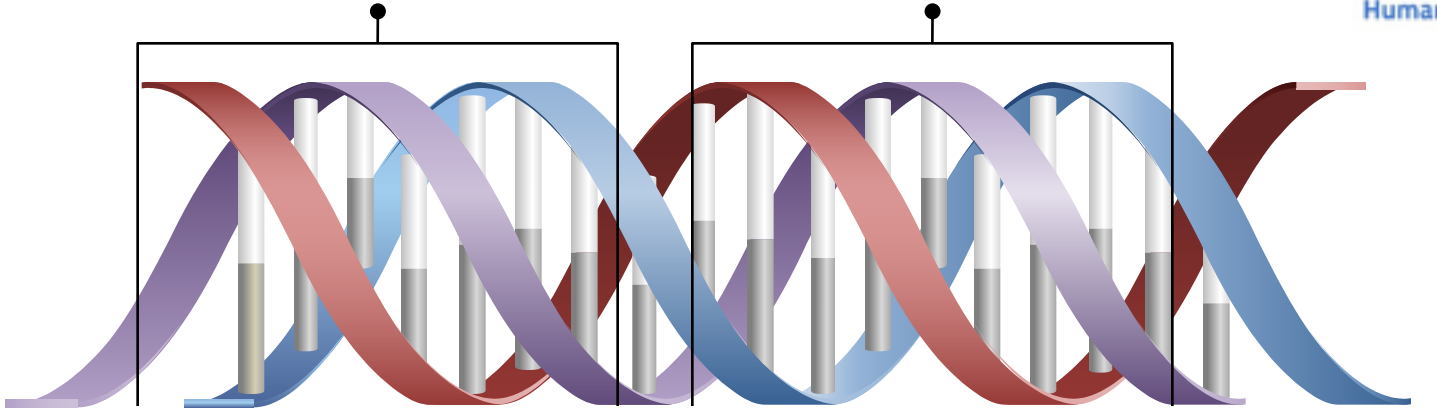
جامعة الملك سعود
King Saud University

Human Genetics 439

Lecture 3

MODE OF INHERITANCE



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MED439

Objectives:

By the end of the lecture, the student should:

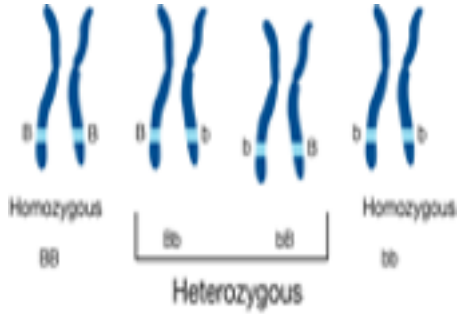
- Assess Mendel's laws of inheritance.
- Understand the bases of Mendelian inheritance.
- Define various patterns of single gene inheritance using family pedigree and Punnett square.

Interpreting the outcomes of Mendel's breeding experiments

- The plant characteristics being studied were each controlled by pair of factors, one of which was inherited from each parent.
- The pure-bred plants, with two identical genes, used in the initial cross would now be referred to as homozygous.
- The hybrid F1 plants, each of which has one gene for tallness and one for shortness, would be referred to as heterozygous.
- The genes responsible for these contrasting characteristics are referred to as *allelomorphs*, or alleles for short.

Genotype

1. **Homozygous (same alleles)**
   - Homozygous for the dominant alleles AA.
   - Homozygous for the recessive alleles aa.



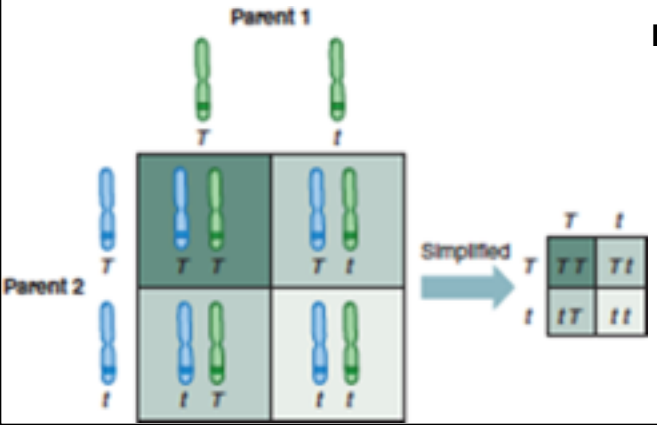
2. **Heterozygous (differ alleles)**
   - Heterozygous is dominant + recessive alleles in the same time Aa.

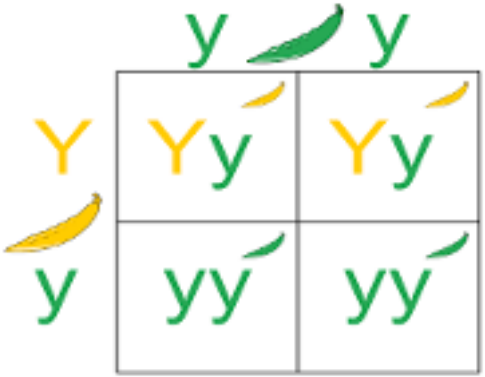
Punnett Square

Each parent can only contribute to one allele per gene

Offspring will inherit 2 alleles to express that gene

These genes are found on the romosomes of gametes of parents





Laws of inheritance

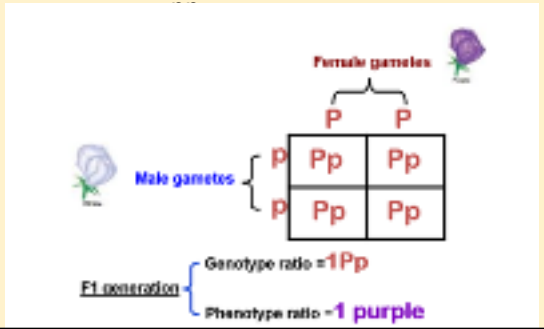
Law of Dominance or Uniformity

COMPLETE DOMINANCE: one allele is dominant to another allele.

- RECALL MENDEL'S 1st EXPERIMENT

CROSS: Pure-bred purple female x White male

P1 generation = PP x



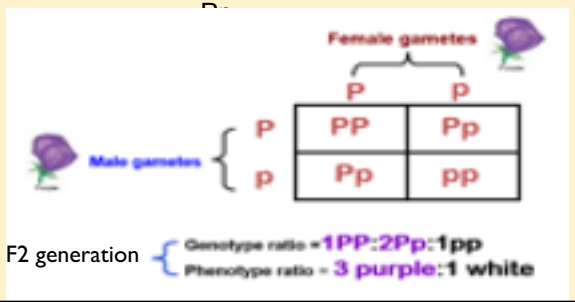
LAW OF SEGREGATION

Definition: the alleles of a given locus segregate into separate gametes.

- RECALL MENDEL'S 2nd EXPERIMENT

CROSS:Two F1 generation offspring with each other

P1 generation = Pp x

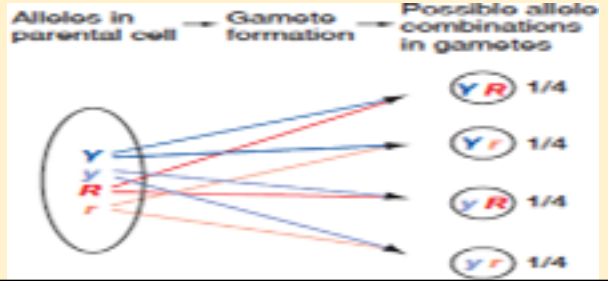


LAW OF INDEPENDENT ASSORTMENT

Definition: During gamete formation, different pairs of alleles segregate independently of each other.

INTERPRETATION:

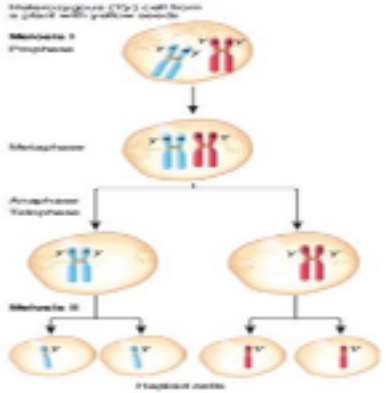
In a dihybrid cross, each pair of alleles assorts independently during gamete formation. In the gametes,Y is equally likely to be found with R or r (that is, Y R = Y r); the same is true for y (that is, y R = y r).As a result, all four possible types of gametes (Y R , Y r , y R , and y r) are produced in equal frequency among a large population.



THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES

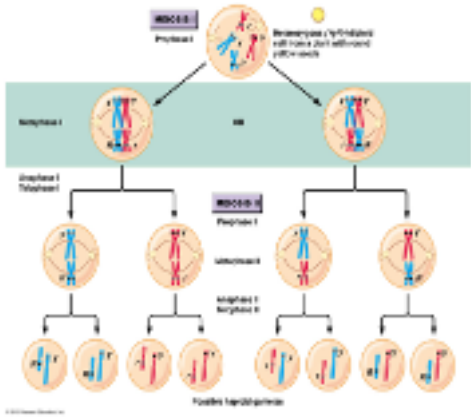
1-Mendel's law of segregation can be explained by the segregation of homologs during meiosis

- The two copies of a gene are contained on homologous chromosomes.
- In this example using pea seed color, the two alleles are Y (yellow) and y (green).
- During meiosis, the homologous chromosomes segregate from each other, leading to segregation of the two alleles into separate gametes.

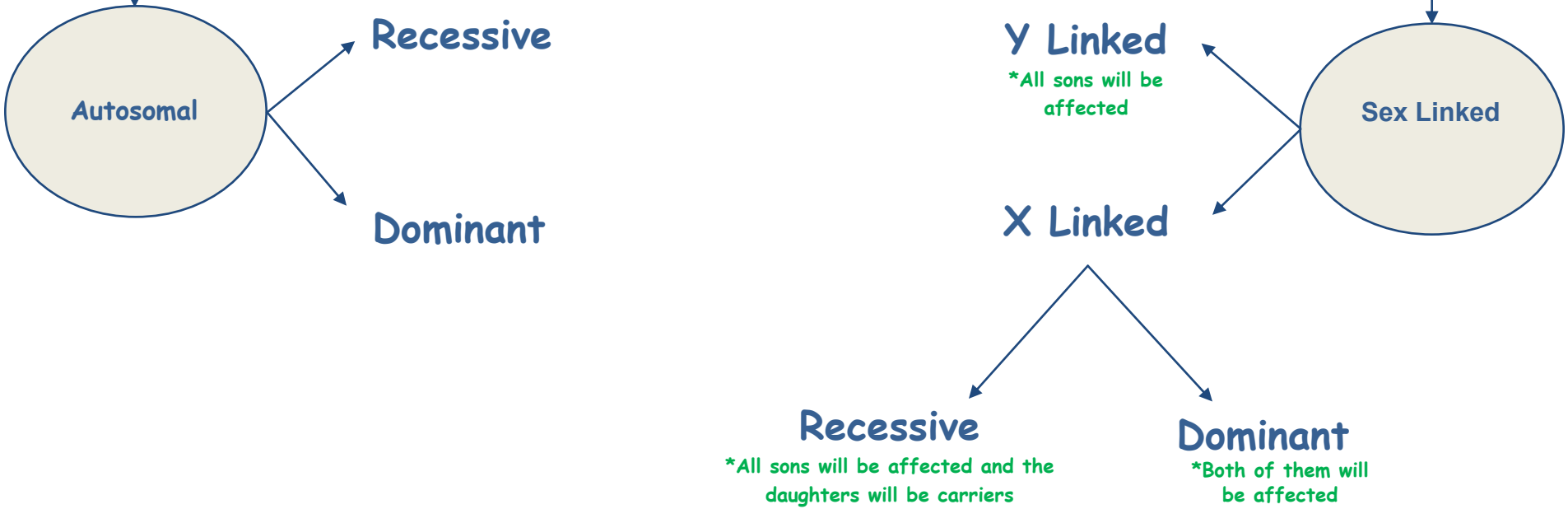


2-Mendel's law of independent assortment can be explained by the random alignment of bivalents during metaphase of meiosis I

- This figure shows the assortment of two genes located on two different chromosomes, using pea seed color and shape as an example (YyRr).
- During metaphase of meiosis I, different possible arrangements of the homologs within bivalents can lead to different combinations of the alleles in the resulting gametes.
- For example, on the left, the dominant R allele has sorted with the recessive y allele; on the right, the dominant R allele has sorted with the dominant Y allele.



MODES OF INHERITANCE FOR SINGLE GENE DISORDERS



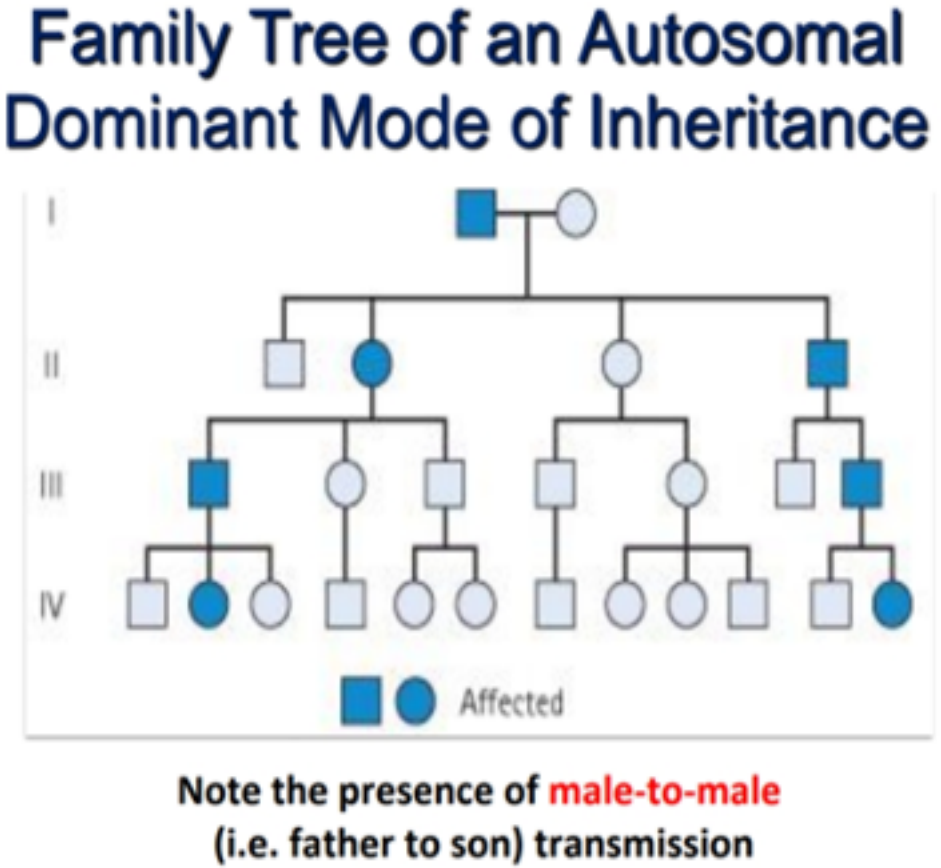
Autosomal Dominant

- The trait (character, disease) appears in every generation.

- Unaffected persons do not transmit the trait to their children.

Examples:

1. **Huntington disease**
2. **Myotonic dystrophy**
3. **Neurofibromatosis type I**
4. **Marfan syndrome**



Autosomal Recessive

- The trait (character, disease) is recessive
- The trait expresses itself only in **homozygous** state
- Unaffected persons (heterozygotes) may have affected children (if the other parent is (heterozygote)
- The parents of the affected child maybe related **(consanguineous)**
- Males and female are **equally** affected
- **Examples: Cystic fibrosis, Phenyketonuria, Sickle cell anaemia, Thalassaemia**

Punnett square showing autosomal recessive inheritance

1) **Both Parents Heterozygous:** *"Aa+Aa"*

- 25% offspring "affected Homozygous" (aa)
- 50% Trait "Heterozygous normal but carrier" (Aa)
- 25% Normal (AA)

Father \ Mother	A	a
A	AA	Aa
a	Aa	aa

1) **One Parent Heterozygous:** *"AA+Aa"*

- 50% normal but carrier "Heterozygous" (Aa)
- 50% Normal (AA)

	A	a
A	AA	Aa
A	AA	Aa

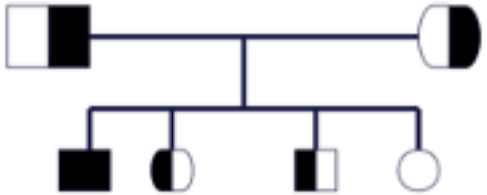
1) **One Parent Homozygous:** *"AA+aa"*

- 100% offspring carriers. (all Aa)

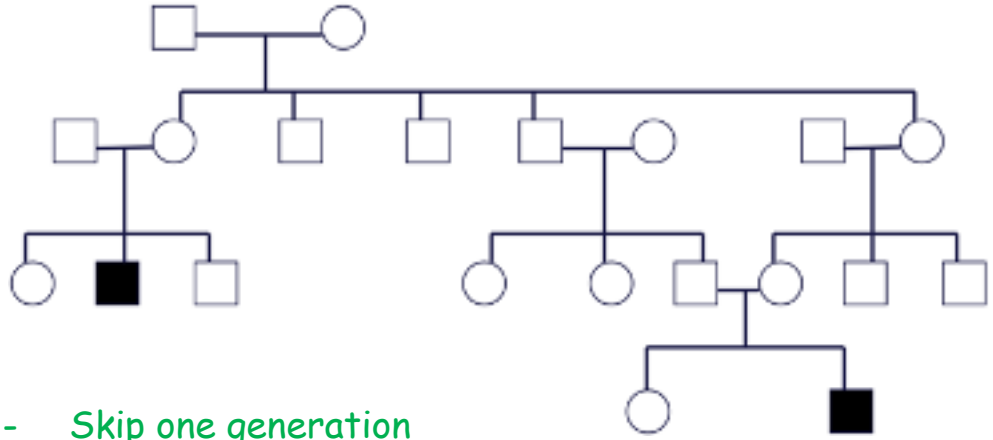
	A	A
a	Aa	Aa
a	Aa	Aa

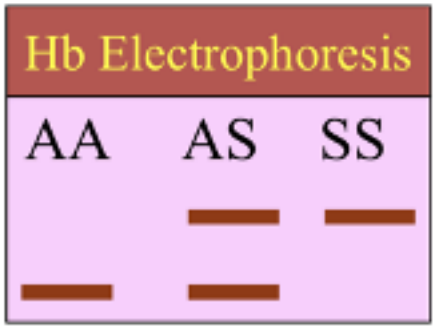
Family tree of an Autosomal recessive disorder

Sickle cell disease (SS)



A family with sickle cell disease - Phenotype





- Skip one generation

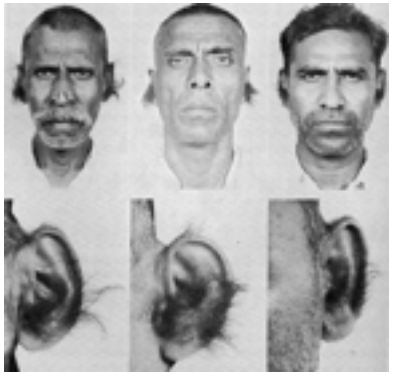
Sex-Linked Inheritance

- This is the inheritance of a gene present on the sex chromosomes.
- The Inheritance Pattern is different from the autosomal inheritance.
- Inheritance differs in males from females.

Two types: X-linked and Y-linked.

Y- linked Inheritance

- The gene is on the Y chromosomes
- The gene is passed from fathers to sons only
- Daughters are not affected
- Hairy ears in India
- Male are Hemizygous the condition exhibits itself whether dominant or recessive



Mother \ Father	X	Y*
X	XX	XY*
X	XX	XY*

X – Linked Inheritance

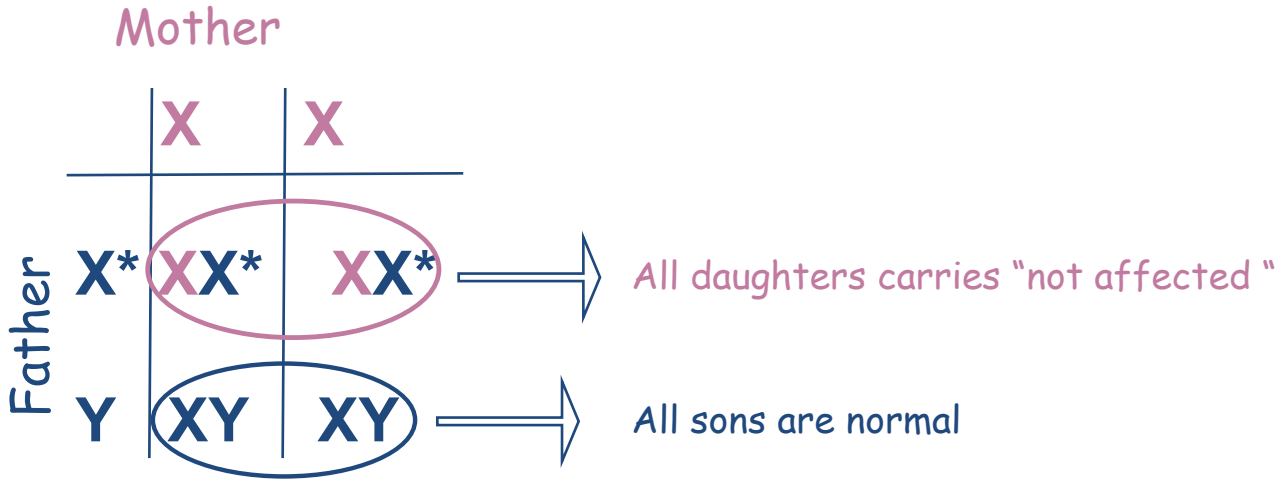
- The gene is present on the X chromosome
- The inheritance follows specific pattern
- Males have one X chromosome, and are hemizygous
- Females have 2 X chromosomes, they may be homozygous or heterozygous
- These disorders may be : recessive or dominant

X – Linked Recessive Inheritance

- The incidence of the X-linked disease is higher in male than in female
- The trait is passed from an affected man through all his daughters to half their sons
- The trait is never transmitted directly from father to sons
- An affected women has affected sons and carrier daughters
- X - Linked Recessive Disorders: Albinism, Fragile X syndrome, Hemophilia, Muscular dystrophy, Retinitis pigmentosa

X – Linked Recessive Inheritance

(1) Normal female "mother" & Affected male "father"



(2) carrier female, normal male

Mother

Father	X*	X
X	XX*	XX
y	X*y	Xy

50% sons are affected
50% daughters carriers

(3) Homozygous female, normal male

-all daughters carriers
-all sons affected

X -Linked Dominant Disorders

- The gene is on X Chromosome and is dominant
- The trait occurs at the same frequency in both males and females
- Hemizygous male and heterozygous females express the disease.

Punnett square showing X – linked dominant type of Inheritance

(1)Affected male and normal female

Mother

Father		X	X
	X*	X*X	X*X
	Y	XY	XY

All daughters affected, all sons normal

(2)Affected female (heterozygous) and normale male

Mother

Father		X*	X
	X	XX*	XX
	Y	X*Y	XY

50% sons and 50% daughters are affected

TAKE HOME MESSAGES

- An accurate determination of the family pedigree is an important part of the workup of every patient
- Pedigrees for single-gene disorders may demonstrate a straight forward, typical mendelian inheritance pattern
- These patterns depend on location of the gene locus on the chromosomal, which may be autosomal or sex chromosome-linked, and whether the phenotype is dominant or recessive

MCQs

Q1)Transmission of genes occurs:

A)Horizontally B)Vertically C)Diagonally D)Between Siblings

Q2)What is the law of dominance:

A)One allele dominant to another B)One allele recessive to another C)Autosomal dominant D)Autosomal Recssive

Q3)Condition resulting in Syndromes happening due to abnormalities in the Y-chromosome is

A)Homozygous B)Sex-linked C)Hemizygous D)Heterozygous

Q4)Changes in the Y chromosome will occur in:

A)Both female and male B)Male only C)Female only D)None

MCQs answers

1) B
2) A
3) C
4) B

Boys team

- Nawaf Alghamdi
- Ahmed Alkhawashki
- Bassam Alasmari
- Rayan Alzahrani
- Khalid AlOsaimii
- Abdulrahman Alswat
- Abdulmalik Mokhtar
- Hadi AlHemsi
- Hesham Alsqabi
- Yazeed Alomar
- Mohammed Benhjji
- Badr Alshahrani
- Homoud Algadheb

Girls team

- Ghaida Alasiri
- Arwa Alqahtani
- Albandri Ahmad
- Aljoharah Albnyan
- Aljohara alshathry
- Alanoud Alshahrani
- Raghad Alasiri
- Renad Alhmidi
- Sara Alharbi
- Taif Almutari
- Abeer Awad
- Ghada Alabdi
- Noura Almassad
- Hind Almutywea

Team Leaders

- Sumo Abdulrahman
- Faisal AlFadel

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