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موقع مذكرات جاهزة للطباعة

Biology

For 1st secondary



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Msc. Of Sciences

 **01013883112**

 **Dr Ahmed Mostafa biology**

Chromosomes and genetic information

⇒ In early 20th century:

- Scientists discovered that genetic information is carried on chromosomes, as they have the same number in the cells of the members of each type of living organisms.
- The genetic information causes the appearance of hereditary traits in living organisms.

⇒ Chromosomes:

- Exist inside the cell nucleus.
- They are found in the form of homologous pairs in somatic and gonads.
- Each chromosome consists of DNA and proteins.

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⇒ Number of chromosomes:

Living organisms have different numbers of chromosomes, while members of the same species have the same number of chromosomes.

⇒ **DNA:** Consists of units called nucleotides.

⇒ **Genes:** A sequence of nucleotides on DNA which represents a codon for building of a certain protein which is responsible for the appearance of a genetic trait.

Types of cells in the body:

	Somatic cells	Reproductive cells (gametes)
Produced by	Mitotic division (mitosis) of somatic cells.	Meiotic division (Meiosis) of the gonads (testis and ovary)
No. of chromosomes	Two pairs of homologous chromosomes. Diploid number (2n)	Half the number of chromosomes in the somatic cells of the organism. Haploid number (n)
Such as	Cells of the skin, muscles, blood And so on.	Sperms (human and animals male gametes) Pollens (plant male gametes) Ovum (female gametes of human, plants and animals)

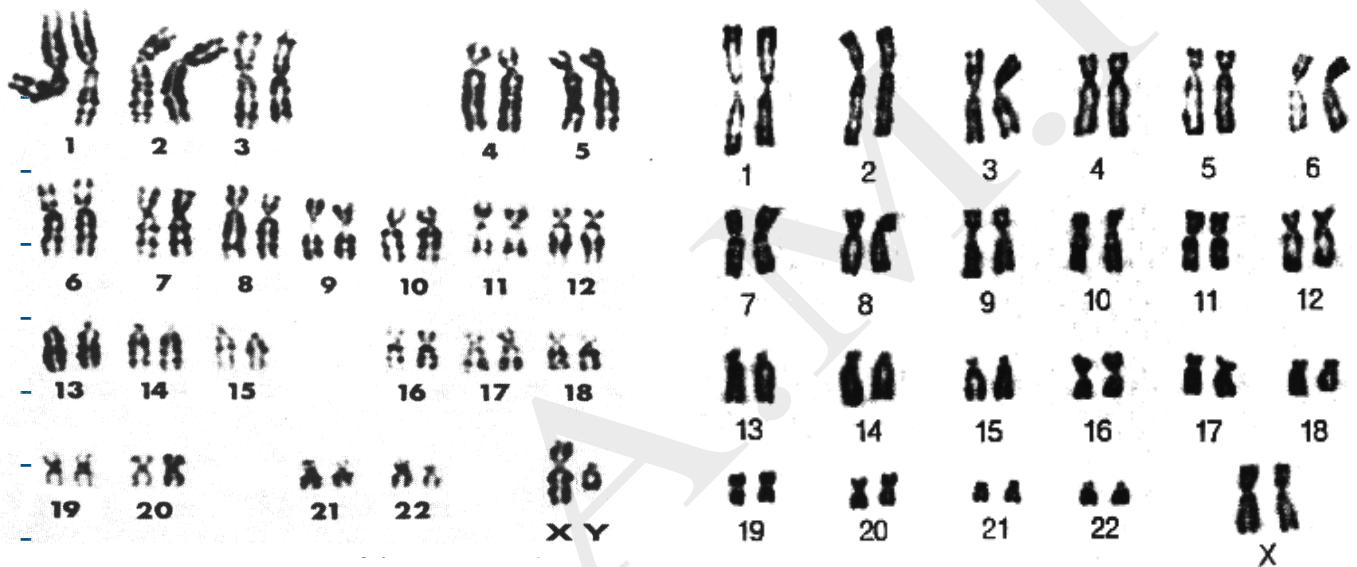
Karyotype

- It is the arrangement of chromosomes in descending order according to their size and numbering them.
- To facilitate the arranging and numbering of chromosomes, they can be coloured with different colours.



→ Human Karyotype:

- Nucleus of each somatic cell contains 46 chromosomes (23 pairs), while that of each gamete cell contains only 23 chromosomes.
- → Chromosomes pairs are arranged in descending order according to their sizes from number (1) to number (23).
- → Chromosomes pairs from (1) to (22) are known as somatic chromosomes
- → Chromosomes pair (23) is called sex chromosomes because this pair carries the genetic information responsible for the determination of sex (male or female).
- → Sex chromosomes pair follows pair (7) in size, but its order is (23)



Karyotype of human male

(44 + XY)

Karyotype of human female

(44+XX)

Karyotype in male:- Sex chromosomes pair in male is non homologous (XY)

Karyotype in female:- Sex chromosomes pair in female is homologous (XX)

Chromosome theory

- → **Scientists Boveri and Sutton put chromosome theory in 1902, which states that:-**
- 1- Chromosomes exist in somatic cells in the form of homologous pairs (2n)
- 2- Gametes contain half the no. of chromosomes in somatic cells as a result of meiotic cell division; where homologous pairs get separated from each other forming two identical groups
- 3- Each pair of chromosomes acts independently when being transferred to gametes.
- 4- After fertilization process, the normal number of chromosomes (2n) comes back
- 5- Each chromosome carries hundreds of genes.
- → A chromosome is made of DNA and protein, DNA carries genes which have the genetic information of living organisms

Somatic cells

- Produced by mitosis.
- They contain the complete number of chromosomes.
- They are called diploid number of chromosomes (2N), i.e they contain two sets of chromosomes.
- Exmples: cells of liver, skin, pancreas, etc.

Reproductive cells (gametes)

- Produced by meiosis of cells of gonads.
- They contain half the number of chromosomes.
- They are called haploid number of chromosomes (N), i.e they contain one set of chromosomes.
- They are two types:
 - Male gametes: sperms (in human and animals) or pollens (in plants)
 - Female gametes (Ova).

⇒ **Gonads:**

- They have diploid number of chromosomes (2n).
- Their cells divide **meiotically** to produce gametes (n).
- They are 2 types of gonads: Male gonads (testis and anthers) and female gonads (ovaries).

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The complete dominance (The Mendelian cases)

- It is a genetic case in which the characteristic is controlled by two genes; one of them is dominant while the other is recessive where the effect of the recessive gene is absent in the presence of the dominant gene.
- It is characterized by the ratio (3 dominant : 1 recessive) in the 2nd generation.

-The Mendelian case is characterized by:

two phenotypes (dominant and recessive) and three genotypes:

Dominant phenotype

Recessive phenotype

May be:

- Homozygous (pure dominant) e.g) AA.
- Heterozygous (hybrid dominant) e.g) Aa.

- Homozygous (Always pure recessive) e.g) aa.

Mendel's first law:

Law of Segregation of Factors:

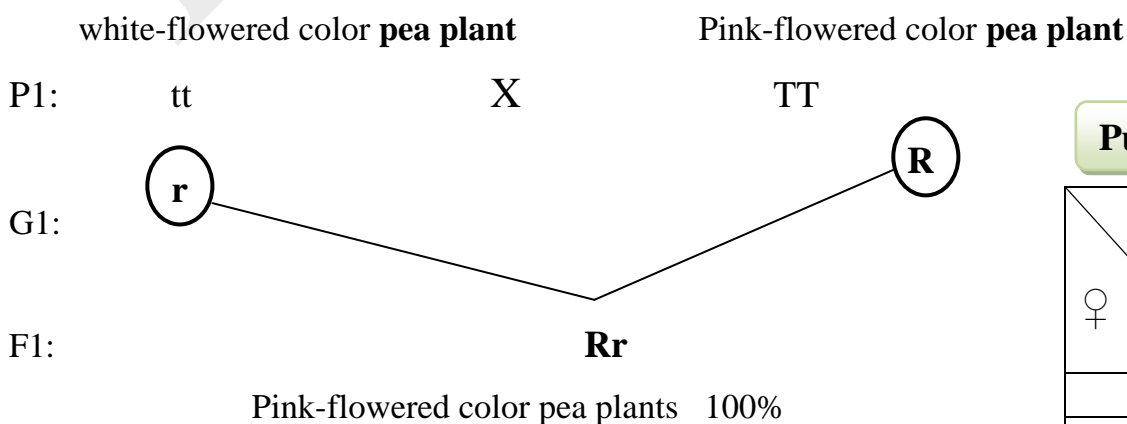
When two pure members that differ in any pair of allelomorphic characteristic are crossed, only the dominant characteristic appears in the F1 generation, while the two characteristics appear in the F2 generation in the ratio of **3 dominant : 1 recessive**.

What is meant by allelomorphic characters?

They are each pair of contrasting (alternating) characteristics (الصفات المتضادة) is called a, such as the seed colour which may be yellow or green, the flower colour which may be pink or white, and the height of the stem which is either tall or short,...etc.

Example

If crossing takes place between a white-flowered color pea plant and another Pink-flowered color pea plant: the **F1** plants of this cross will be all Pink-flowered color pea plants.

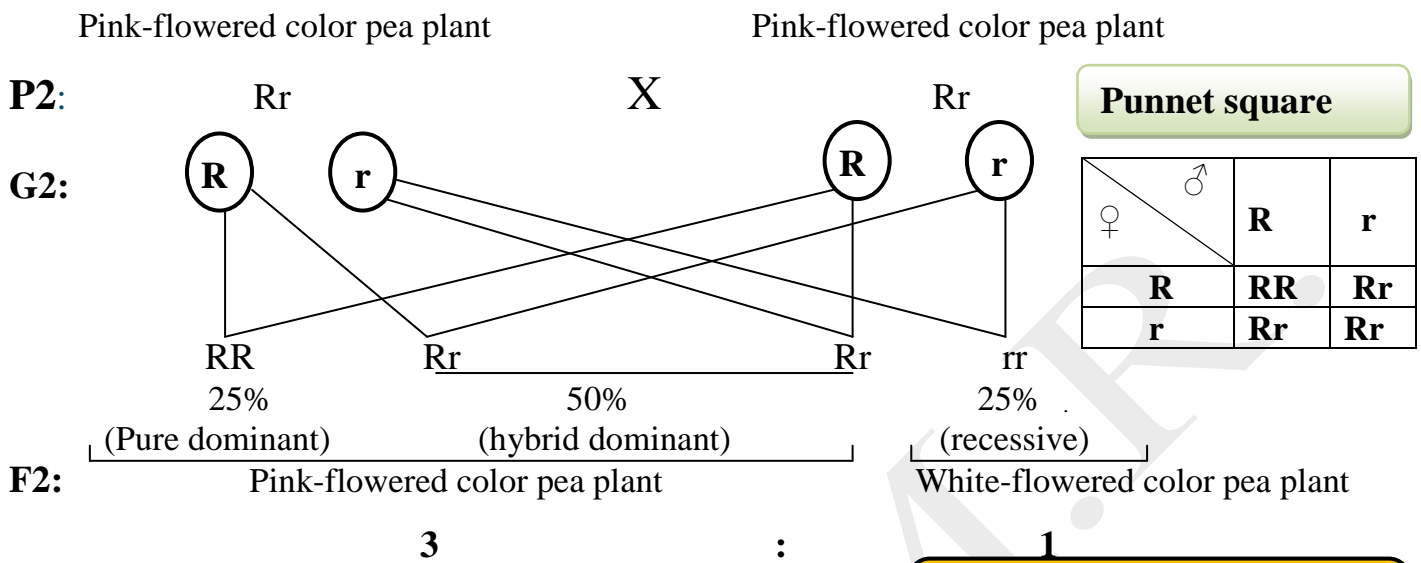


Punnet square

♀	♂	R	R
r		Rr	Rr
r		Rr	Rr



When the individuals of F1 are left for self-pollination, fertilization takes place, and F2 plants are obtained: they are **pink-flowered colour pea plants**, and **white-flowered color pea plants** in the ratio 3 pink-flowered : 1 white flowered.



Punnet square

♂	R	r
♀	R	r
R	RR	Rr
r	Rr	rr

Problems:

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1- What are the genotypes and phenotypes of the plants of first and second generations resulted from crossing of a pure tall-stemed pea plant and another short-stemed pea plant.

2- What are the genotypes and phenotypes of the individuals resulted from mating of a heterozygous black mouse with a pure brown mouse?

The inheritance of some Mendelian characteristics in Man:

Mendel's first law can be applied to some Human characteristics such as hair (dark, and light), (curly, and straight), (dense, and light), the eye colour (brown, and blue), (wide, and narrow), eye-lashes (long, and short), skin colour (normal, and albino), ear lobules (free, and fused), and nose (pointed tip, and flat tip), (curved and straight).

Some abnormalities, and diseases such as deafness (recessive), flat-foot (dominant), high blood pressure (dominant), protrusion of the lower jaw (dominant), short fingers (dominant), memory loss (recessive), and idiocy (recessive).

Mendel's second law (law of independent assortment):

Mendel's first law deals with the inheritance of one allelomorphous characteristic, while Mendel's second law studies the inheritance of two allelomorphous characteristics in the same time.

Mendel's second law: law of the independent assortment of the genetic factors

When two homozygous individuals differing in two (or more) pairs of alleles are crossed, each pair of characteristics is assorted at random, and is inherited independently of the other, and will appear in the F₂ generation in the ratio 3:1

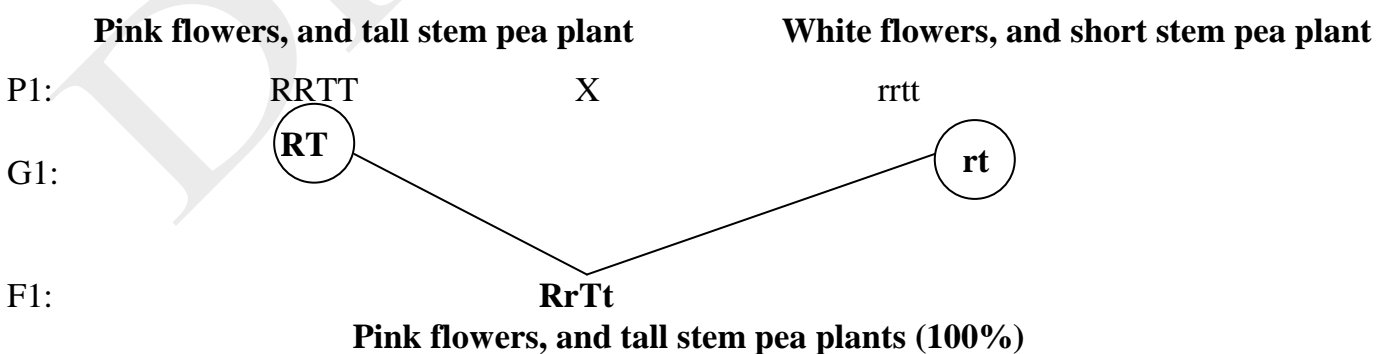
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Example

If crossing takes place between: **one** with pink flowers, and tall stems (two dominant characteristics), whereas **the other** with white flowers, and short stems (two recessive characteristics)

F₁ generation: All the produced pea plants were pink-flowered and tall-stemmed.



Punnet square

	♂	RT	RT
♀	rt	RrTt	RrTt
	rt	RrTt	RrTt



When he left the pea plants of the F1 generation to be self pollinated, The individuals of the F2 will be as follows:

P2: Pink flowers, and tall stem pea plant

Pink flowers, and tall stem pea plant



F2:

♂ \ ♀	RT	Rt	rT	rt
RT	$RRTT$ Pink, Tall	$RRTt$ Pink, Tall	$RrTT$ Pink, Tall	$RrTt$ Pink, Tall
Rt	$RRTt$ Pink, Tall	$RRtt$ Pink, short	$RrTt$ Pink, Tall	$Rrtt$ Pink, short
rT	$RrTT$ Pink, Tall	$RrTt$ Pink, Tall	$rrTT$ white, Tall	$rrTt$ white, Tall
rt	$RrTt$ Pink, Tall	$Rrtt$ Pink, short	$rrTt$ white, Tall	$rrtt$ white, Short

F2 generation :

- Pink-flowered, and tall-stemed plants. 9
- Pink-flowered, and short-stemed plants. 3
- White-flowered, and tall-stemed plants. 3
- White-flowered, and short-stemed plants. 1

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The ratio of the above mentioned individuals of F2 is : 9 : 3 : 3 : 1

N.B)

For the color character:

The number of **pink-flowered plants** was 12, whereas the number of white-flowered plants was 4, thus the ratio between them is 12:4 or (3:1)

For the height character:

The number of the **tall-stemed plants** was 12, whereas the number of short-stemed plants was 4, thus the ratio between them is 12:4 or (3:1)

The interaction of genes (The non- Mendelian cases)

⇒ In 1860, Scientist Gregary Mendel discovered that every hereditary trait is controlled by a pair of genes, which may be dominant or recessive.

⇒ The Mendelian case is called **complete dominance**.

Dominant trait: Trait which appears in all members of 1st generation

Recessive trait: Trait which disappears in all members of 1st generation and appears in 2nd generation at ratio 25%

⇒ After Mendel, Scientists discovered that there are many traits which are not inherited according to Mendel's laws, they are called **Non-Mendilian traits**.

⇒ There are some cases of non-Mendilian traits in which the appearance of hereditary traits is affected by the interaction of genes.

First

1.Lack of Dominance

- It is a genetic case in which the characteristic is controlled by two genes; none of them dominates over the other but the presence of them together leads to the appearance of an intermediate characteristic.

- It is characterized by the ratio (1:2:1) in the 2nd generation.

* Examples of the lack of dominance:

- 1- The inheritance of flower color of Antirrhinum plants.
- 2- The inheritance of blood groups in Man.

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2- The inheritance of flower color of Antirrhinum plants.

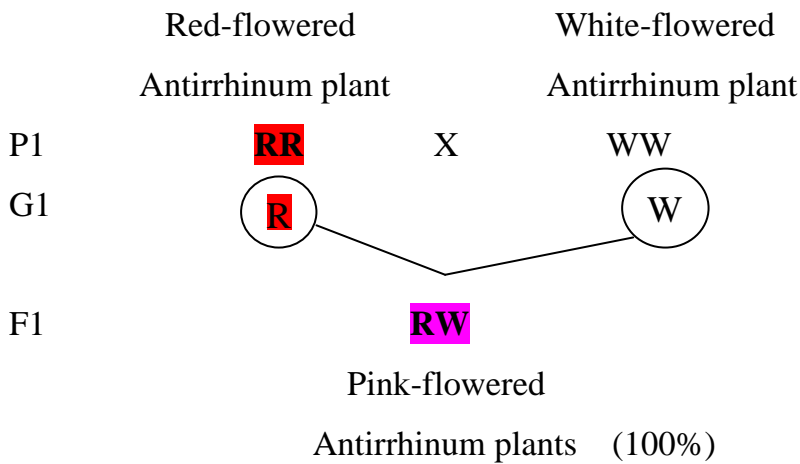
- There are plants of white-flowered color (WW) and others of red-flowers color (RR).
- When crossing is done between the two plants, the F1 individuals didn't have red or white flowers, they had pink flowers. This means that neither the red color of flowers dominates the white color of flowers, nor the reverse.
- When the F1 generation individuals were self-pollinated, the F2 plants were red-flowered, pink-flowered, and white-flowered in the ratio 1 : 2 : 1

This case can be represented by symbols considering:

R: The gene for red flowers and W: The gene for white flowers.



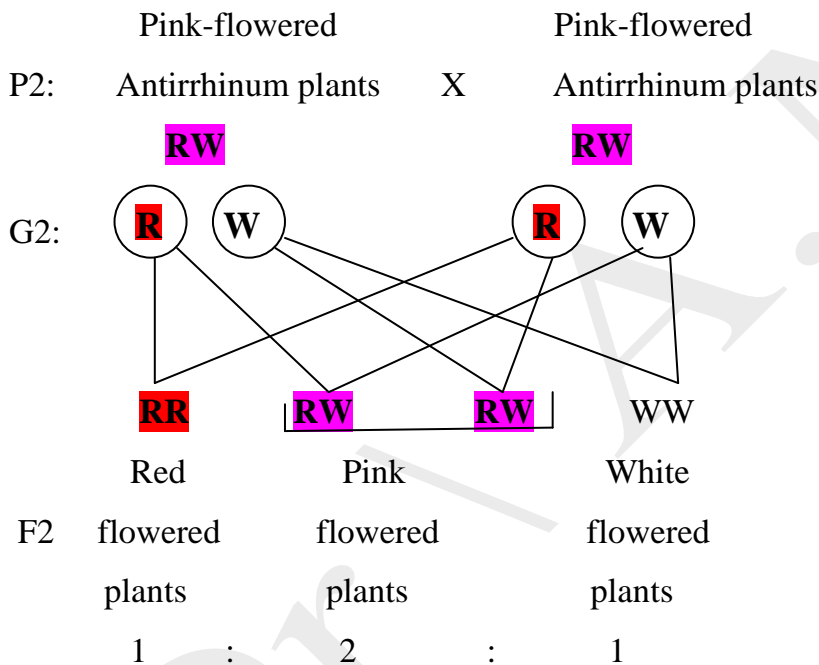
N.B.: Both the two genes are represented by **capital letters** because of the lack of dominance between the two genes.



	♂		
♀		R	R
	W	RW	RW
	W	RW	RW

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To obtain the 2 generation:



	♂		
♀		R	W
	R	RR	RW
	W	RW	WW

Solved problem

There are two types of Andalusian fowls (الدجاج الأندلسي), one of black-color and the other of white-color.

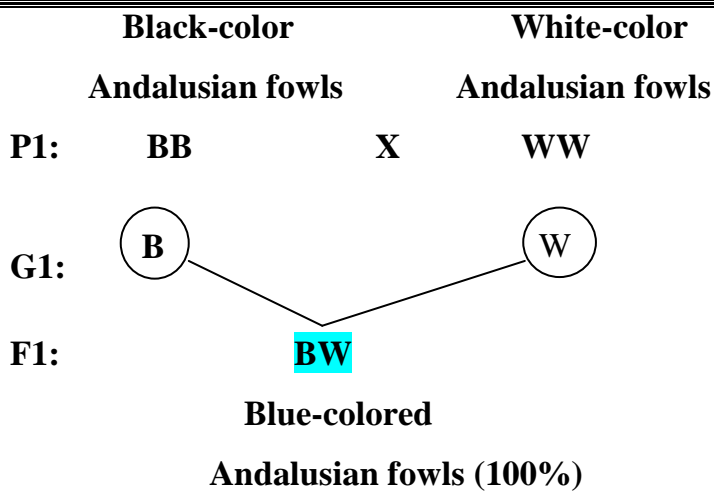
- When crossing takes place between black-colored and white-colored, F1 generation will have blue color.
- While F2 generation will have the three colors black, blue, and white in the ratio 1 : 2 : 1

Explain this case on genetic bases.

This is a case of lack of dominance.

(B) represents the gene for black color, while (W) represents the gene for white color.

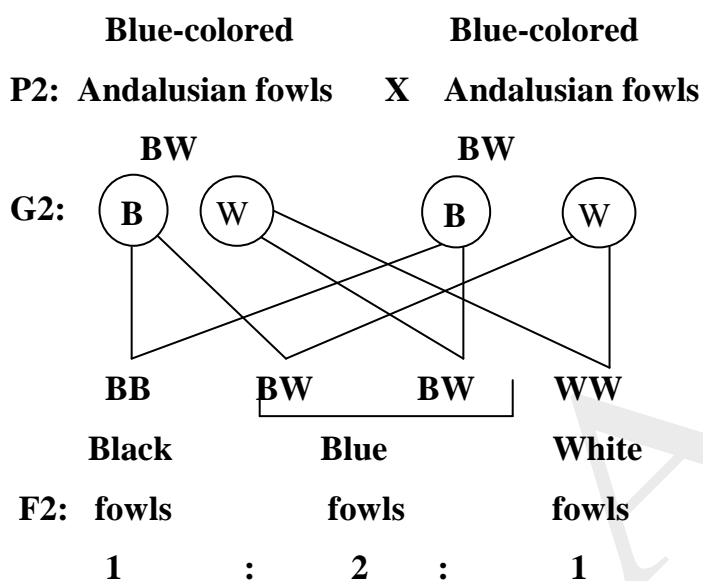




♂ \ ♀	B	B
W	BW	BW
W	BW	BW

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To obtain the 2 generation:



♂ \ ♀	B	W
B	BB	BW
W	BW	WW

Comparison between complete dominance and lack of dominance

	complete dominance	lack of dominance
Number of genes (alleles) control the trait	2 genes (one of them dominates over the other)	2 genes (neither one of gene dominates over the other)
Number of phenotypes	2	3
Number of genotypes	3	3
The phenotype of the heterozygous genotype	Dominant trait appears	Intermediate trait appears
In the 1st generation	The dominant trait appears (100%)	Intermediate trait appears (100%)
In the 2nd generation	3 : 1	1 : 2 : 1
Examples	The inheritance of flower colour and stem length in pea plant	The inheritance of flower color of Antirrhinum plants, and blood groups in Man.



1- The inheritance of blood groups in Man.

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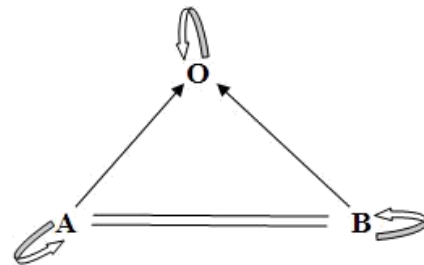
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- As a result of their studies, scientists classified the Human blood into four main groups: - (A), (B), (AB), and (O).
- This classification referred to a genetic classification and a chemical classification.

Genetic classification of blood groups:

- It was also discovered that these blood groups are inherited as multiple alleles (the genetic trait is controlled by more than one pair of allelomorphic genes), where:
 - There are three allelic genes (A), (B), and (O).
 - Each individual inherits only one pair of these alleles.
 - There are six genotypes (AA), (BB), (OO), (AB), (AO), (BO).
- **Relation between the allelic genes is:**
 - (O) is recessive to both (A), and (B) alleles.
 - A case of lack of dominance between (A), and (B) alleles.
- **Phenotypes and genotypes of blood groups:**
 - There are 6 genotypes which are represented by 4 phenotypes.
- **The following table represents the phenotypes and the genotypes of the blood groups:**

4 Phenotypes	6 Genotypes
Blood group A	(AA) & (AO)
Blood group B	(BB) & (BO)
Blood group AB	(AB)
Blood group O	(OO)



Relation between alleles

Notes:

1- The inheritance of blood groups in Human represents 3 genetic cases:

- Complete dominance:** as both the two alleles (A and B) dominates over the (O) allele.
- Lack of dominance:** as the two alleles (A and B) neither one of them dominates over the other and they produce an intermediate trait (AB)
- Multiple alleles,** as there are 3 alleles control the trait (A, B and O) while the individual inherits only two of them (one pair).

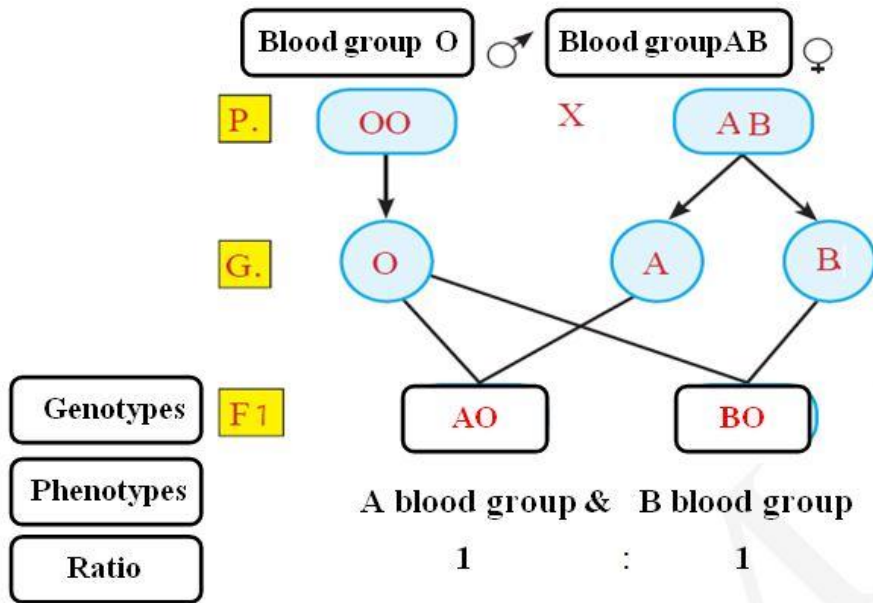
2- The genotypes can be determined by following up the phenotypes of the parents, and

their children, This helps in judging the paternity (الأبوة) of disputed children (الأطفال المتنازع عليهم) or referring mixed children to their real parents.

Example:

- A man of blood group (O) married a woman of blood group (AB), what are the blood groups of their children.

Solution:



Example 2:

To illustrate the case of two newly born children that were mixed up in the hospital.

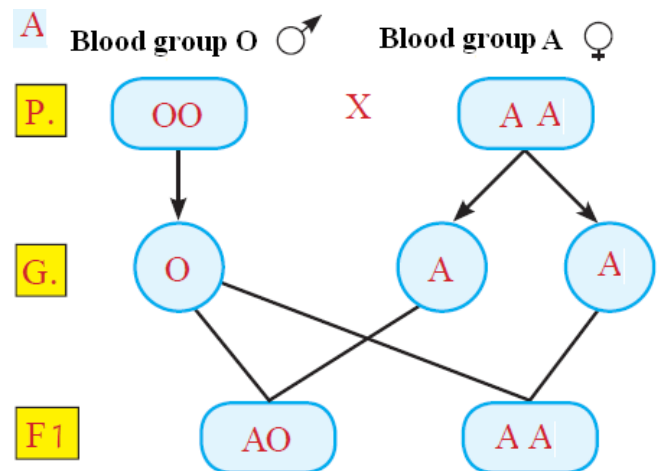
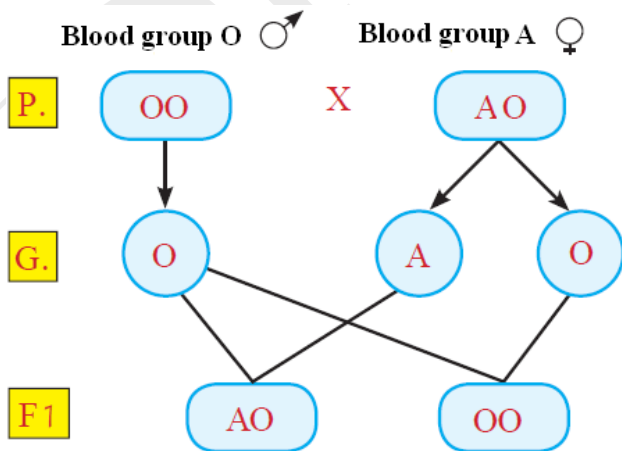
Two fathers argued (تنازعا) about the pedigree (نسب) of a child of **blood type (O)**, if the blood types of both fathers is (O), the blood type of first man's wife is (A), and the second's wife (AB). Which one of those men is more likely to be the father of the child? Why?

Solution:

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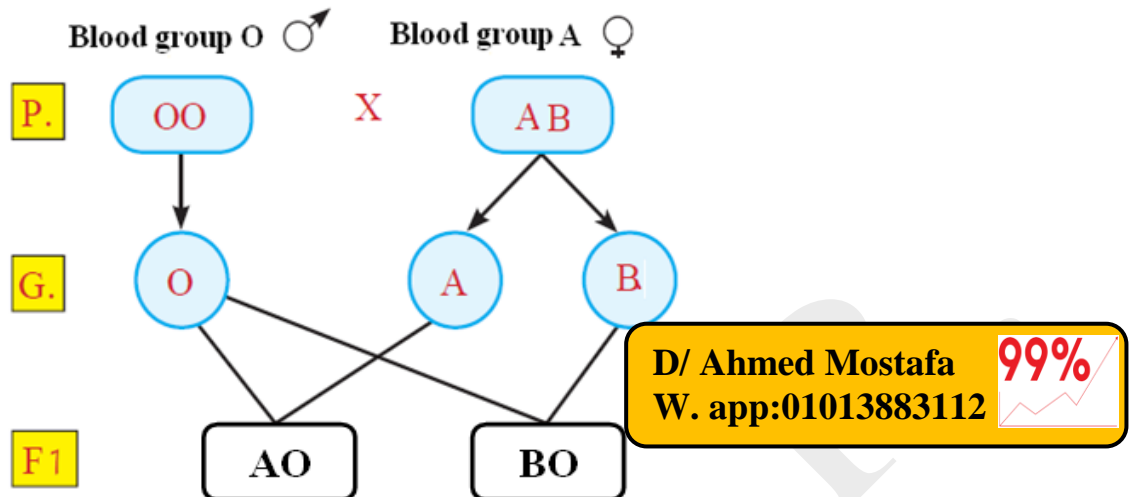
→ Genetic analysis of the first man and his wife:

- The genotype of the man is (OO), while the genotype of his wife is (AO) or (AA)



→ Genetic analysis of the second man and his wife:

- The genotype of the man is (OO), and the genotype of his wife is (AB)



Therefore, the man who is married to the woman having (A) blood group is the father of the child.

Chemical classification of blood groups:

- Blood groups are classified into 4 groups (A, B, AB and O)
- This classification is due to the presence of two types of chemical substances in the blood:

Antigens

- On the surfaces of red blood cells.
- They are two types:

- a
- b

Antibodies

- In the blood plasma.
- They are two types:

- anti-a
- anti-b

Each type of the blood groups has specific antigens and antibodies.

- **In blood group (A):** there is antigen (a) on surfaces of red blood cells, and antibody (anti-b) in the plasma.
- **In blood group (B):** there is antigen (b) on surfaces of red blood cells, and antibody (anti-a) in the plasma.
- **In blood group (AB):** there are antigen (a), and antigen (b) on surfaces of red blood cells, and there are **no antibodies** in the blood plasma.
- **In blood group (O):** there are **no antigens** on surfaces of red blood cells, and there are antibodies (anti-a) and (anti-b) in the blood plasma.

Blood transfusion process

Blood can be transfused between different blood groups according to the antigens and antibodies.

→ The following table summarizes the information about blood groups, and their relation to blood transfusion:

Blood groups	The genotype	Antigen	Antibody	Donor to	Recipient from
A	AA or AO	a	anti-b	A and AB	A and O
B	BB or BO	b	Anti-a	B and AB	B and O
AB	AB	A and B	--	AB	All groups
O	OO	--	anti-b & anti-a	All groups	O

N.B.:

1. Persons with blood group (O) are called **universal donors**, because blood from group (O) can be safely transfused to any other group.
2. Persons with blood group (AB) are called **universal recipients**, because blood can be transfused to them from any other group safely.

Blood group determination

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In order to determine an individual's blood group, we should have the following:

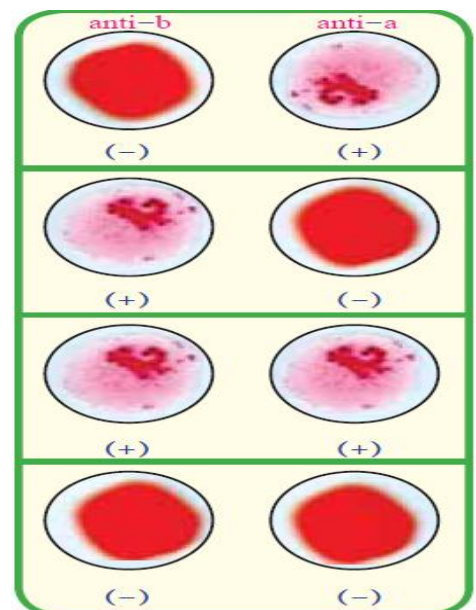
- **anti-a** Serum and **anti-b** Serum.
- Two drops of the individual's blood.

The procedure:

- 1- Put one drop of the individual's blood at the two sides of a microscopic slide.
- 2- Add a drop of anti. (A) Serum on one of the two blood drops, and a drop of anti. (B) Serum on the other drop.
- 3- Observe what happens after stirring separately.

Conclusion:

- If agglutination occurs with **anti-a** serum only:
The blood group is (A)
- If agglutination occurs with **anti-b** serum only:
The blood group is (B)
- If agglutination occurs with both **anti-a** and **anti-b** sera:
The blood group is (AB)
- If agglutination does not occur with any of the two sera:
The blood group is (O)



Possible risks of blood transfusion

There are some risks which the recipient (who takes blood) may be exposed to:-

1- When a person receives blood which is not suitable for its type, **some symptoms appear on him**, such as :-

- Shivering in body.
- Chest pain.
- Blueness.
- irregular heartbeat.
- low blood pressure.
- Shortness of breath.
- Headache

This usually ends with the death of the person

2- **Polluted blood may be transfused to the recipient person, which causes the infection with viruses** (Ex. Hepatitis b virus – AIDS virus)

→ So, blood is being examined before transfusion process in order to make sure it is suitable for the recipient blood, and doesn't carry any pathogenic (disease-causing) organisms.

Importance of the blood groups study

- 1- It helps in the dispute (فض النزاعات) of paternity.
- 2- It is used in blood transfusion.(and also the type of Rh)
- 3- It helps also in Human race taxonomy.

The Rhesus factor (Rh)

They are antigens found on the surface of the RBCs other than the antigens (A) and (B) of the blood groups.

Chemical study of Rhesus factor:

- It was discovered in the blood of a species of monkey known as **Rhesus** before it was discovered in the Human blood.
- These antigens are found in about **85%** of the Human individuals, who are known as **Rh⁺ persons**, whereas the other **15%** are free from these antigens, and they are called **Rh⁻ persons**.

Genetic study of Rhesus factor:

- Production of Rhesus antigens is controlled by **three pairs of genes**.
- These genes are very close in their effect, and they are close to each other **on the pair of chromosomes** that carries them.
- The presence of any pair of these pairs of genes in the dominant state leads to an **Rh⁺ person**. Thus, Rh⁺ persons may be homozygous **Rh⁺ Rh⁺**, that is the three pairs of genes are found in the











dominant state, or heterozygous $Rh^+ Rh^-$, where some of the three pairs of genes are dominant, while the rest are recessive.

- In the Rh^- persons, all his genes are recessive and the person will be homozygous $Rh^- Rh^-$.

Blood types according to blood groups and Rh

The following table summarizes the chemistry of blood group system and Rh factor.

								
Antigens	a, Rh	a	b, Rh	b	a, b, Rh	a, b	Rh	-----
Antibodies	anti-b	anti-b	anti-a	anti-a	-----	-----	anti-b anti-a	anti-b anti-a

Importance of the genetic study of Rhesus

The antigens of Rh factor naturally **has no antibodies in the plasma of persons**, however the body of a person can form antibodies for the Rh factor (anti-Rh) when:

- 1- Transfusion of blood from a positive Rh person to a negative person or,
- 2- During mixing between the blood of a negative Rh mother with a positive Rh baby during giving birth (delivery).

		Donor							
		O-	O+	B-	B+	A-	A+	AB-	AB+
Recipient	AB+								
	AB-								
	A+								
	A-								
	B+								
	B-								
	O+								
	O-								

Example:

If a woman (Rh-) is married to a man (Rh+) their children will be:

- 100% Rh⁺ children, if the father was homozygous (Rh⁺ Rh⁺).
 - 50% Rh⁺ children, and 50% Rh⁻ children, if the father was heterozygous (Rh⁺ Rh⁻).
- * If the fetus is Rh⁺ and the mother is Rh⁻, some of the blood that leaks (يتسرب) from the fetus to the mother's blood during giving birth (الولادة) stimulates it to produce antibodies against the Rhesus factor (**anti-Rh**), normally the first baby is not usually affected.
- The second baby receives an amount of the antibodies that pass to the baby through the placenta and causes disintegration of the his red blood cells, infecting the fetus with sever anemia, which may lead to his death, **unless:**
 - The mother is injected with a **protective serum** after the birth of the first baby (during 72 hours at most) this serum breaks down the red blood cells that leak from the blood of the baby and carry the antigens of Rh before they stimulate the immune system of the mother to produce anti-Rh.

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If the father was (Rh⁺ Rh⁻) the second baby **may be (Rh⁻ Rh⁻)** as his mother, in this case he will not be affected by the Rhesus antibodies of the mother's blood even if his mother did not take the **protective serum**.



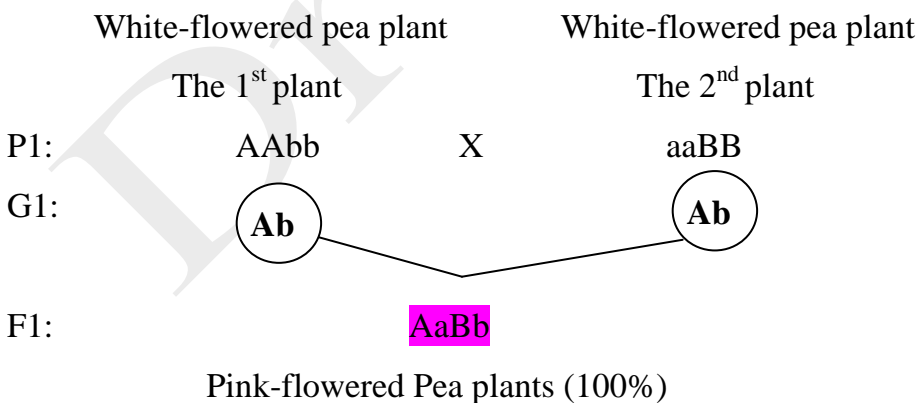
- It is a genetic case in which the trait is controlled by **two pairs of genes**; there must be at least **one dominant gene** from each pair for the characteristic to emerge (appear).
- Each of the two pairs controls the production of a certain enzyme which affects part of the steps of the appearance of the characteristic, if one of the two dominant genes was absent, the steps of the appearance of the characteristic will not be completed.

Example for the complementary genes

Inheritance of the flower color of Pea flower plant (sweet pea):

- When two strains of white-flowered Pea plants were crossed, all the flowers of F1 plants appeared pink.
- In F2: the flowers produced were 9 pink: 7 white, this indicates the presence of two pairs of genes that control the flower color characteristic.
- If the dominant gene in one pair is represented by A, and the recessive gene in that pair is represented a.
- If the dominant gene in the second pair is represented by B, and the recessive gene in that pair is represented by b.
- The genotypes of the white-flowered pea plants (P1) would be (AAbb) and (aaBB). Then the genotype of F1 plants will be (AaBb) as follows:

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	♂		
♀		Ab	Ab
	aB	AaBb	AaBb
	aB	AaBb	AaBb

- The 1st plant is white due to the absence of a dominant B gene.
- The 2nd plant is white due to the absence of a dominant B gene.
- The F1 plants are pink-colored due to the presence of both A and B dominant genes.

The individuals of the F2 will be as follows:

P2: Pink-flowered pea Pink-flowered pea

AaBb X AaBb

	(AB)	(Ab)	(aB)	(ab)	
G2:	(AB)	AABB pink	AABb pink	AaBB pink	AaBb pink
	(Ab)	AABb pink	AAbb white	AaBb pink	Aabb white
	(aB)	AaBB pink	AaBb pink	aaBB white	aaBb white
	(ab)	AaBb pink	Aabb white	aaBb white	aabb white

F2: Pink : White
9 7

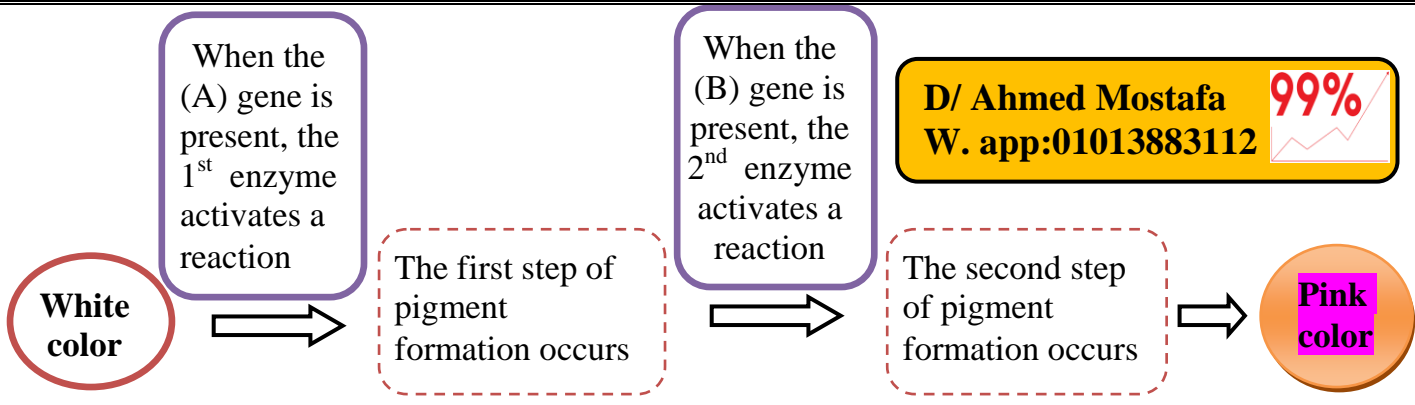
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Conclusions:

- 1- The appearance of the ratio 9 pink: 7 white emphasizes that this characteristic is **influenced by two pairs of genes.**
- 2- The ratio 9:7 is considered as a deviation from the Mendelian ratio 9:3:3:1 **because** the last three genotypes **(3:3:1)** have the same phenotype.
(Note: the Mendelian ratio deals with two pairs of allelomorphic traits)
- 3- **In order for the pink color of flowers to appear,** the two pairs of genes must be present in the dominant state (whether the homozygous or the heterozygous).
- 4- The white color of flowers appears. if only one pair is found in the dominant state, and the other pair in the recessive state or both pairs are found in the recessive state.
- 5- Thus, both pairs of genes participate in the color production. Since, each of them controls the production of a certain enzyme which affects part of the steps of the pigment production.
- 6- If one of the two dominant genes (A or B) was absent, the steps of pigment production will not be completed, and the flowers appear white.

For illustration only:

The validity of the above explanation was confirmed experimentally when extracts of flowers from both strains were mixed together in a test tube, **the pink color** appeared as a result of the presence of all the enzymes, and the completion of the reaction that produces the pigment



Phenotypes	Genotypes	Gametes	No. of gametes	
White-colored flower plants	AAbb	Ab	1	The pink color does not appear (emerge) due to the absence of either the (A) or the (B) genes or both of them.
	Aabb	Ab, ab	2	
	aaBB	aB	1	
	aaBb	aB, ab	2	
	aabb	ab	1	
Pink-colored flower plants	AABB	AB	1	The pink color appears (emerges) due to the presence of both the dominant genes together.
	AaBB	AB, aB	2	
	AABb	AB, Ab	2	
	AaBb	AB, Ab, aB, ab	4	

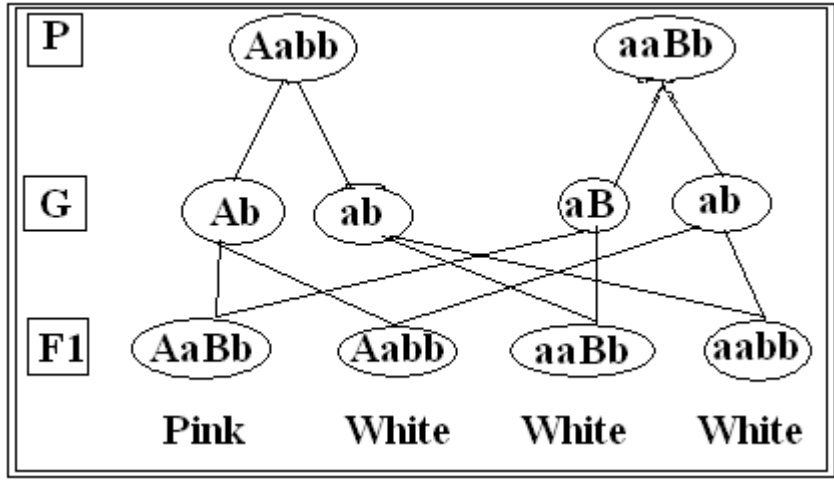
Example (1)

Determine the phenotypes and genotypes of the colour of flowers produced from the copulation (Aabb x aaBb)

Solution

3 : 1

- 3: White-colored flower plants
- 1: Pink-colored flower plants



They are some genes that control some characteristics when found in a pure (homozygous) state (dominant or recessive), lead to the disruption of some vital processes and leading to the death of almost 25% (one fourth) of the offspring at different stages of life.

There are two types of lethal genes

Dominant lethal genes

1- Genes of yellow fur colour in mice

(YY)

2- Genes of Bulldog strain in cattles (DD)

Recessive lethal genes

1- Genes of chlorophyll absence in corn plant

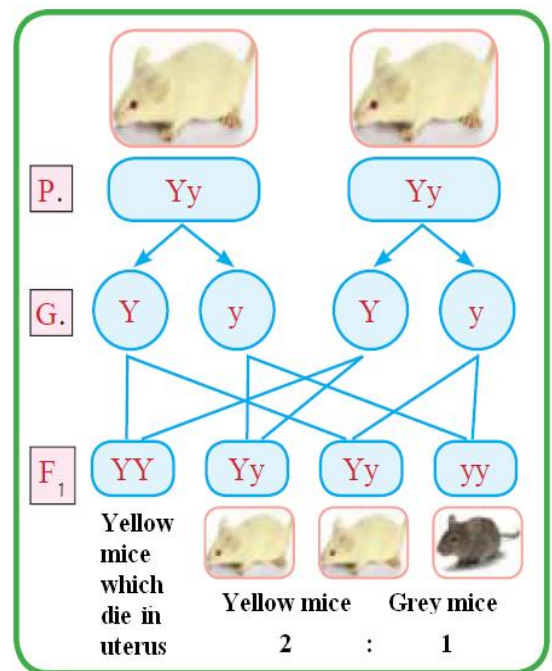
(cc)

2- Genes of infantile dementia in human (aa)

1- Dominant lethal genes

Inheritance of yellow fur colour in mice:

- The fur colour in mice is inherited by 2 genes:
- The gene of yellow fur colour (Y) which dominates over the gene of the grey fur colour (y).
- When two hybrid yellow mice (Yy) mate, the ratio of produced generation is 1:2, where:
→ Pure yellow mice (represent about $\frac{1}{4}$ of the offspring) died because they carry pure lethal dominant pair of gene (YY) which causes the death of mice in mother's uterus.



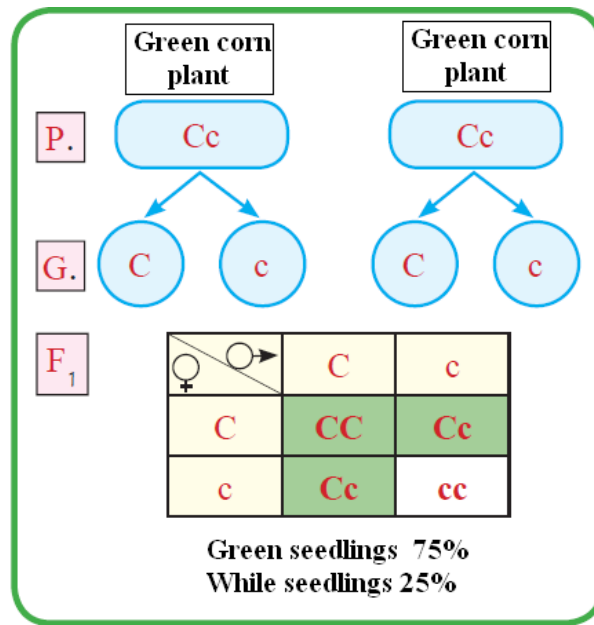
2- Recessive lethal genes

Inheritance of chlorophyll absence trait in corn plant

- When some corn plants self-pollinate, they produce white seedlings which don't have chlorophyll. These seedlings grow for a short time, then they wilt and die. This happens because of a pure recessive lethal gene (cc) which is the gene of absence of chlorophyll.

→ When two recessive lethal genes aggregate together in some individuals, chlorophyll will not be formed in them. Chlorophyll gives the plant its green colour and it is responsible for absorbing light rays to carry out photosynthesis process.

→ Plants carrying (cc) pair of genes are white and not able to perform photosynthesis process which causes their death.



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The effect of environmental conditions on action of some genes

The work of some genes is affected by **the factors surrounding living organisms**, such as:

- Air pollutants
- Lack of oxygen (oxygen deficiency)
- Exposure to radiation.
- Some environmental factors (Ex. Light – temperature).

→ The study of these factors helps us in avoiding the risks resulted from them.

The effect of absence of light on chlorophyll trait in green plants

Experiment 1:

Germinate a group of wheat or corn grains **in an illuminated place** and irrigate the seedlings regularly for several days.

Observation:

Green seedlings grow due to the presence of light factor which is needed by the gene that is responsible for the formation of chlorophyll to show its effect.

Experiment 2:

Germinate a similar group **in a dark place** and irrigate the seedlings regularly for several days.

Observation:

Yellow seedlings grow then wilt and die after a period of time due to the absence of light factor which is needed by the gene that is responsible for the formation of chlorophyll to show its effect.

Conclusion:

→ The gene responsible for chlorophyll formation in green plants needs the light factor to show its effect.

→ **In the absence of the gene** causing the appearance of chlorophyll, the plant can not form the chlorophyll pigment, even if it was placed in the light.

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Notes:

- **The internal cabbage leaves are white-coloured**, because they are not exposed to light needed for appearing the effect of the gene that is responsible for the formation of green chlorophyll pigment, on contrast **the external leaves are green-coloured**, due to their continuous exposure to light which helps in the formation of chlorophyll.
- **When exposing the internal cabbage leaves to light**, they are changed to green, due to the appearance of the effect of green chlorophyll gene.

For illustration:

Another examples that ensure the effect of environmental conditions on the appearance of inherited traits:

- 1- Hymalayan rabbits fur is coloured with a black colour, if it is exposed to cold for a long period of time.
- 2- The wings of Drosophila fly (after pupation):
 - Grow in straight form after the exposure of eggs to a temperature of 16°C.
 - Grow in a curved form after the exposure of eggs to a temperature of 25°C.



- The sex of an individual is determined by a special type of chromosomes called **sex chromosomes**.
 - Usually one or two of these sex chromosomes exist in each cell of the individual, and the rest of the chromosomes are called **autosomes** (somatic chromosomes).
- **In the past**, it was thought that woman controls the sex of her fetus, but when scientists discovered sex chromosomes, they discovered that **man** controls the sex of the fetus (not woman)
- In the nucleus of human somatic cells, there are 23 pairs of chromosomes (46 chromosomes).

There are 2 types of chromosomes in the somatic cells:

Autosomes (somatic chromosomes)

- Their number is 22 pairs (44).
- Identical in males and females.

Sex chromosomes.

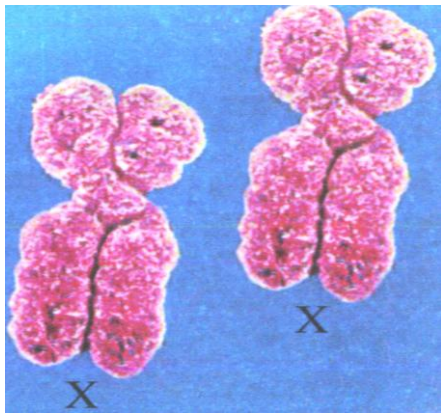
- Their number is 1 pair (2).
- Differ in the males from the females.

The chromosomal structure in the cells of male and female:

	The cells in the female human	The cells in the male human
Chromosomal structure	44 + XX	44 + XY
No. of autosomes	44	44
No. of sex chromosomes	2	2
Types of sex chromosomes	XX symmetric (identical)	XY Asymmetric (not identical)
The gonads	Ovaries	Testes
Gametes	Ova Produced by meiotic division of the ovaries.	Sperms Produced by meiotic division of the testes.
Chromosomal structure of gametes	All of one type (22 + X)	There are 2 types (22 + X) and (22 + Y)

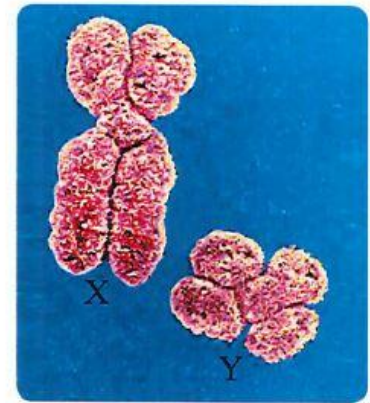
Compare between the sex chromosome X and the sex chromosome Y:

	The sex chromosome X	The sex chromosome Y
Size	Larger than Y	Smaller than X
Genes	Carries other genes that have no relation to sex or sexual development.	Does not carry other genes than those of sex determination in mammals.



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Action of genes responsible for sex determination:

The genes responsible for sex determination and carried on the sex chromosomes X, and Y act only in the first months of Human embryonic development:

- **After six weeks from the beginning of pregnancy**, the embryo begins the production of male hormones if the Y chromosome is present. These hormones induce the gonad tissues (which are undifferentiated) to produce testes, then the rest of the male genital organs is differentiated.
- **After 12 weeks from the beginning of pregnancy, if the embryo does not contain the Y chromosome**, the embryo begins to form ovaries, then the rest of the rest of the female genital organs is differentiated.

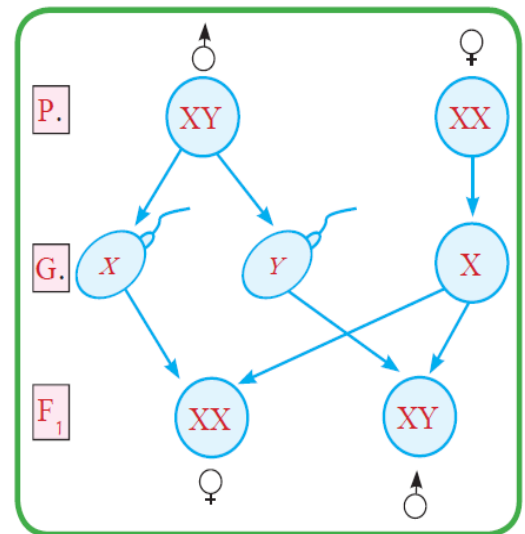
Sex determination in man:

- The chromosome (Y):

is a main constituent of sex determination.

- If the chromosome Y is present, the individual will be a male.
- If it is absent, the individual will be a female.
- There is no one single case in Man or other mammals that **lack the sex chromosome X**, since life requires at least one of such chromosome.

The chromosome Y is not essential for life, since females don't have it.



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There are other systems of sex determination in livings:

Example: Sex determination in turtles, depending on temperature, as:

- The eggs that were laid near the soil surface with high temperature hatch to females.
- The eggs that were laid away from the soil surface with low temperature hatch to males.



Abnormal cases of number of chromosomes in Humans

Abnormality in number of chromosomes:

Reasons:

- Sometimes, during meiosis for gamete formation, mistakes take place.
- Sex chromosomes are not equally distributed, This leads to abnormal cases, **where:**
- In some rare cases, the two sex chromosomes **adhere** closely to each other during meiosis and do not separate, This leads to the presence of the two X chromosomes in one ovum and the absence of the X chromosome from the other.

From the examples of the human abnormal chromosomal cases

1- Klinefelter's syndrome (44+XXY)

Chromosomal structure	44+XXY
No. of chromosomes	47
Sex	Male (due to the presence of (Y) chromosome.
The defective chromosomes	Sex chromosomes
Reason of the case	The fertilization of an abnormal ovum (22 + XX) having two X chromosomes by a normal sperm having Y chromosome (22 + Y)
Reason of the disturbance	The extra X chromosome leads to a disturbance in the balance of the sex determining genes where the female genes that carried on the extra X chromosome express themselves in some way.
Symptoms	A sterile male due to the absence of sperm generating cells in his testes. Tallness. Appearance of some feminine characters, such as the growth of the breast's size.
Case discoverer	Dr. Henry Klinefelter 1942.

2- Turner's syndrome

Chromosomal structure	44 + XO
No. of chromosomes	45
Sex	Female
The defective chromosomes	Sex chromosomes
Reason of the case	The fertilization of an abnormal ovum (22 + O), having no X chromosome, by a normal sperm having X chromosome (22 + X)
Reason of the disturbance	The lackage of X chromosome including the genes of unsexual characters that carried on it, leading to the production of a female with several deformation.
Symptoms	A female who does not reach puberty, due to the lack of sufficient amount of hormones. Shortness. The presence of some congenital defects in the heart and the kidneys.
Case discoverer	Dr. Turner in 1938.

3- Turner's syndrome

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Chromosomal structure	The males: 45 + XY The females: 45 + XX
No. of chromosomes	47
Sex	Male or female
The defective chromosomes	Somatic chromosomes (autosomes)
Reason of the case	The fertilization of a normal gamete by an abnormal gamete (ovum or sperm) which carries a pair of chromosomes no. (21)
Reason of the disturbance	The presence of three copies of the chromosome no. (21).



Symptoms

- Growth retardation
- - Mental retardation.
- - The back of head is flate.
- Small ears.
- Oval face
- Shortness
- Convex eyes.
- Short fingers and toes.

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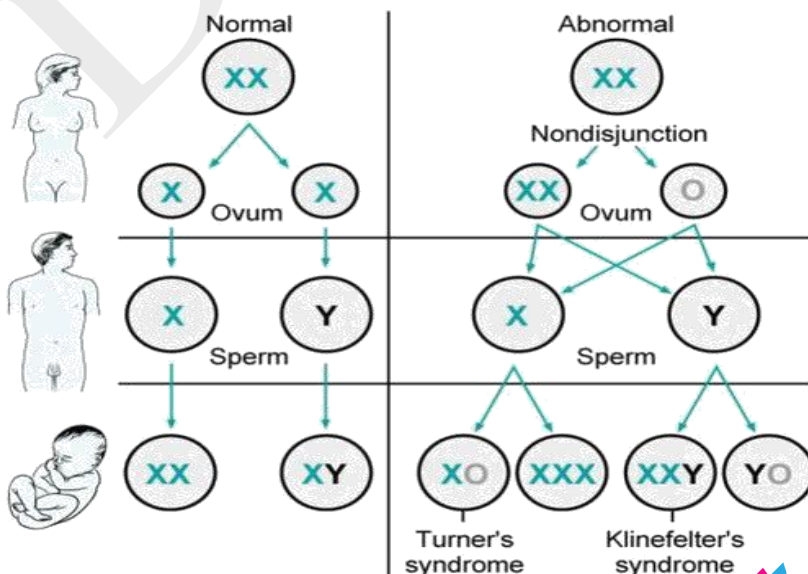
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Chromosomal structure	44 + XXX
No. of chromosomes	47
Sex	Female
The defective chromosomes	Sex chromosomes
Reason of the case	The fertilization of an abnormal ovum (22 + XX) having two X chromosomes, by a normal sperm having X chromosome (22 + X)

Summary

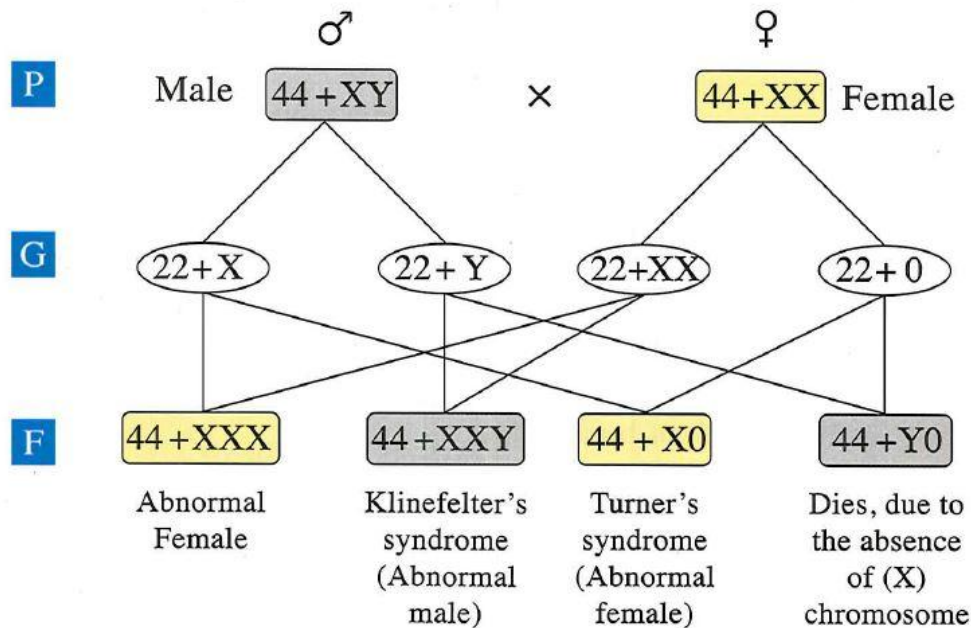
Characteristics of the individual	Chromosomal structure	Sex
Normal male	46: 44+XY	Male
Normal female	46: 44+XX	Female
Down's syndrome (Mongolism)	47: 45+XY	Male
Down's syndrome (Mongolism)	47: 45+XX	Female
Klinefelter's syndrome	47: 44+XXY	Male
Turner's syndrome	45: 44+X	Female
Multiple sex genes	47: 44+XXX	Female
Multiple sex genes	48: 44+XXXX	Female



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Example

When a normal man married to a normal woman, the genetic analysis that shows the possibilities of giving birth to abnormal individuals after fertilization is, as the following



Sex-linked traits

Case discoverer: Thomas Morgan.

- He found that the genes of some somatic traits are carried on the sex chromosomes.
- He named these traits as sex-linked traits.

They are somatic traits that their genes are located on the sex chromosomes and their appearance is not affected by the sex hormones.

Examples:

- 1- In Drosophila insect: eye colour.
- 2- In human: color blindness, haemophilia, muscle atrophy and short-sightedness.

The inheritance of Drosophila eye colour

Experiment:

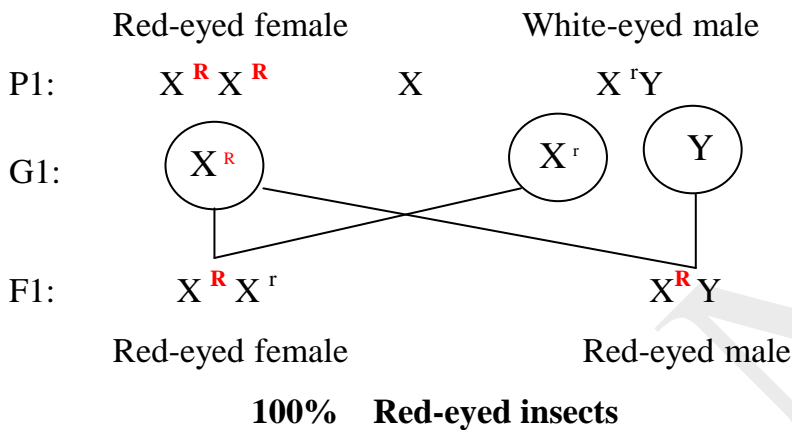
- Morgan crossed a white-eyed male with a red-eyed female, the F1 generation was all red-eyed. So that the red colour of eyes is **dominant** over the white colour of eyes.
- By inbreeding the F1 individuals, he got red-eyed and white-eyed insects in the ratio 3:1.

However, the white-eyed insects were all **males**.

Conclusion:

- Morgan explained the appearance of white-eyed males in the F1 generation by assuming that the gene for the white colour of the eye is recessive and the genes of eye color is carried on the sex chromosome X, while the sex chromosome Y does not carry the the other allele of the gene, Thus, the presence of one gene of the white colour of eyes is sufficient for males to be white-eyed.
- Morgan called this case sex-linked inheritance to refer to traits that are determined by the genes located on the sex chromosome X.

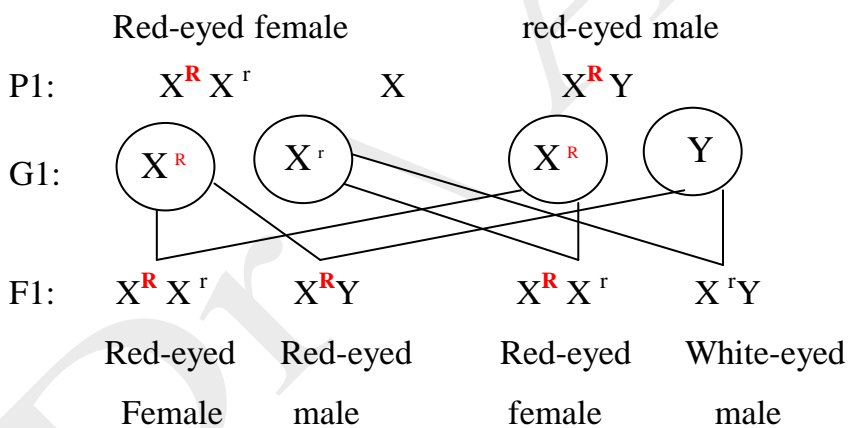
Morgan represented the crossing between white-eyed males, and red-eyed females as follows:



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By inbreeding F1 individuals, red-eyed insects and white-eyed insects appear in the ratio 3:1, where the white colour of eyes appears in males only as follows:

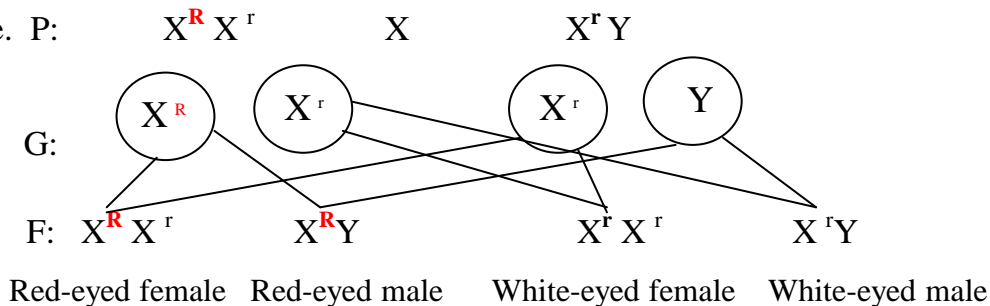


Problem:

How can you obtain a white-eyed female Drosophila insect, explain on genetic bases.

Solution:

By crossing a hybrid red-eyed female Drosophila (its father was a white-eyed) with a white-eyed male. P:



Sex-linked character in human

- In Man, the genes of some characters are carried on X chromosome, such as the genes of:
 - Color blindness.
 - Haemophilia.
 - Muscle atrophy.
 - Short-sightedness.

Enrichment information

Sex chromosome (Y) in male human carries some genes that related to somatic characters without the presence of corresponding genes to them on the sex chromosome (X), such as the gene that is responsible for the appearance of hair on ear margins in males. So, these traits are restricted only to males.

In this case:

- The male has only one gene for each trait, this gene is carried on the sex chromosome (X), he will inherit this gene for his daughters only.
- The female has two genes for each trait, one on each of the two sex chromosomes (X), she will inherit these genes for her daughters and sons.

The inheritance of Colour blindness

Colour blindness is a case causes **the inability to distinguish the colours**, especially the red and green colours.

The gene of colour blindness is recessive and is carried on sex chromosome (X).

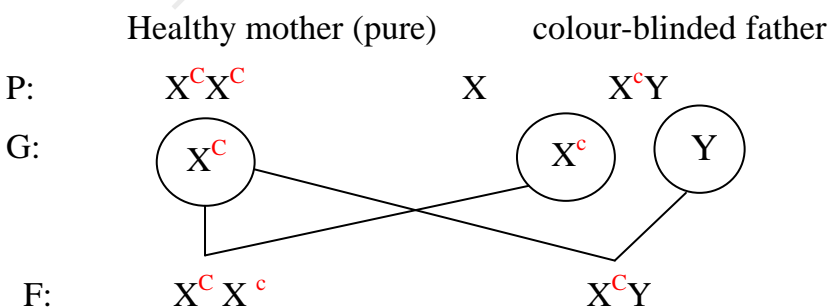
The gene of colour blindness is symbolized as (X^c), while the normale gene is (X^C)

	Normal vision	Colour blinded	No. of genes
Males	$X^C Y$ No carrier males	$X^c Y$	Have only one gene for the trait (always pure)
Females	$X^C X^C$ (Pure) and $X^C X^c$ (carrier)	$X^c X^c$	Have two genes for the trait

Example 1:

What would happen when a colour blinded man married to a normal vision woman (Pure)

Solution:



Healthy female (hybrid) carries the gene of the disease

Healthy male has only one normale gene

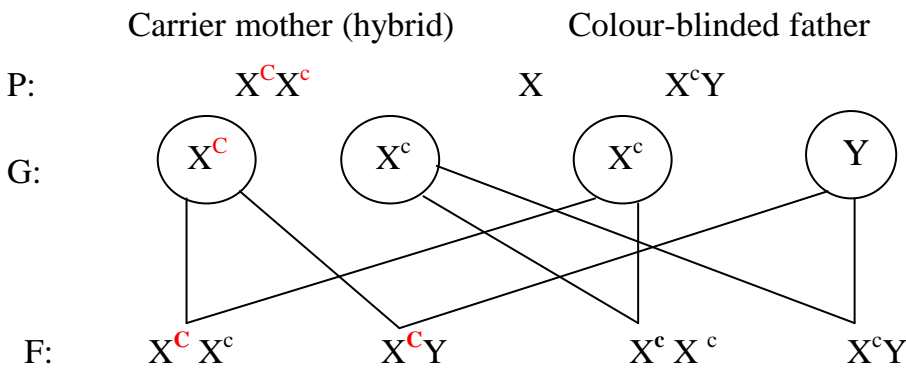
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Example 2:

What would happen when a colour blinded man married to a woman that is carrier to the gene of colour blindness (hybrid).

Solution:



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Healthy female (hybrid) carries one gene of the disease

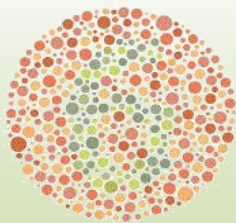
Healthy male has one normal gene

Infected female (Pure) carries two genes of the disease

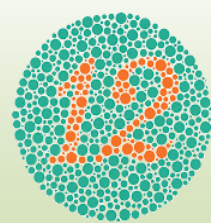
Infected male carries one gene of the disease

Test your eyes

Can you see the number in both the first and second circles



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The inheritance of Haemophilia (Blood liquidity)

Haemophilia is a genetic case causes blood liquidity, due to the absence (lack) of some substances that are necessary for the blood clotting.

The gene of haemophilia is recessive and is carried on sex chromosome (X).

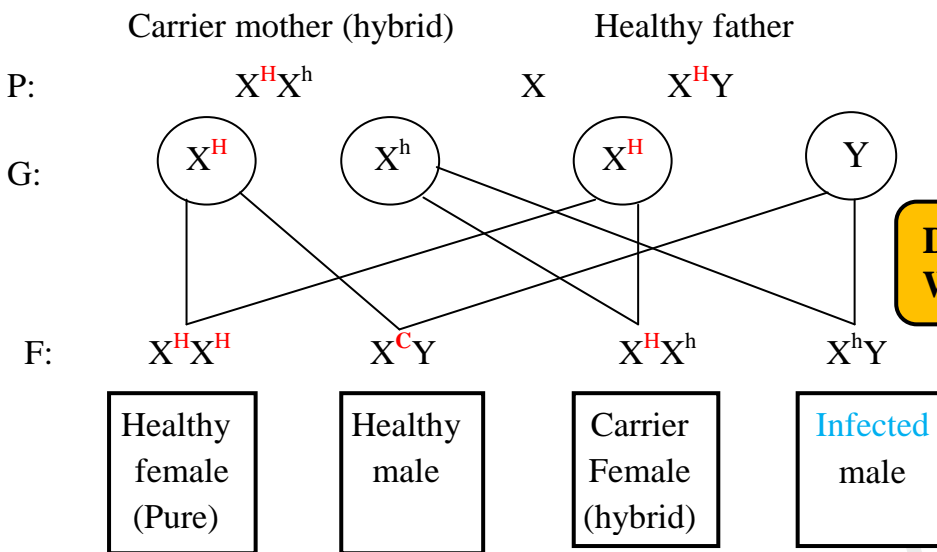
The gene of haemophilia is symbolized as (X^h), while the normale gene is (X^H)

	Normal blood	Infected	No. of genes
Males	$X^H Y$ No carrier males	$X^h Y$	Have only one gene for the trait (always pure)
Females	$X^H X^H$ (Pure) and $X^H X^h$ (carrier)	$X^h X^h$	Have two genes for the trait

Example 2:

What would happen when a healthy man married to a haemophilic carrier woman.

Solution:



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- Pedigree studies (دراسات سجل النسب الوراثي) indicated that these two characteristics are common among males while they are recessive and rarely occur in females.

Sex linkage follows the same system as that seen in case of colour of eyes in Drosophila:

- The female passes these traits to her sons and daughters.
- The sick male passes these traits to his grandsons through his daughters.
- Sons inherit the sex linked gene from their mothers.
- Sex-linked trait (as Colour blindness and Haemophilia) appears in daughters when they gain this gene from both parents (this requires that the mother should be a carrier, whereas the father would be infected).



Sex-influenced traits

They are genetic traits that their genes are carried on somatic chromosomes (autosomes) not sex chromosomes but the action of these genes is influenced by the sex hormones that are secreted from the gonads of adult males and females.

i.e, **The sex of the individual acts to modify the dominance of some traits.**

Examples:

- Genetic baldness trait in humans.
- The presence of horns trait in cattles.

The inheritance of genetic baldness trait in humans

- Genetic baldness spreads among males of some families more than in females.
- This trait is controlled by a dominant gene which is influenced by masculinity (male) hormones (هرمونات الذكورة).
- This gene causes hair falling.
- The baldness appears in the male in the presence of **one gene only due to the presence of male hormones**, whereas in females it requires the presence of the **two genes** where the hair of the female head falls down.

Genotype Sex	Pure genotype (B ⁺ B ⁺)	Hybrid genotype (B ⁺ B)	Pure genotype (BB)
Male :	Male suffers from baldness, due to the : - Presence of two dominant genes. - Masculinity hormones.	Male suffers from the baldness, due to the : - Presence of one dominant gene. - Masculinity hormones.	Normal hair.
Female :	Female suffers from genetic hair falling, due to the : - Presence of two dominant genes. - Absence of masculinity hormones.	Normal hair, although the presence of one dominant gene, but it doesn't express its effect, due to the absence of masculinity hormones.	Normal hair.

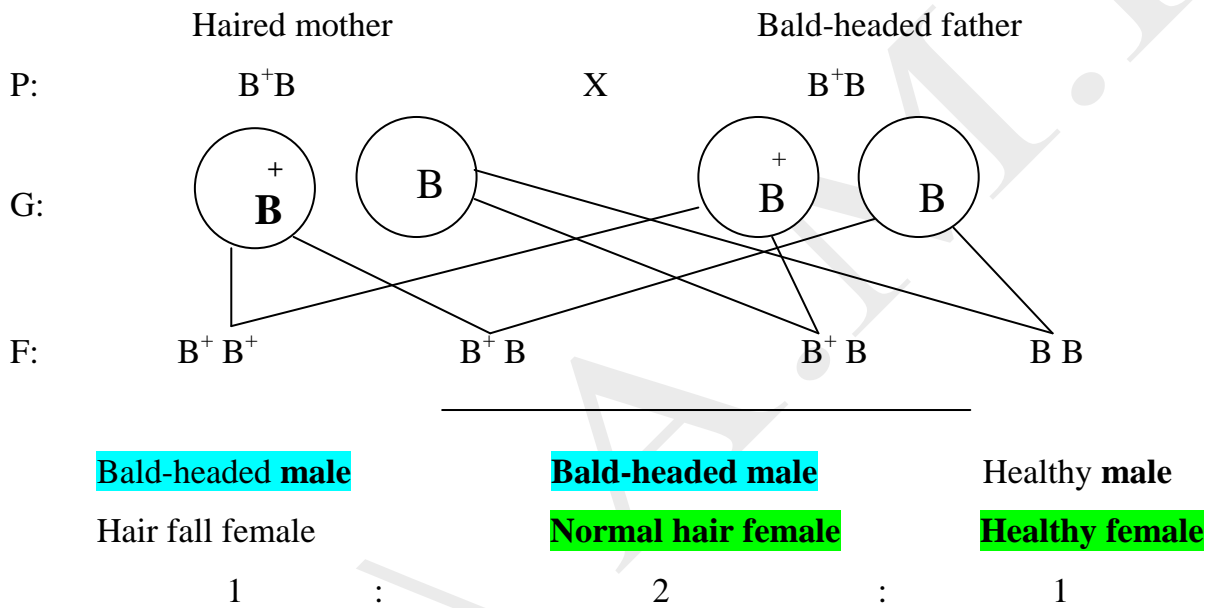
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Example:

- When a bald man marries a normal woman, both of them heterozygous, the results will be as follows:



Sex-limited traits

They are traits whose appearance is constricted (limited) on one sex only, due to the difference in sex hormones of each sex.

Examples:

- **Milk production** trait is limited on females only, because they have certain sex hormones which help the gene to express its effect.
- The ability of birds to **lay eggs** is limited on females only.
- **The appearance of beards** is limited on males only, it is one of the secondary sex character in male humans.

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Medical examinations before marriage

They are a group of medical tests which are carried out by the persons who will get married.

Causes of the medical tests for the persons who will get married:

- 1- To make sure that they are free from:
 - **Infectious diseases** (such as hepatitis and Acquired immunity deficiency syndrome (AIDS))
 - **Genetic diseases** (Such as Thalassemia).
- 2- Giving the medical council about the probability of the transmission of the previous diseases to the other partner or offspring in the future.
- 3- Providing the choices or alternatives to who will get married to help them in planning for a healthy family.

Importance of the medical tests for the persons who will get married:

- Giving birth to healthy children
- Limiting the spreading out of genetic diseases, congenital deformities (disorders) and mental retardation.
- Avoiding social, psychological and financial problems resulted from taking care of children with genetic diseases.

Science, Technology and Society

1) The DNA fingerprint (DNA typing)

It is sequences of of the genetic material (DNA) that repeat themselves several times in the living organism.

→ In 1984, Dr. Alec Jeffreys (genetics scientist Leicester university in London) published a research in which he explained that:

The genetic material (DNA) may be repeated several times inside the living organism.

→ In 1985, he discovered that every person has unique sequences. These sequences cannot be the same in any two persons (except for identical twins).

→ He named them **human DNA fingerprint (DNA fingerprint)**

Importance of the genetic fingerprint:

In medical field:

- Used in the study of genetic diseases and operations of tissues transplantation.

In forensic medicine:

- It helps in recognizing deformed corpses (dead bodies).

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- Tracking the missing children.
- Judging issues of ancestries.
- Exoneration (تبرئة) or condemnation (ادانة) of persons from killing and ravishment (الاعتداء) crimes.

2) human Genome:

It is all the genes that found in the nucleus of each somatic cell

- The number of genes in each cell is estimated between 60000 to 80000 genes that are carried on the 23 pairs of chromosomes, these genes are responsible for the presence of enormous number of human character.

Discovering the human genome:

- In 1953, scientists Watson and Creek discovered that genes are carried on a double helix of DNA.
- In 1980, the idea of the human genome appeared and the number of identified human genes by scientists was only 450 genes.
- In the middle of eighties, the number of identified genes was doubled three times to reach about 1500 genes.

Uses of human genome:

- Identifying the genes causing the genetic diseases through drawing a good genetic map that identifies the location of genes on the chromosomes accurately.
- The study of evolution of living organisms by comparing the human genome with other ones of the other organisms.
- Breed improving through identifying the genes of diseases in the fetus before its delivery and act to improve them.
- Manufacturing drugs without side effects.

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