



THE LOGIC OF MENDEL

All of us have some characters similar to our parents or grand parents. Many of us likes to look like or behave like our parents. Our grand parents, most of them are farmers, selected the best seeds for their next season. All of them knew that the best seeds have the best characters. Characters are passed to next generation through seeds. Seeds comes from flowers. Bees visit flowers for honey. Flowers contain honey fine powders.

Here we are trying to understand logically, how a normal man like Mendel who had lowest grades in Biology could do such a meticulously planned experiment? Above all he had failed for a teaching certificate in natural sciences! On the other hand Mendel studied physics, mathematics and chemistry along with important aspects of biology. This mathematical back ground enabled Mendel to plan his experiments, draw out a theory and experimentally evaluate it.

We are logically going to understand what brought Mendel to understand the concept. Remember to consider the context that are looking at this from Mendel's perspective. He knew nothing about meiosis, never considered chromosomes or whether they had anything to do with what he was investigating. The word "gene" was invented after his death. All he did was breed peas, and do some high quality thinking. All this done between 1856 and 1863!

It is also interesting to see what is Mendel's previous knowledge? Sometimes early in his life he worked as a gardener and done the beekeeping. He also studied practical and theoretical philosophy and physics at the University of Olomouc, Czech republic. He happened to see the research of hereditary traits of plants and animals, in the department of natural history and agriculture. He also worked as a substitute high school teacher. He failed in getting a certificate for teacher and send to University of Vienna for further study on a sponsorship. He returned and again worked as physics teacher, but failed on the oral part of examination. Finally he was taken the superior priest of the monastery.

Mendels work can be divided into the following steps: (A) Preparation for experiments (B) Choice of experimental material (C) Planning and execution of experiment (D) Interpretation of experimental results and (E) Further testing of his observations.

A. Preparation for Experiments

How do to an experiment? The laboratory should be accessible! That is Mendel approached the head of the department of Natural history and agriculture where he was working as a priest for permission to use the 2 hectar experimental garden intended to study differences in plants. His colleges conducted studies on the heredity of sheep. Why he wanted the entire full 2 hecters rather a few cents or pots ? If we need to have a reliable results, the sample size should be very high ! Therefore how much land you need to cultivate around 20,000 pea plants? Ask your grandfather!

B. Choice of the experimental material

First he decided to work on plants? Do you know why? Plants attain reproductive age very soon compared to many animals, the number of plantlets from a plant will be very high, Many plants

can be grown together and above all plants reproduce through seeds which we will get from flowers!. Now the question is which plant? Shall we take a mango tree? Here comes the advantage of critical observation. The feasibility. It is Pea plants, because very easy to cultivate, flowers are big (think why?), number of plantlets produced from one flower will be more and above all its generation time is less. But the most important thing? What is the aim of the experiment? To study variation in plants? So we need to study variation. Examples of variation? Flower color, seed color, seed size and shape, flower position etc. So the experimental plant should have easily recognizable variations. From the Mendel's observations it is the Pea.

C. Planning and execution of the experiment

Selection of variations: He selected the following characters: 1. Seed shape [Spherical Vs Wrinkled]; 2. Seed color [Yellow Vs Green], 3. Flower color [Purple Vs white], 4. Pod shape [inflated vs constricted], 5. pod color [green pod vs yellow], 6. Flower position [Axial vs Terminal] and 7. Plant size [Tall Vs Dwarf] (**The first logic of Mendel**: Easy recognizable separate characters for observation -that had well-defined, contrasting alternative traits)

Why he selected seven characters? How did he do? He selected seven variations simultaneously in each experiment or separate experiments for separate variations? What you feel? He conducted different experiments for different characters first of all to see that whether he is getting the same kind of results in each case, secondly it is easy to follow the inheritance of one character at a time rather all together. That was the **second logic** of Mendel.

This third logic of Mendel from the knowledge he may got from his practical knowledge on agriculture and theoretical knowledge he got from mathematics. Does Mendel randomly selected seeds? We should know which characters seeds are inherited? We are studying variation, should we select seeds which show consistent inheritance or variable inheritance? The choice should be of consistent inheritance. He knew that it is possible to raise plants with consistent inheritance by crossing sibling plants. That is Mendel used well defined seeds as the starting material in all his experiments involving thousands of plants. So if we want to test the fuel efficiency of 10 different bikes, we should use the same experimental conditions in all the cases against one variability. This is the spectacular logic he applied and the basis of Mendel's success.

The **fourth logic is experimental**. How to do controlled breeding? We want to cross a tall plant with a dwarf plant. So we need to ensure that only the pollen grain of the tall plant is falling on the stigma of the dwarf plant and vice versa. How to do this? Simply bag the flower bud, cut the stamens of the experimental flower, take the pollen grain from the other flower using a brush, "paint" on the stigma of the initial flower, bag it again to avoid unwanted entry of other pollen grain. (See the animation). Get the seeds from the flower and observe for the variation intended to study!

Here is the **fifth logic**. He just crossed only once. All other experiments involved self pollination. ~~he~~e, he allowed mixing of two different characters only once. Why not twice or thrice? As simple as this, first let us start from simple things! So do the cross only once, avoid complexity and evaluate the results.

Now comes the **sixth and final logic**. Looking behind a character how many generations we should follow after crossing? Again comes the simplicity and reproducibility. Just followed the first generation after crossing.

So in short he started with the same experimental plan is as follows:

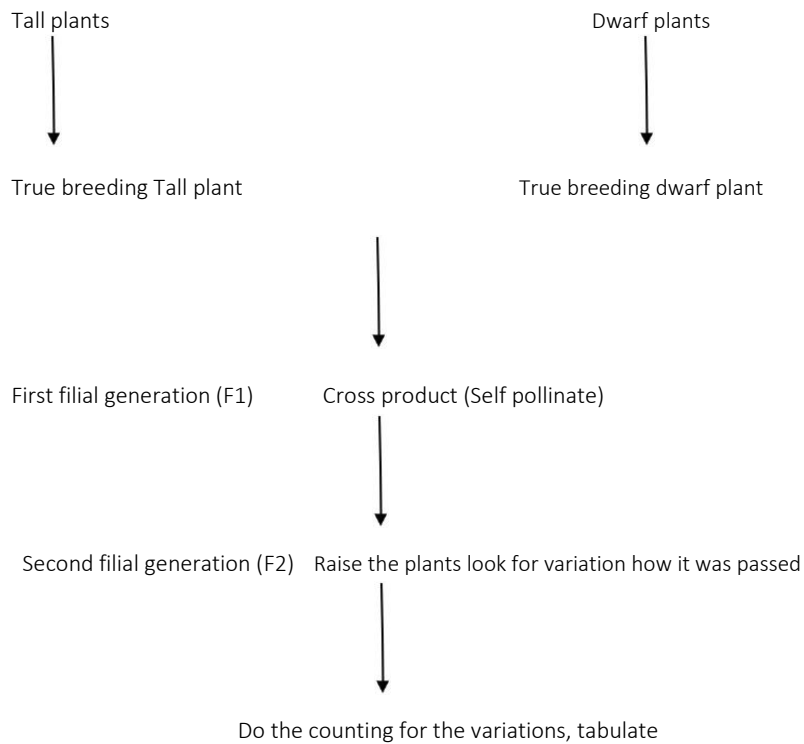


Figure 1

The seven variations observed in Pea plant that Mendel followed: Are they easily recognizable?

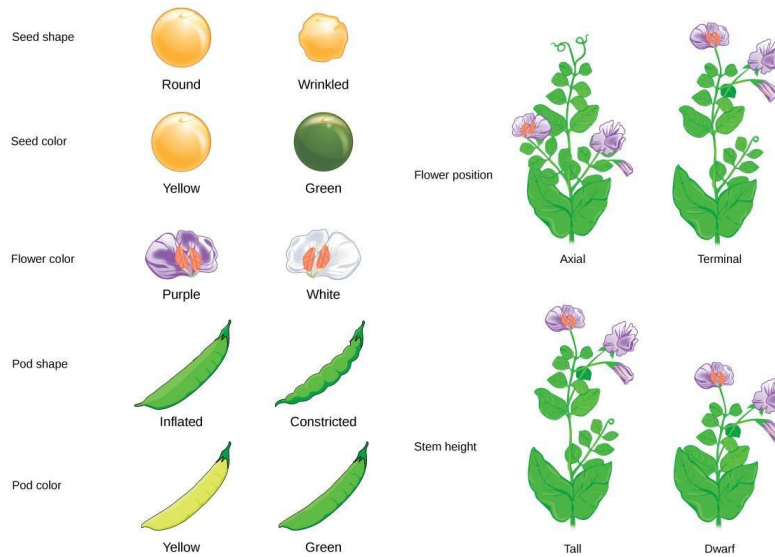


Figure 2 Flower structure of Pea: Observe the reproductive parts of the flower.

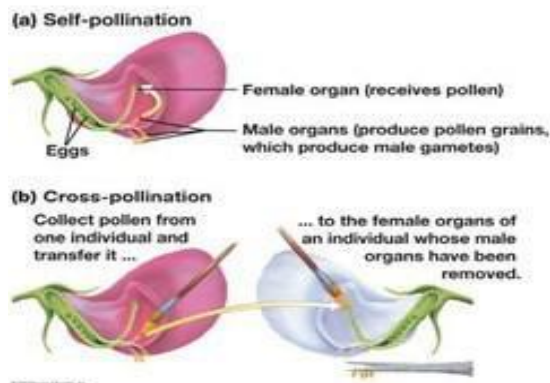


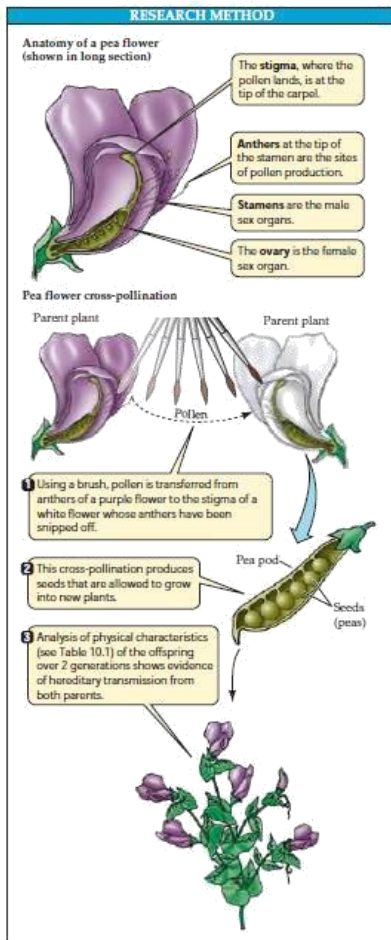
Photo of Mendel's ~~Experimental Choice~~ Model i.e. Green Peas:



Mendel's controlled experiments and elucidation of the results

THE RESEATRCH METHOD OF MENDEL

All of us now that pollen grains of one flower falls on the stigma of another flower or same flower. From the stigma the male nucleus of the pollen grain reaches the ovary and fertilizes an egg to produce a zygote. In plants fertilized zygote produces the seed and the ovary produces the fruit. The problem faced by Mendel is controlled pollination. Suppose he want to cross a tall plant with a dwarf plant. In this case the pollen grains of dwarf plant only should fall on the stigma of the dwarf plant or vice versa. How to achieve this? (Solve this problem with the help of the following figure and the animation provided)
























10.1 A Controlled Cross between Two Plants Plants were widely used in early genetic studies because it is easy to control which individuals mate with which. Mendel used the garden pea (*Pisum sativum*) in many of his experiments.

EXPERIMENTS RESULTS OF MENDEL

We have seen what Mendel's logics were and how he executed the experiments. After his experiments the results were carefully tabulated by him. Analyze the following table showing the tabulation of Mendel

10.1 Mendel's Results from Monohybrid Crosses

PARENTAL GENERATION PHENOTYPES		F ₁ Phenotypes	F ₂ GENERATION PHENOTYPES			
DOMINANT	RECESSIVE		DOMINANT	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

What you find here?

- (A) Only one character appeared in the F₁ generation
- (B) Both characters were appeared in the F₂ generation, but not in equal percentage
- (C) A character which disappeared in the F₁, reappeared in F₂
- (D) The results are consistent in all the seven characters
- (E) There is no blending of characters

The analysis of the results clearly indicate the following

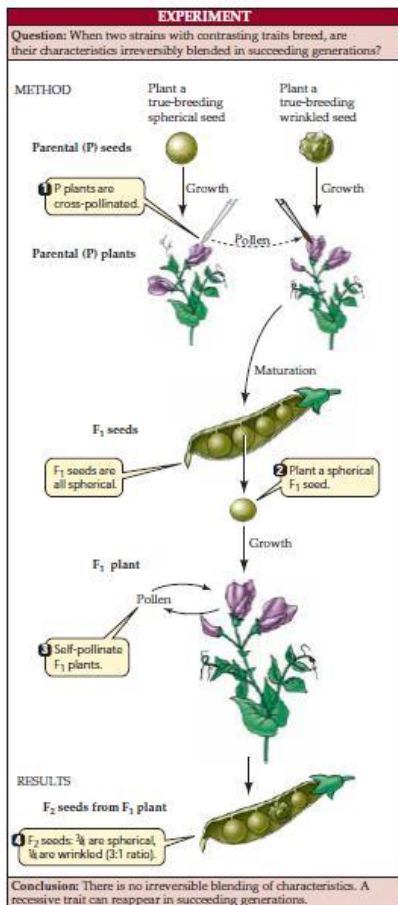
The character which appeared in the F₁ is having the higher percentage in F₂ i.e. almost three times to that of the disappeared character in F₁. What you will conclude from these results? The character which is appeared in the F₁ is also **dominating** in F₂ also. This is the **Dominant character**. The other or alternative character which is disappearing in the F₁ generation is also less in number compared to dominant character. This is **Recessive** character. (Law of Dominance).

Many of us, including Mendel, expected a blended phenotype in F₁. But it didn't happen. Why? Can't the characters blend? The experimental results in all the seven characters studied by Mendel didn't showed any blending (i.e. when we cross a Tall and Dwarf plant we can expect a plant intermediate between tall and dwarf; but it didn't ~~happend~~happen). This further means that some "units" which functions as discrete particles are responsible for characters. Since the character which disappeared in the F₁ reappeared in F₂ it is logic to conclude that these units occur as pairs. It means that two discrete units are responsible for the "Tall" character. Otherwise any given individual can be homozygous (Dominant/Dominant or Recessive/Recessive) or heterozygous (Dominant/Recessive). We can express the alternate forms of characters (dominant or recessive) as alleles. Also we know at present that character means a gene. So alleles are alternate forms (in fact variables) of a gene. They can be

represented as any letter forms (similar to polynomials). For example a tall plant can have TT or Tt. A dwarf plant can be represented as tt. This binomial expression is the genotype. In conclusion the “units” (or factors) are discrete, they never blend and are responsible for passing character from one generation to next ie inheritance.

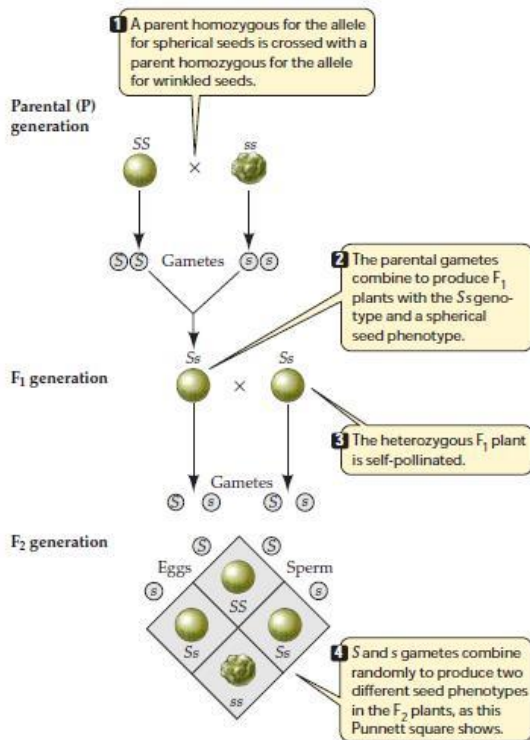
Why two units of inheritance? Not three or four? We have father and mother. i.e we have characters both from our father and mother. A character is represented by two units. One unit inherited from father and one from mother. So the gamete contains one unit. This is the core idea of Mendel’s inheritance. At present the “units” proposed by Mendel is known as gene.

The following figures illustrates a typical Mendelian cross.



10.3 Mendel's Experiment 1 The pattern Mendel observed in the F₂ generation— $\frac{3}{4}$ of the seeds wrinkled, $\frac{1}{4}$ spherical—was the same no matter which strain contributed the pollen in the parental generation.

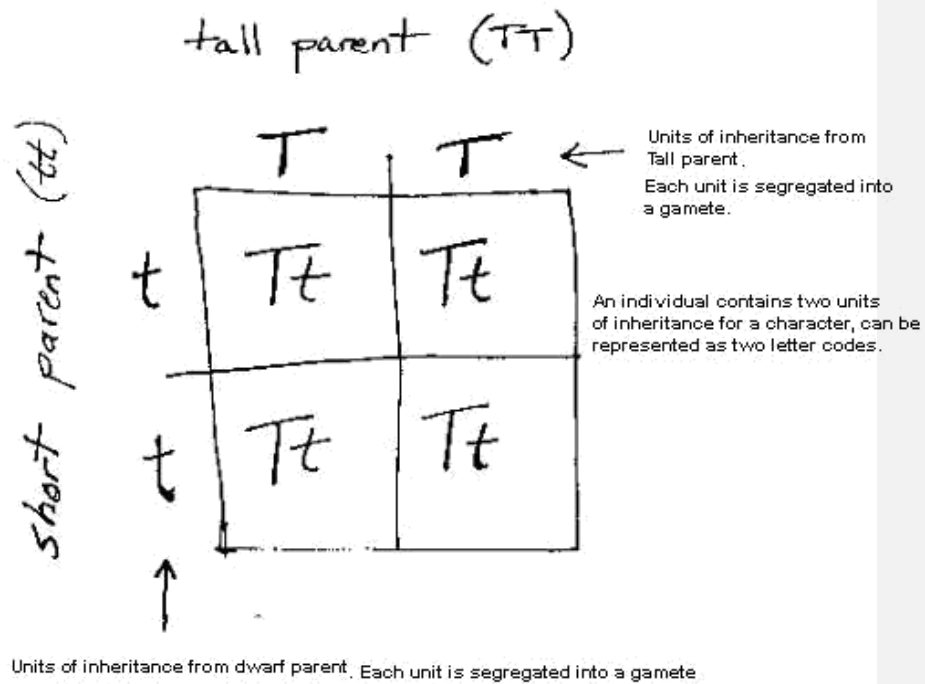
Now study the following cross which can be represented in the form of a Punnett square (A simple grid representing all possible gametes and combinations. Given the credit to Reginald Crundall Punnett, a British geneticist)



10.4 Mendel's Explanation of Experiment 1 Mendel concluded that inheritance depends on factors from each parent, and that these factors are discrete units that do not blend in the offspring.

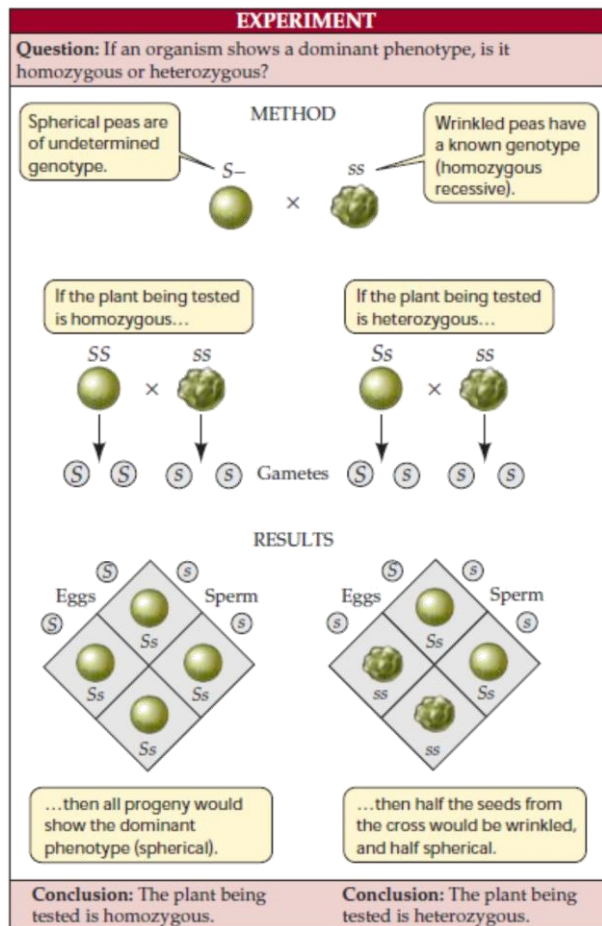
In conclusion the units of inheritance are never blended, but segregated independently during reproduction (Law of segregation). This is the second core idea of Mendel's theory.


Another illustration is here for you regarding the cross between a Tall plant and a Dwarf plant. This illustration represents Mendel's core ideas.



Verification of Mendel's hypothesis:

He did verification experiments for his hypothesis, as illustrated below.



 **10.6 Homozygous or Heterozygous?** An individual with a dominant phenotype may be homozygous or heterozygous. Its genotype can be determined by crossing it with a homozygous recessive plant and observing the phenotypes of the progeny produced. This procedure is known as a test cross.

Similarly the following figure illustrates a dihybrid test cross

Mendel has modelled one experiment, he executed and finally he tested it. This type of cross is known as a test cross (Crossing the unknown genotype with the recessive parent). It can predict the genotype to be tested based on the phenotypic ratio of the cross output.

Probability laws govern Mendelian inheritance: The Study of dihybrid cross

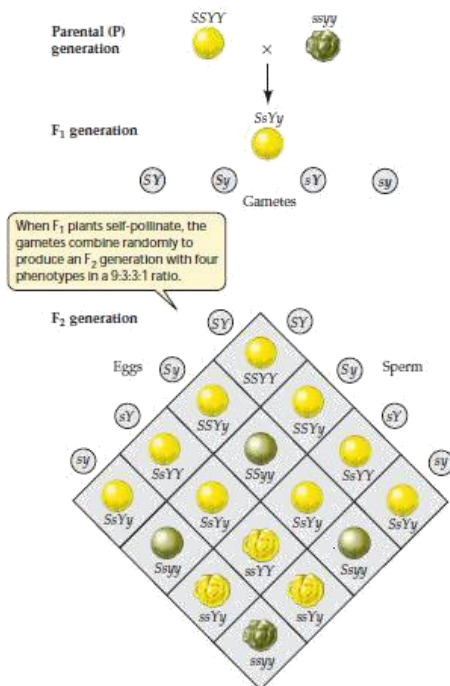
Initially Mendel did all his experiments by analyzing only one character at a time – monohybrid cross. Based on his results, he has tested his hypothesis of dominance and segregation. After this he wanted to study the inheritance of two characters at a time – the dihybrid cross

The experiment is planned in such a way to analyze the following:

Whether the alleles maintain the association they had in the parental generation: For this he crossed pure breeding spherical seed and yellow seed color pea plant with a wrinkled seed green seed color pea plant. If the alleles maintain the association, he is expecting only the parental types in the F₂ generations (Why not F₁ generation?)

If the alleles maintain the association, the F₁ gametes will be SY and sy. As a result the probability of Spherical and yellow seed peas: wrinkled and green seed peas will be 3:1 (i.e. only two phenotypes). If they segregate independently he was expecting four different phenotypes.

The experiment and the results are illustrated as below



10.7 Independent Assortment The 16 possible combinations of gametes in this dihybrid cross result in 9 different genotypes. Because S and Y are dominant over s and y , respectively, the 9 genotypes result in 4 phenotypes in a ratio of 9:3:3:1. These results show that the two genes segregate independently.

He didn't get a 3:1 ratio in F₂ generation. New types were obtained in F₂. It means that the alleles didn't maintain the same association as seen in the parental types, rather they assorted independently (Law of Independent Assortment).

Mendel and his Mathematics predictions.

You have a 1 rupee coin and 5 rupee coin. probability of getting a tail in both cases? (independent?)

You are going to toss it together. What is the (Are the two events linked to each other or independent?)

Probability of getting a 1 rupee tail = $\frac{1}{2}$. Probability of getting a 5 rupee tail = $\frac{1}{2}$

Hence the probability of getting both tail = $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ ie i.e. 25%.

In a homozygote (SS), the probability of producing a S gamete is 1

In a heterozygote (Ss), the probability of producing a S gamete is $\frac{1}{2}$ and s gamete is also $\frac{1}{2}$

Now consider the F2 generation. The probable gametes here are S and s.

Hence the probability of getting SS is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ ie i.e. 25% are homozygous dominant

The probability of getting ss is $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ ie i.e. 15% are homozygous recessive

Adding probabilities: What is the probability of getting Ss and sS?

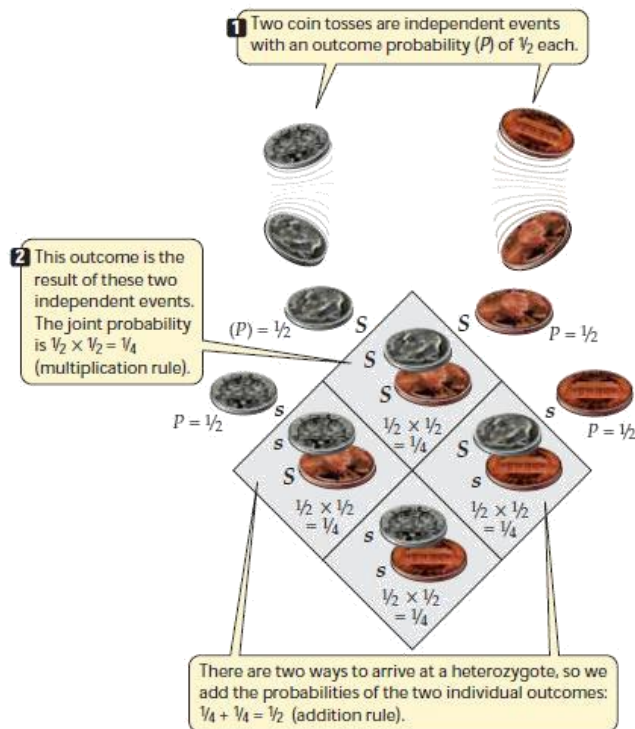
Probability of Ss (S from sperm and s from egg) = $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$

Probability of sS (s from sperm and S from egg) = $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$

Both Ss and sS are heterozygotes and will have the same phenotype. Hence added probability is $\frac{1}{4} + \frac{1}{4} = \frac{1}{2}$ ie 50% will be heterozygotes.

Now can we calculate the probabilities in dihybrid cross?

In F2 generation, the probabilities are illustrated below



10.9 Using Probability Calculations in Genetics The probability of any given combination of alleles from a sperm and an egg appearing in the offspring of a cross can be obtained by multiplying the probabilities of each event. Since a heterozygote can be formed in two ways, these two probabilities are added together.

Now what is the probability of getting an SS homozygote? i.e. $\frac{1}{4}$

The probability of getting heterozygote (i.e. Ss or sS) is $\frac{1}{4} + \frac{1}{4} = \frac{1}{2}$

The added probability (i.e. spherical seed) = $\frac{3}{4}$

Now calculate the probability of yellow seed using the above reasoning? It will be $\frac{3}{4}$

Hence what is the added probability of getting a spherical seed and yellow seed = $\frac{3}{4} \times \frac{3}{4} = \frac{9}{16}$

(Since both events are independent i.e. independent assortment)

What will be the probability of getting ~~aaa~~ yellow and wrinkled seed?

Probability of yellow seed = $\frac{3}{4}$

Probability of wrinkled seed = $\frac{1}{4}$

Hence the added probability = $\frac{3}{4} \times \frac{1}{4} = \frac{3}{16}$

Using the same logic it is easy to calculate the probability of wrinkled yellow seed is $\frac{3}{16}$ and wrinkled green seed is $\frac{1}{16}$.

Mendel did all these statistical problems. Because of his mathematical knowledge, he could easily predict, the ratio obtained in F₂ generation of monohybrid and dihybrid crosses are simply a statistical event and the factors are independent of each other.

You should understand both; i.e. doing a genetic problem by using probability and by using a Punnett square.

One of Mendel's major contributions to the science of genetics was his use of the rules of statistics and probability to analyze his masses of data from hundreds of crosses producing thousands of plants. His mathematical analyses led to clear patterns in the data, and then to his hypotheses. Ever since Mendel, geneticists have used simple mathematics in the same ways that Mendel did.

It is also possible to test the dihybrid genotypes as illustrated below. Similar to monohybrid test cross, it is possible to predict the genotype of a phenotype by crossing with a true recessive parent. The prediction is based on the characteristic phenotypic ratio we will get in this cross.

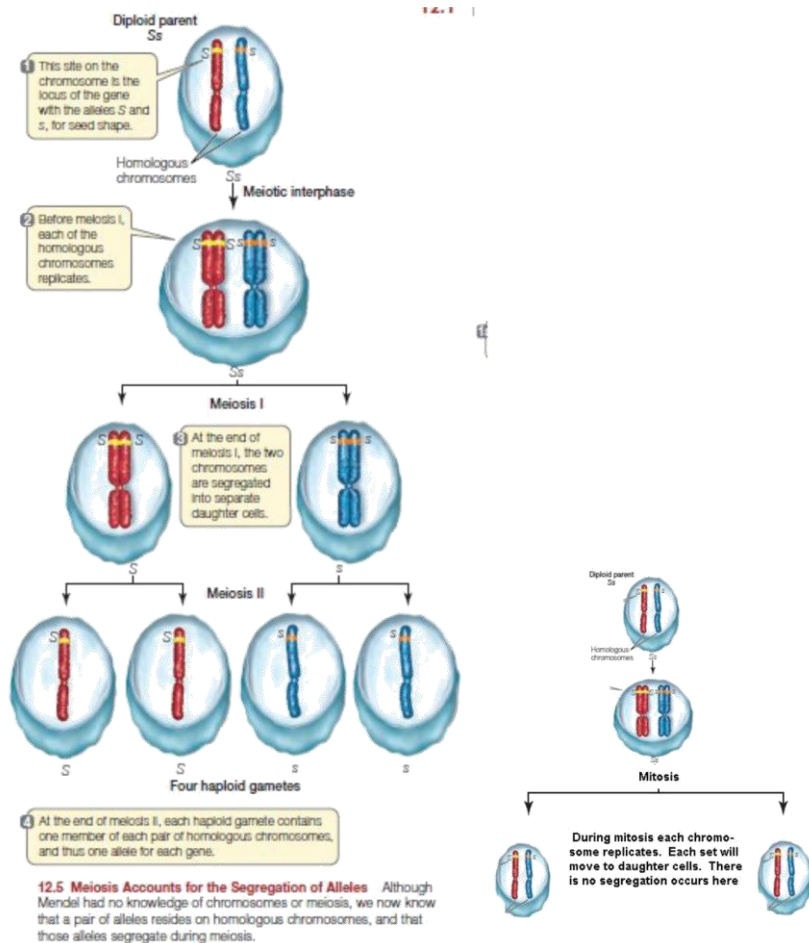
Test cross Genotypes	[Double heterozygous F ₁ Dihybrid] <i>Yellow Round</i> YyRr				[Double homozygous recessive parent] <i>Green Wrinkled</i> yyrr			
	↓ ↓ ↓ ↓				↓			
Types of Gametes	YR	Yr	yR	yr	yr			
Test cross Progeny	YR	Yr	yR	yr				
	YyRr	Yyrr	yyRr	yyrr				
	<i>Yellow</i>	<i>Yellow</i>	<i>Green</i>	<i>Green</i>				
	<i>Round</i>	<i>Wrinkled</i>	<i>Round</i>	<i>Wrinkled</i>				
	1	1	1	1				
	25%	25%	25%	25%				

Mendel experiments: Did he predicted chromosomes?

(Meiosis accounts for segregation)

The segregation of Mendelian factors is because of meiosis

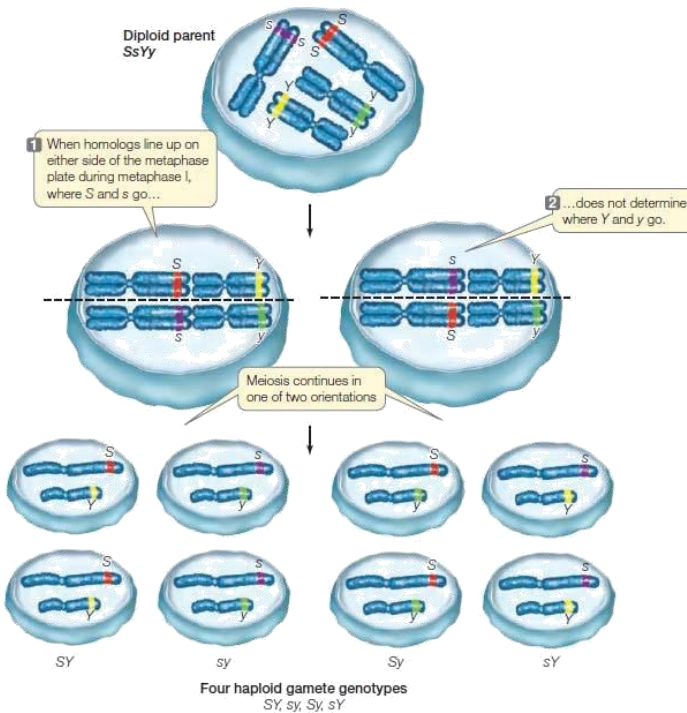
Mendel proposed mechanisms of heredity. Mendel had no knowledge of chromosomes or meiosis. But he speculated that cells contained some type of **factor** that carried traits from one generation to the next. The scientific importance of Mendel's work remained unrecognized for several years. Most probably Mendel believed that for each character there is a factor. Currently we know that this factor is a gene (or an allele) that is located on a chromosome. They show characteristic segregation and independent assortment are due to meiosis (Illustrated below).



So if we have a cell with a genotype Ss it should produce two types gametes, i.e. one type with S and the another type with s. Mendel said the two alleles will segregate. Our current knowledge is that meiosis accounts for segregation. This is exactly matches what Mendel speculated from his results. He also proved that each factor segregates when traits passed from one generation to another generation.

What ~~happens~~happens in mitosis? A cell with genotype Ss just produces two daughter cells with the same genotype as illustrated above and there is no segregation.

Now you will see how alleles assort independently during meiosis when we consider a dihybrid cross



Mendelian principles doesn't applies to all cases of inheritance.

Whether all the inheritance follows Mendelian pattern? Human have several traits like hair pattern, skin color, tasting ability, shape of ear and so on. How many chromosomes we have? 23 pairs. So if Mendel's rules we apply, we should have only 23 chromosomes. Hence it becomes clear that a chromosome can contain more than one factor. Now we have to think that who is the luckiest Man? It is Mendel. He selected seven characters. Each character was regulated by a gene and they were located in seven different chromosomes. Now we know that Pea plant has seven chromosomes. Suppose if the traits selected by Mendel resides on the same chromosome, he will not get a 9:3:3:1 ratio as expected.

In short if we get a ratio of 9:3:3:1, we can assume that the genes we selected are located on different chromosome. If we are not getting this ratio, then genes ~~may~~ be on same chromosome. The genes on the same chromosome means that they are linked.

The seven chromosomes of *Pisum sativum*. Luckily the seven genes for the selected traits by Mendel was located on seven different chromosomes leading to his success in ~~modeling~~ modelling his hypothesis and successful testing. If it was not, Mendel might have failed in his efforts. Hence many people believes that Mendel was the luckiest person.

Morgan and his *Drosophila*: Mendel's hypothesis is rejected in Morgan's experiments

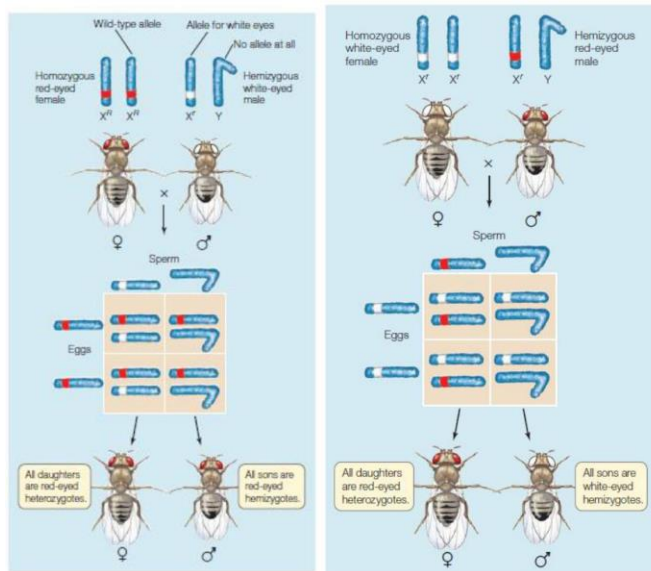
[Chromosomal theory, Connecting Mendel to Morgan, Linkage and crossing over]

It was Thomas Hunt Morgan and his students of Columbia University who did pioneering works to explain many of this from the beginning of 1909. He selected *Drosophila melanogaster*, the fruit fly as the experimental organism. He selected fruit fly because of its small size, easy to grow and breed and its short generation time.

His preliminary experiment is illustrated below:

We know that in mammals and many other animals sex is determined by chromosomes ~~i.e.~~ XX will be female and XY will be male. Now let us think that if the genes for the trait is present in the X chromosome or Y chromosome will it appear on both sexes? Can we determine which chromosome (X or Y) carries a specific gene for a specific trait?

Many experiments were conducted by Morgan to answer these questions.



In the above cross, the normal allele is red, the recessive allele is white. Red is dominant over white. Whenever the white male is crossed with a true breeding **red male-female** the result is both ~~son~~ male and female flies are red eyed. Whenever a red male is crossed with a true breeding white female all son-male offspring's are white eyed and daughters-female flies are red eyed.

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So the gene for the trait eye color in *Drosophila* resides on X linked chromosome. This inheritance is X linked recessive. We can show X linked inheritance in the pedigree chart illustrated below.

Did you note three things (1) ~~in~~ females, both X chromosomes should carry the recessive allele for the expression of white eye color. Hence this is ~~an~~ X linked recessive trait. (2) Males have only one X chromosome. Hence the trait will express even if the X chromosome contains the recessive allele (3) X linked recessive traits are more frequently occurs in males compared to females. The reason is that males have only one X chromosome. Hence the recessive allele will express.

Linked genes notes should appear first. The figure shown above is related to Morgan's experiment related to X-Linked inheritance. Which is repeated again in page no 28. Kindly note and do the modifications.

Notes related to Morgan's Linkage experiment was missing.

12.18 Some Alleles Do Not Assort Independently

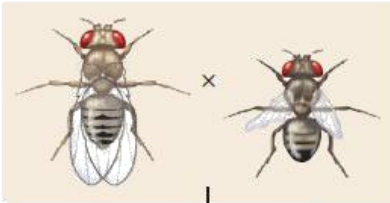
Morgan's studies showed that the genes for body color and wing size in *Drosophila* are linked, so that their alleles do not assort independently.

HYPOTHESIS Alleles for different characteristics always assort independently.

METHOD

Parent (P)

BbVgvg
Wild type
(gray body,
normal
wings)
♀



bbvgvg
(black body,
vestigial
wings)
♂

RESULTS

F₁

Genotypes

<i>BbVgvg</i> Wild type	<i>bbvgvg</i> Black vestigial	<i>Bbvgvg</i> Gray vestigial	<i>bbVgvg</i> Black normal
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Expected
phenotypes

575	575	575	575
-----	-----	-----	-----

Observed
phenotypes
(number of
individuals)

965	944	206	185
-----	-----	-----	-----

Parental
phenotypes

Recombinant
phenotypes

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

...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).

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

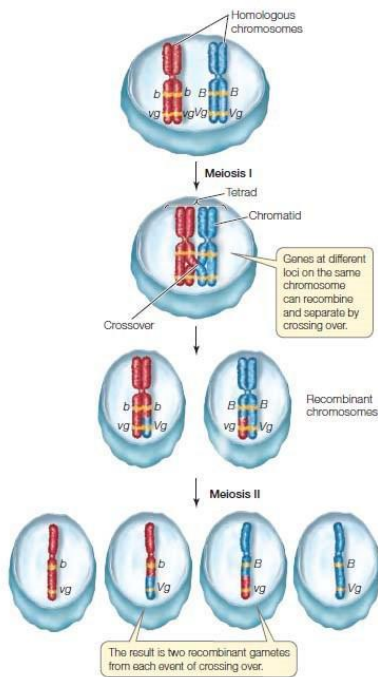
Non parental types or recombinant types = 391

Recombination frequency = (Recombinant types/Total individuals) X 100 = (391/2300) X 100 = 17%

(We can also write recombination frequency as 0.17 assuming that maximum recombination is 1)

Now we have to see why the new phenotypes (non-parental phenotypes) occurs?

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.



Towards the genetic map

The probability of recombination between two loci increases with distance. Morgan's found recombination frequencies of many genes through experiments and used this frequencies to construct a genetic map or mapping the genes. A genetic map tells the distance between two genes. The following illustration helps us to find how to do a genetic map. It is measured in terms of centi-morgan^s or cM.

- 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 $AABB \times aabb$, 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:

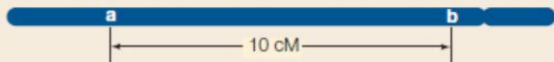
450 $AaBb$, 450 $aabb$, 50 $Aabb$, and 50 $aaBb$.
 (parental types) (recombinant types)

- 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

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 $AACC \times aacc$, obtain an F_1 generation, and test cross it, obtaining

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

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

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



4 How far apart are the *b* and *c* genes?

We make a cross $BBCC \times bbcc$, obtain an F_1 generation, and test cross it, obtaining

490 $BbCc$, 490 $bbcc$, 10 $Bbcc$, and 10 $bbCc$

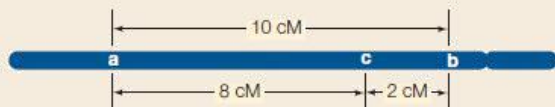
Recombinant frequency (*b* to *c*) = $(10 + 10)/1,000 = 0.02$

Map distance = $100 \times \text{recombinant frequency} = 100 \times 0.02 = 2 \text{ cM}$



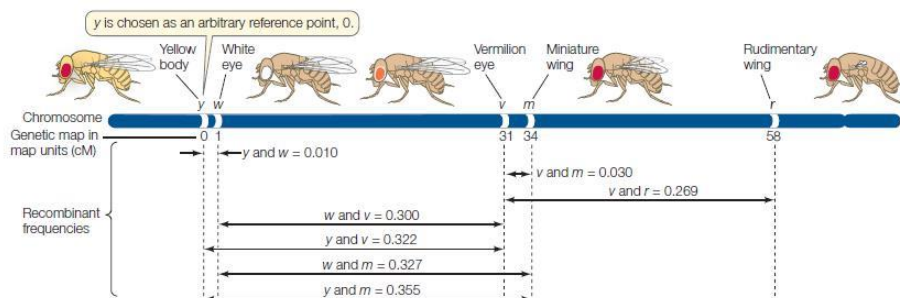
5 Which of the three genes is between the other two?

Because *a* and *b* are the farthest apart, *c* must be between them.



These numbers add up perfectly. In most real cases, they will not add up perfectly because of multiple crossovers.

The recombination frequencies can be used for making genetic maps. Morgan's group conducted several crosses in *Drosophila*. After finding out the frequency he was able to apply for construction of genetic map because the more the distance between two loci, the more will be the recombination. It means the distance between genes can be calculated based on this. The unit is cm (Centimorgan) or map units (1cM = 1 map unit). The following illustration shows an illustration of genetic mapping by Morgan.

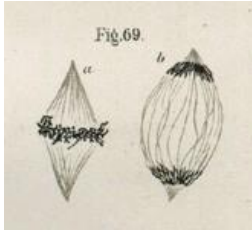


Walther Flemming, the grand father of mitosis discovered chromosomes. He says chromosomes splits longitudinally!

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Flemming was a German military physician. He found cells contains the colored genetic material, the chromosomes (Chrome = color; some = body). This is in fact the factor represented by Mendel. Even he discovered that chromosomes splits longitudinally during cell division (His illustration is given below). This is what happens during mitosis. We ~~we~~ know, in meiosis the longitudinal splitting happens after crossing over.



The chromosomal theory was not the work of a single scientist by Mendel or Morgan. Many people experimented over decades on it. Indeed, the first logic steps were initiated during 1860 by the mathematician Mendel and evolutionary biologist Charles Darwin. The probable mechanism of transmission from one generation to next was speculated by the discovery of chromosomes by Walther Flemming, a ~~german~~German biologist. Now to connect between chromosomes and heredity. This was done by Boveri, Sutton and Morgan during the dawn of 20th century. Thus the chromosomal theory came out which experimentally proved that chromosomes are responsible for transmission of trait from one generation to next.

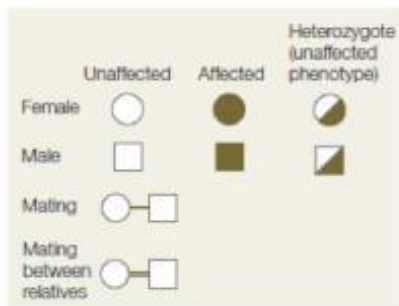
In fact Mendel was a Physicist (and philosopher), Darwin was a naturalist, Morgan was a zoologist. Above all Flemming was a military physician!!

INHERITANCE AND PEDIGREE

Life has evolved on earth gradually. Imagine we are producing and marketing a machine without any modifications over the years. What will happen? The machine is not satisfying the changing demands of the people over the time. It means nobody prefers to buy it and ultimately the product has to be withdrawn. Now comes the logic. Why life preferred two genders? Male and Female? It would have been simple for the life if only one gender is existing and all of them will reproduce. Isn't it? Do you agree?

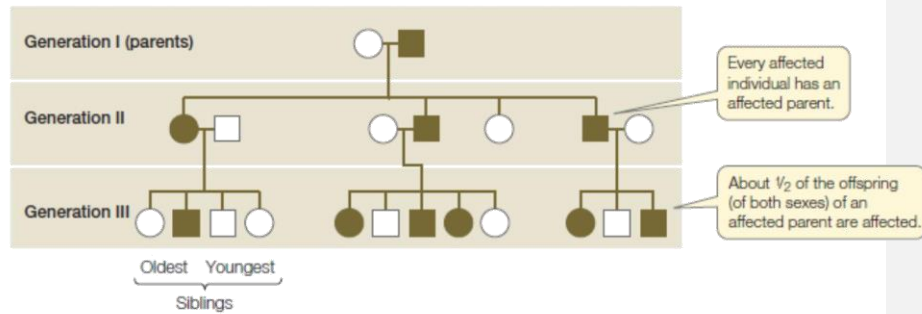
Most of the life forms have two different sex, what we say is a male and a female or a + strain and a – strain. The advantage lies on the recombination event. During sexual reproduction, the chromosome number of the gametes are reduced into half through meiosis. We have seen that meiosis accounts for segregation and assortment. We have also seen the non-parental genotypes appeared in the F₂ generation of dihybrid cross. It means sexual reproduction gives an opportunity for variation through meiotic recombination. Hence life systems are not static, they are dynamic. They are evolving. The most perfect life machine will always be preferred by the nature. Others will disappear, the survival of the fittest. It means the best character is inherited over the generations.

Now let us go through the different types of inheritance. Before discussing that we should see how to represent the inheritance in the form of a diagram. This is known as pedigree. The basic rules of pedigree chart is give below.

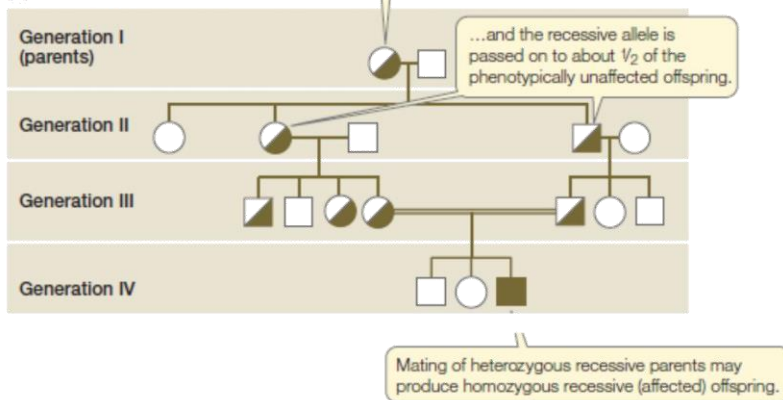



As we have seen with the Mendel and Morgan inheritance can be dominant or recessive


(A) Dominant inheritance



(B) Recessive inheritance

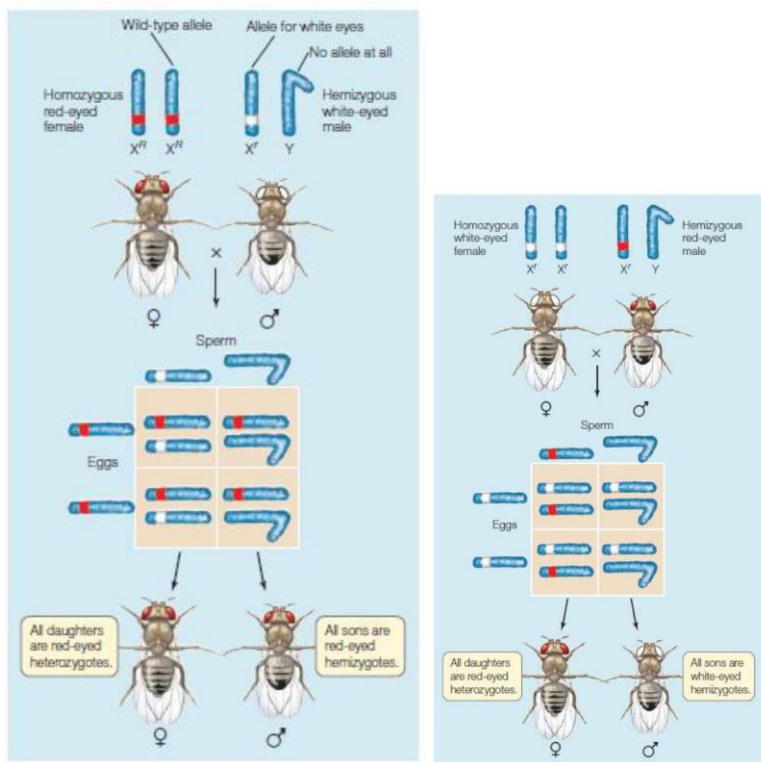


Now calculate the probability of the children getting affected in a cross . The male can be homozygous dominant or heterozygous. If he is homozygous dominant, the probability of an unaffected child is zero. This probability will be 50% if he is heterozygous in each child birth. Now see the

probability in the case of a recessive inheritance . The probability of getting the child having the recessive character is zero, because we know that the expression of a recessive character occurs only in case of homozygous condition of the alleles. But the unaffected child may be a carrier of the allele, even though he or she is not expressing the trait. The above types of inheritance illustrated here appears on both sexes. Hence the factor or the gene is resided on the autosomes. So an autosomal inheritance can be autosomal dominant or autosomal recessive.

We know that in mammals and many other animals sex is determined by chromosomes ie XX will be female and XY will be male. Now let us think that if the genes for the trait is present in the X chromosome or Y chromosome will it appear on both sexes? Can we determine which chromosome (X or Y) carries a specific gene for a specific trait?

Many experiments were conducted by Morgan to answer these questions.

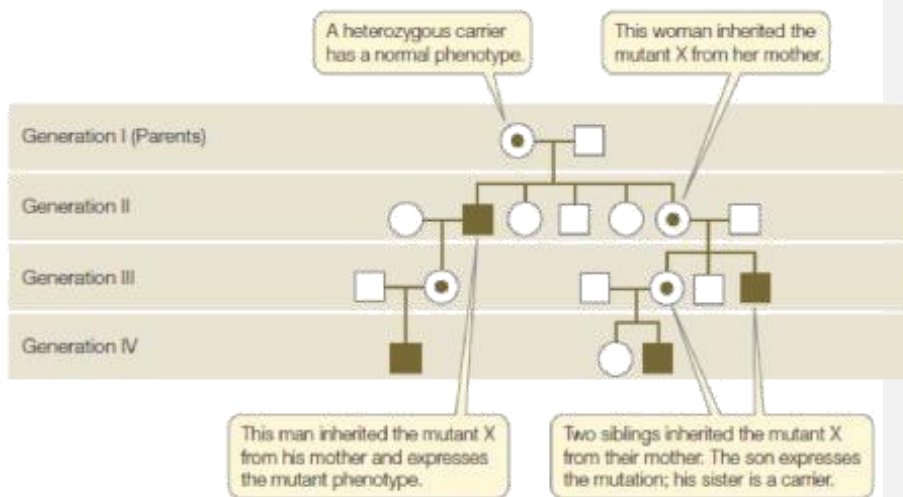


In the above cross, the normal allele is red, the recessive allele is white. Red is dominant over white. ~~When ever~~ Whenever the white male is crossed with a true breeding red ~~male female~~ the result is both son and daughters are red eyed. Whenever a red male is crossed with a true breeding white female all son are whited eyed and daughters are red eyed. So the gene for the trait eye color in *Drosophila* resides on X linked chromosome. This inheritance is X linked recessive. We can show X linked inheritance in the pedigree chart illustrated below.

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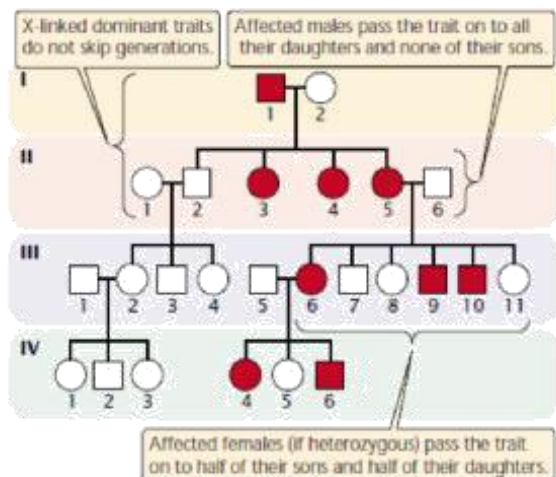
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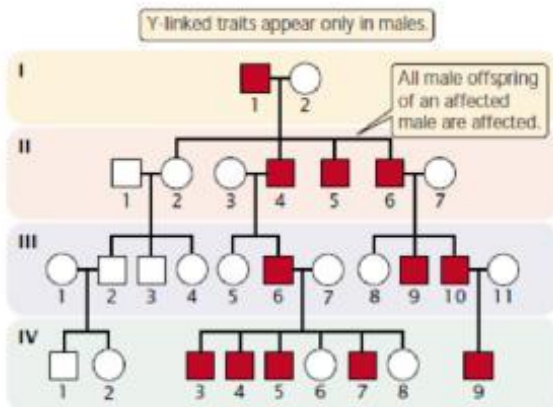
Did you noted three things (1) In females, both X chromosomes should carry the recessive allele for the expression of white eye color. Hence this is an X linked recessive trait. (2) Males have only one X chromosome. Hence the trait will express even if the X chromosome contains the recessive allele (3) X linked recessive traits are more frequently occurs in males compared to females. The reason is that males have only one X chromosome. Hence the recessive allele will express.

In some other cases, X linked characters may appear in dominant pattern also. Illustration is given below



6.9 X-linked dominant traits affect both males and females. An affected male must have an affected mother.

Some of the traits only appear in males because the gene for this trait are located on Y chromosome. They are passed by father to all of his sons, but not daughter. The illustration is given below



6.10 Y-linked traits appear only in males and are passed from a father to all his sons.

The following table explains the features of different patterns of inheritance

Table 6.1 Pedigree characteristics of autosomal recessive, autosomal dominant, X-linked recessive, X-linked dominant, and Y-linked traits		
Autosomal recessive trait <ol style="list-style-type: none"> 1. Appears in both sexes with equal frequency. 2. Trait tends to skip generations. 3. Affected offspring are usually born to unaffected parents. 4. When both parents are heterozygous, approximately 1/4 of the offspring will be affected. 5. Appears more frequently among the children of consanguine marriages. 	<ol style="list-style-type: none"> 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 dominant trait <ol style="list-style-type: none"> 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. 4. Affected mothers (if heterozygous) will pass the trait on to 1/2 of their sons and 1/2 of their daughters.
Autosomal dominant trait <ol style="list-style-type: none"> 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. 	X-linked recessive trait <ol style="list-style-type: none"> 1. More males than females are affected. 2. Affected sons are usually born to unaffected mothers; thus, the trait skips generations. 3. A carrier (heterozygous) mother produces approximately 1/2 affected sons. 4. Is never passed from father to son. 5. All daughters of affected fathers are carriers. 	Y-linked trait <ol style="list-style-type: none"> 1. Only males are affected. 2. Is passed from father to all sons. 3. Does not skip generations.

Diseases or traits can pass from one generation to another generation. Many of them follow typical Mendelian inheritance. The following table gives few examples

Commented [vP3]: Notes related to autosomal dominance autosomal recessive (pedigree) is missing.

Trait or disease	Type of inheritance
Color blindness	X linked recessive
Hemophilia	X linked recessive
Huntington disease	Autosomal dominant
Sickle cell anemia	Autosomal recessive
Tongue rolling	Autosomal dominant
Hand clasping	Clasp your hands together. Notice whether your left or your right thumb is on top. If the left thumb is on top you have the dominant trait (C), the right thumb is recessive
Alport syndrome	X linked dominant
ADP/ATP translocase	The gene responsible for this enzyme that moves ADP into and ATP out of mitochondria has been linked to Y chromosome