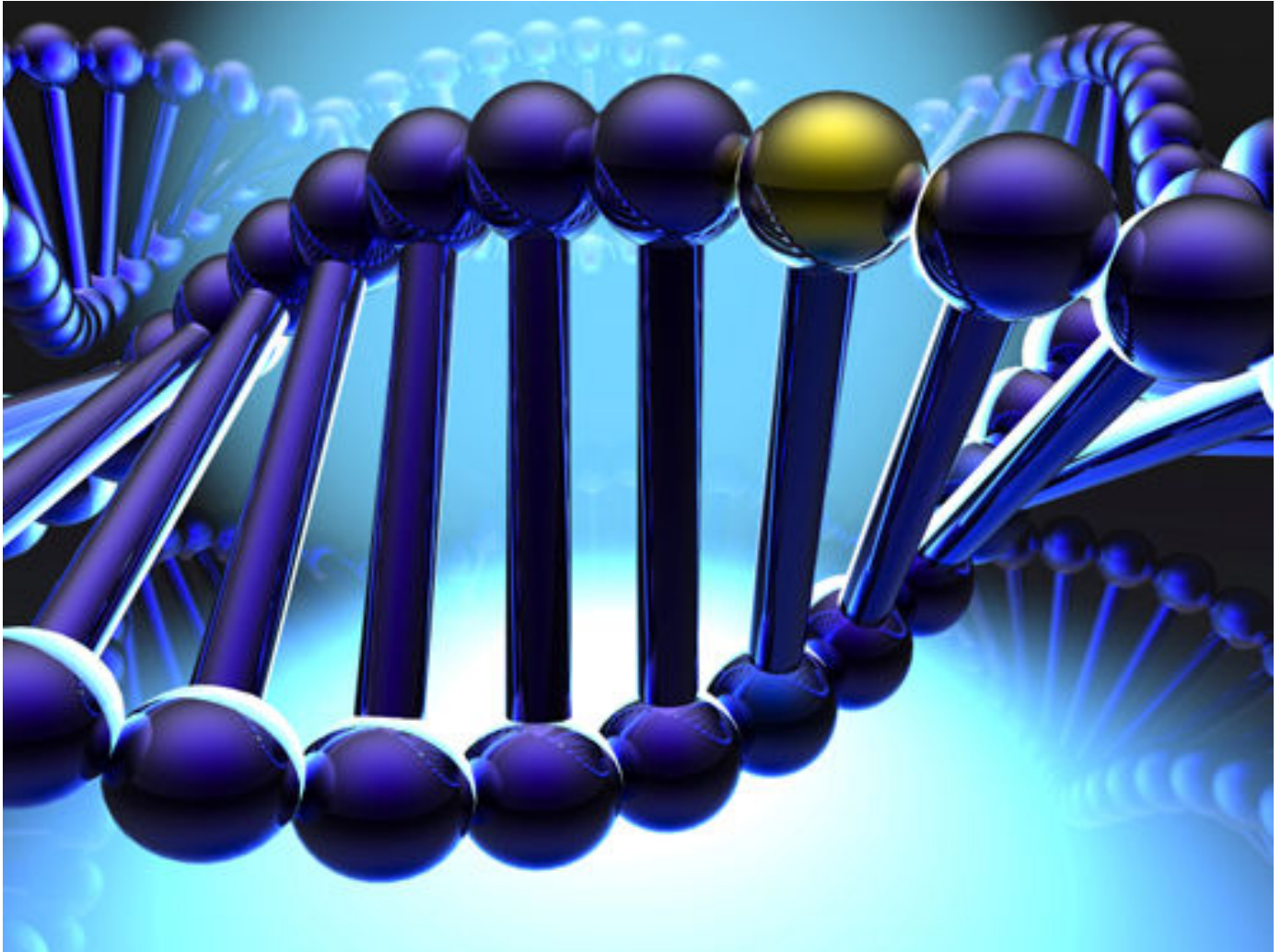


## Chapter (3) Inheritance of traits

### *Lesson (1) Chromosomes and genetic information*



→ In early 20<sup>th</sup> century, scientists discovered that genetic information is carried on chromosomes, this genetic information causes the appearance of hereditary traits in living organisms.

→ Chromosomes exist inside cell nucleus, they are found in the form of homologous pairs in somatic and reproductive cells.

## Karyotype

**Karyotype:** The arrangement of chromosomes of cell nucleus in descending order according to their size and number.

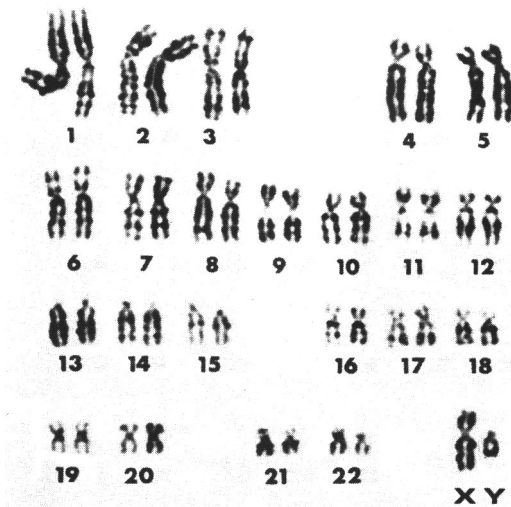


Fig. (1) Karyotype of human male

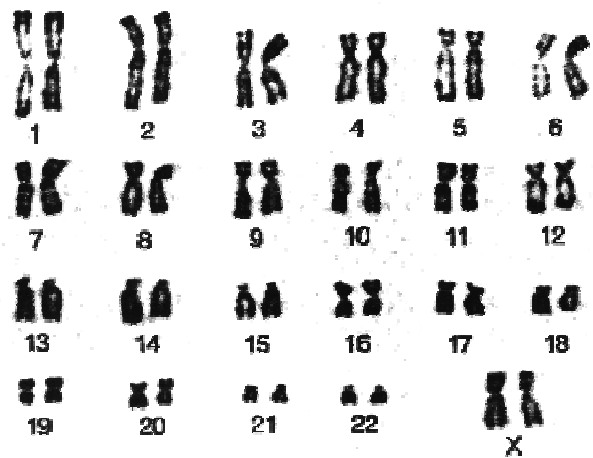


Fig. (2) Karyotype of human female

### Number of chromosomes:-

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

**Somatic cells:** Each somatic cell has two pairs of homologous chromosomes (one pair is inherited from father while the other from mother). They are called **diploid cells (2n)**

**Reproductive cells (gametes) (sperms – ova – pollen grains):** Each gamete has one pair of chromosomes. A gamete has half the number of chromosomes of somatic cell. Gametes are known as **haploid cells (n)**

→ In human, 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 in male:-**

Sex chromosomes pair in male is non homologous (XY) (See fig (1))

### **Karyotype in female:-**

Sex chromosomes pair in female is homologous (XX) (See fig (2))

**(N.B:** The constant number of chromosomes in all members of human kind proved that chromosomes carry genetic information responsible for the appearance of hereditary traits)

Species	Number of chromosomes
Human	46
Chicken	32
Cat	38
Drosophila	8
Dog	78
Tobacco	48
Gorilla	48
Onion	16
Potato	48
Peas	14
Frog	26
Wheat	48

## **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

→ A gene consists of a series of nucleotides, it forms protein codes which are responsible for the appearance of traits in living organisms.

---

**Remember**

**Fertilization:** The fusion of male gamete (n) with female gamete (n) forming zygote of complete number of chromosomes (2n)

---

**Mendel's laws in light of chromosome theory**

---

**Remember**

**Mendel's second law (law of independent assortment of hereditary factors):**  
When two individuals bearing one or more contrasting pairs of hereditary factors copulate, each factor is inherited independently of the other factors.

---

We studied in 3<sup>rd</sup> prep that :-

- In Meiotic cell division, genes separate from each other forming gametes, but they meet again after fertilization process forming zygote of (2n) of chromosomes.
- Dominant trait appears in all members of 1<sup>st</sup> generation at ratio 100%
- Dominant and recessive traits appear in members of 2<sup>nd</sup> generation at ratio 3 dominant : 1 recessive

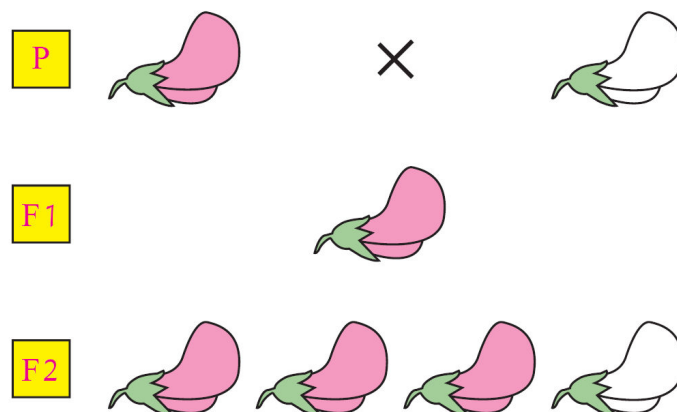


Fig. (3) The appearance of pink roses (dominant) and white rose (recessive)

→ When two individuals bearing one or more pairs of contrasting genes copulate, every gene is inherited independently of the others.

→ The assortment (distribution) of genes in gametes is independent because every gene exists on an independent chromosome.

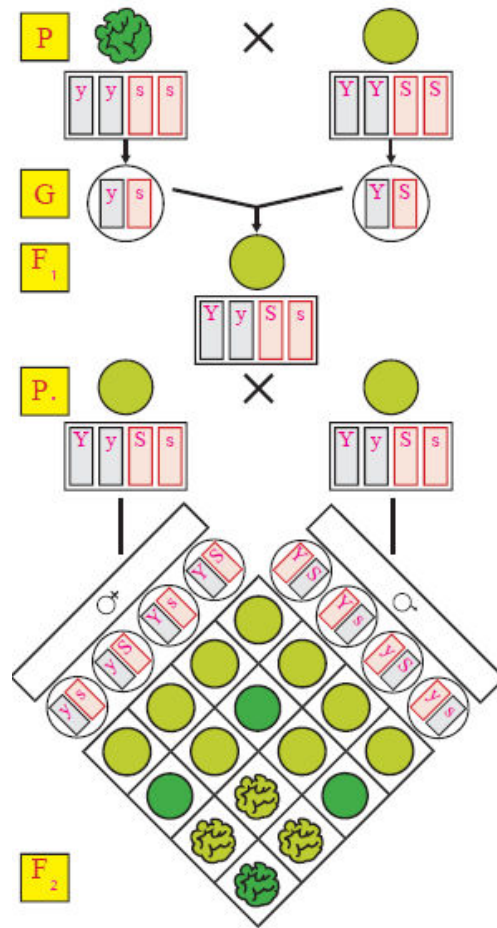


Fig. (4) Independent assortment of genes

We conclude from the previous figure that:-

- Yellow colour and smooth shape of pea are from dominant traits
- Green colour and sinuous shape of pea are from recessive traits
- Each gene of each hereditary trait is inherited independently of the others
- 100% of the Individuals of 1<sup>st</sup> generation are yellow-coloured and smooth-shaped
- Individuals of second generation have ratio of 1 : 3 : 3 : 9

### Linkage and crossing over

Scientist **Thomas Morgan** discovered in 1911 while studying the inheritance of traits in drosophila insect (fruit fly) that:-

- Genes of Drosophila exist on only four pairs of chromosomes.
- Each chromosome pair carries hundreds of genes
- Genes on the same chromosome are inherited as a single unit

→ This contradicts with Mendel's second law of the independent assortment of hereditary factors because Mendel's law states that hereditary factors (genes) are



inherited independently of the other factors, while Thomas Morgan supposed that genes on the same chromosome are inherited as a single unit

→ This means that Mendel's second law is not a general law.

→ Morgan supposed that the linkage of genes with each other is because they exist on the same chromosome. He also supposed that the linkage force between linked genes depends on the distances between them.

### Types of linkage between genes:-

It depends on the distances between genes, there are two types of linkage.

#### 1- Complete linkage

Linkage in which two genes - on the same chromosome - are so close to each other that they cannot get separated.

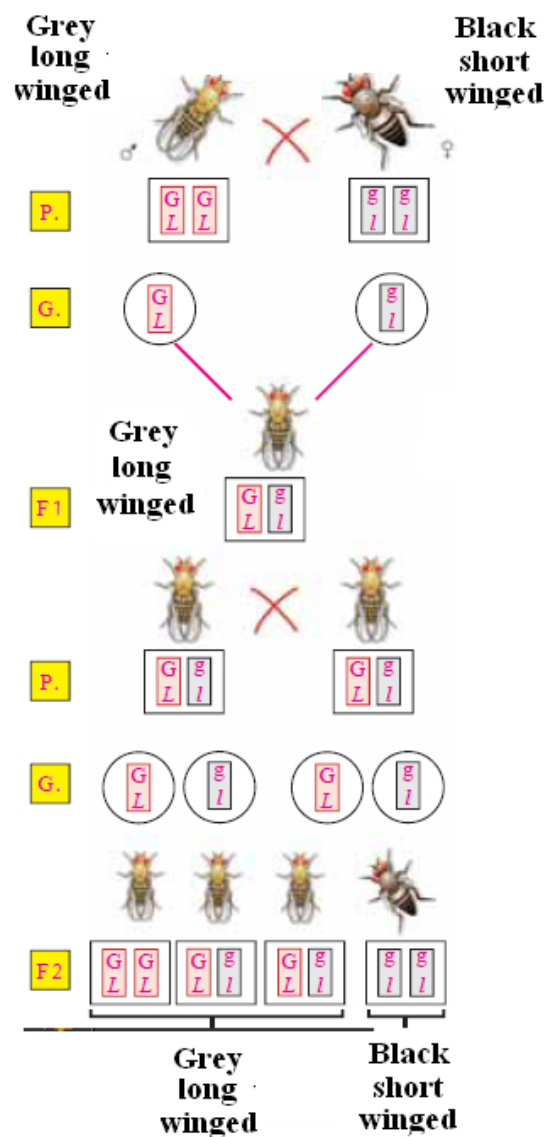


Fig. (5) Complete linkage in *Drosophila*

→ In the previous figure, we'll find that genes **G** and **L** are inherited as single unit and didn't separate (as Mendel's second law states)

→ Complete linkage causes the constancy of genes, and hence the constancy of the inheritance of hereditary traits

## 2- Incomplete linkage

→ In some cases, genes on the same chromosome don't stay linked together forever.

→ Genes on the same chromosome separate from each other and get exchanged with genes of another chromosome during meiotic cell division; which causes the appearance of new traits in the offspring. This phenomenon is called "crossing over"

### Crossing over:-

→ It occurs during Prophase I stage of meiosis

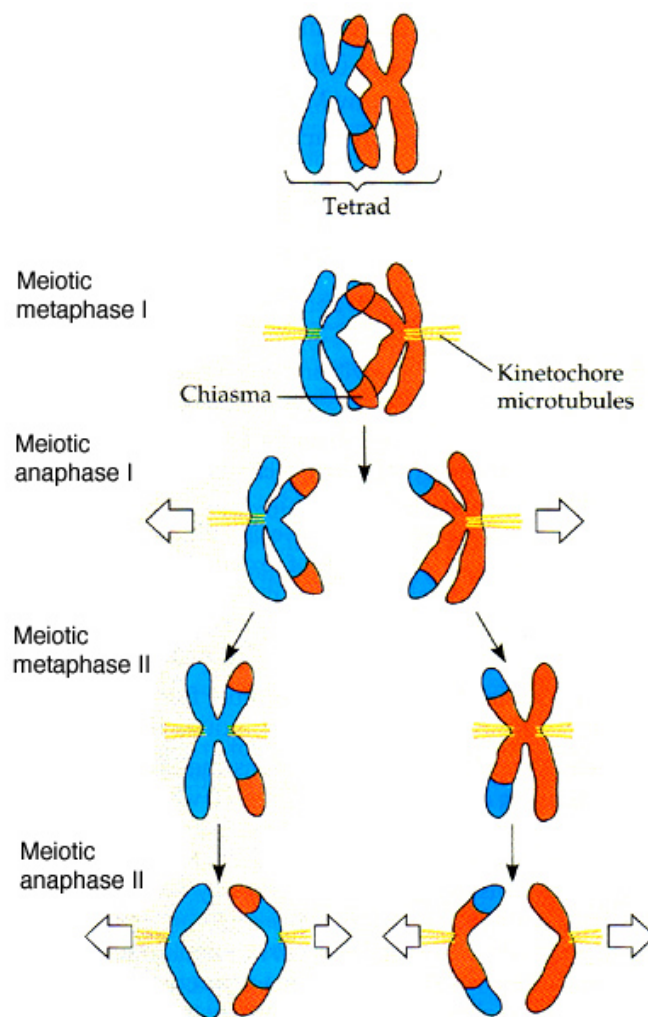


Fig. (6) Crossing over phenomena

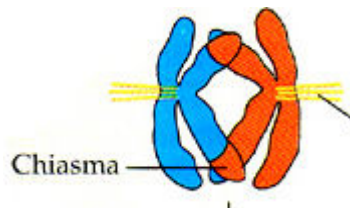
### **Prophase stage I**

→ Homologous chromosomes approach to each other forming **Tetrad**, which consists of 4 chromatids.



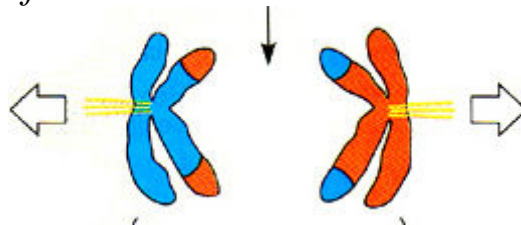
→ The internal chromatids of homologous chromosomes pairs coil around each other (cross-over) at one or more points called **Chiasma**, where the breakage takes place.

→ The internal parts of intertwined chromatids of homologous chromosomes pairs are exchanged, which is known as **Crossing over**



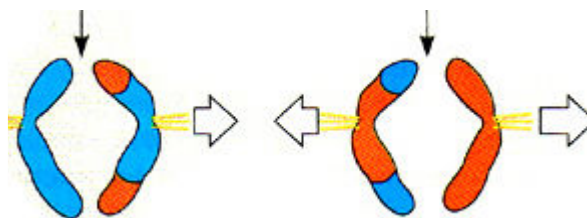
### **Anaphase I stage**

→ Pairs of homologous chromosomes separate from each other after crossing over forming two groups of chromosomes.



### **Anaphase II stage**

→ Chromatids gets separated from each other, they are called "building chromosomes". Then, they are distributed randomly among gametes.



→ Chromatids which underwent crossing over are called "**New chromosomes**". While chromatids which didn't undergo crossing over are called "**Parental chromosomes**". Thus, gametes contain new and parental chromosomes



→ Crossing over is an incomplete linkage of genes which causes the change of hereditary traits with certain ratios

→ Chances of crossing over occurrence increase by the increase of the distance between genes.

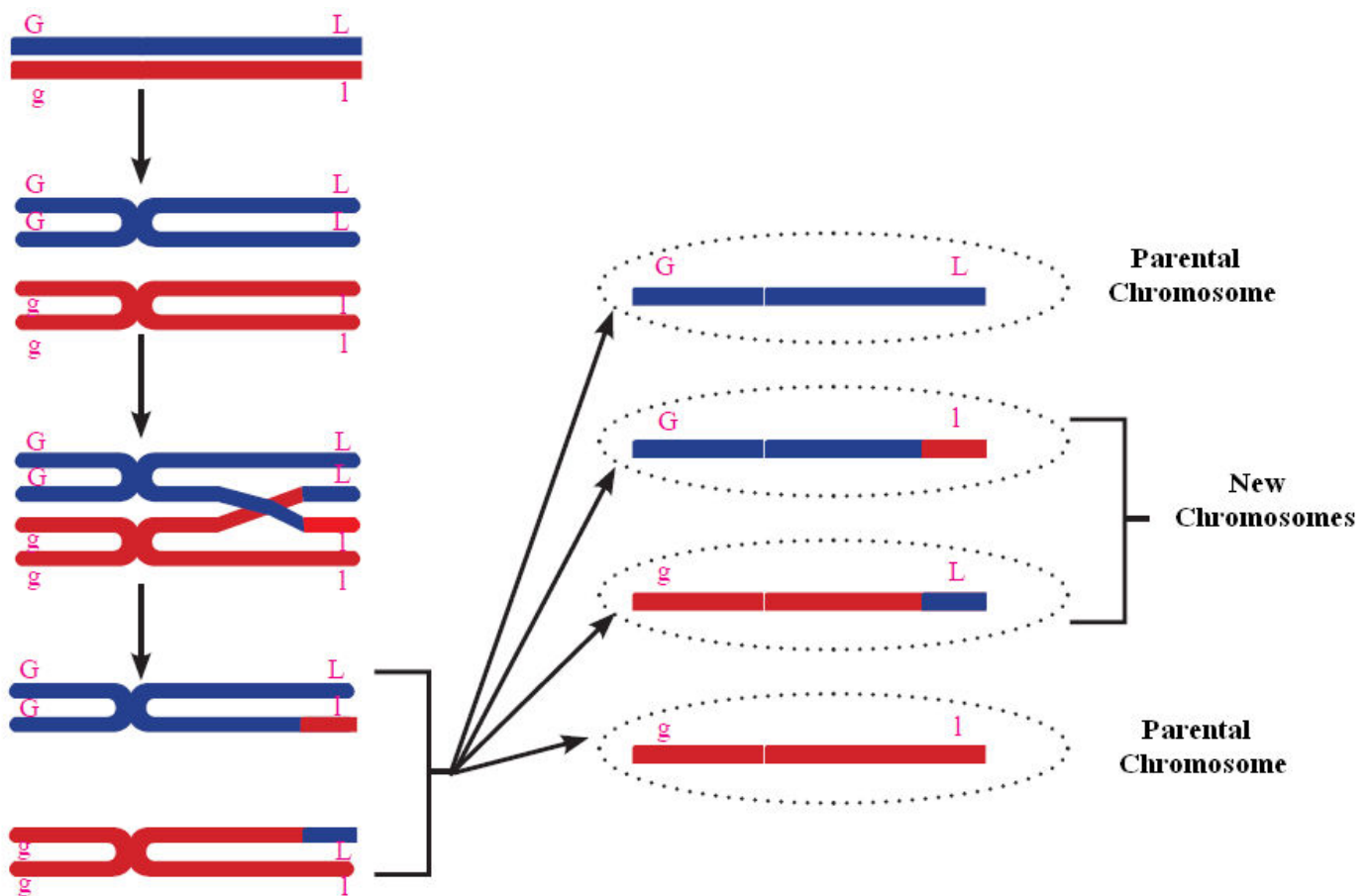


Fig. (7) Crossing over and chromosomes

→ In the previous figure, the two new chromosomes are (GL) and (gl), crossing over result in the formation of 4 chromatids, which are:-

(GL) → Parental chromosome

(Gl) → New chromosome

(gl) → Parental chromosome

(gL) → New chromosome

### Importance of crossing over:-

It causes the variation of hereditary traits of the members of same species, which helps them in the adaptation with environment conditions, and the development and continuation of their life

## Chromosomal maps

→ Scientists could identify the locations of genes on chromosomes by studying the ratios of crossing over phenomenon, which is known as **Chromosomal maps**.

## Definitions of lesson (1)

**Karyotype:** The arrangement of chromosomes in cells in descending order according to their size and number.

**Chiasma:** The points of connection of internal chromatids in homologous chromosomes pairs.

**Crossing over:** The exchange of parts of internal chromatids of homologous chromosomes pairs.

## Give reasons for

**1- Chromosomes pair (23) is called sex chromosomes**

Because this pair carries the genetic information responsible for the determination of sex (male or female).

**2- The assortment (distribution) of genes in gametes is independent according to Mendel's second law**

Because every gene exists on an independent chromosome

**3- Thomas Morgan's hypothesis on inheritance contradicts with Mendel's second law of the independent assortment of hereditary factors**

Because Mendel's law states that hereditary factors (genes) are inherited independently of the other factors, while Thomas Morgan supposed that genes on the same chromosome are inherited as a single unit.

**4- The importance of crossing over phenomenon**

Because it causes the variation of hereditary traits of the members of same species, which helps them in the adaptation with environment conditions, and the development and continuation of their life.

## Questions

**1- Choose the correct answer**

1- In human, nucleus of each somatic cell contains ..... pairs of chromosomes

A- 23    B- 46    C- 37    D- 15

2- In human, nucleus of each somatic cell contains ..... of chromosomes

A- 46    B- 37    C- 23    D- 18

3- Chromosomes pair number ..... is called sex chromosomes.

A- 15    B- 17    C- 9    D- 23

4- Sex chromosomes pair in male is .....

A- XX    B- XY    C- YY    D- XXY

5- Sex chromosomes pair in female is .....

A- XX    B- XY    C- YY    D- XXY

6- Scientist/s ..... discovered that genes on the same chromosomes are not inherited independently, but as single unit.

A- Gregory Mendel    B- Boveri and Sutton    C- Thomas Morgan    D- Darwin

7- Scientist/s ..... formulated the chromosomal theory.

A- Boveri and Sutton    B- Thomas Morgan    C- Mendel    D- Leeuwenhoek

8- Homologous chromosomes approach to each other forming tetrad in stage ..... of meiosis

A- Prophase I    B- Prophase II    C- Anaphase I    D- Anaphase II

9- The points of connection of internal chromatids in homologous chromosomes pairs is called .....

A- Chiasma    B- Chromosome    C- Centromere    D- Centrosome

10- In gametes, chromosomes which didn't undergo crossing over are called ....

A- New chromosomes    B- Building chromosomes    C- Parental chromosomes  
D- Chromatids

## **2- Write the scientific term**

1- The arrangement of chromosomes in cells in descending order according to their size and number.

2- The points of connection of internal chromatids in homologous chromosomes pairs.

3- The exchange of parts of internal chromatids of homologous chromosomes pairs with their genes.

4- Chromosomes which didn't undergo cross over in gamete cells during meiosis.

5- Chromosomes which didn't undergo cross over in gamete cells during meiosis.

## **3- Write one work of each one of the following scientists**

1- Boveri and Sutton

2- Thomas Morgan

## **4- Write short notes about**

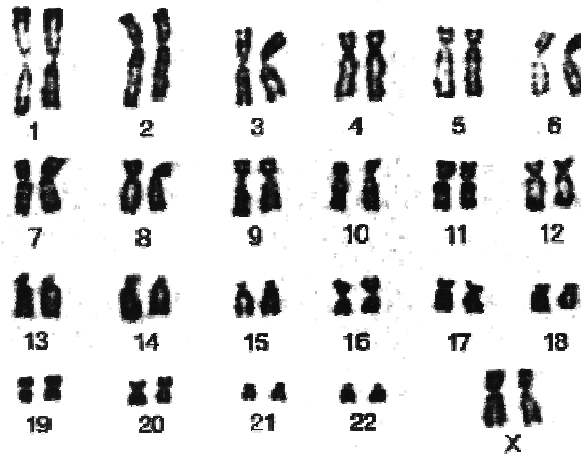
1- Chromosomal theory

2- Chromosomal maps

3- Complete linkage

4- Karyotype

5- The following figure describe Karyotype of a cell, answer the questions

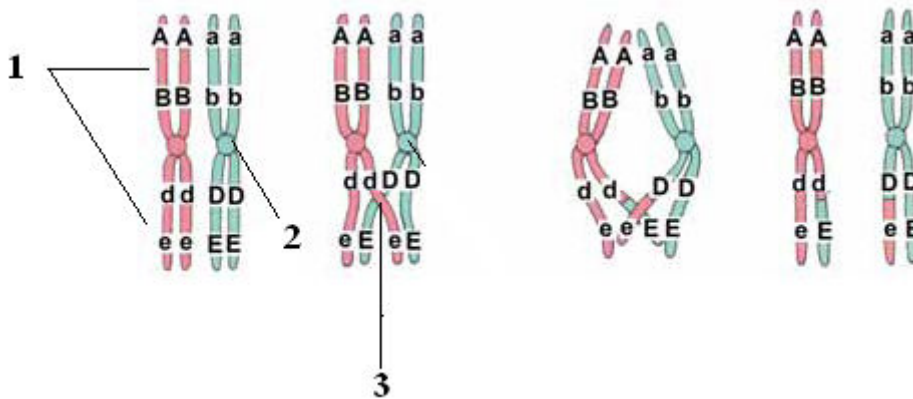


1- What does this Karyotype describe, a somatic cell or gamete? Why?

2- What is the sex of the person carrying this Karyotype ? Why?

3- What is the number of somatic and sex chromosomes?

6- The following figure describe a very important phenomenon



1- What is the name of this phenomenon, when does it happen?

2- Label the numbers (1), (2), (3)

3- Mention the genotypes of gametes resulted from this phenomenon.

## The Answers

1- Choose the correct answer

1- 23

6- Thomas Morgan

2- 46

7- Boveri and Sutton

- |       |                         |
|-------|-------------------------|
| 3- 23 | 8- Prophase I           |
| 4- XY | 9- Chiasma              |
| 5- XX | 10- Parental chromosome |

**2- Write the scientific term**

- |                  |                         |
|------------------|-------------------------|
| 1- Karyotype     | 4- Parental chromosomes |
| 2- Chiasma       | 5- New chromosomes      |
| 3- Crossing over |                         |

**3- Write one work of each one of the following scientists**

- 1- **Boveri and Sutton:** They put chromosomal theory in 1902
- 2- **Thomas Morgan:** He noticed while studying *Drosophila* in 1911 that genes on the same chromosome are inherited as one unit not independently

**4- Write short notes about**

- 1- See page (3)
- 2- See page (9)
- 3- See page (6)
- 4- See page (2)

**5- The following figure describe Karyotype of a cell, answer the questions**

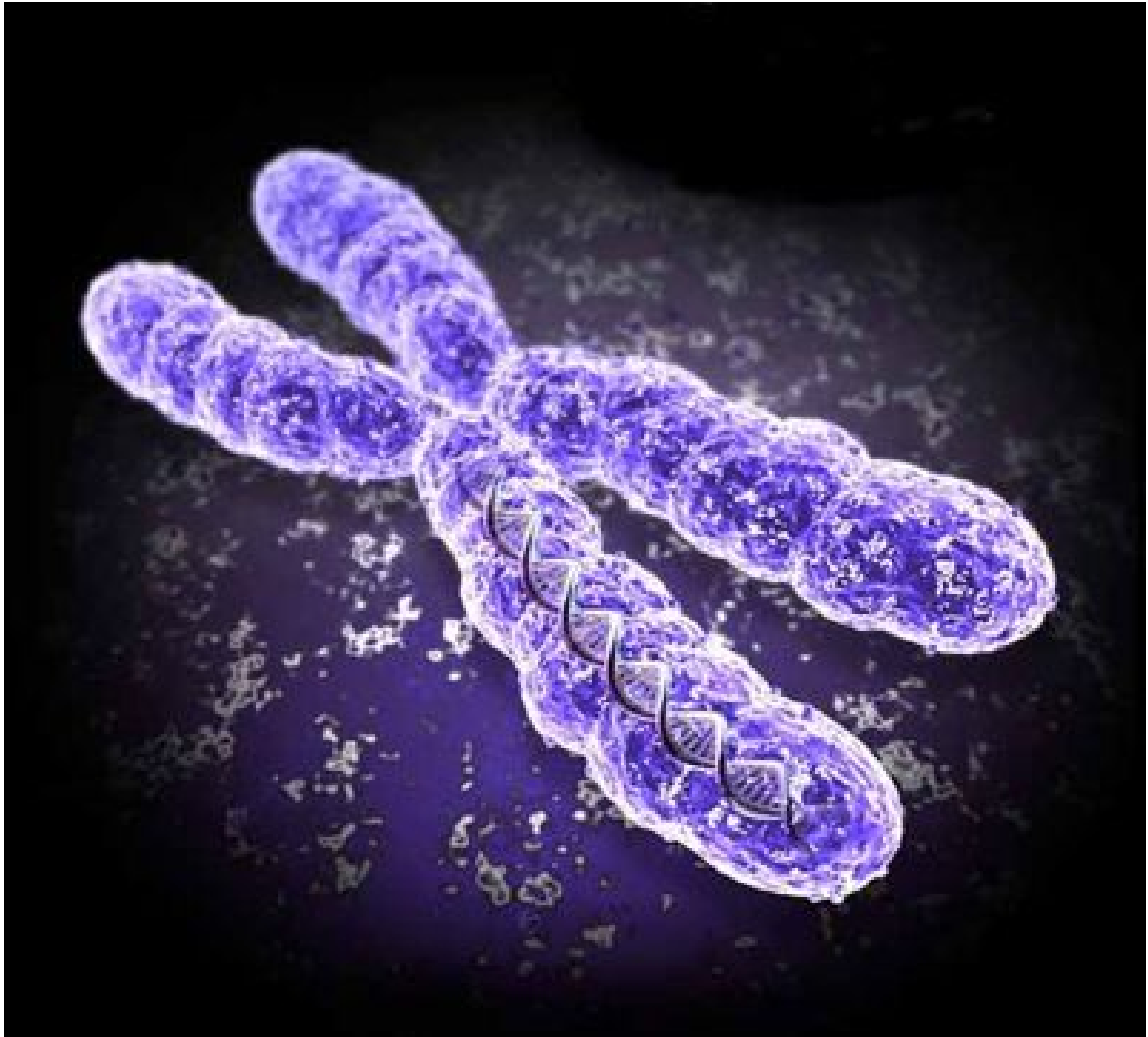
- 1- Somatic cell, because it contains 23 pairs of chromosomes (diploid cell  $2n$ )
- 2- Female, its sex chromosome is homologous (XX)
- 3- Somatic chromosomes: 44 (22 pairs) Sex chromosomes: 2 (1 pair)

**6- The following figure describe a very important phenomenon**

- 1- Crossing over, prophase I of meiosis
- 2- (1) Chromatid (2) Centromere (3) Chiasmus
- 3- (ABde) (ABdE) (abDe) (abDE)



*Lesson (2)*  
*The interaction of genes*



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

---

**Remember**

**Dominant trait:** Trait which appears in all members of 1<sup>st</sup> generation

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

---

→ 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.

→ **Examples of genes interaction:** Lack of dominance – Complementary genes – Lethal genes.

---

**Remember**

→ Each pair of contrasting traits is called "Allelomorphic traits"

→ According to Mendel, when two pure individuals (one of them carries a dominant trait, while the other carries recessive trait) copulate, Only the dominant trait appears in the individuals of 1<sup>st</sup> generation, while it appears with recessive trait in the individuals of 2<sup>nd</sup> generation at ratio 3 dominant : 1 recessive

---

**First: Lack of dominance**

→ Flowers of **Four o'clock plants** have three colours: **Red** – White – **Pink**



Fig. (1) Four o'clock plant

→ When a plant with white flowers (WW) copulates with another one of red flowers (RR), they produce 100% plants with pink flowers (RW) in the first generation.

→ In 2<sup>nd</sup> generation, red, pink and white flowers appear at ratio 1:2:1

We conclude from the following figure:-

→ The colour of flower trait (Red or white) is controlled by two genes which do not dominate over each other.

→ Those two gene interact together, and every opposite gene participates in the formation of a new trait (RW).

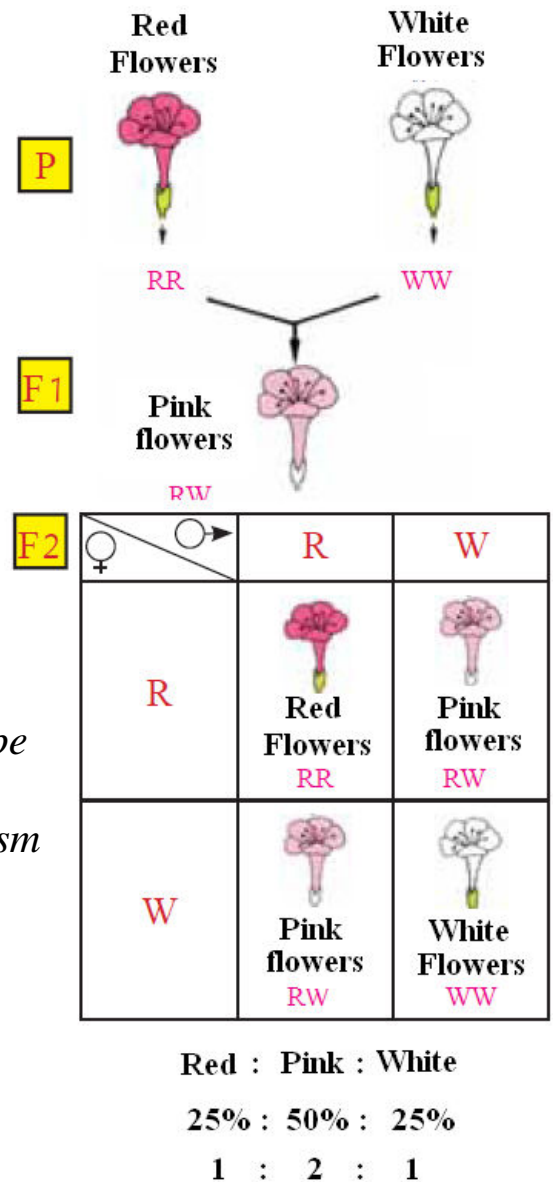
→ These results contradict with Mendel's laws because no hereditary factor (gene) dominates over the other one, which forms plants of ratio 1:2:1 instead of 1:3 in the 2<sup>nd</sup> generation. But the two genes interact forming new gene, which is known as **lack of dominance**

→ **Note:** in lack of dominance case, the phenotype describe genotype

Genotype: The structure of gene in living organism

Phenotype: The hereditary trait appearing on Living organism.

The gene responsible for eye colour is genotype, While the colour of eye trait is phenotype



### Inheritance of blood groups in human

Although the components of blood are constant in all humans, they have different blood groups

→ Blood transfusion process depends on **blood group** and **kind of Rhesus factor**

→ **Blood groups trait** is controlled by three kinds of genes called **Alleles**, these alleles are denoted by (A – B – O). Each person inherits only one pair of Alleles.

→ This pair of alleles exist on **chromosome pair (9)** in all humans.

---

**Allele (For illustration only) :** Alternative forms of the same gene which produce different effects.

---

### Genetic classification of blood groups

Blood groups share three inheritance types, which are:-

#### **- Alleles multiplicity:-**

→ Blood groups trait is controlled by 3 alleles (A – B – O)

→ Human inherits only one pair of them

<b>Blood group</b>	<b>Genotype (genetic structure)</b>
A	AA or AO
B	BB or BO
AB	AB
O	OO

**- Complete dominance:** Both of genes (A) and (B) dominate over gene (O)

**- Lack of dominance:** There is no dominance between genes (A) and (B), but they participate in the formation of a new group (AB)

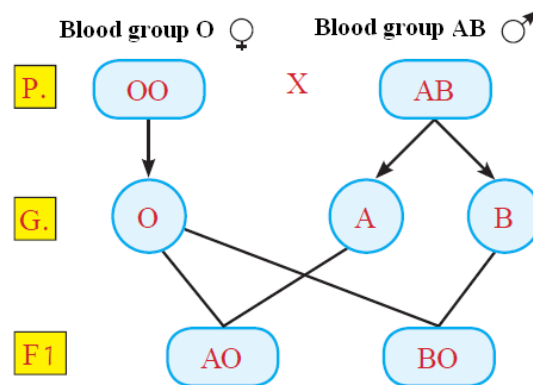


Fig. (2) Blood group inheritance

→ From the previous figure, we conclude that 50% of children will have blood group (A), and the other 50% will have blood group (B)

### **Chemical classification of blood groups:-**

Blood groups (A – B – AB – O) classification depends on the kind of two substances in blood, which are:-

#### **Antigens:-**

They are chemical substances which exist on the surface of red blood cells (RBC), they have two kinds:-

1- Antigens a

2- Antigens b

#### **Antibodies:-**

They are antibodies of antigens which exist in blood plasma, they have two kinds:-

1- anti-a

2- anti-b

Blood Group	Antigens	Anti Bodies
A	a	anti-b
B	b	anti-a
AB	a - b	-----
O	-----	anti-a anti-b

## ***The importance of blood groups***

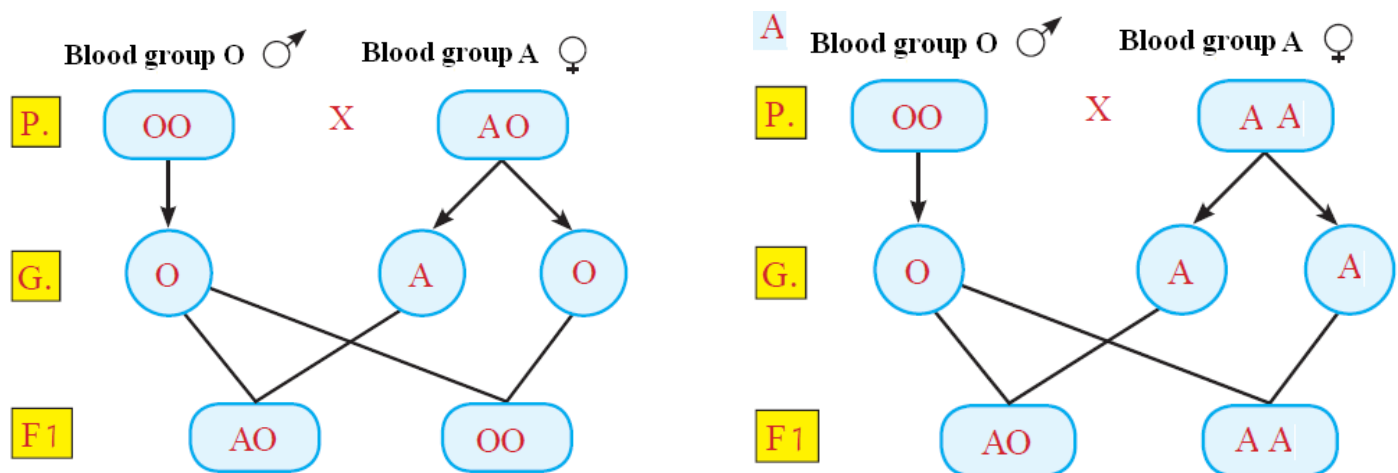
- 1- Solving problems of the determination of paternity (parents of children) and pedigree of children (blood groups deny pedigree but don't prove it)*
- 2- Determination of blood transfusion processes between individuals.*
- 3- Used in the study of human races classification and evolution*

### ***Example***

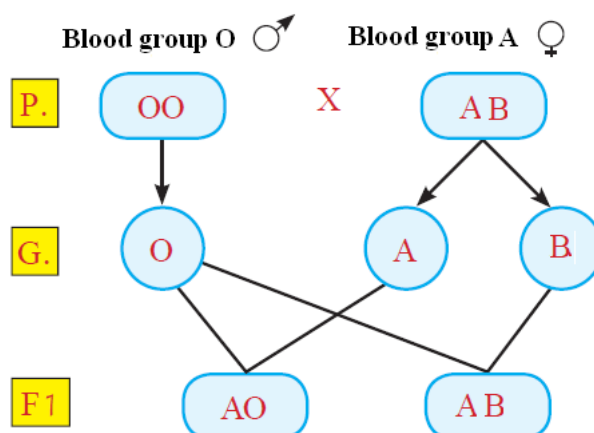
*Two fathers argued about the pedigree of a child of blood type (O), if the blood types of both fathers is (O), the blood type of first man's wife is (A), and the second's wife (AB). Which one of those men is more likely to be the father of the child? Why?*

### ***Solution***

*→ Genetic analysis of the first man and his wife (N.B: genotype of blood type (A) is (AO) or (AA) – Genotype of blood type (O) is (OO))*



*→ Genetic analysis of the second man and his wife (N.B: genotype of blood type (AB) is (AB) – genotype of blood type (O) is (OO))*



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



## **Blood transfusion process**

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

→ The following table describes blood groups and the probability of transfusing blood between them.

Blood Group	Genotype		Antigens	Antibodies		Gives blood to	Receives blood from
A	AO	AA	a	anti-b		A , AB	A , O
B	BO	BB	b	anti-a		B , AB	B , O
AB	AB		a-b	-----		AB	All blood groups
O	OO		-----	anti-a	anti-b	All blood groups	O

From the previous table, we conclude that:-

→ Blood group (AB) is called **Universal Recipient** because it can receive blood from all groups, as it doesn't have any antibodies.

→ Blood group (O) is called **Universal Donor** because it can give blood to all groups, as it doesn't have any antigens.

## **Determination of blood types**

→ We said that antigens have antibodies (Ex. Antigen a has antibody a)

→ We can determine the type of blood by the reactions which occur between antigens and antibodies and the occurrence of blood coagulation (clotting)

### **Steps of determining blood type**

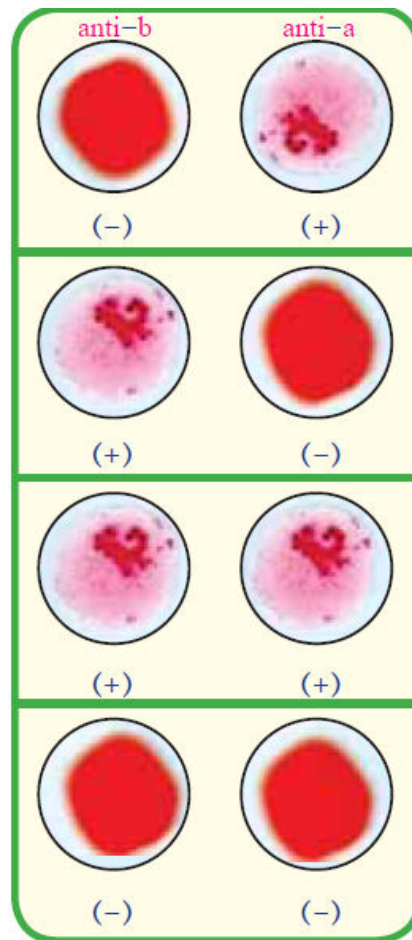
1- Sample of blood is taken from the person.

2- We put two drops of blood on two clean glass sheets

3- We add (anti-a) antibodies to the first blood drop and (anti-b) antibodies to the second one.

4- We compare the result to the following table

<b>First blood drop (anti-a)</b>	<b>Second blood drop (anti-b)</b>	<b>Blood type</b>
- Coagulation (+)	- No coagulation ( - )	A
- No coagulation ( - )	- Coagulation (+)	B
- Coagulation (+)	- Coagulation (+)	AB
- No coagulation ( - )	- No coagulation ( - )	O



*Fig. (3) Blood coagulation and blood group*

### ***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*

## ***Rhesus factor (Rh)***

→ Blood carries two kinds of antigens, which are:-

- Antigens of blood group (a – b)
- Rhesus factor antigens

→ 85% of humans have rhesus factor antigens in their blood, they are denoted by (Rh+) – positive rhesus factor

→ 15% of humans don't have rhesus factor antigens in their blood, they are denoted by (Rh-) – negative rhesus factor

→ The inheritance of rhesus factors antigens is controlled by three pairs of genes carried on one chromosome pair.

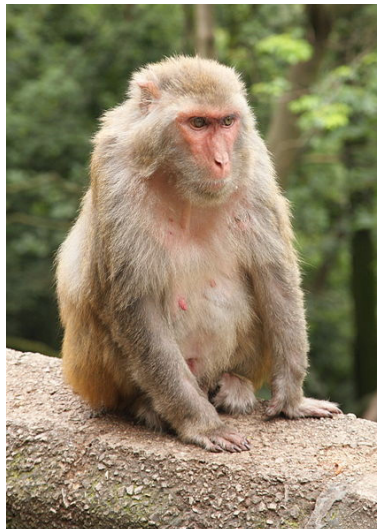
→ A person becomes positive Rh factor (Rh+) if it has one or more dominant genes in each pair, while it becomes negative Rh factor (Rh-) if all its genes are recessive

---

### **For illustration only**

→ Rhesus factor was discovered in 1940 in "Rhesus monkeys" species

→ Rhesus monkeys are native to southeast, central and south Asia



---

### ***The importance of rhesus factor:-***

We should determine rhesus factor in blood before blood transfusion process and marriage; in order to prevent the formation of antibodies of rhesus factor antigens, which breaks red blood cells.

### ***Rhesus factor role in pregnancy***

→ If a (Rh+) man married a (Rh-) woman, and the fetus inside uterus of woman is (Rh+) like his father, a part of the fetus blood mix with that of mother.

→ Immune system of pregnant mother produces antibodies of Rh factors antigens.

→ If mother becomes pregnant with another baby, antibodies (which were produced from pregnancy of the first fetus) transfer from mother to her second fetus through placenta, which breaks its red blood cells causing acute anemia to him, which may lead to its death.

**Protective procedure:-**

If we discovered the difference of Rh factor in mother right before delivery, we give her vaccine within 72 hours after every delivery in order to protect the next fetus. This vaccine breaks the amount of blood mother had taken from her fetus (which contains Rh+) before her immune system forms antibodies of Rh antigens.

## Complementary genes

---

**Complementary genes:** Genes which interact with each other causing the appearance of a hereditary trait

---

→ The inheritance of complementary trait is controlled by two pairs of genes.

→ Dominant trait appears if every pair of genes has a least one dominant gene. Otherwise, opposite recessive trait will appear.

**Flowers colours of sweet pea plant**

The inheritance of flower colour trait in sweet pea plant is an example of complementary genes.

→ Pink colour of flower is the dominant trait, while white colour of flower is the recessive trait

→ Colour trait is controlled by two pairs of genes, dominant genes are denoted by (A) and (B), while recessive genes are denoted by (a) and (b)



Fig. (4) Pink flower of sweet pea plant (Dominant trait)

→ When two white flowers ( $AA\ bb$ ) copulate, 100% pink flowers are produced in the first generation, while both pink and white flowers appear in the second generation at ratio 9:7

→ The appearance of pink colour (dominant trait) in sweet pea flowers depends on the aggregation of one or more dominant genes in each pair because both dominant genes participate in the formation of pink colour, as each gene controls the production of certain enzyme influencing the formation of pink colour.

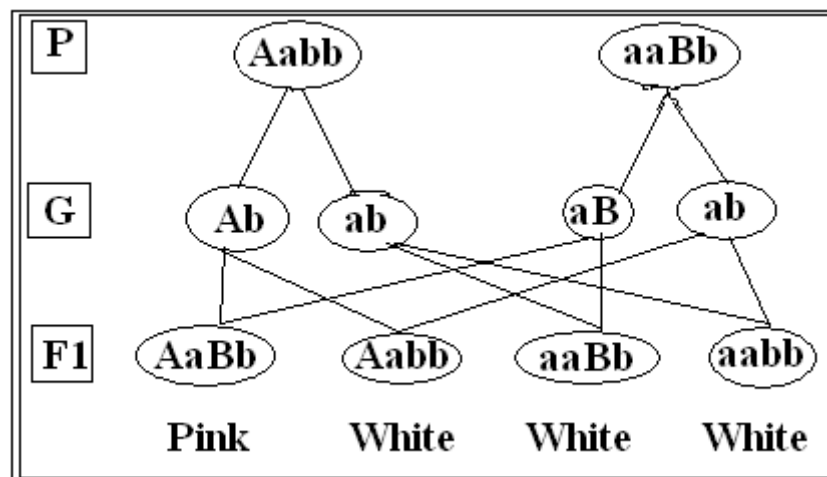
→ This example proves the complementation of genes, as we can get the dominant trait from two fathers each one of them carries the recessive trait.

→ The ratio of 2<sup>nd</sup> generation according to Mendel's second law is 1:3:3:9, while that according to complementary genes (non-Mendilian traits) is 7:9

### Example (1)

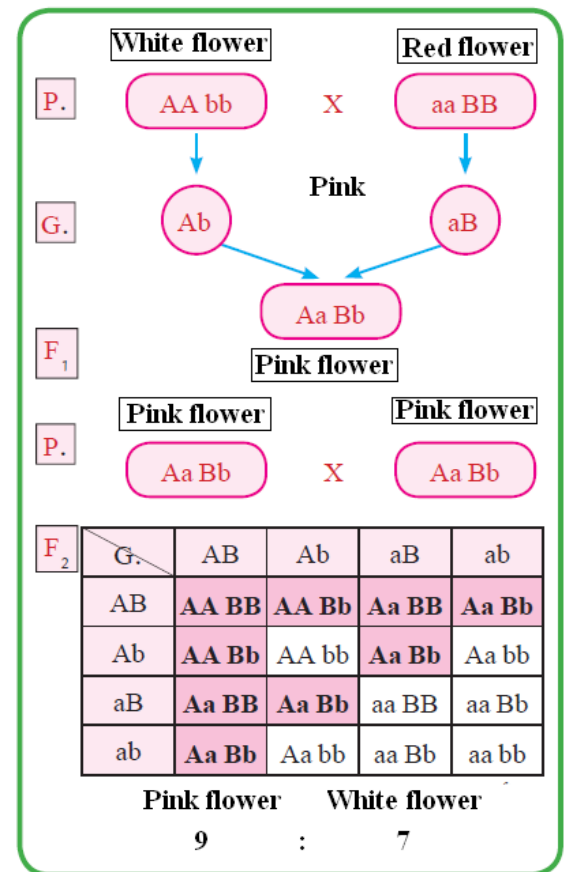
Determine the phenotypes and genotypes of the colour of flowers produced from the copulation ( $Aabb \times aaBb$ )

### Solution



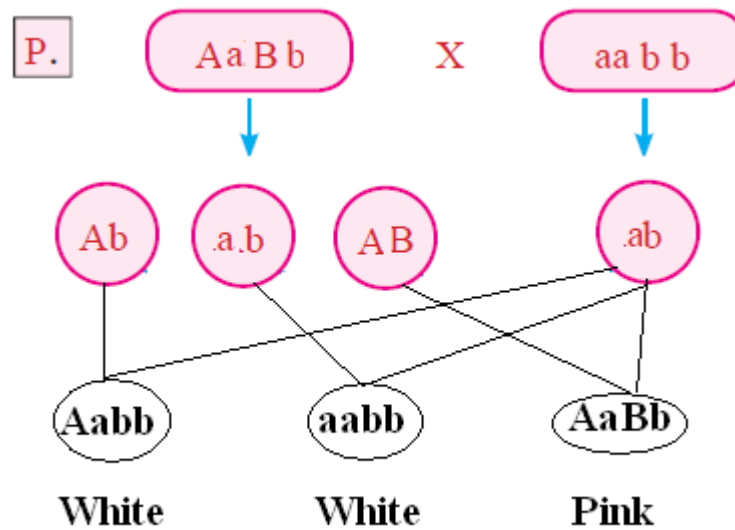
### Example (2)

Determine the phenotypes and genotypes of the colour of flowers produced from the copulation ( $AaBb \times aabb$ )





## Solution



## Lethal genes

**Lethal genes:** They are genes which cause the death of the living organisms if they are present in homozygous (pure) state.

→ Lethal genes stop biological processes inside living organisms at different ages.

### Kinds of lethal genes

**1- Dominant lethal genes:** (Ex. Yellow colour gene in mice – Bulldog race in cows)

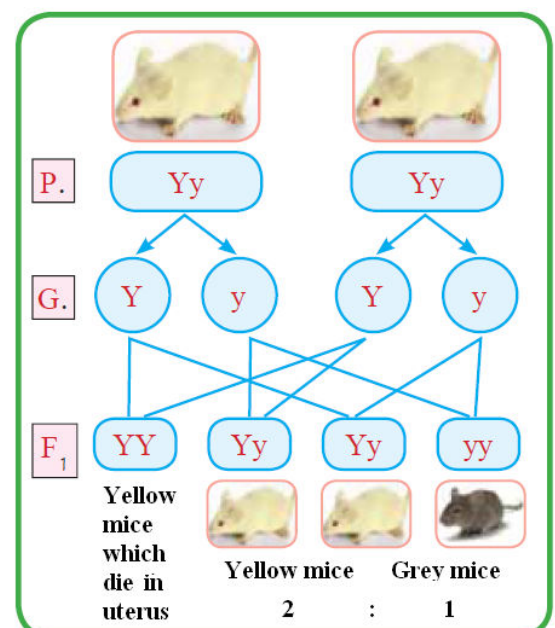
**2- Recessive lethal genes:** (Ex. The absence of chlorophyll in corn – infantile dementia in human)

### Inheritance of colour trait in mice

→ When two hybrid yellow mice copulate, the ratio of product generation is 1:2.

→ Pure yellow mice die because they carry pure lethal dominant pair of gene (YY) which causes the death of mice in uterus.

→ These results contradicts with Mendel's first law of the segregation of factors because when two hybrid yellow mice copulate, they produce yellow and grey mice at ratio 3:1 according to Mendel's first law, while they produced yellow and grey mice at ratio 2:1



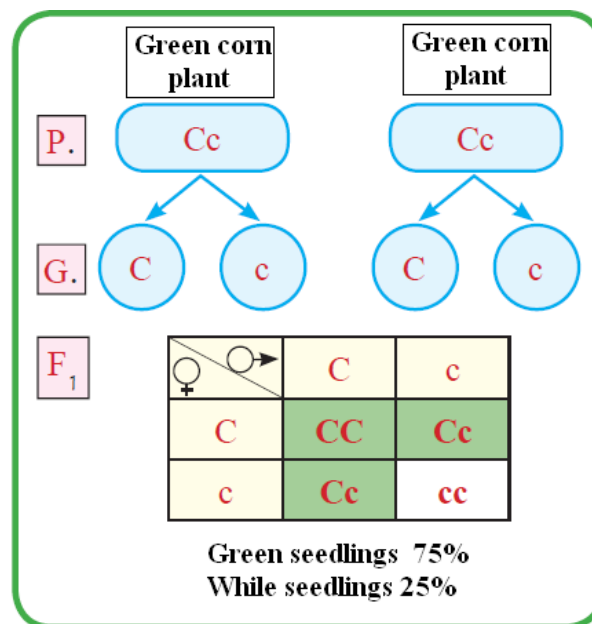
→ The ratio of dead yellow mice is 25% of the product generation

### ***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 wither and die. This happens because of a pure recessive lethal gene (cc)

→ 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.



### ***The effect of environmental conditions on some genes work***

→ The work of some genes is affected by the factors surrounding living organisms (**Ex.** Air pollutants – lack of oxygen – radiation) and environmental factors (**Ex.** Light – temperature).

→ The study of factors affecting genes help us avoid the risks resulted from them.

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

→ The gene responsible for chlorophyll formation in green plants needs light factor, so that the effect of gene appears.

→ Green plant cannot form chlorophyll if the gene responsible for it is absent, even if it was put in light

## Definitions of lesson (1)

**Lack of dominance:** A form of inheritance in which no genes dominate over the other one, but they interact forming new trait

**Antigens:** They are chemical substances which exist on the surfaces of red blood cells, they play an important role in blood transfusion process.

**Antibody:** They are antibodies of antigens which exist in blood plasma, they play an importance role in blood transfusion process

**Rhesus factor:** A kind of antigens which exists on the surfaces of red blood cells of most humans, its inheritance is controlled by three pairs of genes which are carried on one chromosome pair.

**Complementary genes:** Genes which interact with each other causing the appearance of a hereditary trait.

**Lethal genes:** Genes which obstruct growth and cause death at different ages when they exist in pure (homozygous) form

## Give reasons for

**1- When two individuals different in one pair of hereditary traits copulate, the second generation ratio is 1:2:1 not 1:3**

Because the genes of those different traits do not dominate over each other. So, they interact with each other forming new trait, which appears in 2<sup>nd</sup> generation with the two opposite traits at ratio 1:2:1 (not 1:3 – as Mendel laws state – because of the lack of dominance)

**2- The importance of blood groups**

Because they :-

- Solve problems of the determination of paternity (parents of children) and pedigree of children (blood groups denies pedigree but don't prove it)
- Determine blood transfusion processes between individuals.
- Are used in the study of human races classification and evolution

**3- Blood group (O) is a universal donor, while blood group (AB) is a universal recipient**

Blood group (O) is a universal donor because it has both antigen-a and antigen-b and doesn't have any antibodies, which makes it capable of giving blood to all groups. While blood group (AB) is a universal recipient because it has both anti-a and anti-b and doesn't contain any antigens, which makes it capable of receiving blood from all types.

**4- Giving blood of inconvenient group to a recipient person is very dangerous.**  
*Because giving blood to a person of a blood group unsuitable for his blood causes the break of red blood cells, which cause sickness, chest pain, irregular heartbeat, blueness, headache, and even death!!*

**5- Blood should be examined before transfusion process**

*To make sure it is convenient for the blood of recipient person and doesn't contain any disease-causing organisms (Ex. Viruses)*

**6-The importance of determination of rhesus factor in blood**

*Because it is very important to determine it before blood transfusion process and marriage; in order to prevent the formation of antibodies of Rh factor antigens, which breaks red blood cells.*

**7- Woman who was pregnant with a baby of different rhesus factor should take vaccine within 72 hours after every delivery**

*To break up the amount of blood mother had taken from her first fetus – which contains Rh+ - before her immune system forms Rh antibodies. Which protects her second fetus.*

**8- Inner cabbage leaves are not green-coloured**

*Because the gene responsible for chlorophyll formation in green plants (which give them its green colour) needs light; so that its effect will appear. But we find that inner cabbage leaves are not exposed to light.*

### **What happens when**

**1- Transfusing blood from a man of group (AB) to another one of group (A)**

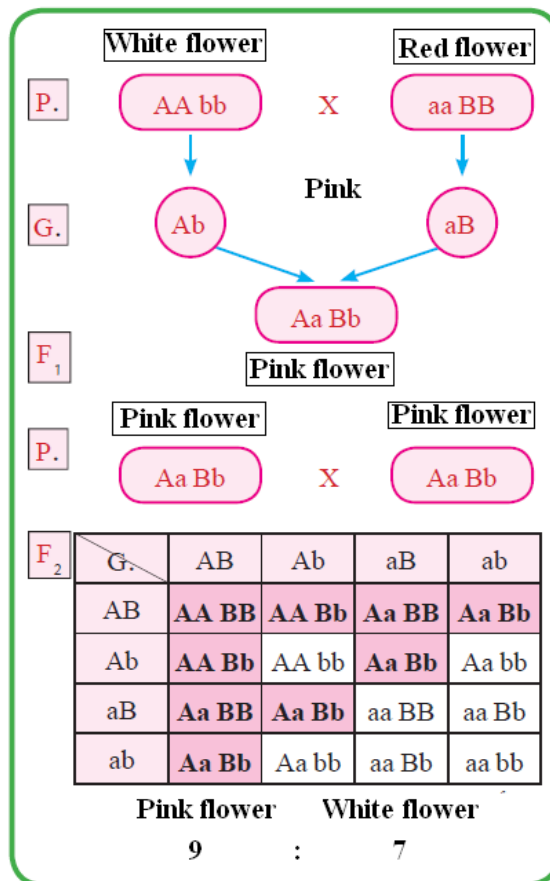
*This will break red blood cells of the recipient person because his blood produces anti-b for antigens-B of blood group (AB), which causes shivering in body, chest pain, blueness, irregular heartbeat, headache, low blood pressure*

**2- (Rh-) woman married (Rh+) man (with respect to the first and second babies)**

*When the woman becomes pregnant with the first baby (which is Rh+), a part of his blood transfers from him to his mother, which stimulates her immune system to produce antibodies of Rh factor antigens. If mother wasn't given vaccine after delivery of the first baby, and became pregnant again with another baby, Rh+ blood transfers from mother to her second baby through placenta, which breaks up his red blood cells and causes him acute anemia and even death.*

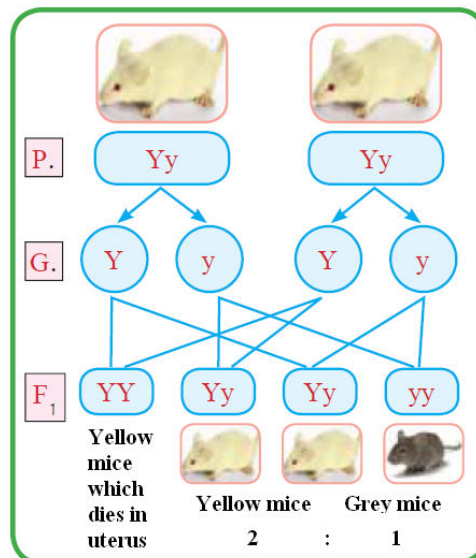
**3- Two sweet pea plants with white flowers whose genotypes are (aaBB) and (AAbb) copulate (first and second generations)**

100% pink flowers are produced in the first generation, while both pink and white flower appear in the second generation at ratio 9:7



#### 4- Breeding two yellow mice (Yy)

Hybrid yellow and black mice are produced at ratio 2:1 respectively, pure yellow mice (YY) - which represent 25% of the generation – die in uterus before being born.



#### 5- Planting corn plant seedlings in a dark place

Seedlings lose their green colour due to the lack of chlorophyll, as the gene responsible for chlorophyll formation is activated only by light (which is absent)



# Questions

## **1- Choose the correct answer**

1- in lack of dominance case, the ratio of 2<sup>nd</sup> generation resulted from the copulation of two individuals different in one pair of opposite traits is .....

A- 1:3    B- 1:2:1    C- 7:9    D- 1:2

2- When a man of blood group (AB) marries a woman of blood group (O), the ratio of children which have blood group (O) is .....

A- 0%    B- 50%    C- 25%    D- 75%

3- Blood group which has both types of antigens is.....

A- A    B- O    C- AB    D- B

4- Blood group which has both types of antibodies is.....

A- A    B- O    C- AB    D- B

5- Blood group which is known as "Universal recipient" is

A- A    B- O    C- AB    D- B

6- Blood group which is know as "Universal donor" is.....

A- A    B- O    C- AB    D- B

7- Trait of flower colour of sweet pea plant is an example on .....

A- Lack of dominance    B- Alleles multiplicity    C- Complementary genes  
D- Lethal genes

8- The inheritance yellow mice colour is an example on .....

A- Lack of dominance    B- Alleles multiplicity    C- Complementary genes  
D- Lethal genes

9- ..... gene is an example on recessive lethal genes

A- Yellow colour of mice    B- Infantile dementia    C- Turner's syndrome  
D- Bulldog race in cow

10- ..... is an example on dominant lethal genes

A- Yellow colour of mice    B- Infantile dementia    C- Turner's syndrome  
D- Bulldog race in cow

## **2- Write the scientific term**

1- A form of inheritance in which no genes dominate over the opposite one, but they interact forming new trait

2- chemical substances which exist on the surfaces of red blood cells, they play an important role in blood transfusion process.

3- A kind of antigens whose inheritance is controlled by three pairs of genes which are carried on one chromosome pair.

4- Genes which interact with each other causing the appearance of a hereditary trait.

5- Genes which obstruct growth and cause death at different ages when they exist in pure (homozygous) form

**3- Write short notes about:-**

1- Dangers of blood transfusion

2- Rhesus factor

3- Lethal genes

4- Complementary genes

**4- Compare between**

1- Blood types (A) and (B)

2- Lethal and complementary genes



**5- Rationalize the following cases on a genetic basis**

1- A man of blood group (A) married a woman of blood group (B) and bore a child of blood group (O)

2- A woman whose blood group is (AB) has a son of the same blood group, what are the probable genotypes of the father?

3- Breeding four o'clock plant with red flowers with another one of pink flowers.

**6- The following table illustrates the generation resulted from the breeding of two sweet pea plants, then answer the following questions**

 \ 	AB	--	aB	ab
--	(1)	AABb	(2)	AaBb
--	(3)	AAbb	(4)	Aabb

1- What are the genotypes of (1), (2), (3), (4)


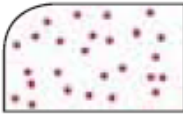
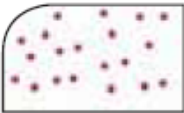





2- Find the genotypes of the parents

3- What is the percentage of white flowers in this generation?

4- What is the colour of flowers produced from the breeding of plant (4) with (3)

**7- If your blood group is (A) and you need blood transfusion, which blood groups are suitable for you? Why?**

### 8- Answer the following question

Group	anti-a	anti-b
.....		
.....		
.....		
.....		

- 1- Complete the previous table mentioning blood groups
- 2- Which blood group has both types of antigens?
- 3- Which blood group has both types of antibodies?

## Answers

### 1- Choose the correct answer

- 1- 1:2:1
- 2- 0%
- 3- AB
- 4- O
- 5- AB
- 6- O
- 7- Complementary genes
- 8- Lethal genes
- 9- Infantile dementia
- 10- Yellow colour of mice

### 2- Write the scientific term

- 1- Lack of dominance
- 2- Antigens
- 3- Rhesus factor
- 4- Complementary genes
- 5- Lethal genes

### 3- Write short notes about:-

1- Giving blood to a person of an inconvenient group causes headache, shortness of breath, chest pain, irregular heartbeat, blueness, shivering in body and usually ends with death. Transfusing polluted blood to a person may cause viral infection (Ex. AIDS – Hepatitis B)

2- Rhesus factor is a kind of antigens which exist on the surfaces of red blood cells of 85% of humans, its inheritance is controlled by 3 pairs of genes which exist on one chromosome pair.

3- Lethal genes are genes which obstruct growth and cause death to living organisms if they are present in pure (homozygous) form. There are recessive lethal genes (such as the genes causing the absence of chlorophyll in corn plants and infantile dementia in humans) and dominant lethal genes (such as the genes of yellow colour of mice and bulldog race in cows)

4- Complementary genes are genes which interact together forming a certain trait. The appearance of this trait is controlled by two pair of genes, there must be at least one dominant gene in each pair so that the dominant trait appears. Otherwise, recessive trait will appear. The flower colour of sweet pea plant is an example of complementary genes

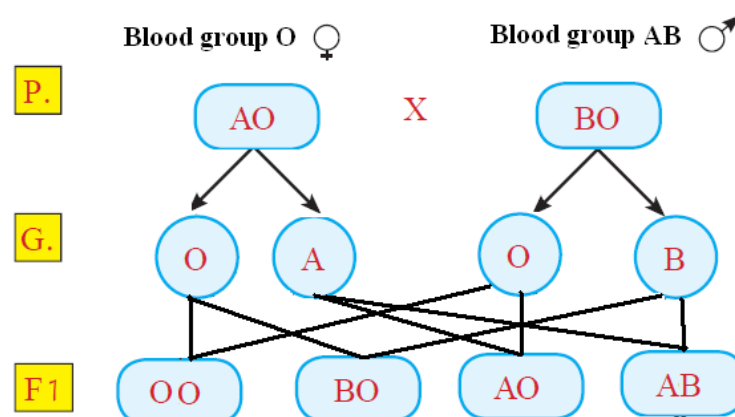
#### 4- Compare between

<b>P.O.C</b>	<b>Blood type (A)</b>	<b>Blood type (B)</b>
<b>Genotype</b>	AA – AO	BB – BO
<b>Antibody</b>	Anti-b	Anti-a
<b>Antigen</b>	Antigen-a	Antigen-b
<b>Receives blood from</b>	Blood types (A) and (O)	Blood types (B) and (O)

<b>Lethal genes</b>	<b>Complementary genes</b>
- They are genes which obstruct growth and cause death to living organisms if they are present in a pure form - Ex. Infantile dementia gene in humans	- They are genes which interact together forming hereditary trait - Ex. Flower colour trait of sweet pea

#### 5- Rationalize the following cases

##### **Case (1)**



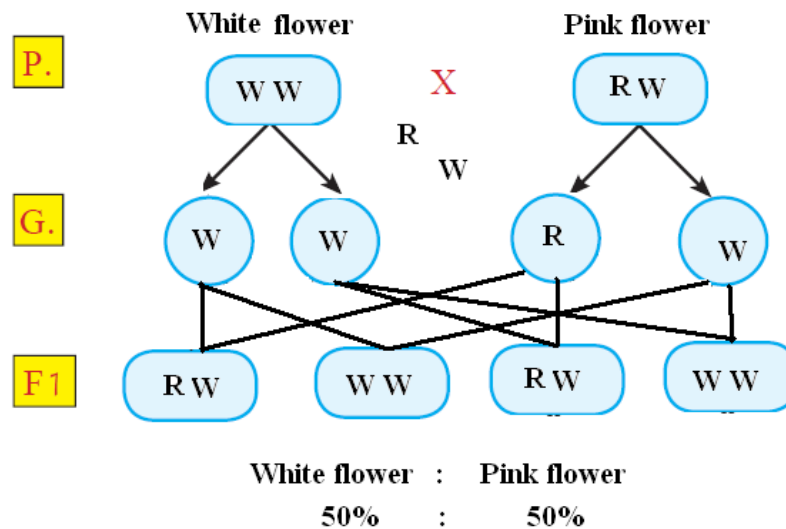
### Case (2)

Genes forming blood type (AB) are (A) and (B)

Thus, father should have at least one of those genes in his blood type

Probable genotypes of father are (AO) – (AA) – (AB) – (BO) – (BB)

### Case (3)



**6- The following table illustrates the generation resulted from the breeding of two sweet pea plants, then answer the following questions**

♀ \ ♂	AB	Ab	aB	ab
AB	(1)	AABb	(2)	AaBb
Ab	(3)	AAbb	(4)	Aabb

1- (1) AABB

(2) AaBB

(3) AABb

(4) AaBb

2- Genotype of 1<sup>st</sup> parent: AaBb

Genotype of 2<sup>nd</sup> parent: AABb

3- 25%

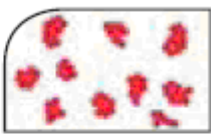
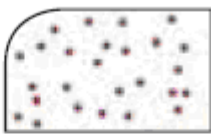
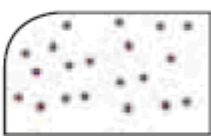

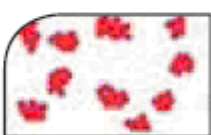
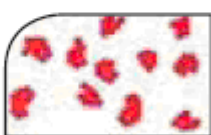


4- 75% Pink flowers

25% white flowers

<b>P.</b>		<b>Pink flower</b>			<b>Pink flower</b>	
		<b>Aa Bb</b>		X	<b>AA Bb</b>	
<b>F<sub>1</sub></b>		<b>G.</b>	<b>AB</b>	<b>Ab</b>	<b>aB</b>	<b>ab</b>
		<b>AB</b>	<b>AA BB</b>	<b>AA Bb</b>	<b>Aa BB</b>	<b>Aa Bb</b>
		<b>Ab</b>	<b>AA Bb</b>	<b>AA bb</b>	<b>Aa Bb</b>	<b>Aa bb</b>
		<b>Pink flower</b>			<b>White flower</b>	
		<b>6</b>		:	<b>2</b>	
		<b>3</b>		:	<b>1</b>	

7- Blood of groups (A) and (O) will be suitable because group (A) has antigen-a and anti-b like my own blood group, and (O) group has no antigens and both a-anti and b-anti

8-

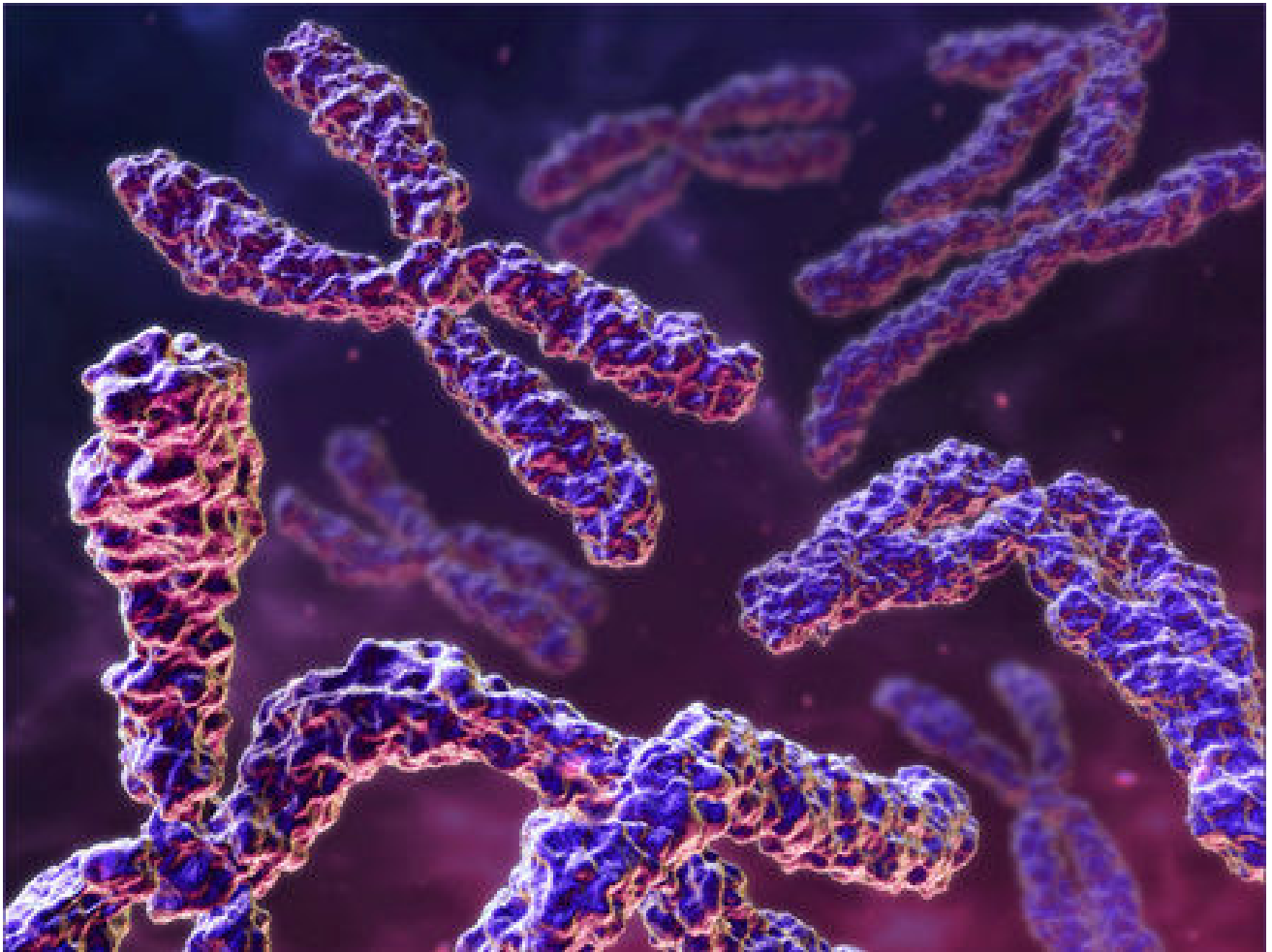
Group	anti-a	anti-b
<b>B</b>		
<b>A</b>		
<b>O</b>		
<b>AB</b>		

2- (AB) group

3- (O) group



### *Lesson (3) Genetic inheritance and diseases*



→ In the past, it was thought that woman controls the sex of her fetus, but in the middle of 20<sup>th</sup> century, when scientists discovered sex chromosomes, they discovered that man controls the sex of the fetus (not woman)

### Sex determination in human

Human body has 23 pairs of chromosomes, they are divided into:-

**1- Somatic chromosomes:** They are 22 chromosomes pairs which are similar in both sexes (male and female)

**2- Sex chromosomes:** 1 chromosome pair, which determines the sex of human.

**- Female cell:** it has 22 pairs of somatic chromosomes, and a homologous pair of sex chromosomes (XX + 44)

**- Male cell:** It has 22 pairs of somatic chromosomes and a non-homologous pair of sex chromosomes (XY + 44)

→ Chromosomes (X) and (Y) are different in shape and size

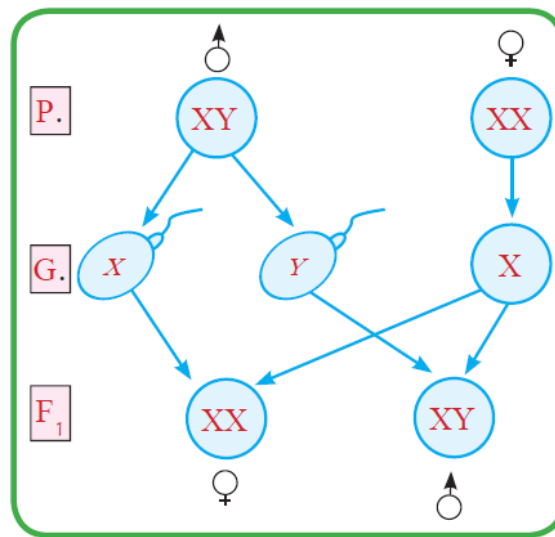


Fig. (5) Probabilities of sex determination of fetus

→ Gametes are formed from the meiotic cell division of gonads (testes in male and ovaries in female). So, gametes have half the number of chromosomes of somatic cells (Haploid number  $n$ )

→ Male produces two kinds of sperms with equal ratios – a kind of sperms carrying chromosomes (Y) and another one carrying chromosomes (X).

→ Female produces one kind of ova which carry chromosome (X)

→ If an ovum (X+22) was fertilized by sperm (X+22), a female fetus is formed (XX + 44)

→ If an ovum (X+22) was fertilized by sperm (Y+22), a male fetus is formed (XY+44)

*Therefore, sperms determine the sex of fetus, not ovum*

→ *Genes carried on chromosomes (X) and (Y) are responsible for sex determination.*

→ *6 weeks after conception (pregnancy), the fetus carrying (Y) chromosome begins secreting hormones which stimulate the undifferentiated tissues of gonads to form the tests and male sex organs.*

→ *12 weeks after conception (pregnancy), the fetus which doesn't carry (Y) chromosome begins forming the two ovaries, and then female sex organs differentiate.*

## **Abnormal chromosomal cases in human**

### ***Klinefelter's syndrome***

→ *This syndrome (case) was discovered by **Dr. Henry Klinefelter** in 1942.*

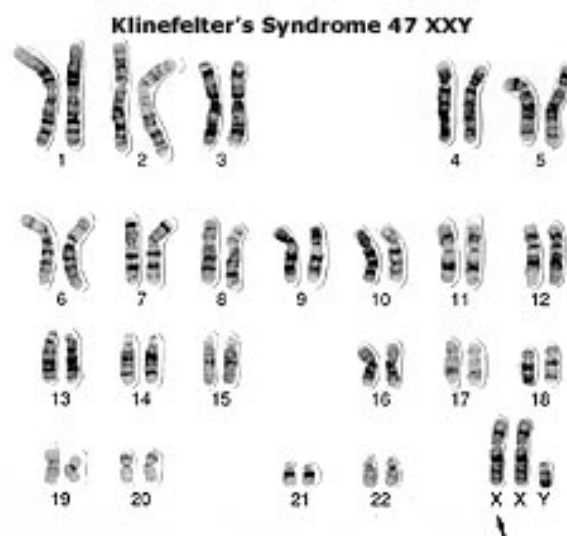
#### **Causes of Klinefelter's syndrome**

→ *When an abnormal ovum ( $XX + 22$ ) – **Instead of ( $X+22$ )** – is fertilized by a sperm ( $Y+22$ ), forming a fetus ( $XXY + 44$ ) **instead of ( $XY+44$ )**. The extra (X) chromosome causes disorder in sex hormones.*

→ *This syndrome appears in males only*

#### **Symptoms**

- 1- Infertile male due to the absence of the cells which produce sperms.*
- 2- Mental retardation*
- 3- The appearance of feminine characteristics (Ex. Enlargement of breasts)*
- 4- Tall stature*
- 5- Growth of limbs more than average rate*



*Fig. (6) Karyotype of Klinefelter's syndrome*

## Turner's syndrome

### Causes of Turner's syndrome

→ When an abnormal ovum ( $O+22$ ) – *Ovum without X chromosome* – is fertilized by a sperm ( $X+22$ ), forming a fetus ( $XO+44$ ) *fetus with only one X chromosome (instead of two)*

→ This syndrome appears in females only

### Symptoms

- 1- Short stature
- 2- Woman doesn't hit puberty due to the lack of hormones.
- 3- Slow mental development
- 4- congenital disorder of heart and kidneys

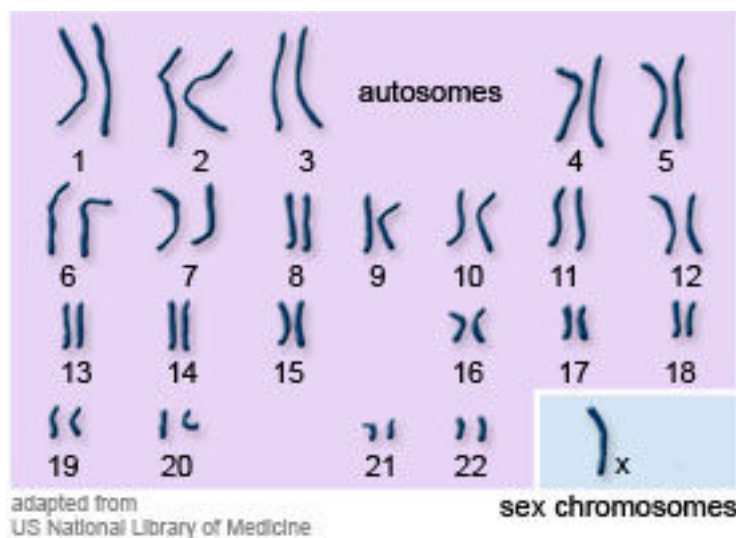


Fig. (7) Karyotype of Turner's syndrome

## Down syndrome (Mongolism)

→ Mongolism was discovered by British doctor Down in 1866

### Causes of down syndrome

→ caused by the fertilization of a gamete (sperm of ovum) gamete carrying a complete number of chromosomes in pair 21, which forms a fetus carrying 3 chromosomes in pair (21) *instead of 2 chromosomes*.

→ Baby with down syndrome may be a male ( $XY+45$ ) or female ( $XX+45$ )

### Symptoms of down syndrome

- 1- Delayed growth
- 2- Short stature
- 3- Oval face
- 4- Flat head back
- 5- Short fingers and toes

- 6- Narrow eyes
- 7- Small ears
- 8- Mental retardation

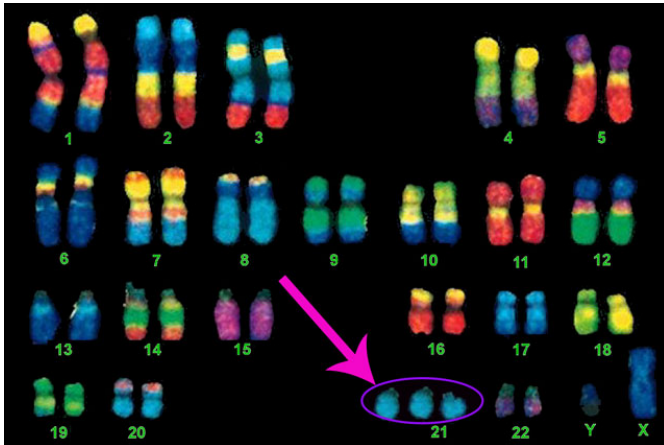


Fig. (7) Karyotype of down syndrome



(8) A girl with down syndrome

## Sex-linked traits

**Sex-linked traits:** They are traits whose genes are carried on sex chromosomes, but their appearance is not affected by sex hormones

→ Scientist Thomas Morgan was the first to discover sex-linked traits while studying eye colour trait in *Drosophila* insect. He crossed white-eyed male  $\frac{RR}{XX}$  *Drosophila* insects with red-eyed female  $\frac{r}{Xy}$  *Drosophila* insects.

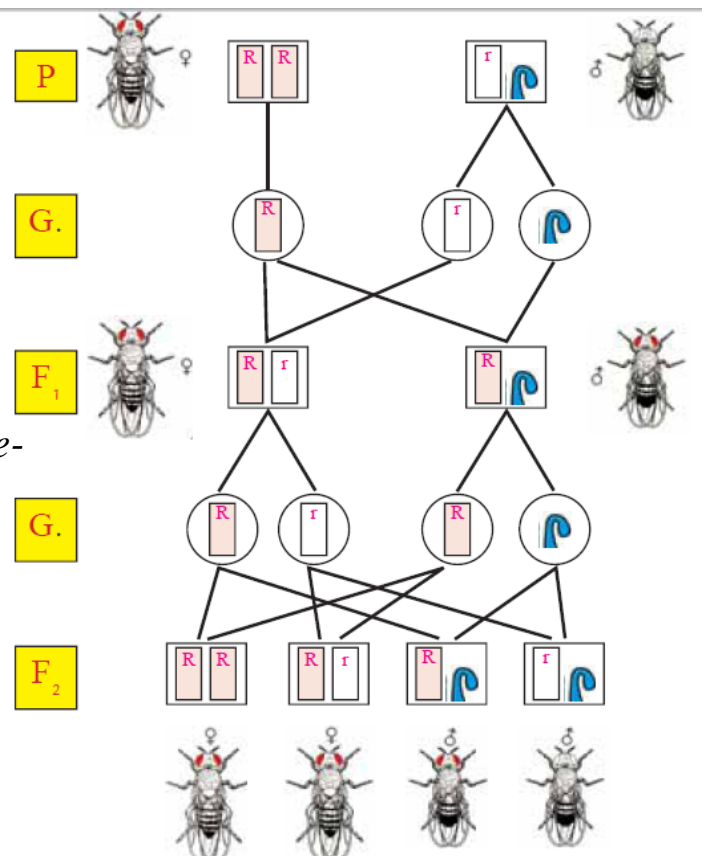
→ 100% of the individuals of 1<sup>st</sup> generation are red-eyed, which means that red eyes trait is dominant

→ Red-eyed and white-eyed insects were formed in 2<sup>nd</sup> generation at ratio 3:1

→ This trait is not a Mendelian one  
Because Morgan discovered that all white-eyed insects are males!!!!

→ Morgan stated that these genes are carried on sex chromosome (X), while there are few genes on chromosome (Y)

**Eye colour of *Drosophila* insect is an Example on sex-linked traits**





## Sex-linked traits in human

Chromosome (X) in human carries many somatic traits (Ex. Hemophilia – colour blindness – short sight – Muscle atrophy). These traits are inherited from the father to his daughters only because he gives them chromosome (X)

### Colour blindness

→ Colour blindness is caused by a recessive gene on sex chromosome (X).

→ This gene causes the inability to differentiate between colours, especially **green** and **red**

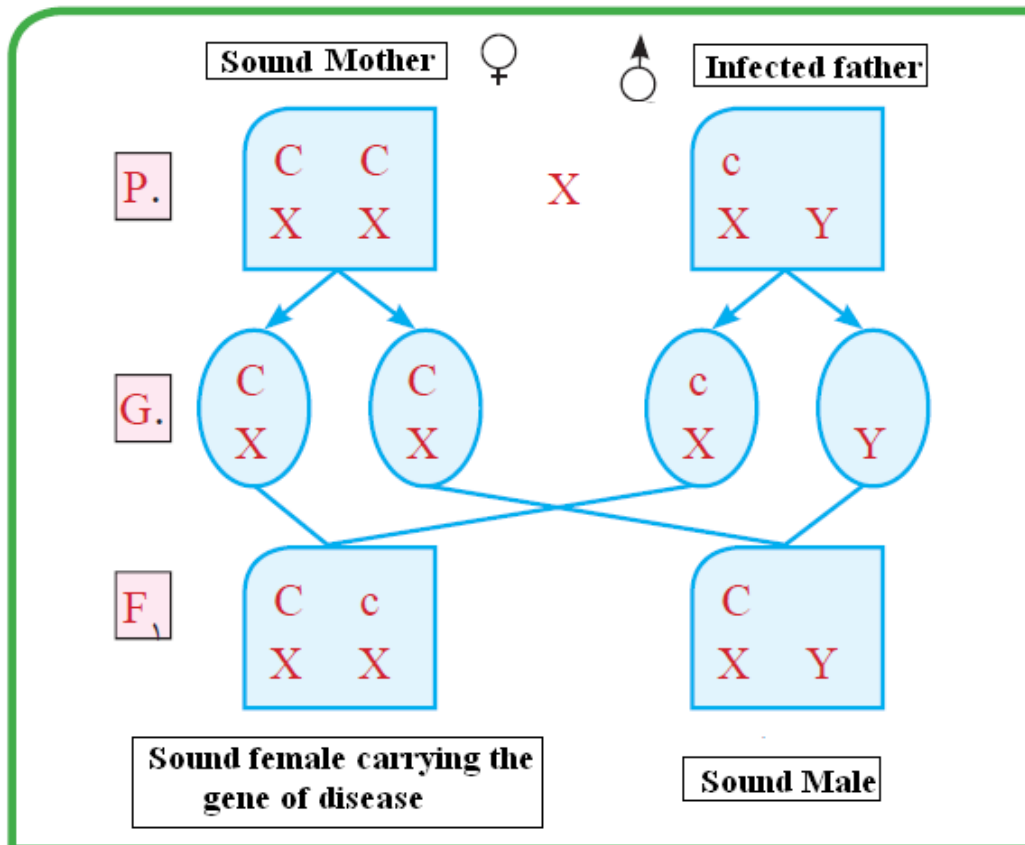


Fig. (9) Inheritance of colour blindness in human

In males, colour blindness trait is represented by only one gene because chromosome (Y) doesn't carry this trait, while it is represented by a pair of genes in females because they have a pair of sex chromosome (XX)

→ A male doesn't give this trait to his sons, because they inherit from him chromosome (Y) not chromosome (X) (which carries colour blindness trait)

→ Sons and daughters can inherit this trait from mother because they inherit chromosome (X) from her (which carries colour blindness trait)

→ Male can give this trait to his grandsons through his daughters.



## Hemophilia

**Hemophilia:** Genetic disease which causes the inability of body to control blood clotting (coagulation) process – the process which stops bleeding

→ Hemophilia is caused by a recessive gene carried on chromosome (X), this gene disables the body to control blood clotting process because some substances which are essential for blood clotting process are not formed

→ Hemophilia may cause death, especially in childhood stage

## Sex-influenced traits

**Sex-influenced traits:** They are traits whose genes are carried on somatic chromosomes, and their appearance is affected by sex hormones

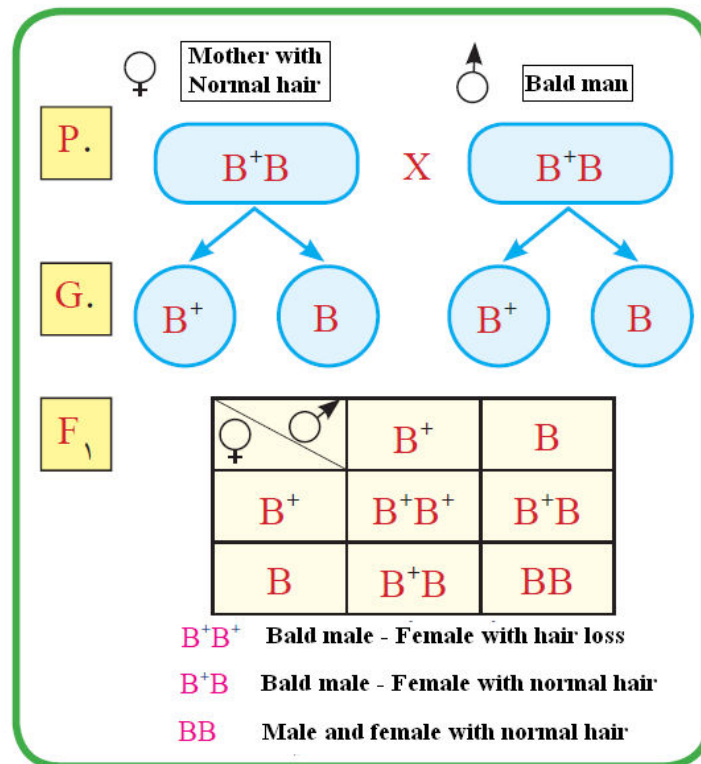
→ Sex-influenced traits are affected by male and female sex hormones

**Examples:-**

- 1- Horns trait in cattle
- 2- Baldness trait in human
- 3- Shortness of index finger trait in human

### Baldness

The following figure describes the inheritance of hair loss trait in human



→ Baldness trait appears due to the existence of a dominant gene responsible for hair loss, this gene is affected by male sex hormone only.

→ The appearance of baldness trait is different in males and females.

→ baldness appears in males in cases of pure gene ( $B+B+$ ) and hybrid gene ( $B+B$ ) due to the influence of male sex hormone on them

→ Hair loss trait in females appears only in case of pure gene ( $B+B+$ )

→ People with genetic structure ( $BB$ ) have normal hair



Fig. (10) Hereditary baldness in men



Fig.(11) hair loss in women

## Sex-limited traits

---

**Sex-limited traits:** They are traits which appears in one of the two sexes only due to the difference in sex hormones

---

Sex-limited genes are responsible for the appearance of certain traits in each sex.

**Examples:-**

→ Milk production trait is limited on females only because they have sex hormones which stimulate the gene of this trait

→ Secondary sex characteristics in humans (Ex. The appearance of beards in men)

→ The ability of birds to lay eggs.

## Methods of study in human genetics

### A- Pedigree (family tree)

Scientists find it hard to study the inherited traits and their transfer to human due to:-

- The long time period between one generation and another.

- The small number of individuals resulted from every marriage

So, scientists study some hereditary traits in human by studying the pedigree (family tree) of some families

---

**Pedigree:** Hereditary data represented in the form of chart diagram which explains how a certain trait is inherited, and used in tracking different hereditary traits

---

### Importance of pedigree

1- It explains how a certain trait is inherited in a certain family

2- It is used to trace different hereditary traits (especially those linked to genetic diseases and disorders)

3- It is used for prediction of the probability of appearance of these traits in coming generations.

### The following chart describes pedigree of a family

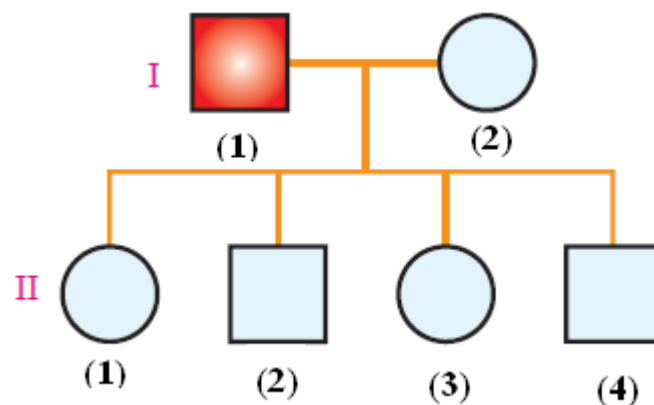


Fig. (12) Pedigree chart

- Male individuals are denoted by squares

- Female individuals are denoted by circles

- Mating (marriage) is symbolized by a line joining between the circle and square

- Offspring is symbolized by a vertical line extending from that of marriage

- Every generation is given a Roman number ( I – II – III – V...etc)

- Every individual is given a number, in order to facilitate finding it

- Shaped which are studies are coloured (in the previous chart, father (1) of generation (I) is the individual being studied)

### The study of genetic cases in human

**1- Albinism** ( also called **Albino** or **Sun enemy**)

---

**Albinism:** Genetic disorder which causes the absence of melanin pigments from the cells of hair, eye lashes and skin.

---

→ Albinism is caused by the recessive gene (a)

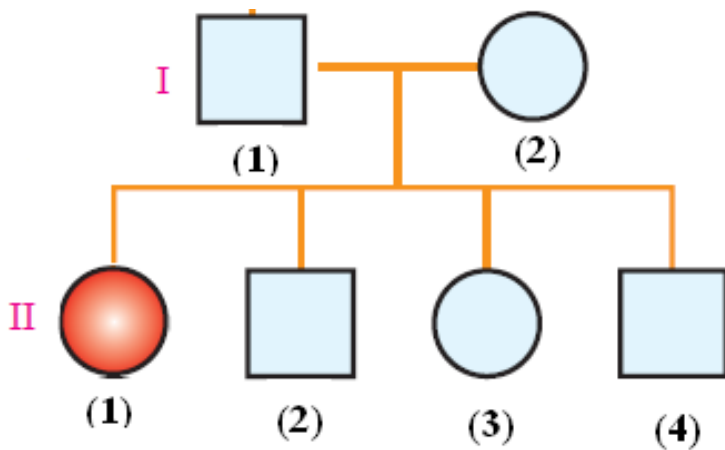


Fig. (13) Pedigree chart of albinism

Fig. (14) Child with Albinism

**Probable genotypes of this family members are:-**

Generation (I)

1- Aa 2- Aa

Generation (II)

1- aa 2- AA or Aa 3- AA or Aa 4- AA or Aa

## 2-Polydactyl

---

**Polydactyl:** Genetic disorder in which human has sixth fingers (instead of five), it is caused by a dominant gene

---



Fig. (14) Polydactyl genetic disorder

## **B- Gene maps (DNA analysis)**

→ Scientists succeeded in creating a map of all human genes.

→ This map can help us determine the diseases with by which human may be infected in the future.

## **C- Amniotic fluid testing during pregnancy**

---

**Amniotic fluid:** The fluid which surrounds the fetus in a pregnant female

---

→ Examining the chromosomes of fetus cells in amniotic fluid helps us determine the diseases caused by the increase or decrease of the no. of chromosomes (Ex. Down syndrome – Klinefelter's syndrome – Turner's syndrome), as we can get photos of these chromosomes and make Karyotype for the fetus.

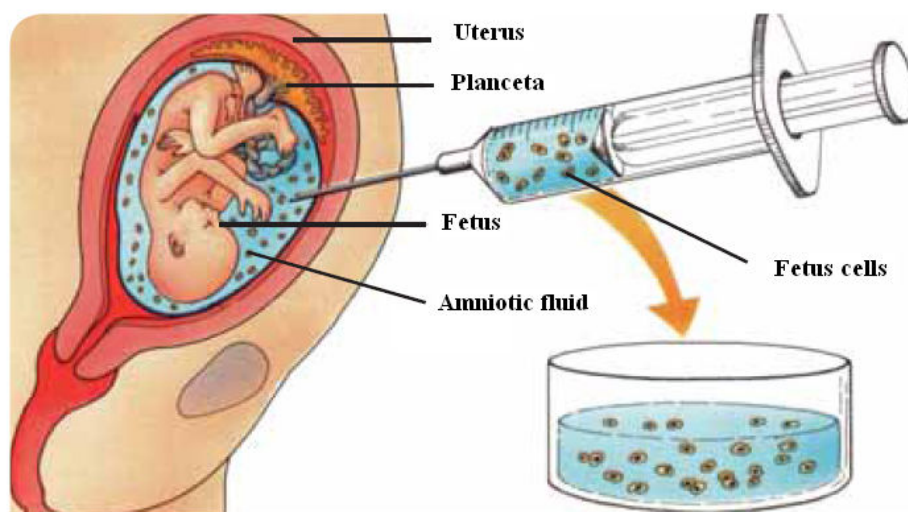


Fig. (15) Amniotic fluid testing

## **Medical tests before marriage**

→ Medical test before marriage is a group of medical tests which are carried out by the couple before they marry

→ Medical tests make sure they are not infected by contagious diseases ( such as hepatitis and Acquired immunity deficiency syndrome (AIDS) ) or genetic diseases (Such as Thalassemia), which predicts the probability of the transfer of the disease to the other mate or children , and gives choices and advises to the couple in order to help them make a healthy family.

### **Factors of genetic diseases spread**

1- Relatives marriage

2- Getting married without carrying out medical tests



## **The importance of medical tests**

*Medical tests help in:-*

- *Having healthy children*
- *Limiting the spread of genetic diseases, congenital disorders and mental retardation*
- *Avoiding social, psychological and financial problems resulted from taking care of children with genetic diseases.*

## **Science, Technology and society**

### ***The DNA fingerprint (DNA typing)***

→ In 1984, **Dr. Alec Jeffreys** (genetics scientist Leicester university in London) published a research in which he explained that the hereditary material may be repeated many times. After one year, he discovered that every person has unique sequences. These sequences cannot be the same in any two persons (except for identical twins). "Alec" patented his discovery in 1985 and called these sequences **The DNA fingerprint**

---

**The DNA fingerprint:** A method used to recognize a person by the comparison of his DNA sequences

---

### **The importance of DNA fingerprint**

**Field of medicine:-**

- *Used in the study of genetic diseases*
- *Used in tissues transplantation process*

**Field of forensics**

- *It helps in recognizing deformed dead bodies and tracking lost children*
- *It helped courts begin investigations in crimes whose offenders were unknown.*
- *It acquitted hundreds of people who were accused of rape and murder crimes and convicted others*
- *It plays an important role in parentage cases*

### ***Human genome***

→ Human genome contains all genes which exist in the nucleus of somatic cell

→ There are from 60,000 to 80,000 genes in nucleus, these genes are carried on 23 pairs of chromosomes. These genes cause the appearance of all these numerous human traits.

→ In 1953, scientists Watson and Creek discovered that genes are carried on a double helix of DNA.



→ In 1980, the idea of genome emerged. In that time, the number of discovered human genes was only 450 genes, in middle 80's the number of discovered genes became 1500. Scientists aimed to draw a genome map by determining the locations of genes on chromosomes, which would help them determine genes causing genetic diseases

### **The importance of genome**

Scientists aim to use genome in :-

- The manufacture of drugs without side effects
- The study of evolution of living organisms by comparing human genome to that of other organisms
- Improving offspring by determining the genes causing diseases in the fetus before its birth and improving them.

### **Definitions of lesson (1)**

**Klinefelter's syndrome:** Genetic disorder caused by the increase of sex chromosome (X) in some males (XXY + 44)

**Turner's syndrome:** Genetic disorder caused by the decrease of chromosome (X) in some females (XO+44)

**Down's syndrome:** Genetic disorder caused by the existence of an additional chromosome in chromosome pair (21)

**Sex-linked traits:** They are traits whose genes are carried on sex chromosomes , but their appearance is not affected by sex hormones.

**Sex-influenced traits:** They are traits whose genes are carried on somatic chromosomes, and their appearance is affected by sex hormones.

**Hemophilia:** Genetic disease which causes the inability of body to control blood clotting process – the process which stops bleeding

**Sex-limited traits:** They are traits which appear in one of the two sexes only due to the difference in sex hormones

**Albinism:** Genetic disorder which causes the absence of melanin pigments from the cells of hair, eye lashes and skin

**Pedigree:** Diagram which represents hereditary data which explains how a certain trait is inherited, and used in tracing different hereditary traits

**DNA fingerprint:** A method used to recognize a person by the comparison of his DNA sequences

## **Give reasons for**

### **1- A Sperm determines the sex of fetus not ovum**

*Because there are two kinds of sperms – a kind carrying (X) sex chromosome and another one carrying (Y) sex chromosome, while ova have only one kind (which carries X sex chromosome). When a sperm carrying (Y) chromosome fertilizes an ovum of (X) chromosome, a male fetus (XY) is formed, while when a sperm of (X) chromosome fertilizes an ovum, a female fetus (XX) is formed.*

### **2- A male with Klinefelter's syndrome is infertile and has feminine traits**

*Because it has an additional (X) sex chromosome in his Karyotype, which causes the disorder of sex hormones, as there are female sex hormones on (X) chromosome*

### **3- A female with Turner's syndrome cannot hit puberty**

*Due to the lack of female sex hormones responsible for puberty, as this female has only one (X) sex chromosome instead of two.*

### **4- Klinefelter's syndrome affects males only**

*Because Klinefelter's syndrome is caused when an abnormal ovum having 2 (X) chromosome (XX+22) is fertilized by a sperm (Y+22), which forms a male zygote of Karyotype (XXY+44)*

### **5- Turner's syndrome affects females only**

*Because Turner's syndrome is caused when an abnormal ovum with no X chromosomes (O+22) is fertilized by a sperm (X+22), which forms a female zygote of Karyotype (XX+44).*

### **6- Eye colour of Drosophila insect is from sex-linked traits**

*Because eye colour trait of Drosophila insect exists on sex chromosomes, and not affected by sex hormones*

### **7- Colour blindness is more common in males than females**

*Because colour blindness trait gene is carried on one (X) sex chromosome in males, while it has two genes carried on (XX) sex chromosomes in females. And as we know, a males carries only one (X) chromosome, while a woman carries two (X) chromosomes, so colour blindness is more common in men.*

### **8- Males cannot inherit colour blindness from their fathers**

*Because they inherit sex chromosomes (Y) from father, not chromosome (X) on which colour blindness gene is carried.*

### **9- Both females and males can inherit colour blindness from their mothers**

*Because both of them inherit chromosome (X) from mother – the chromosome on which colour blindness gene is carried*

**10- A man with genotype (B+B) is infected with baldness, while a female with the same genotype has normal hair**

*Because baldness is from sex-influenced traits, so its gene is only affected by male sex hormones*

**11- Scientists find it hard to study the inherited traits and how they transfer to human.**

*Due to:-*

- The long time between a generation and another one
- The small number of individuals resulted from every marriage

**12- Pedigree is from the most important methods in human genetics**

*Because:-*

- 1- It explains how a certain trait is inherited in a certain family
- 2- It is used to trace different hereditary traits (especially those linked to genetic diseases and disorders)
- 3- It is used for prediction of the probability of appearance of these traits in coming generations

**13- The importance of DNA analysis (gene map)**

*Because this map can help us determine the diseases with by which human may be infected in the future.*

**14- The spread of genetic diseases and disorders**

*Due to:-*

- 1- Marriage of relatives
- 2- Getting married without carrying out medical tests

**15- The importance of carrying out medical tests before marriage**

*Because medical tests help in:-*

- Having healthy children
- Limiting the spread of genetic diseases, congenital disorders and mental retardation
- Avoiding social, psychological and financial problems resulted from taking care of children with genetic diseases

**16- DNA fingerprint plays an important role in the field of medicine**

*Because:-*

- It is used in the study of genetic diseases
- It is used in tissues transplantation process

### ***17- Amniotic fluid testing is very important during pregnancy***

*Because it helps us determine the diseases caused by the increase or decrease of the no. of chromosomes (Ex. Down syndrome – Klinefelter's syndrome – Turner's syndrome), as we can get photos of these chromosomes and make Karyotype for the fetus.*

### ***18- The importance of DNA fingerprint in forensic science***

*Because:-*

- It helps in recognizing deformed dead bodies and tracking lost children*
- It helped courts begin investigations in crimes whose offenders were unknown.*
- It acquitted hundreds of people who were accused of rape and murder crimes and convicted others*
- It plays an important role in parentage cases*

### ***19- The importance of genome in human genetics***

*Because scientists aim to use genome in :-*

- The manufacture of drugs without side effects*
- The study of evolution of living organisms by comparing human genome to that of other organisms*
- Improving offspring by determining the genes causing diseases in the fetus before its birth and improving them*

## ***What happens when***

### ***1- A sperm (Y+22) fertilizes an ovum (X+22)***

*A male zygote (XY+44) is formed*

### ***2- A sperm (X+22) fertilizes an ovum of the same Karyotype***

*A female zygote (XX+44) is formed*

### ***3- A sperm (Y+22) fertilizes an abnormal ovum (XX+22)***

*A male fetus (XXY+44) suffering from Klinefelter's syndrome is formed, which makes him infertile and causes the appearance of some feminine traits, tall stature, growth of limbs more than average rate and mental retardation due to the disorder of sex hormones.*

### ***4- A sperm (X+22) fertilizes an abnormal ovum (O+22)***

*A female fetus (XO+22) suffering from Turner's syndrome is formed, which makes her unable to hit puberty due to the lack of hormones and causes short stature, slow mental development, and congenital disorders in heart and kidneys.*

### ***5- The fertilization of a gamete carrying a complete pair of chromosome in pair (21)***

*A male fetus (XY+45) or female fetus (XX+45) suffering from Down syndrome is formed (because of having 3 copies of chromosome 21), which causes mental retardation, short stature, oval face, flat head back, short fingers and toes, small ears and narrow eyes*

## **Comparison**

<b><i>Point of comparison</i></b>	<b><i>Sex-linked traits</i></b>	<b><i>Sex-influenced traits</i></b>	<b><i>Sex- limited traits</i></b>
<b><i>Definition</i></b>	<i>They are traits whose genes are carried on sex chromosomes , but their appearance is not affected by sex hormone</i>	<i>They are traits whose genes are carried on somatic chromosomes, and their appearance is affected by sex hormone</i>	<i>They are traits which appears in one of the two sexes only due to the difference in sex hormone</i>
<b><i>Examples</i></b>	<i>Eye colour of Drosophila</i>	<i>Baldness / Hemophilia</i>	<i>Milk production n females</i>

<b><i>Point of comparison</i></b>	<b><i>Down syndrome</i></b>	<b><i>Klinefelter's syndrome</i></b>	<b><i>Turner's syndrome</i></b>
<b><i>Karyotype</i></b>	<i>(XX + 45) or (XY+45)</i>	<i>(XXY + 44)</i>	<i>(XO+44)</i>
<b><i>Causes</i></b>	<i>The fertilization of a gamete carrying a complete pair of chromosome (21)</i>	<i>The fertilization of an abnormal ovum (XX+22) by a sperm (Y+22)</i>	<i>The fertilization of an abnormal ovum (O+22) by a sperm (X+22)</i>
<b><i>Symptoms</i></b>	<ul style="list-style-type: none"> <li>- Mental retardation</li> <li>- Narrow eyes</li> <li>- Short stature</li> <li>- Short toes and fingers</li> <li>- Flat head back</li> <li>- Delayed growth</li> <li>- Oval face</li> <li>- Small ears</li> </ul>	<ul style="list-style-type: none"> <li>- Infertile male</li> <li>- The appearance of feminine traits (growth of breast)</li> <li>- Tall stature</li> <li>- Mental retardation</li> <li>- Overgrowth of limbs</li> </ul>	<ul style="list-style-type: none"> <li>- Female's inability to hit puberty</li> <li>- Congenital disorders in kidneys and heart</li> <li>- Short stature</li> </ul>

# Questions

## 1- Choose the correct answer

1- The Karyotype of male cell is .....

A-  $XX + 44$     B-  $XY + 44$     C-  $XO + 44$     D-  $XXY + 44$

2- The Karyotype of female cell is .....

A-  $XX + 44$     B-  $XY + 44$     C-  $XO + 44$     D-  $XXY + 44$

3- Karyotype of Klinefelter's syndrome is .....

A-  $XXY + 44$     B-  $XO + 44$     C-  $YO + 45$     D-  $XY + 45$

4- Karyotype of Turner's syndrome is .....

A-  $XXY + 44$     B-  $XO + 44$     C-  $YO + 45$     D-  $XY + 45$

5- Down syndrome is caused by the fertilization of an ovum ( $X + 22$ ) with sperm

A-  $X + 23$     B-  $Y + 23$     C-  $X + 22$     D-  $Y + 22$

6- ..... affects males only

A- Turner's syndrome    B- Klinefelter's syndrome    C- Down syndrome  
D- Hepatitis

7- ..... affects females only

A- Turner's syndrome    B- Klinefelter's syndrome    C- Down syndrome  
D- Hepatitis

8- ..... is also known as Mongolism

A- Turner's syndrome    B- Klinefelter's syndrome    C- Down syndrome  
D- Hemophilia

9- The eye colour of *Drosophila* insect is an example on ..... traits

A- Sex-linked    B- Sex-influenced    C- Sex-limited    D- Mendelian

10- Colour blindness trait is an example on ..... traits

A- Sex-linked    B- Sex-influenced    C- Sex-limited    D- Mendelian

11- When a sound male and a female infected with colour blindness marry, the appearance of this case will be in .....

A- All males    B- All females    C- Half the males    D- Half the females

12- Hemophilia is an example on ..... traits

A- Sex-linked    B- Sex-influenced    C- Sex-limited    D- Mendelian

13- Growth of beards in males is an example on ..... Traits

A- Sex-linked    B- Sex-influenced    C- Sex-limited    D- Mendelian



14- Genetic disorder which causes the absence of melanin pigments in human body is called .....

A- Albinism    B- Hemophilia    C- Polydactyl    D- Baldness

15- DNA fingerprint was discovered by scientist .....

A- Gregory Mendel    B- Watson and Creek    C- T. Morgan    D- Alec Jeffreys

## **2- Write the scientific term**

1- Genetic disorder caused by the increase of sex chromosome (X) in some males (XXY + 44)

2- Genetic disorder caused by the decrease of chromosome (X) in some females (XO+44)

3- Genetic disorder caused by the existence of an additional chromosome in chromosome pair (21)

4- They are traits whose genes are carried on sex chromosomes , but their appearance is not affected by sex hormones.

5- They are traits whose genes are carried on somatic chromosomes, and their appearance is affected by sex hormones.

6- Genetic disease which causes the inability of body to control blood clotting process – the process which stops bleeding

7- They are traits which appear in one of the two sexes only due to the difference in sex hormones

8- Genetic disorder which causes the absence of melanin pigments from the cells of hair, eye lashes and skin

9- Diagram which represents hereditary data which explains how a certain trait is inherited, and used in tracing different hereditary traits

10- A method used to recognize a person by the comparison of his DNA sequences

## **3- Write short notes about**

1- Sex-linked traits

2- Sex-influenced traits

3- Sex- limited traits

4- Turner's syndrome

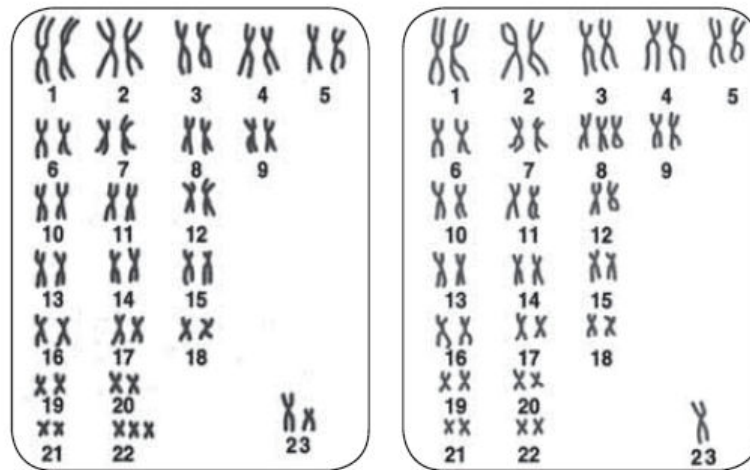
5- Klinefelter's syndrome

6- Down syndrome

7- Genome

8- DNA fingerprint

**4- Study the following figure and answer the questions**

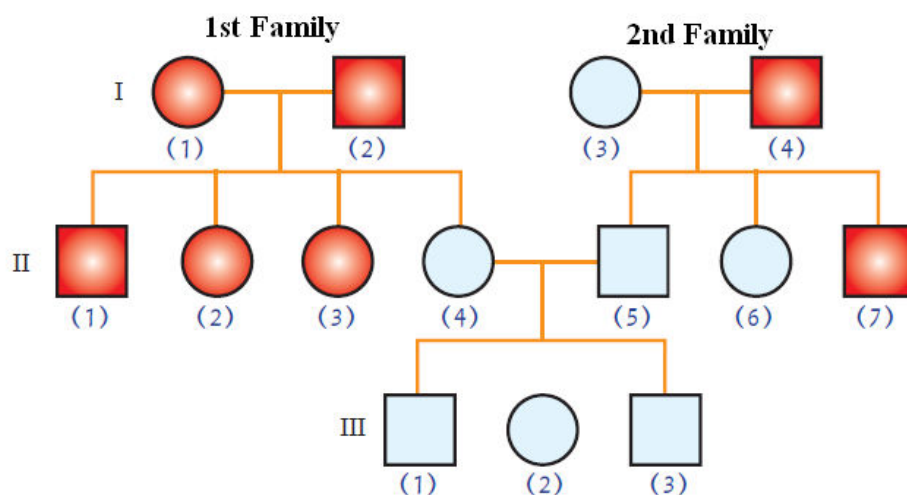


Karyotype (1)

Karyotype (2)

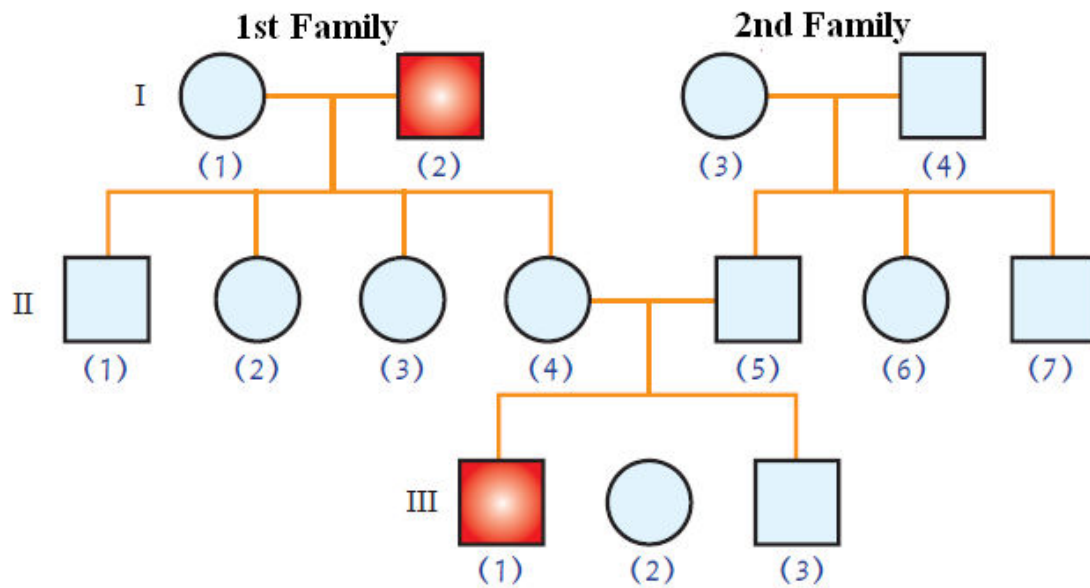
- 1- What is the number of chromosomes in karyotypes (1) and (2)?
- 2- What are the sexes of the individuals carrying karyotypes (1) and (2)?
- 3- What is the name, causes and symptoms of the abnormal condition in karyotype (1)?
- 4- What is the name, causes and symptoms of the abnormal condition in karyotype (2)?

**5- The following pedigree chart describe the ability to roll tongue trait, answer the questions**



- 1- Is this trait dominant or recessive? Why?
- 2- What are the genotypes of the following individuals
  - I (1)
  - I (3)
  - II (3)
  - II (5)
  - III (1)

**6- The following pedigree charts describe the inheritance of Hemophilia trait, answer the following questions**



1- Is this trait dominant or recessive?

2- Write the genotypes of the following individuals

I (1)

I (2)

II (1)

II (5)

III (1)

## The Answers

**1- Choose the correct answer**

1- XY+44

2- XX + 44

3- XXY + 44

4- XO +44

5- Y + 23

6- Klinefelter's syndrome

13- Sex-limited

15- Alec Jeffreys

7- Turner's syndrome

8- Down syndrome

9- Sex-linked

10- Sex-linked

11- All males

12- Sex-linked

14- Albinism

## **2- Write the scientific term**

- |                               |                       |
|-------------------------------|-----------------------|
| 1- Klinefelter's syndrome     | 6- Hemophilia         |
| 2- Turner's syndrome          | 7- Sex-limited traits |
| 3- Down syndrome              | 8- Albinism           |
| 4- Sex-linked chromosomes     | 9- Pedigree chart     |
| 5- Sex-influenced chromosomes | 10- DNA fingerprint   |

## **4- Study the following figure and answer the questions**

- 1- Karyotype (1): 47 chromosomes – Karyotype (2): 45 Chromosomes
- 2- Karyotype (1): Male – Karyotype (2): Female
- 3- Name: Down syndrome  
Cause: Fertilization of a gamete having a complete pair of chromosomes 21  
Symptoms: Mental retardation – Delayed growth – Oval face – Narrow eyes – Short stature – Small ears – Flat head back – Short fingers and toes
- 4- Name: Turner's syndrome  
Cause: Fertilization of an abnormal ovum ( $XO+22$ ) by a sperm ( $X+22$ )  
Symptoms: Short stature – Inability to hit puberty – Congenital disorders in heart and kidneys

## **5-**

1- Rolling tongue trait is dominant because it appeared in all members of 1<sup>st</sup> generation in the first family at ratio 100%, and 2<sup>nd</sup> generation at ratio 75%

2-

- $I(1) \rightarrow Bb$   
 $I(3) \rightarrow bb$   
 $II(3) \rightarrow BB \text{ or } Bb$   
 $II(5) \rightarrow bb$   
 $III(1) \rightarrow bb$

## **6-**

1- Recessive

2-

- $I(1) \rightarrow AA$   
 $I(2) \rightarrow aa$   
 $II(4) \rightarrow Aa$   
 $II(5) \rightarrow Aa$   
 $III(1) \rightarrow aa$

## General test on chapter (3)

*Answer four questions only*

### **Question (1)**

**A- Choose the correct answer**

1- The points of connection of internal chromatids in homologous chromosomes pairs is called .....

A- Chiasma    B- Chromosome    C- Centromere    D- Centrosome

2- In gametes, chromosomes which didn't undergo crossing over are called ....

A- New chromosomes    B- Building chromosomes    C- Parental chromosomes  
D- Chromatids

3- When a sound male and a female infected with colour blindness marry, the appearance of this case will be in .....

A- All males    B- All females    C- Half the males    D- Half the females

4- Blood group which is known as "Universal donor" is.....

A- A    B- O    C- AB    D- B

5- Trait of flower colour of sweet pea plant is an example on .....

A- Lack of dominance    B- Alleles multiplicity    C- Complementary genes  
D- Lethal genes

**B- Give reasons for**

1- Turner's syndrome affects females only

2- The importance of blood groups

3- The importance of crossing over phenomenon

4- Colour blindness is more common in males than females

### **Question (2)**

**A- Write the scientific term**

1- Genes which interact with each other causing the appearance of a hereditary trait.

2- A form of inheritance in which no genes dominate over the opposite one, but they interact forming new trait

3- chemical substances which exist on the surfaces of red blood cells, they play an important role in blood transfusion process.

4- Genetic disorder caused by the decrease of chromosome (X) in some females (XO+44)

5- Genetic disorder caused by the existence of an additional chromosome in chromosome pair (21)

**B- What happens when**

1- Breeding two yellow mice (Yy)

2- A sperm (Y+22) fertilizes an abnormal ovum (XX+22)

3- Two sweet pea plants with white flowers whose genotypes are (aaBB) and (AAbb) copulate (first and second generations)

4- Transfusing blood from a man of group (AB) to another one of group (A)

**Question (3)**

**A- Match**

(A)	(B)
1- Flower colour of sweet pea plant	A- Lack of dominance
2- Colour blindness in human	B- Lethal genes
3- Yellow colour of mice	C- Complementary genes
4- Flower colour of four o'clock plant	D- Sex-linked traits
5- Milk production in females	E- Sex-influenced traits
	F- Sex-limited traits

**B- Write short notes about**

1- Chromosomal theory

2- Rhesus factor

3- Pedigree

4- Sex-limited traits

**Question (4)**

**A- Correct the underlined words**

1- The genetic disorder caused by extra (X) sex chromosome in males is Down syndrome

2- DNA fingerprint was discovered by Thomas Morgan

3- Hemophilia is an example on sex-influenced traits

4- The appearance of chlorophyll is affected by the factor of temperature

5- The ratio of 2<sup>nd</sup> generation in case of complementary genes is 1:3:3:9

**B- Compare between**

1- Blood types (A) and (B)

2- Lethal and complementary genes

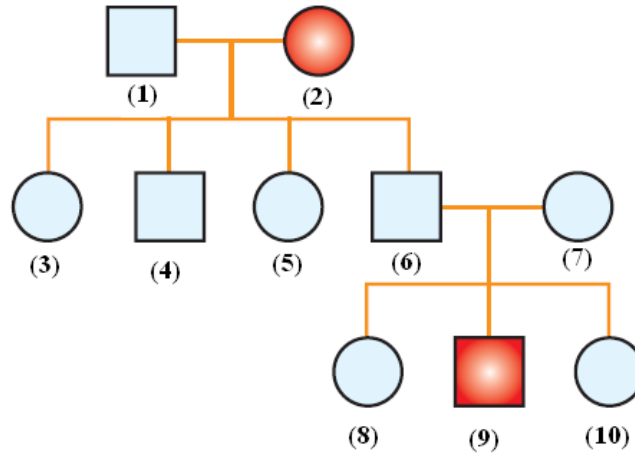
3- Klinefelter's and Turner's syndrome



#### 4- Antigens a and anti-a

##### Question (5)

**A-** The following figure describes the pedigree chart for the trait of freckles presence , answer the questions



1- Is this trait dominant or recessive? Why?

2- What are the probable genotypes of individuals (1), (2), (6), (7), (9)

##### **Solution**

##### Question (1)

**A- Choose the correct answer**

1- Chiasma

2- Parental chromosomes

3- All males

4- O

5- Complementary genes

**B- Give reasons for**

1- Because Turner's syndrome is caused when an abnormal ovum with no X chromosomes ( $O+22$ ) is fertilized by a sperm ( $X+22$ ), which forms a female zygote of Karyotype ( $XX+44$ ).

2- Because they :-

- Solve problems of the determination of paternity (parents of children) and pedigree of children (blood groups denies pedigree but don't prove it)
- Determine blood transfusion processes between individuals.
- Are used in the study of human races classification and evolution

3- Because it causes the variation of hereditary traits of the members of same species, which helps them in the adaptation with environment conditions, and the development and continuation of their life.

4- Because colour blindness trait gene is carried on one (X) sex chromosome in males, while it has two genes carried on (XX) sex chromosomes in females. so colour blindness is more common in men because they should have only one recessive gene to be infected with colour blindness .

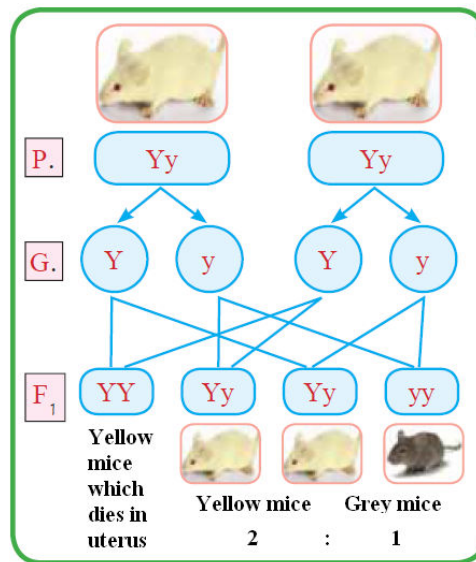
### **Question (2)**

#### **A- Write the scientific term**

- 1- Complementary genes
- 2- Lack of dominance
- 3- Antigens
- 4- Turner's syndrome
- 5- Down syndrome

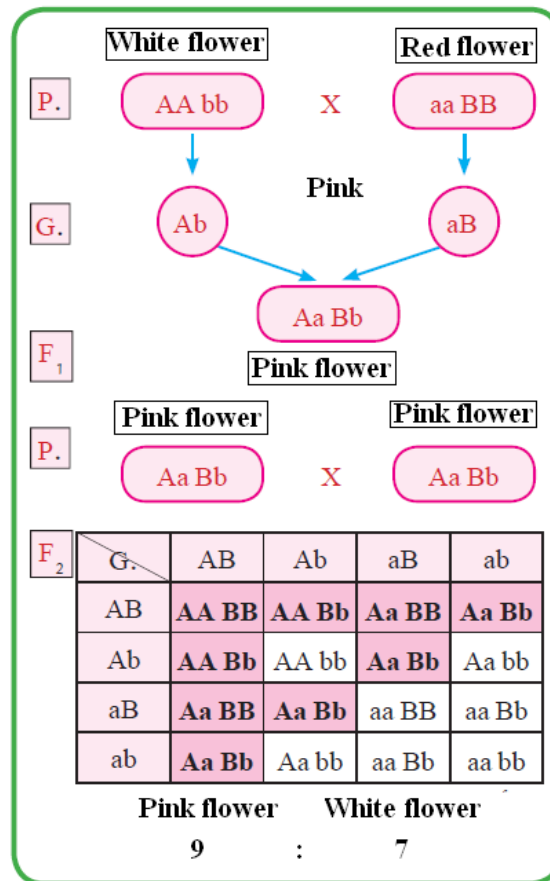
#### **B- What happens when**

1- Hybrid yellow and black mice are produced at ratio 2:1 respectively, pure yellow mice (YY) - which represent 25% of the generation – die in uterus before being born.



2- This causes Klinefelter's syndrome, which affects males and make them infertile.

3- 100% pink flowers are produced in the first generation, while both pink and white flower appear in the second generation at ratio 9:7



4- This will break red blood cells of the recipient person because his blood produces anti-b for antigens-B of blood group (AB), which causes shivering in body, chest pain, blueness, irregular heartbeat, headache, low blood pressure

### Question (3)

#### **A- Match**

1- C      2- D      3-B      4-A      5-F

#### **B- Write short notes about**

1- 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 transferring to gametes.
- 4- After fertilization process, the normal number of chromosomes (2n) comes back
- 5- Each chromosome carries hundreds of genes.

2- Rhesus factor is a kind of antigens which exist on the surfaces of red blood cells of 85% of humans, its inheritance is controlled by 3 pairs of genes which exist on one chromosome pair.

3- Hereditary data represented in the form of chart diagram which explains how a certain trait is inherited, and used in tracing different hereditary traits

4- They are traits which appear in only one of the two sexes due to the difference of hormones, such as the growth of beard trait in men and milk production trait in women.

#### **Question (4)**

##### **A- Correct the underlined words**

1- Klinefelter's syndrome

2- Dr. Alec Jeffreys

3- Milk production trait in women

4- Light

5- 7:9

##### **B- Compare between**

<b>Blood group (A)</b>	<b>Blood group (B)</b>
<ul style="list-style-type: none"> <li>- It has antigens-a</li> <li>- It has antibodies (anti-b)</li> <li>- Its genotype is AA or AO</li> <li>- Receives blood from groups (A), (O)</li> <li>- Gives blood to groups (A) and (AB)</li> </ul>	<ul style="list-style-type: none"> <li>- It has antigens-b</li> <li>- It has antibodies (anti-b)</li> <li>- Its genotype if BB or BO</li> <li>- Receives blood from groups (B), (O)</li> <li>- Gives blood to groups (B), (AB)</li> </ul>

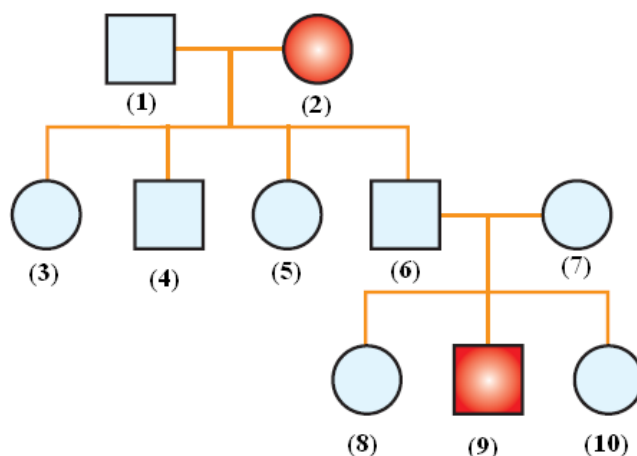
<b>Lethal genes</b>	<b>Complementary genes</b>
<ul style="list-style-type: none"> <li>- They are genes which cause the death of living organisms if they are present in pure form because they stop its biological processes</li> </ul> <p>Ex. Yellow colour of mice trait</p>	<ul style="list-style-type: none"> <li>- They are genes which interact together forming new trait</li> </ul> <p>Ex. Flower colour of sweet pea plant</p>

<b>Klinefelter's syndrome</b>	<b>Turner's syndrome</b>
<ul style="list-style-type: none"> <li>- Its karyotype is (XXY+44)</li> <li>- Caused due to the fertilization of an abnormal ovum (XX+22) by a sperm (Y+22)</li> <li>- If affects males only</li> <li>- It causes infertility, tall stature, ..etc</li> </ul>	<ul style="list-style-type: none"> <li>- Its karyotype is (XO+44)</li> <li>- Caused due to the fertilization of an abnormal ovum (O+22) by a sperm (X+22)</li> <li>- If affects Females only</li> <li>- It causes inability to attain puberty</li> </ul>

<i>Antigens-a</i>	<i>Antibodies-a</i>
<i>They exist in blood groups (A) and (AB)</i>	<i>They exist in blood groups (B) and (O)</i>

**Question (5)**

**A- The following figure describes the pedigree chart for the trait of freckles presence , answer the questions**



1- This trait is recessive because it disappeared in all members of 1<sup>st</sup> generation

2-

(1) AA

(2) aa

(6) Aa

(7) Aa

(9) aa