HUMAN GENETICS

Color index:
- Important
- Slides
- Drs' notes
- Explanation
- New terminology

LECTURE : 3

Mode of inheritance

EDITION FILE

OBJECTIVES

By the end of this lecture, students should be able to:

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

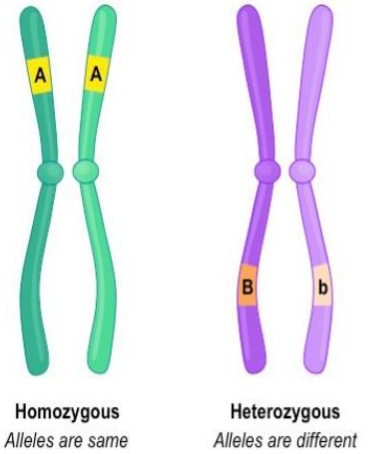
INTERPRETING THE OUTCOMES OF MENDEL'S BREEDING EXPERIMENTS:

- The plant characteristics being studied were each controlled by a pair of **FACTORS** (allels) , one of which was inherited from each parent.
- The pure-bred plants, with two identical genes, used in the initial cross.
- The hybrid F1 plants, each of which has one gene for tallness and one for shortness.
- The genes responsible for these contrasting characteristics are referred to as *allelomorphs*, or **ALLELES** for short.

Genotype:	*AA*	*Bb*	*cc*
	Homozygous for the dominant allele	Heterozygous	Homozygous for the recessive allele

Homozygous The same allele

Heterozygous different alleles

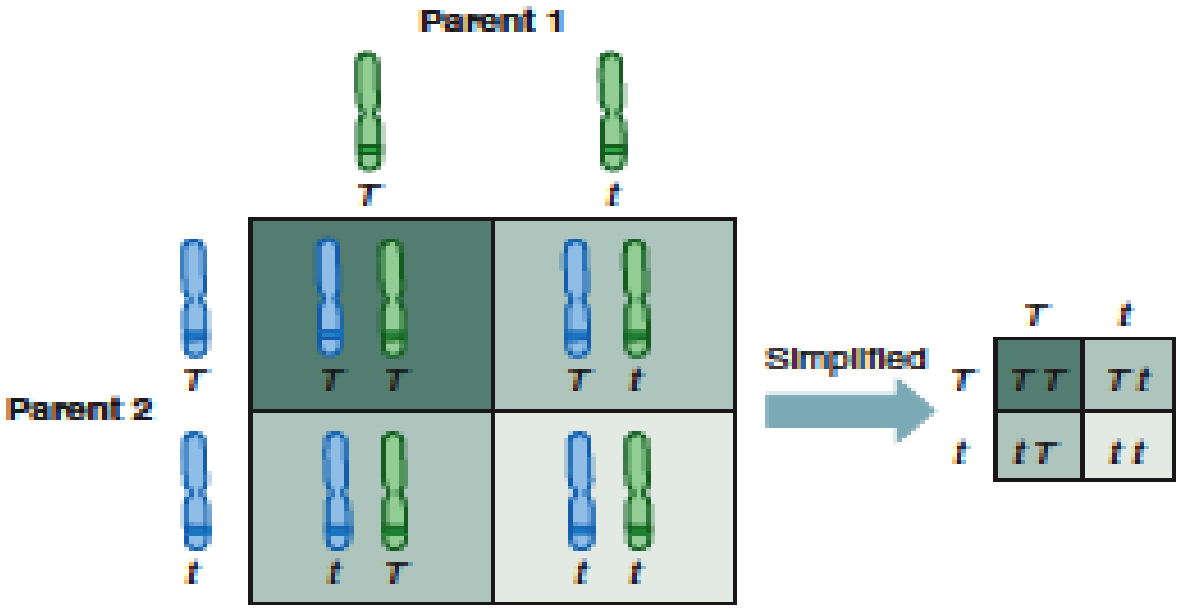


PUNNETT SQUARE

- Each parent can only contribute one allele per gene.
- These genes are found on the chromosomes of gametes of parents.
- Offspring will inherit **2 alleles** to express that gene.

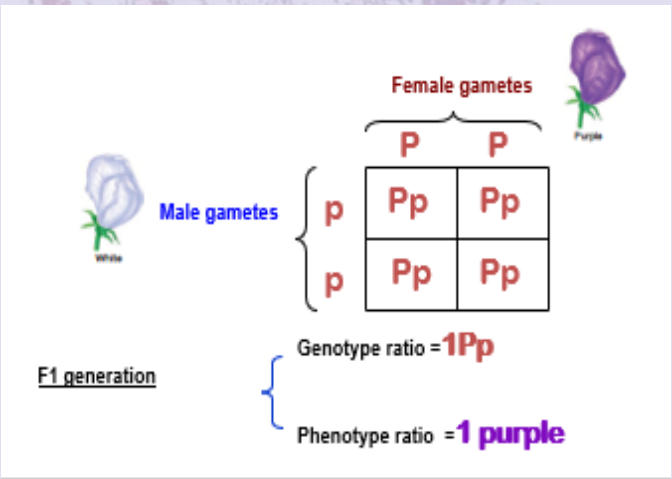
A Punnett Square Can Be Used to Predict the Outcome of simple genetic crosses. A Punnett square illustrates how alleles combine in offspring, To construct a Punnett square, you must know the genotypes of the parents. With this information, the Punnett square enables you to predict the types of offspring the parents are expected to produce and in what proportions.

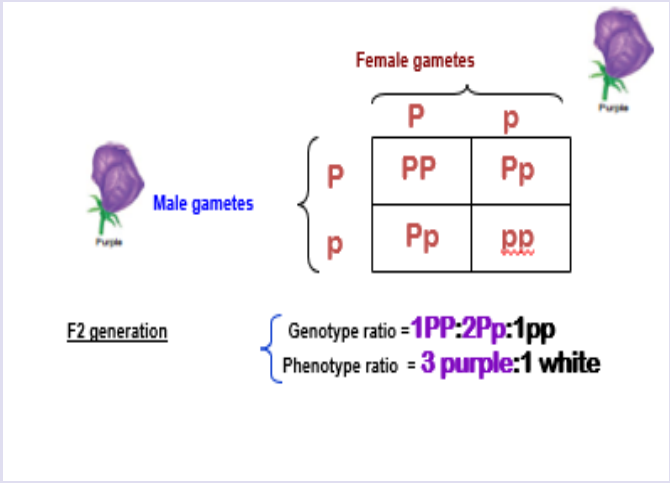
Allele: one of a pair of genes that appear at a particular location on a particular chromosome and control the same characteristic, such as blood type or colorblindness



LAW OF DOMINANCE OR UNIFORMITY & LAW OF SEGREGATION

Law of Dominance or Uniformity	Law of segregation
➢ **COMPLETE DOMINANCE** - one allele is dominant to another allele. ➢ RECALL MENDEL'S 1st EXPERIMENT	RECALL MENDEL'S 2nd EXPERIMENT
CROSS: Pure bred purple female x White male	CROSS: <u>Two F1 generation offspring with each other</u>
P1 generation = <u>PP</u> x <u>pp</u>	P1 generation = <u>Pp</u> x <u>Pp</u>



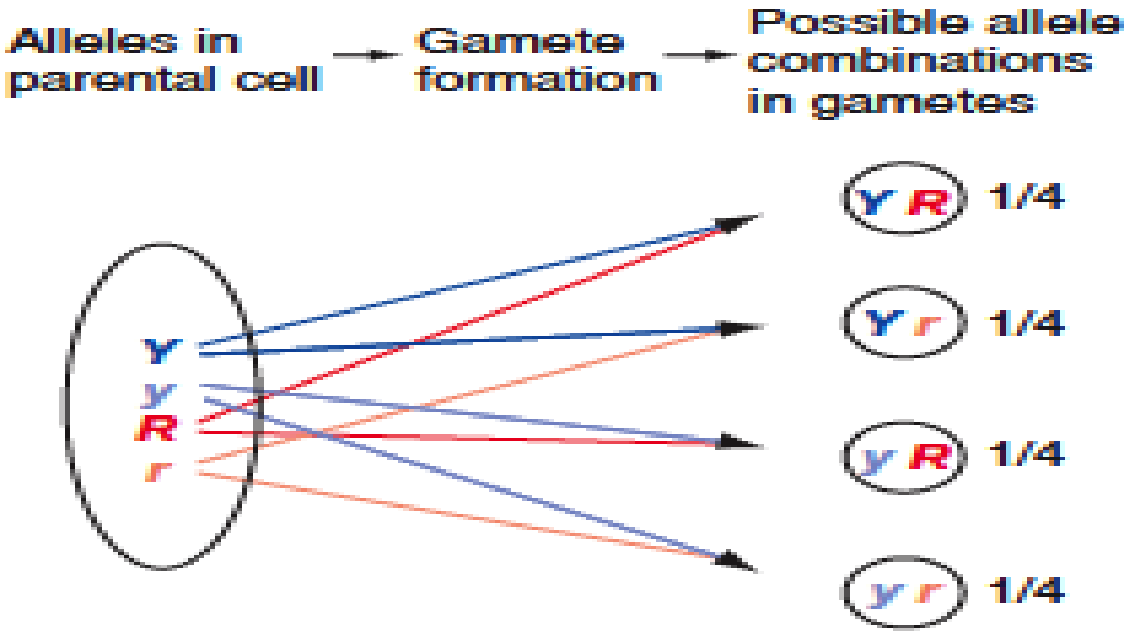


Q1: Mixing the homozygous will result as ….?
- all heterozygous

Q2: Mixing the heterozygous will result as…..?
- half of offspring homozygous and the other half is heterozygous

LAW OF INDEPENDENT ASSORTMENT

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



هذي السلايد للشرح فقط وليست موجودة في المحاضرة

Mendel law of dominance:

- There are recessive and dominant alleles
- In a cross of parents that are pure for contrasting traits, only one form of the trait will appear in the next generation

All offspring will be heterozygous and express only the dominant trait.

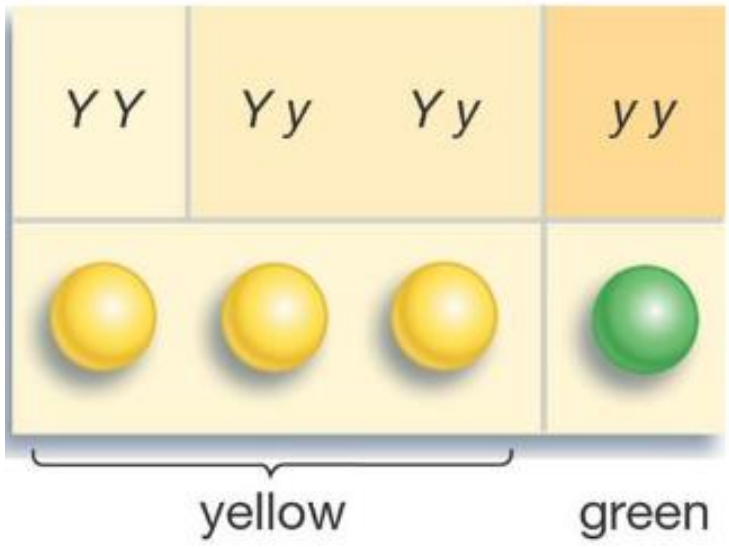
Mendel's law of segregation states:

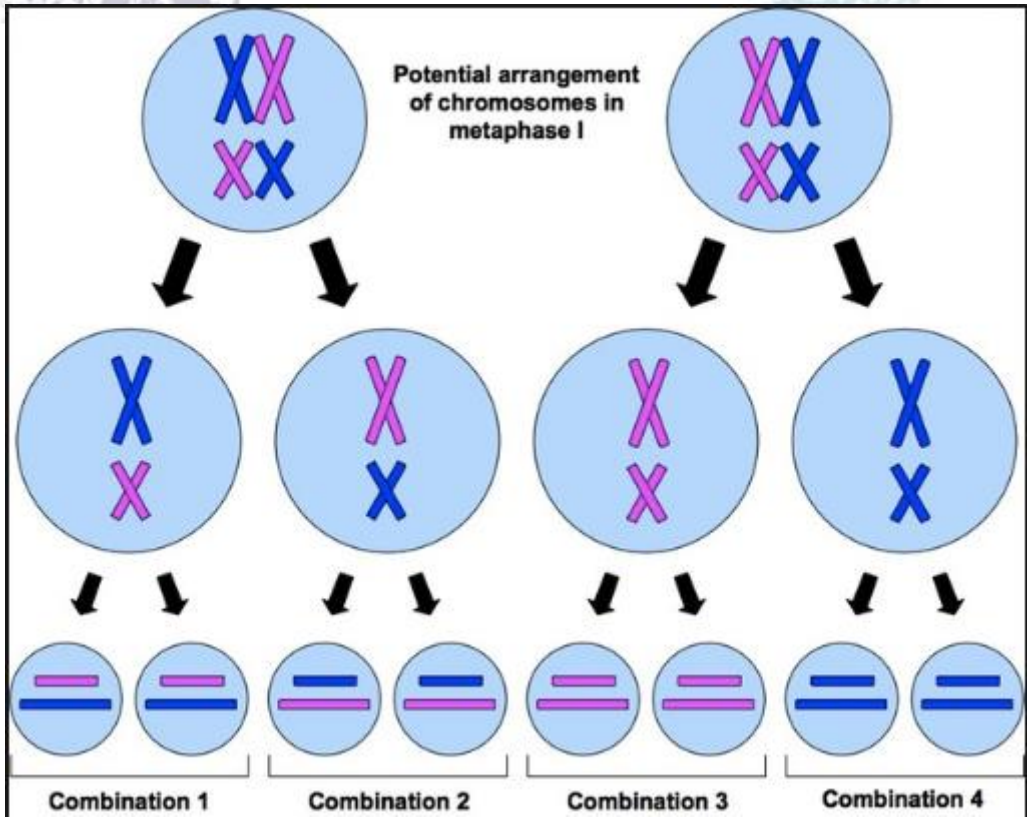
The two copies of a gene segregate (or separate) from each other during transmission from parent to offspring.

Therefore, only one copy of each gene is found in a gamete. At fertilization, two gametes combine randomly, potentially producing different allelic combinations

Mendel law of independent assortment :

The inheritance pattern of trait will not effect the inheritance pattern of another



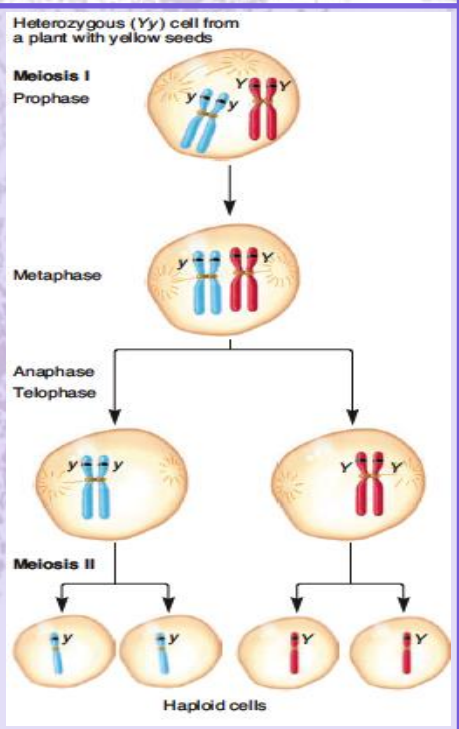


THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES

How chromosomal transmission is related to the patterns of inheritance observed by Mendel?

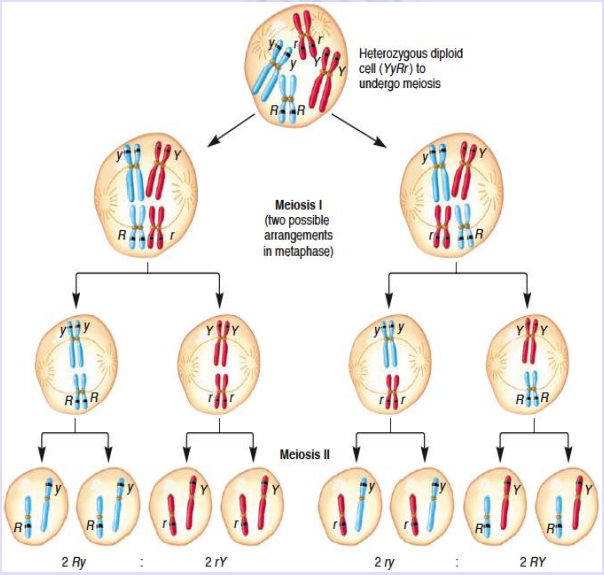
Mendel's law of segregation can be explained by the segregation of homologs during meiosis

- 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.



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

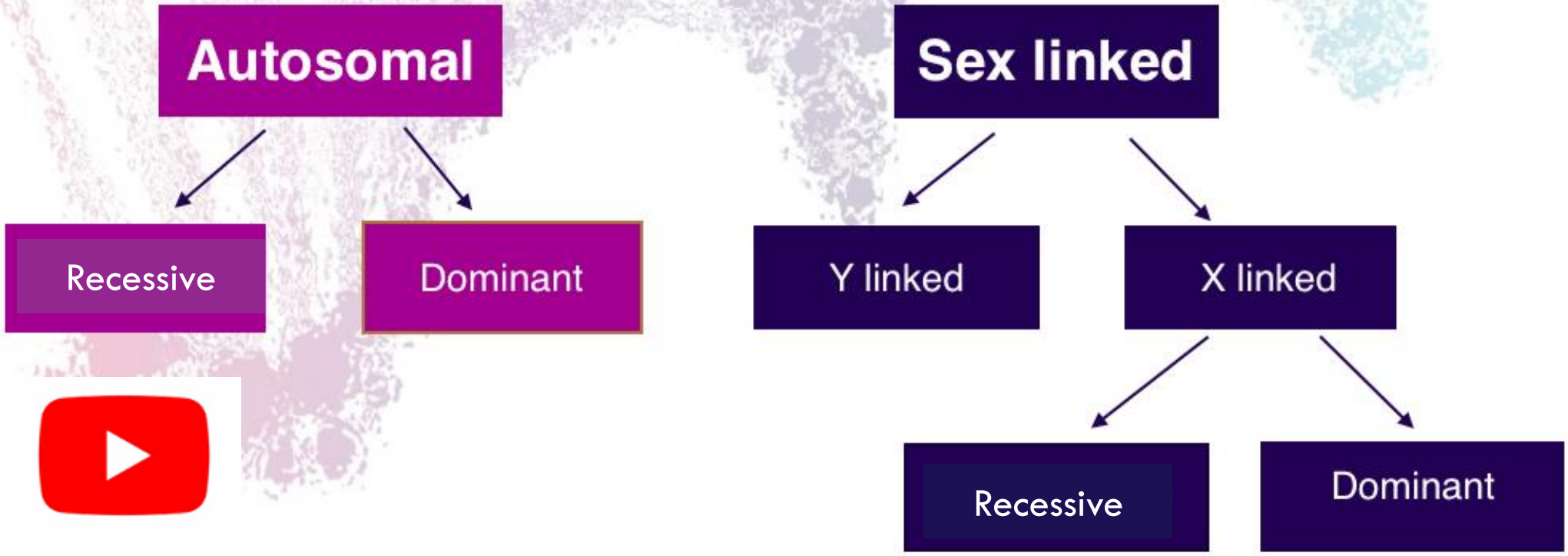
- This figure shows the assortment of two genes located on two different chromosomes, using pea seed color and shape as an example (YyRr).
- During metaphase of meiosis I, different possible arrangements of the homologs within bivalents can lead to different combinations of the alleles in the resulting gametes.

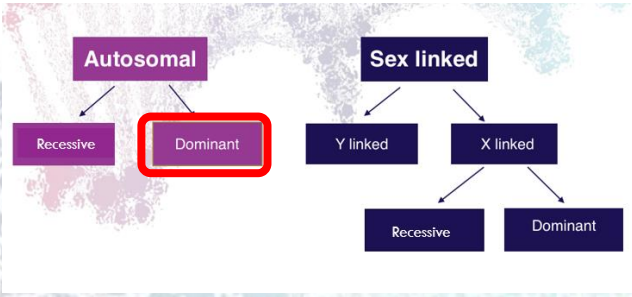


- For example, on the left, the dominant R allele has sorted with the recessive y allele; on the right, the dominant R allele has sorted with the dominant Y allele.

Important

MODES OF INHEREITANCE FOR SINGLE GENE DISORDERS .

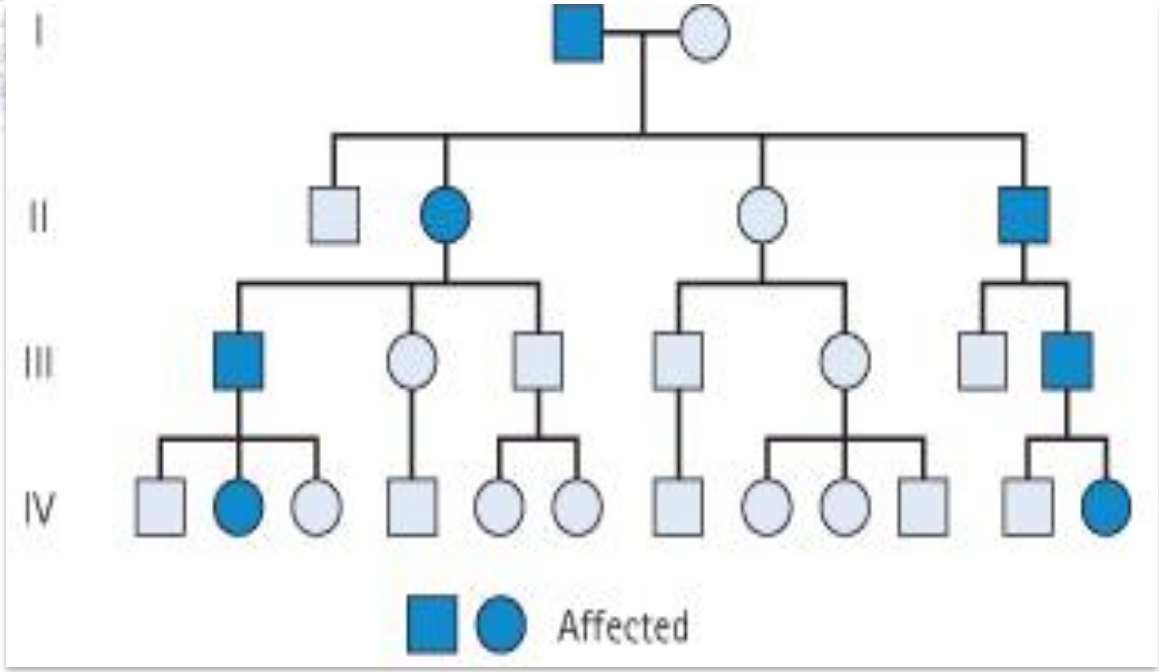




1- AUTOSOMAL DOMINANT

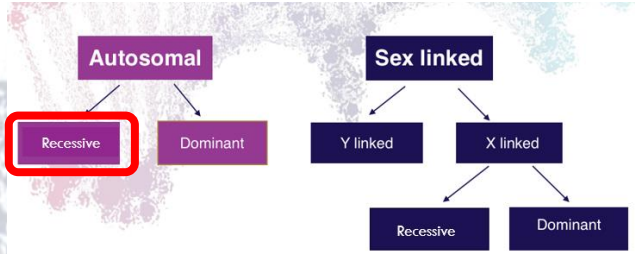
- The trait (character, disease) appears **in every generation.**
- Unaffected persons do not transmit the trait to their children.
- Examples: Huntington disease, Myotonic dystrophy, Neurofibromatosis type 1, Marfan syndrome etc.

Family tree of an autosomal dominant mode of inheritance



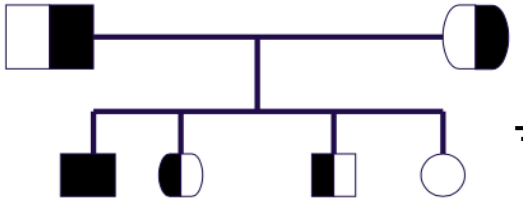
This pedigree is dominant because in EVERY generation there is effected individuals

2-AUTOSOMAL RECESSIVE



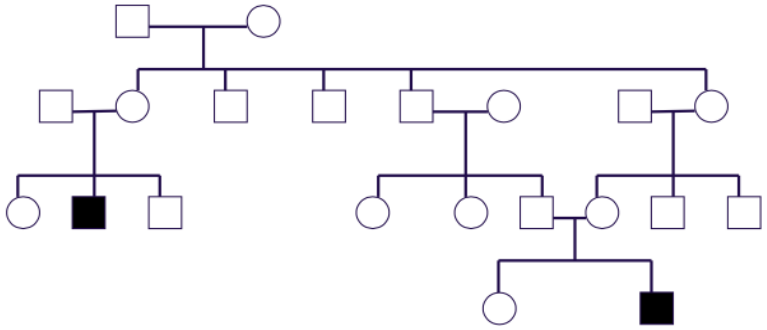
- The trait (character, disease) is recessive
- The trait expresses itself only in homozygous state
- Unaffected persons (heterozygotes) may have affected children (if the other parent is heterozygote)
- The parents of the affected child maybe related (consanguineous)
- Males and female are equally affected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

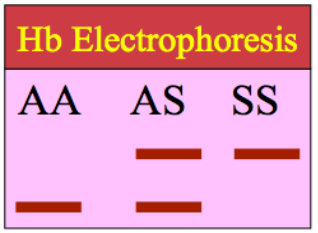
(Family tree of an Autosomal recessive disorder
Sickle cell disease (SS)



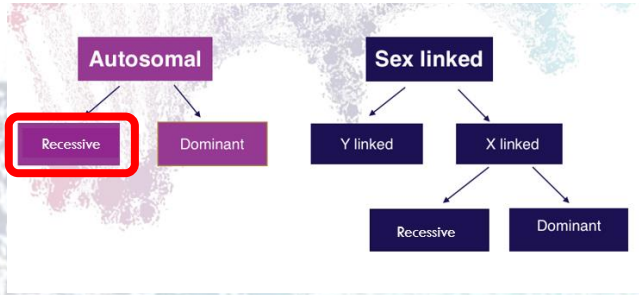
mother \ father	A	S
A	AA	AS
S	AS	SS

A family with sickle cell disease -Phenotype





PUNNETT SQUARE SHOWING AUTOSOMAL RECESSIVE



Mother

Father

	A	a
A	AA	Aa
a	Aa	aa

	A	a
A	AA	Aa
A	AA	Aa

	A	A
a	Aa	Aa
a	Aa	Aa

1-) Both Parents Heterozygous:

- 25% offspring "affected Homozygous"
- 50% Trait "Heterozygous normal but carrier"
- 25% Normal

2-) One Parents Heterozygous:

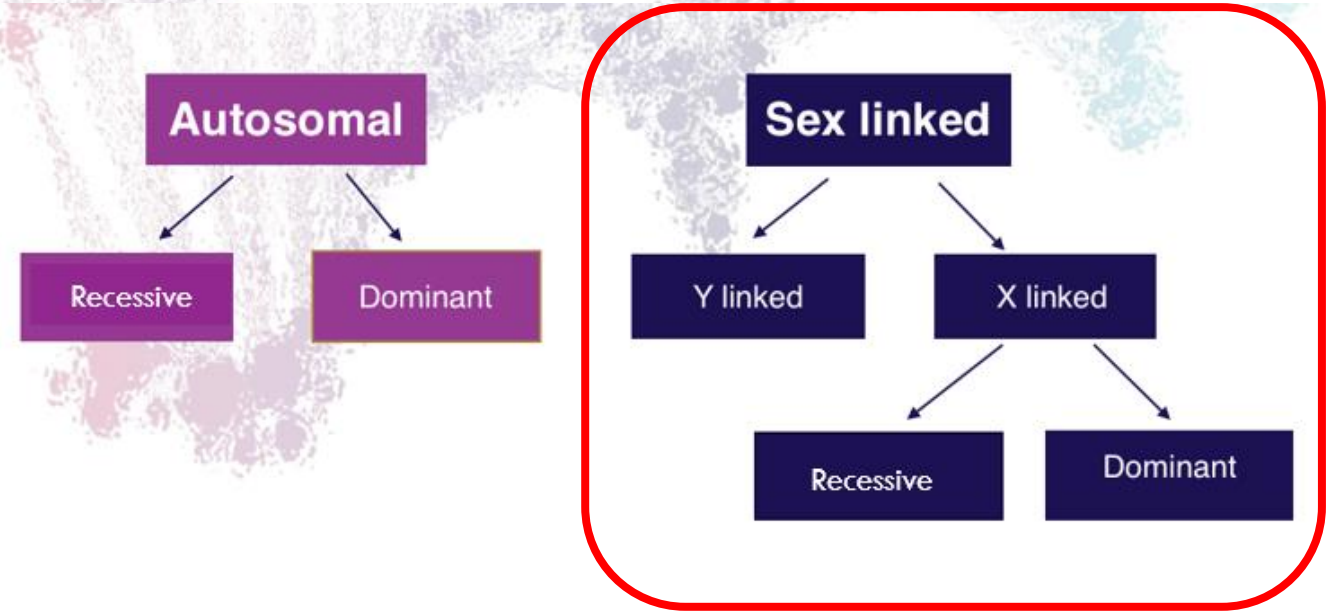
- 50% normal but carrier "heterozygous"
- 50% normal

3-) One Parents Homozygous:

- 100% offspring is carriers.

SEX- LINKED INHERITANCE

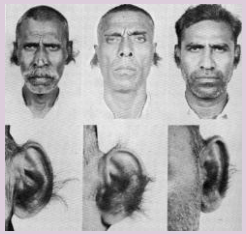
- This is the inheritance of a gene present on the sex chromosomes.
- The Inheritance Pattern is different from the autosomal inheritance.
- Inheritance differs in males from females.



Y-linked

- The gene is on the Y chromosomes
- The gene is passed from fathers to sons only
- Daughters are not affected

Example :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:

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

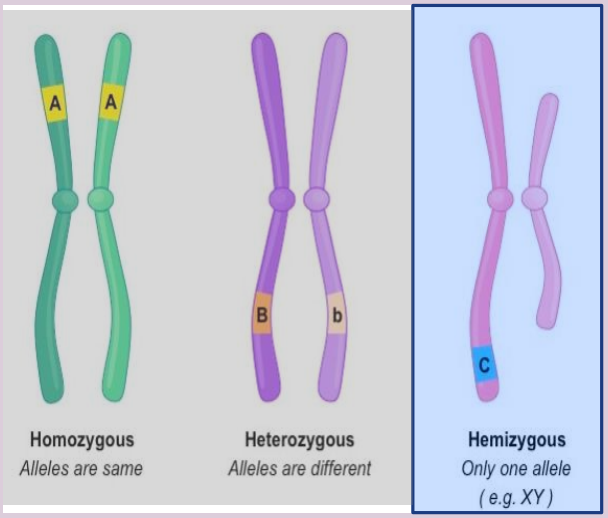
X-linked Recessive Inheritance :

- The incidence of the X-linked disease is higher in male than in female. Because males have only one X chromosome, so they can't be *healthy carriers*, either effected or healthy.
- 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. Because the father passes only his Y chromosome to all his sons, so he can't pass the mutation to them.
- An affected women has affected sons and carrier daughters.

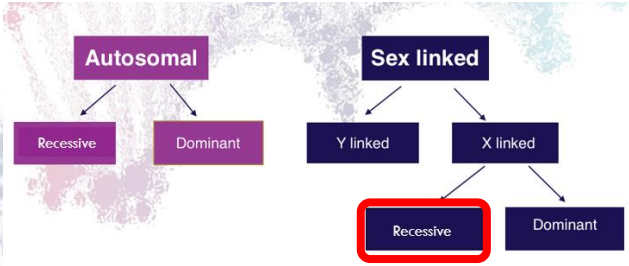
X - Linked Recessive Disorders: Albinism, Fragile X syndrome, Hemophilia, Muscular dystrophy, Retinitis pigmentosa.

X-linked Dominant Inheritance:

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



X – LINKED RECESSIVE INHERITANCE



(1) Normal female, affected male

Mother

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

All sons are normal

All daughters carriers "not affected"

(2) Carrier female, normal male:

Mother

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

50% sons affected

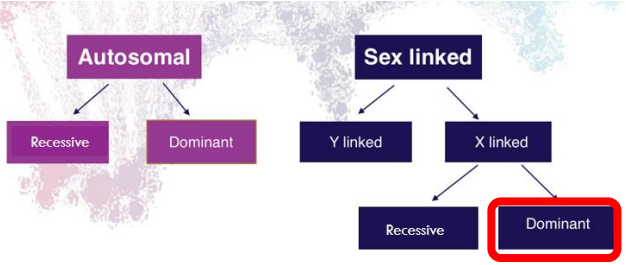
50% daughters carriers

(3) Homozygous female, normal male:

	X*	X*
X	X*X	X*X
Y	X*Y	X*Y

- All daughters carriers.
- All sons affected.

X – LINKED DOMINANT 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 normal male:

Mother

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

50% of sons are effected
50% of daughters are affected

TAKE HOME MESSAGE:

- 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 straightforward, 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

MCQ

1-the pure-bred plants, with two identical genes, Used in the initial cross would now be referred To as:
A. Homozygous
B. Heterozygous
C. Genetic twin

2-a person with these alleles (TT) is known to be:
A. Homozygous recessive
B. Homozygous dominant
C. Heterozygous

3-the physical expression of a character is called:
A. Phenotype
B. Morphology
C. Genotype

4-A trait or disorder determined by a gene on one of the sex chromosomes is said to be:
a-autosomal inheritance
b-recessive inheritance
c-sex-linked inheritance

5-an individual inherits .... allele for each gene (character)
A. 2
b. 3
c. 4

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

تمّ بحمدِ الله

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