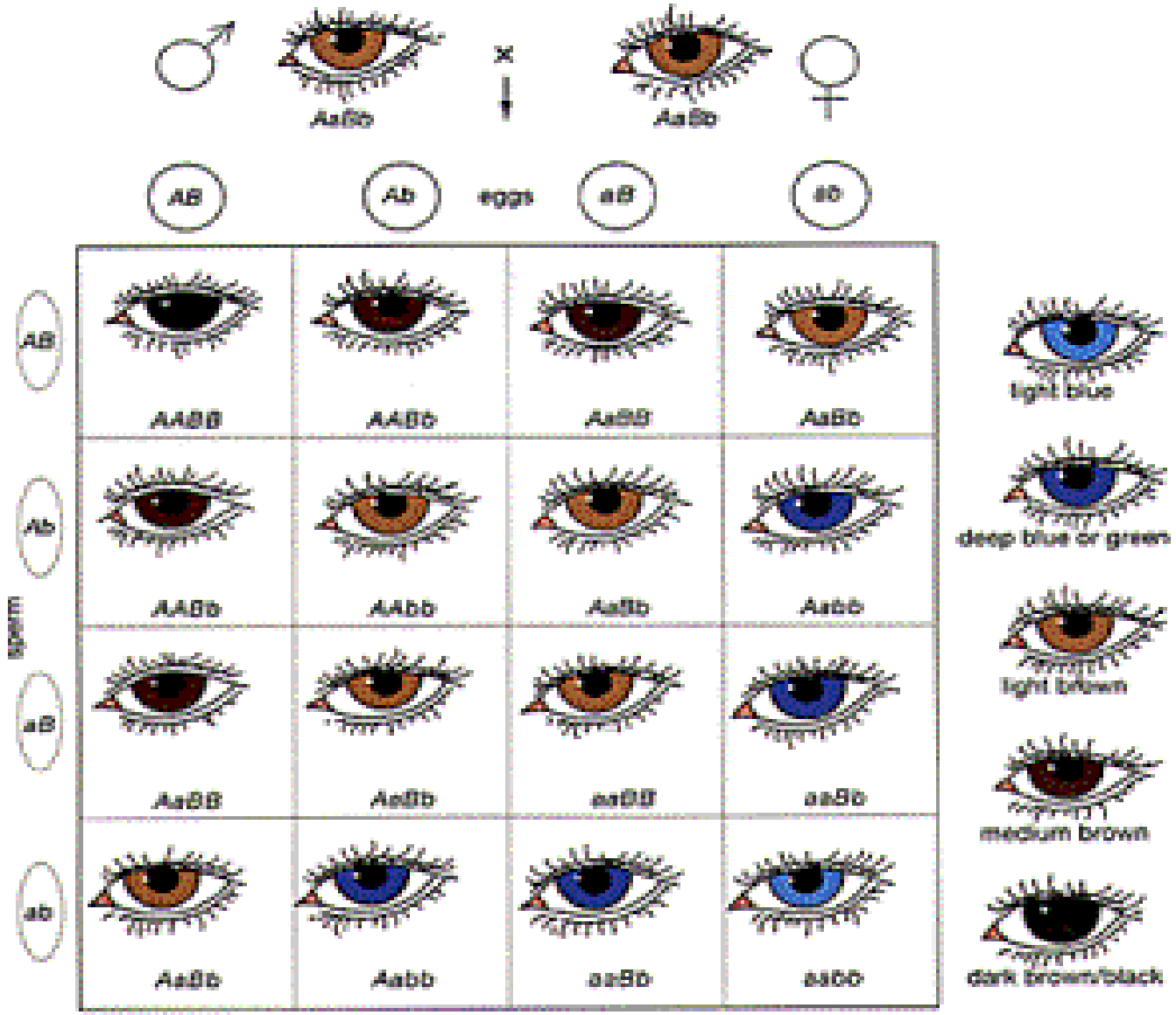


What is Polygenic Inheritance (Multiple Gene Inheritance)?

- When many genes are responsible for a trait. This is an **exception to Mendel's Rule**
- For example, humans are not just tall or short. Sometimes traits vary between two extremes, such as tall and short

- When two or more independent genes affect a characteristic, it is called polygenic or multiple-gene inheritance.

- Other examples are: eye color, skin color


AaBb
x
AaBb
AB
Ab
eggs
aB
ab
sperm
AABB
AABb
AaBB
AaBb
AABb
AAbb
AaBb
Aabb
AaBB
AaBb
aaBB
aaBb
AaBb
Aabb
aaBb
aabb
light blue
deep blue or green
light brown
medium brown
dark brown/black

What are Multiple Alleles?

- A trait that is determined by more than two alleles (ex. Human blood types)
- Human blood types are determined by 3 alleles
- These alleles control the production of antigens on the surface of the blood cells
  - Allele I^A produces antigen A
  - Allele I^B produces antigen B
  - Allele i produces no antigen

- ABO Genotypes/Phenotypes Compatibility
  - **Type O blood** can donate blood to anyone (universal donor)
  - **Type AB blood** can receive blood from anyone (universal recipient)

Note:

- I^A and I^B are said to be co-dominant
- I^A is dominant over i
- I^B is dominant over i

Human Blood Types:

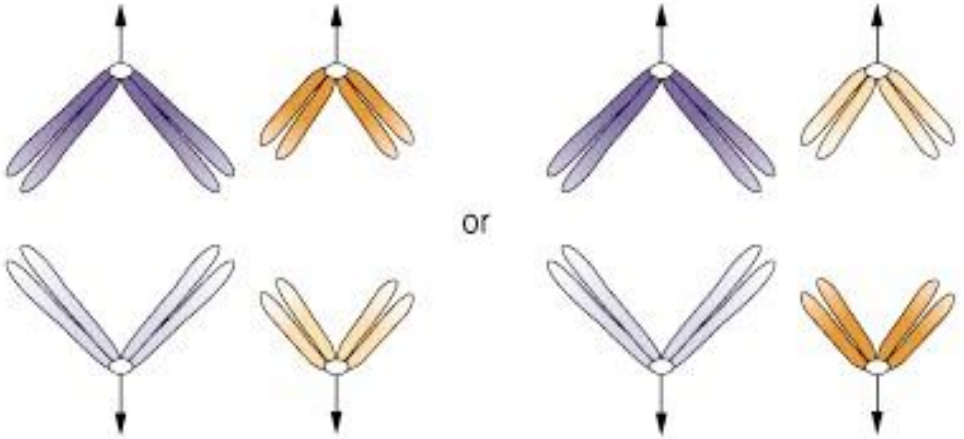
Phenotype (blood type)	Genotypes
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B
O	ii

Post- Mendel

- Theodor Boveri and Walter Sutton started to study **chromosomes**

What observations did Boveri and Sutton make?

1. Chromosomes occur in pairs
2. The pairs segregate during the anaphase I stage of meiosis
3. Chromosomes line up independently of one another (metaphase)

	Mendel's factors	Chromosomes
Pairing	*A* *a*	
Segregation	*A* *a*	
Independent assortment	*A* *B* or *A* *b* *a* *b* *a* *B*	

What is the Chromosome theory of inheritance?

1. Mendel's 'factors' (genes) are carried on chromosomes

2. It is the segregation and independent assortment of chromosomes during meiosis that accounts for the patterns of inheritance

- *Video: Punnett Squares and Sex-Linked Traits*
- https://www.youtube.com/watch?v=h2xufrHWG3E&safe=active

What is Sex-Linked Inheritance?

- The transmission of genes located on one of the sex chromosomes, either **X or Y**

- A gene that is located on the X chromosome only is called **X-linked**.
- A gene that is located on the Y chromosome only is called **Y-linked.**

Most of the known sex-linked traits are **X-linked.**

Sex-Linked Traits in Fruit Flies

- Thomas Morgan (1910) studied eye color in fruit flies
- (Scientific name *drosophila*)

- He discovered that eye color is an *X-linked trait. This was the first time that a gene had been linked to a specific chromosome!*

- ***When doing Punnett squares for these traits, the alleles are written on the X chromosome (for x-linked traits) and no allele is written with the Y chromosome. When the genotypic and phenotypic ratios are written, the sex is always included***

- He also discovered that **white eyes** are a **recessive trait** and **red eyes** are **dominant.**

Writing Sex-Linked Traits

For fruit flies:

- Remember, eye colour is x-linked.
- Red is dominant (R)
- White is recessive (r)

Possible Female genotypes: X^RX^R, X^RX^r, X^rX^r

Possible Male genotypes: X^RY, X^rY

- ***When doing Punnett squares for these traits, the alleles are written on the X chromosome (for x-linked traits) and no allele is written with the Y chromosome. When the genotypic and phenotypic ratios are written, the sex is always included***

Example: In fruit flies, eye color is X-linked and therefore is not carried on the Y chromosome. Red (R) is dominant to white eyes (r). Show a punnett square for a cross between a **white eyed male and a homozygous Red eyed female**. State the phenotypic ratio of the F1 generation.

P:	Male	x	Female
	X^rY		X^RX^R
Gametes:	X^r or Y		X^R

F1 Punnett Square:

	X^R
X^r	X^RX^r
Y	X^RY

Phenotypic Ratio: ½ girls with Red eyes : ½ boys with Red eyes

Red-Green Colour-blindness

- Red Green color blindness is a sex-linked condition.
- The gene loci are on the non-homologous region of the **X-chromosomes**.
- Red Green color blindness is more common in males than in females.

Why?

- This is due to the fact that men only have one X chromosome (XY). Subsequently, genes on that chromosome are usually expressed in the male phenotype even if they are recessive since there are no corresponding genes on the Y chromosome in most cases.
- - In women, a recessive allele on one X chromosome is often masked in their phenotype by a dominant normal allele on the other. This explains why women are frequently carriers of X-linked traits but more rarely have them expressed in their own phenotypes.

Writing Genotypes for Colour-blindness

Normal is Dominant (N)

Colour blind is recessive (n)

Possible female genotypes: X^NX^N, X^NX^n, X^nX^n

Possible male genotypes: X^NY, X^nY

Haemophilia

- Haemophilia is a recessive, sex-linked genetic disorder.
- Persons suffering from haemophilia are unable to produce clotting factor.
- The haemophiliac allele (X^h) is recessive to the normal allele (X^H).
- The gene is located on the non-homologous region of the x-chromosome.
- Haemophilia is more common in men than women.
- *Since (until recently) the prognosis for survival was poor and haemophiliac males did not survive to pass on the allele to their daughters (it's on the X-chromosome). Therefore, female haemophilia was rare.*

- *Since (until recently) the prognosis for survival was poor and haemophiliac males did not survive to pass on the allele to their daughters (it's on the X-chromosome). Therefore, female haemophilia was rare.*

Writing alleles for Haemophilia

- H- Normal (Dominant)
- h- haemophilic (recessive)

Normal Females: Normal Male:

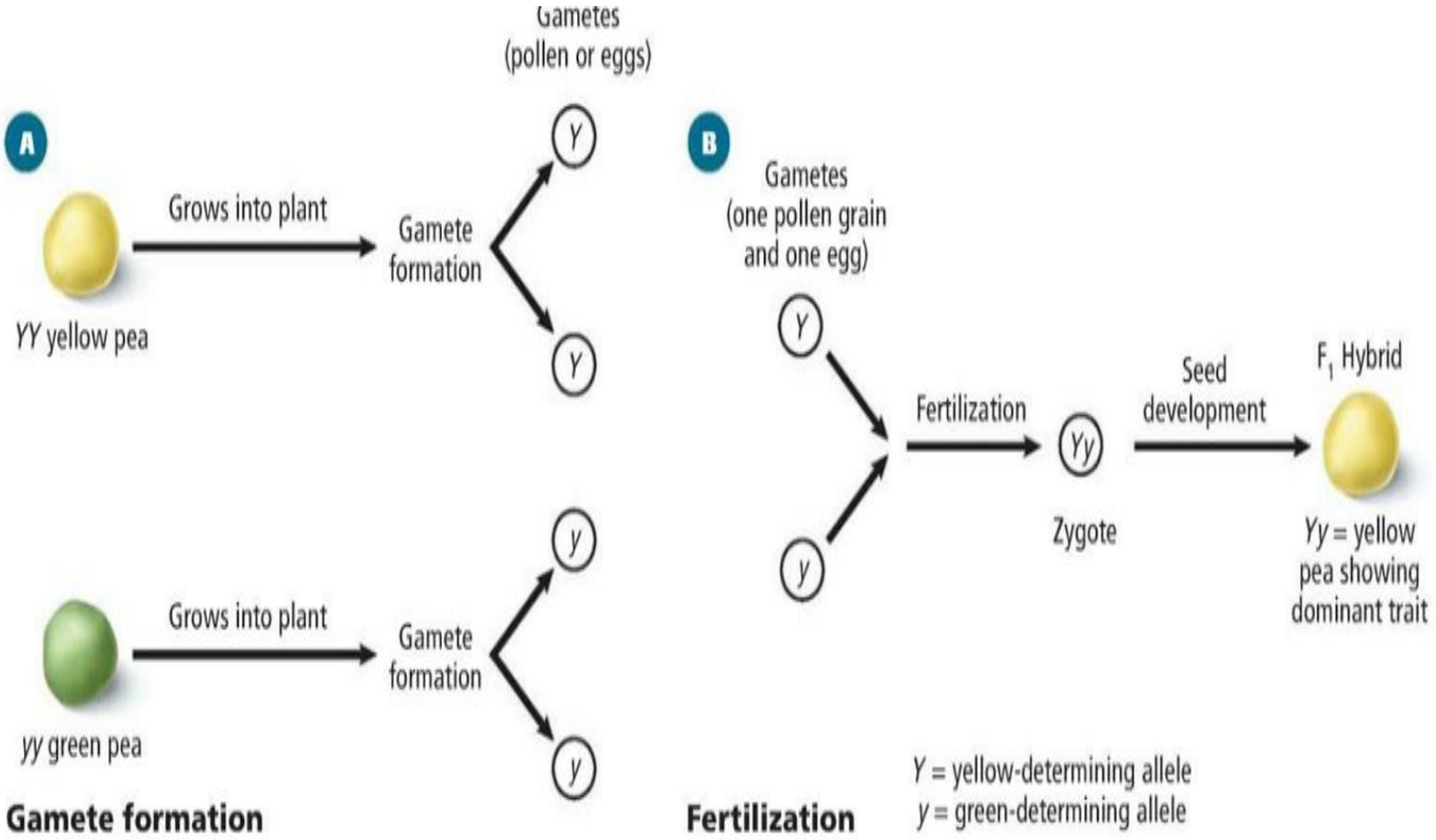
X^HX or X^HX^h X^HY

Hemophilia Females: Hemophilic Male:

X^hX^h X^hY

Gene-Chromosome Theory

1. Genes exist at specific sites arranged in linear fashion along chromosomes.
2. When pairs of homologous chromosomes separate during gamete formation, they form two gametes.
3. Each gamete will contain a separate allele for each trait.
4. During fertilization, chromosomes from one gamete will combine with another gamete.


A
Gametes
(pollen or eggs)
Grows into plant
Gamete
formation
Y
Y
YY yellow pea
Grows into plant
Gamete
formation
y
y
yy green pea
Gamete formation
B
Gametes
(one pollen grain
and one egg)
Y
y
Fertilization
Yy
Zygote
Seed
development
F1 Hybrid
Yy = yellow
pea showing
dominant trait
Fertilization
Y = yellow-determining allele
y = green-determining allele

Gene-Linkage

- Mendel's law says ALL genes are inherited independently of one another YET some traits (and thus genes) are often inherited together, like red hair and freckles. This is because some genes are located very close together on a chromosome and are **unlikely** to be separated during a crossing over event! These genes are said to be linked.

- *Therefore* : **genes on chromosomes** which are **very close together** will almost always be **inherited together.**

- *And* : **genes** located **far apart** are more likely to be **separated during a crossing over event**. The likelihood of crossing over increases with the distance between the two genes

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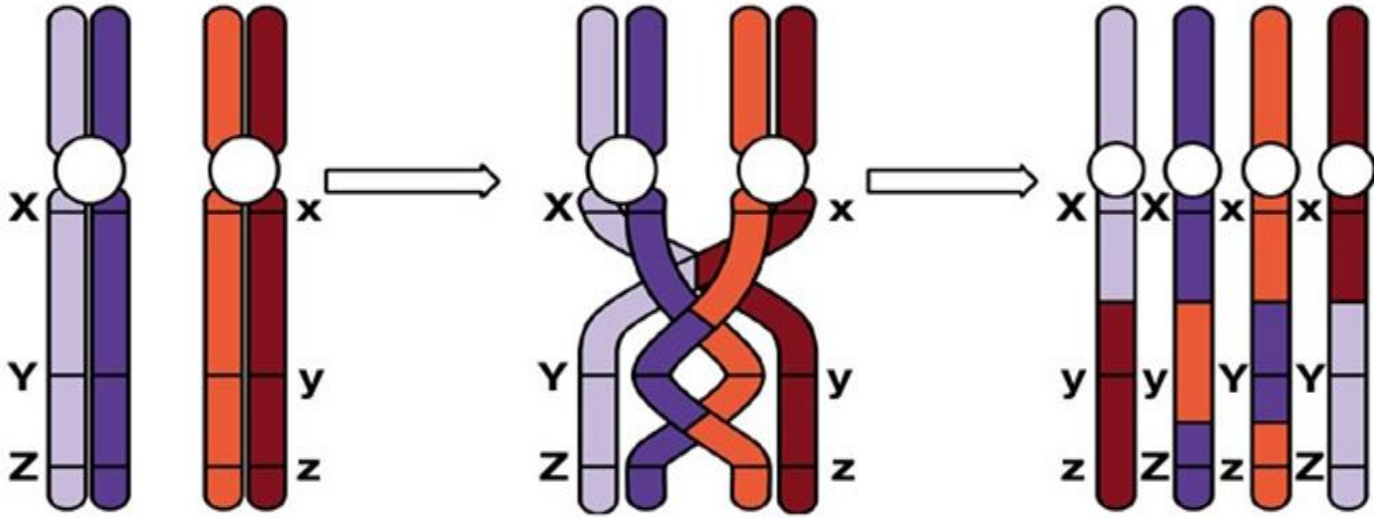
Crossing over during meiosis



- If crossing over does not take place, genes that are located on different chromosomes will assort independently while genes that are located on the same chromosome will be inherited together. This is the **MODERN VERSION OF THE LAW OF INDEPENDENT ASSORTMENT**