Modes of Inheritance

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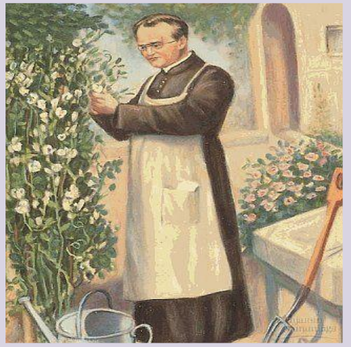

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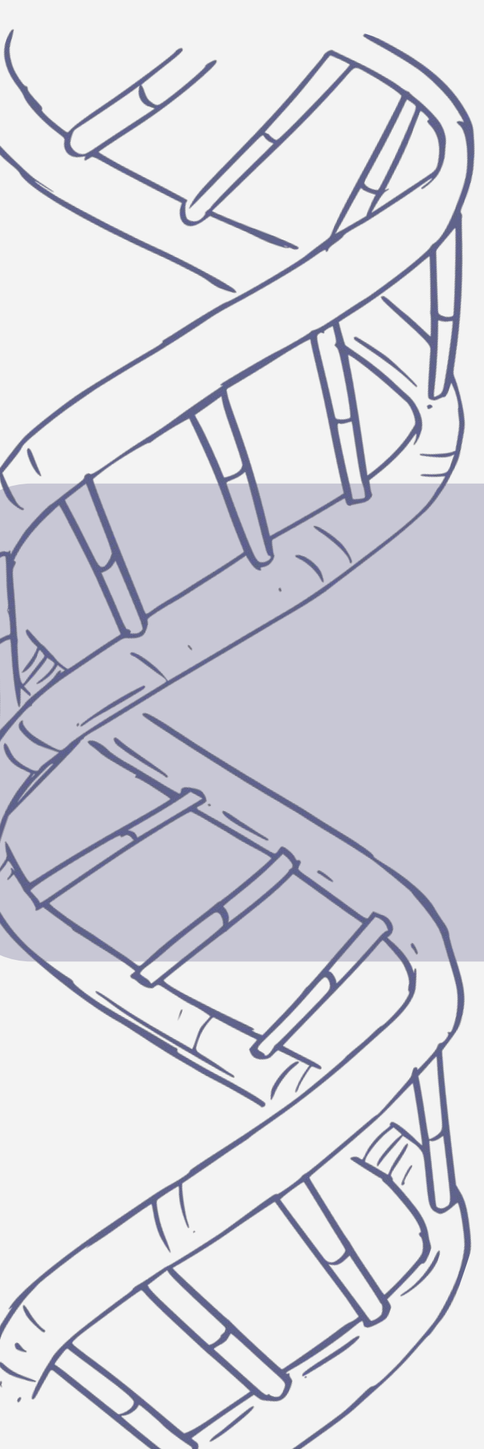
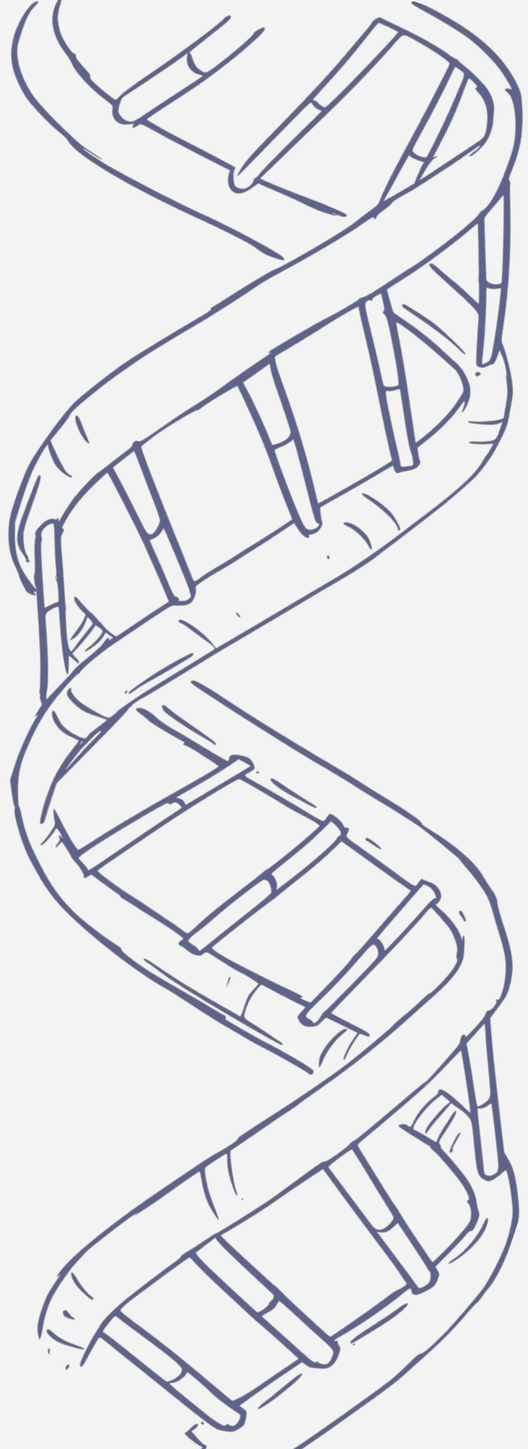


“Success is the sum of small efforts, repeated.”

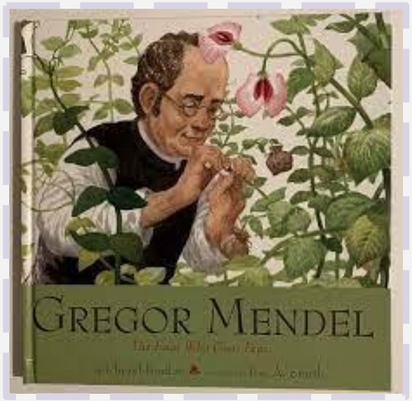


Objectives

- 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 (tools that help us to understand how genes are passed from one generation to another)

Father of genetics

Born in 1822
He was a monk and teacher.



Discovered some of the basic laws of heredity.

He published his work entitled “Experiments on Plant Hybrids” in 1866 However, it was largely ignored.

He died in 1884 with his work still unnoticed, his work was rediscovered in 1900.

Interpreting the outcomes of Mendel's breeding experiments:

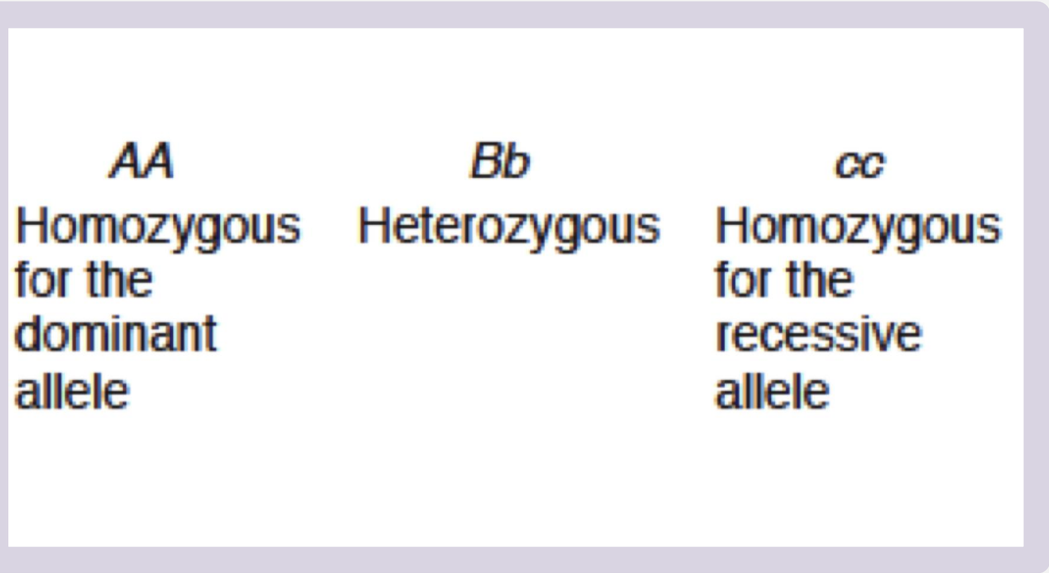
- The plant characteristics being studied were each controlled by a pair of **factors** (genes) , one of which was inherited from each parent.
- The pure-bred plants, with two identical genes, used in the initial cross would now be referred to as **homozygous**.
- The hybrid F1 plants, each of which 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.

Team 434: An individual inherits 2 alleles for each gene (character), one from each parent. If the 2 alleles are the same, the individual is homozygous for that gene. If the alleles are different, the individual is heterozygous.

Genotype

Homozygous (same alleles)
For **dominant** alleles e.g. **AA**. For **recessive** alleles e.g. **aa**.

Heterozygous (different alleles)
It's combination of recessive and dominant alleles e.g. **Aa**.



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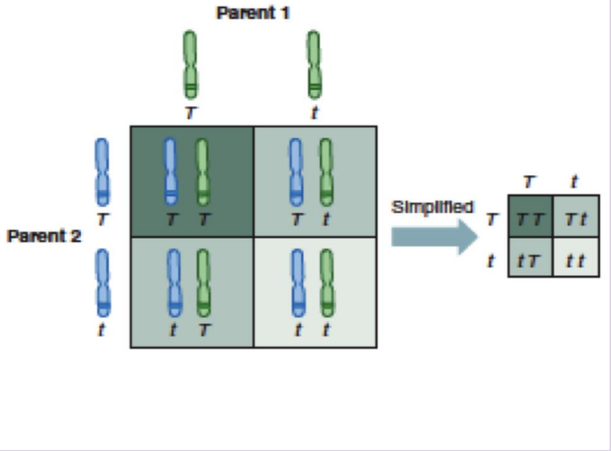
[سورة الشرح: 5,6] .

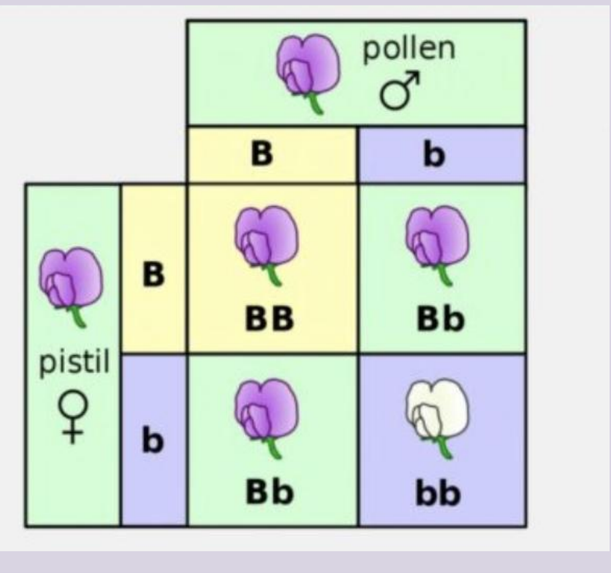
Punnett square

1 Each parent can only contribute one allele per gene.

2 These genes are found on the chromosomes of gametes of parents.

3 Offspring will inherit 2 alleles to express that gene.

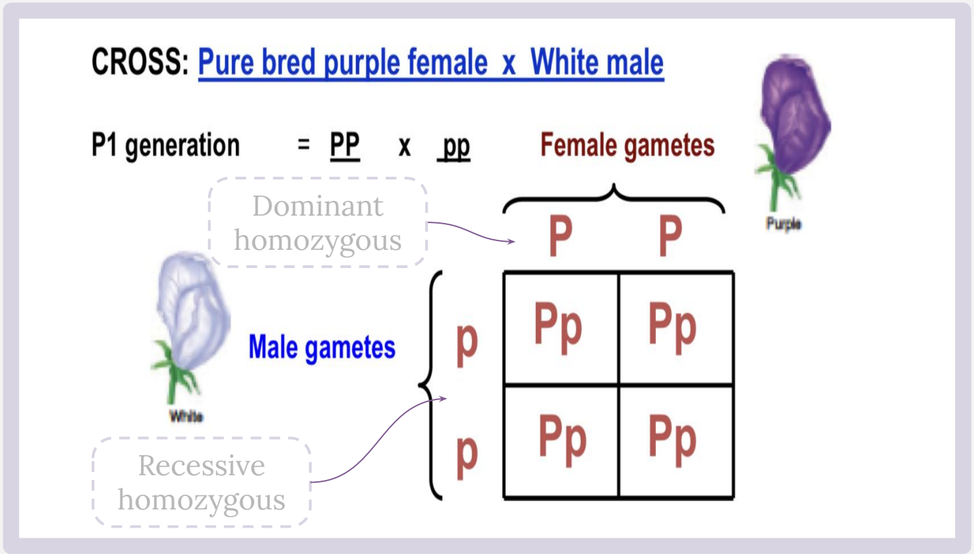




Law of Dominance or Uniformity

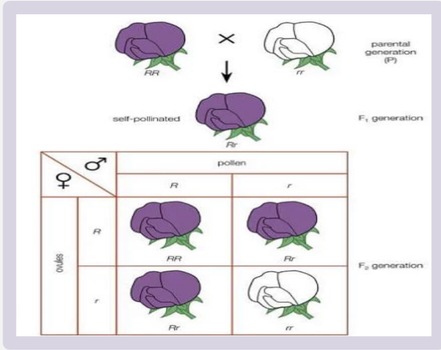
Team 437:
In a cross of parents that are pure for contrasting traits, only one form of the trait will appear in the next generation
All offspring will be heterozygous and express only the dominant trait.

1. COMPLETE DOMINANCE: one allele is dominant to another allele
2. Recall Mendel's 1st experiment.
3. CROSS: Pure-bred purple female x White male.
4. P1 generation = PP x pp
5. F1 generation
   - **Genotype:** ratio = 1Pp , percentage = 100% Pp
   - **Phenotype:** ratio = 1 purple , percentage = 100% purple



Law of Segregation:

For every trait, there are two alleles that get segregated and only one of these is passed to the offspring



law of segregation:

Definition:
During the formation of gamete, each gene separates from each other so that each gamete carries only one allele for each gene.
the alleles of a given locus segregate into separate gametes. (Team 439)

Recall
Mendel's 2nd experiment.

Cross
Two F1 generation offspring with each other.

P1 generation
= Pp x Pp

F1 generation

Genotype:

ratio = 1PP: 2Pp:1pp ,

PCT = 25% PP, 50% Pp, 25% pp

Phenotype:

ratio = 3 purple:1 white,

PCT = 75% purple, 25% white

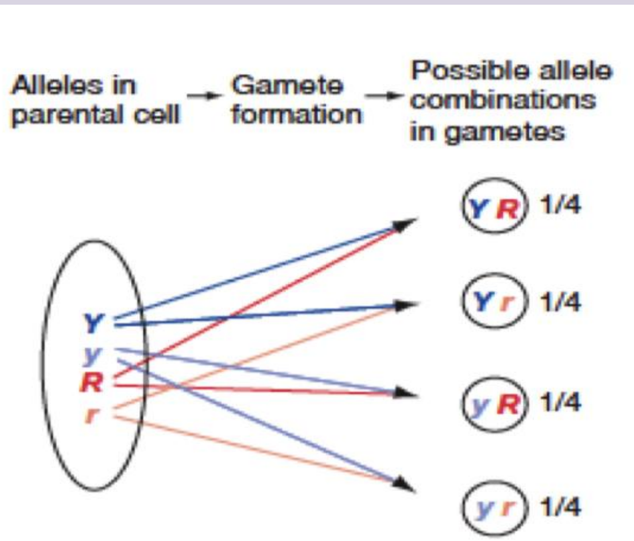
Law of Independent Assortment

Definition

- During gamete formation, different pairs of alleles (different traits) segregate independently of each other

Interpretation

- In a dihybrid cross(which means studying 2 traits together) , 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.

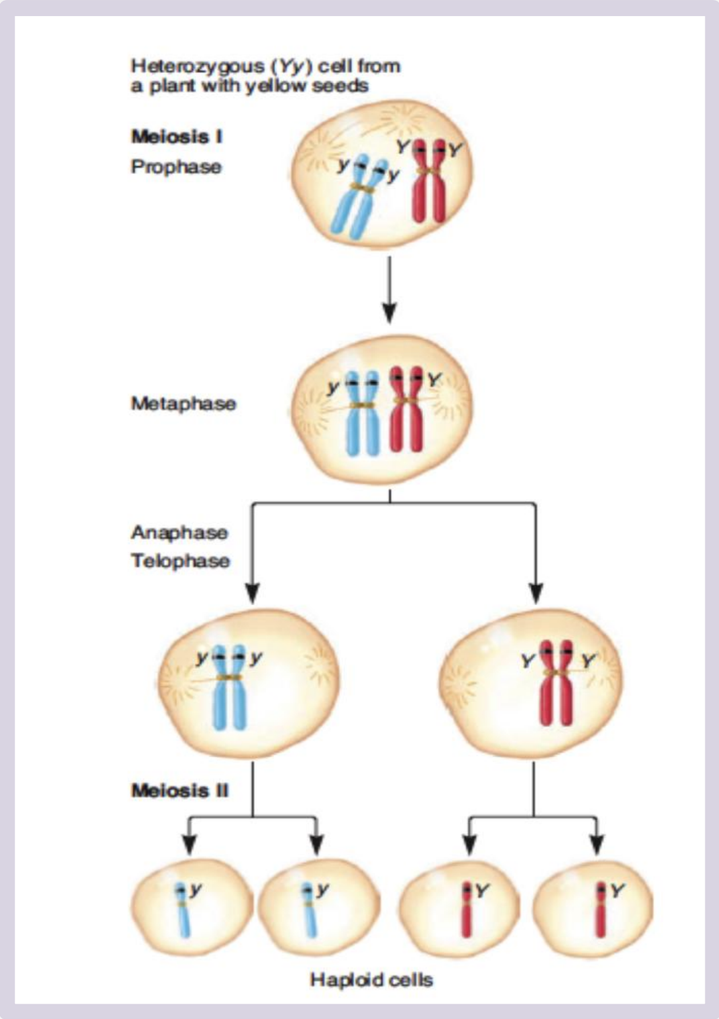


Team 436: - The alleles for different genes usually separate and inherited independently of one another. - So, in dihybrid crosses you will see more combinations of the two genes.

THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES

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

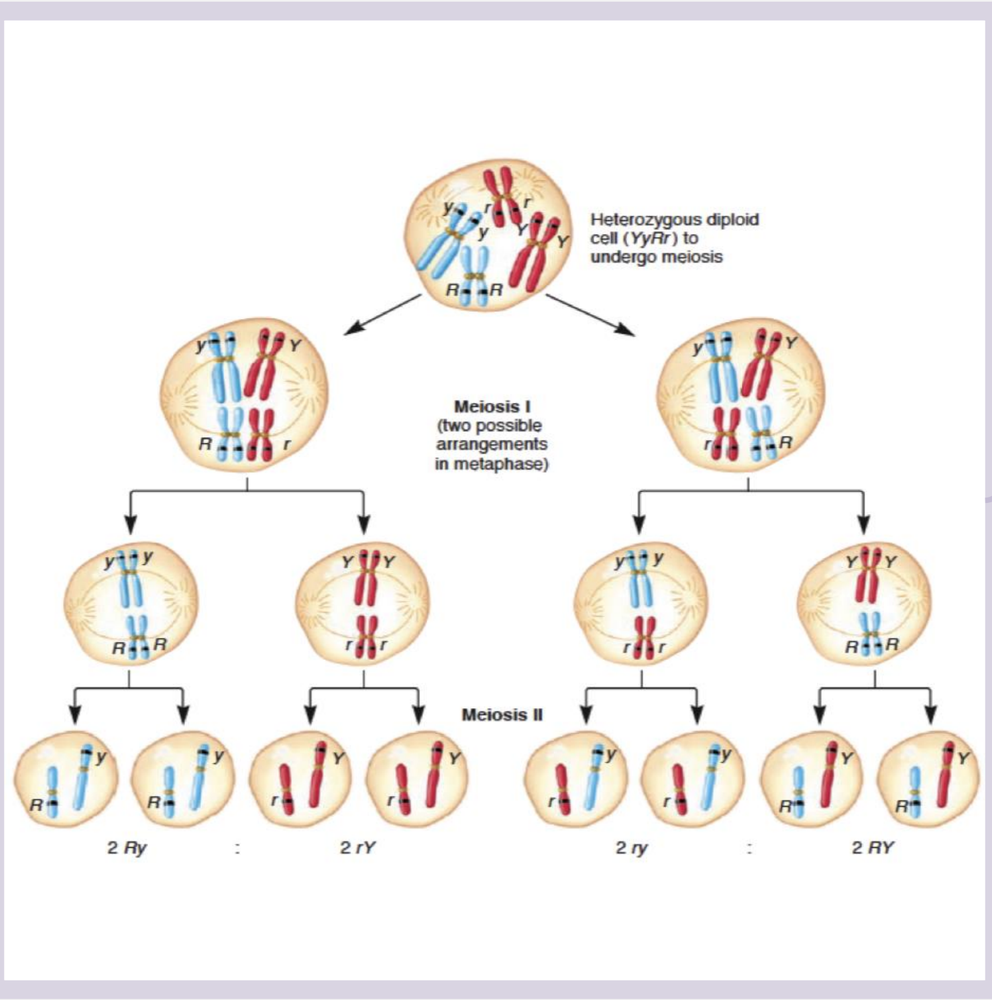
Mendel's law of segregation can be explained by the segregation of homologs during meiosis



- The two copies of a gene are contained on homologous chromosomes. (chromosomes carrying the same gene/coding for the same trait)
- 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.

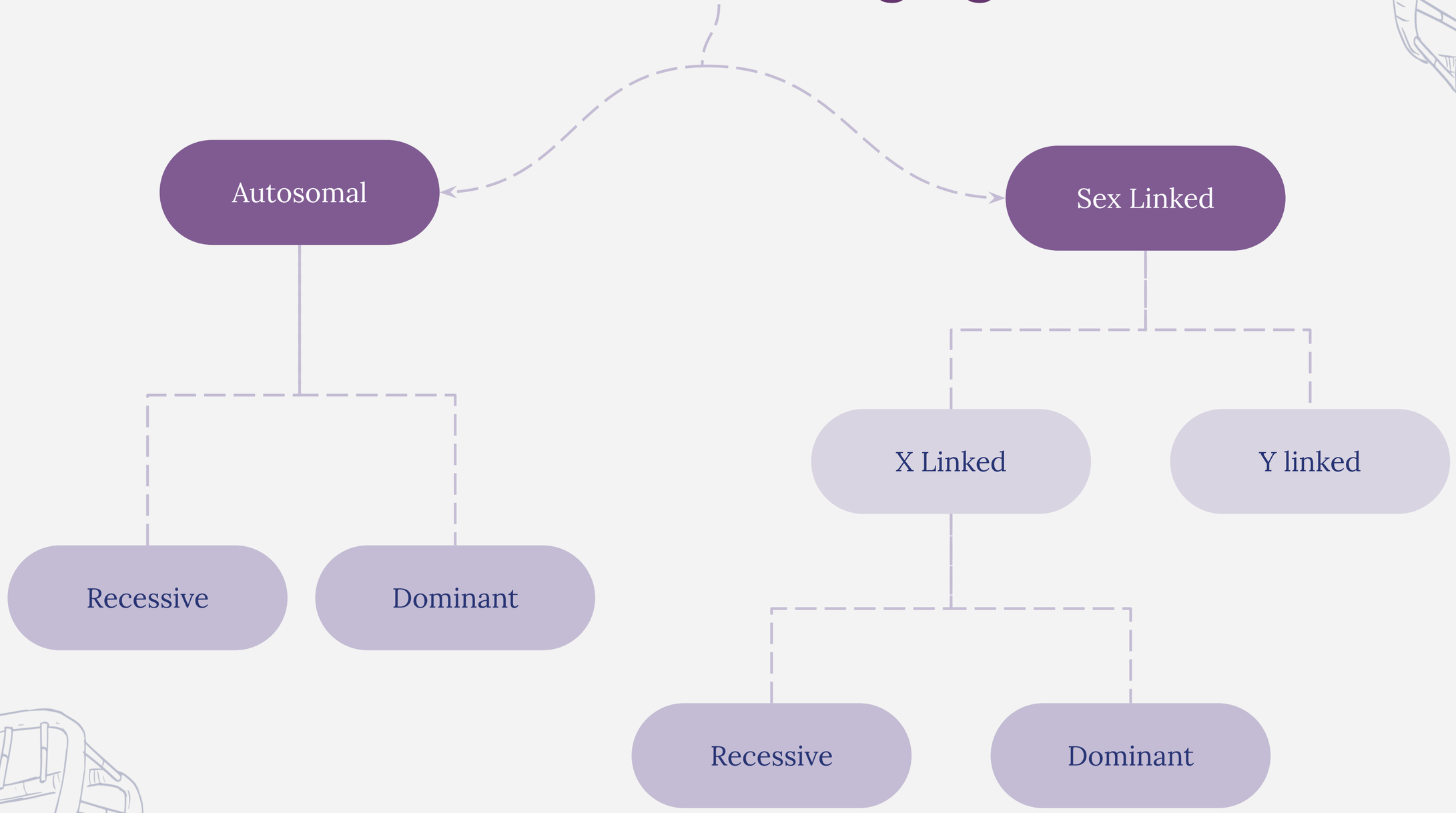
Mendel's law of Independent assortment can be explained by the random alignment of bivalents during metaphase of meiosis I

doctor's note:
Bivalents mean having two sets of chromosomes/ diploid.

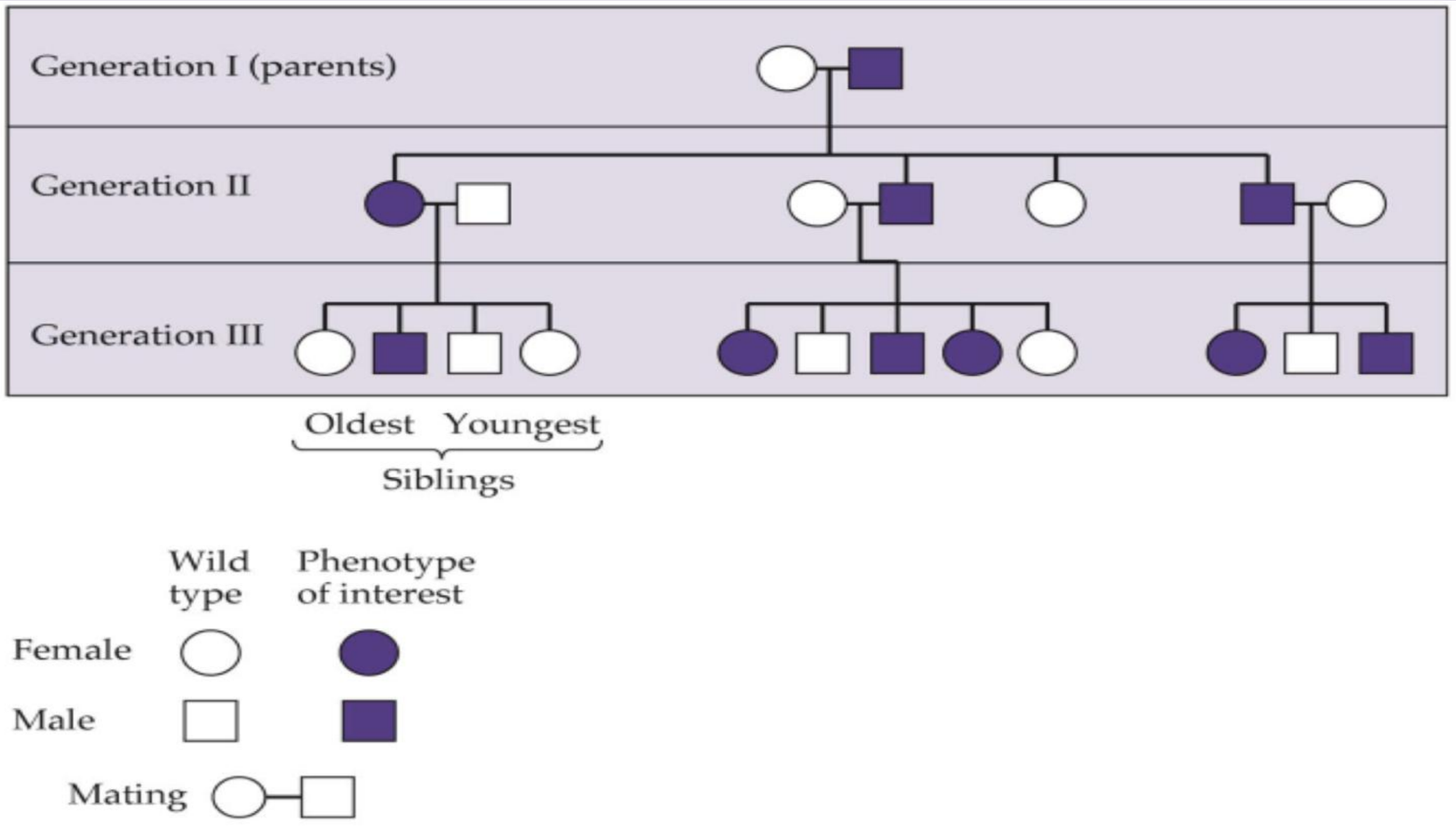


- This figure shows the assortment of two genes located on two different chromosomes, using **pea seed color and shape** as an example (**Yy** (for the color) **Rr** (for the shape)).
  - Y—> yellow color (dominant)
  - y —> green color (recessive)
  - R —> Round shape (dominant)
  - r —> wrinkled shape (recessive)
- 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 disorder

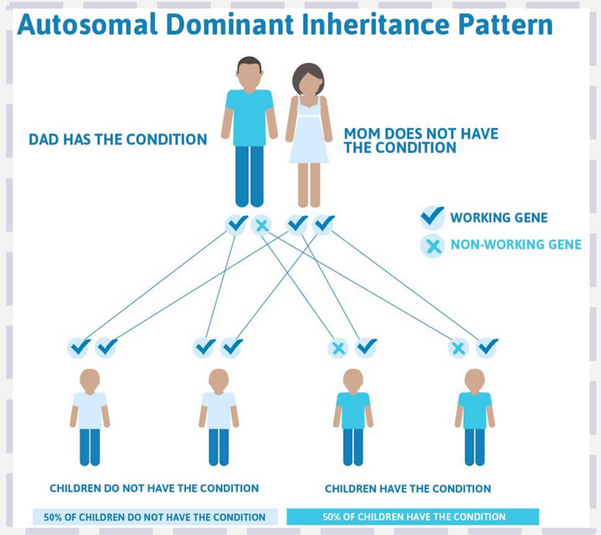


A Pedigree Analysis

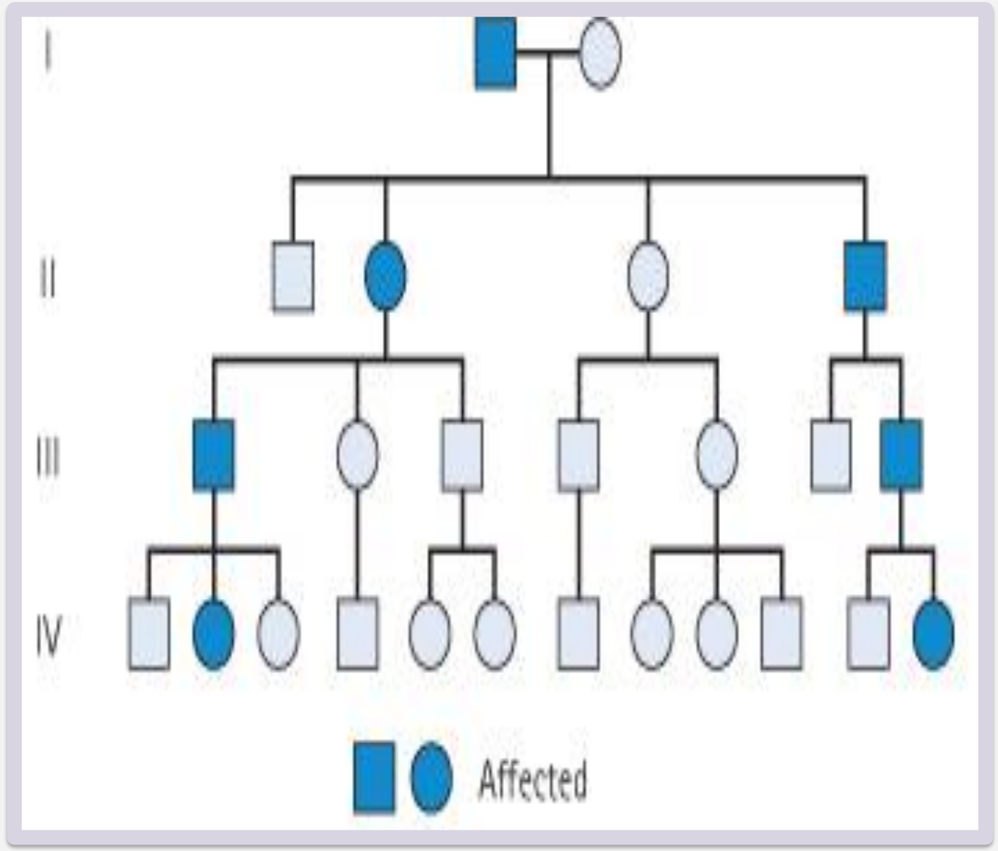


Autosomal Dominant

- 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



Note the presence of male-to-male (i.e. Father son) transmission (this is a key feature for Autosomal Dominant)

Autosomal Recessive

1-The trait (character, disease) is recessive (when both genes are recessive)

2-The trait expresses itself only in homozygous state

3-Unaffected persons (heterozygotes) may have affected children (if the other parent is heterozygous)

4-The parents of the affected child may be related (consanguineous)

5-Males and female are equally affected

• Examples:

Cystic fibrosis, Phenylketonuria, Sickle cell anaemia, Thalassaemia etc.

1) Both Parents Heterozygous:

25% offspring "affected Homozygous recessive"

50% Trait "Heterozygous normal but carrier"

25% Normal Homozygous Dominant

Father \ Mother	**A**	**a**
A	AA	Aa
a	Aa	aa

2) One parent Heterozygous:

(Mother)

and the other is normal

50% normal but carrier "Heterozygous"

50% Normal

Father \ Mother	**A**	**a**
A	AA	Aa
A	AA	Aa

3) One parent Homozygous:

(Father)

and the other is normal

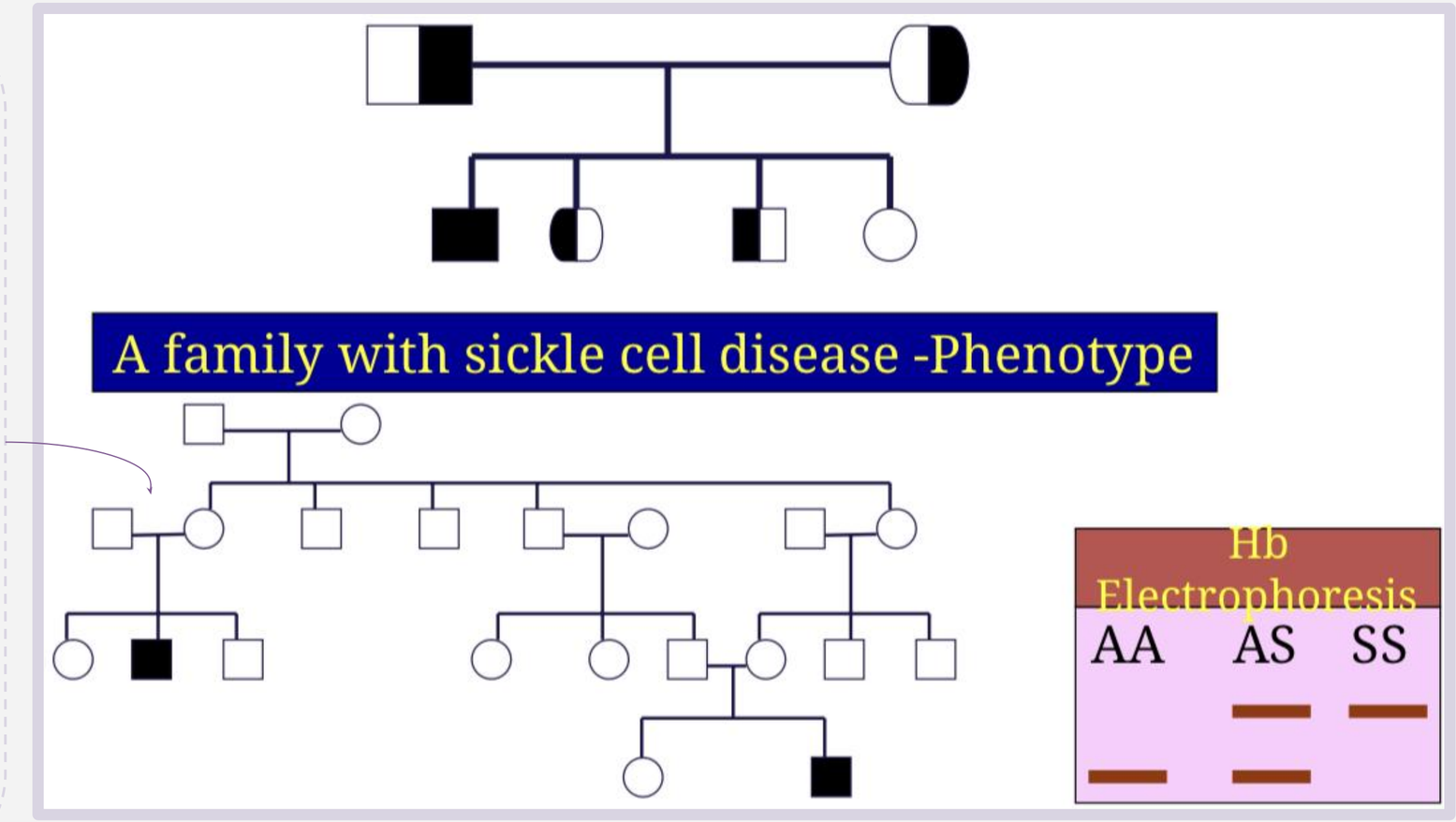
100% offspring carriers.

Father \ Mother	**A**	**A**
a	Aa	Aa
a	Aa	Aa

Autosomal Recessive

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

Unaffected parents have affected an offspring here (it's the condition in which one parent is **heterozygous** and the other parent is also **heterozygous**) so you immediately recognize that this is an autosomal recessive pedigree

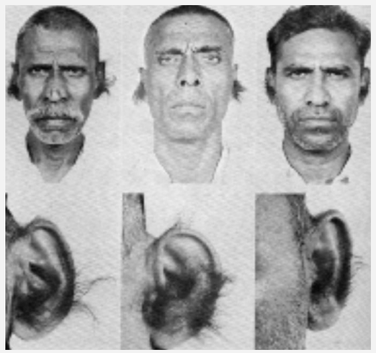


Sex-Linked Inheritance

- 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.

There are 2 types of Sex-Linked Inheritance:

Father / Mother	X	*Y
X	XX	*XY
X	XX	*XY



1) 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(it means only one gene is responsible for the expression of a disease), and the condition exhibits itself whether dominant or recessive

2) 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**

Sex-Linked Inheritance

1) 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)
- 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

1) Normal Female, Affected Male

All sons are normal

All daughters are carriers “not affected”

Mother / Father	X	X
*X	X*X	X*X
Y	XY	XY

2) Carrier Female, Normal Male

50% of sons are affected

50% of daughters are carriers “not affected”

Mother / Father	*X	X
X	*XX	XX
Y	X*Y	XY

3). Homozygous Female, Normal Male

All sons are affected

All daughters are carriers “not affected’

Mother / Father	*X	*X
X	X*X	X*X
Y	X*Y	X*Y

Sex-Linked Inheritance

2) 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.

Homozygous females will definitely express the disease

1- Normal Female, Affected Male

All sons are normal

All daughters are affected

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

2-Affected Female (heterozygous), Normal Male

50% of sons are affected

50% of daughters are affected

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

Take Home Message

- An Accurate Determination Of The Family Pedigree is an important part of the workup of every patient

- Pedigree 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

MCQs:

1 The law of Mendel which describes the separation of two alleles independently is:			
A- Law of Independent assortment.	B- Law of Segregation.	C- Law of Dominance.	D- Law of Codominance
2 What is the law of dominance?			
A-Autosomal Dominant	B-Autosomal Recessive	C-One allele dominant to another	D-One allele recessive to another
3 Hairy ears is India is an example of?			
A-X-linked Inheritance	B-Y-Linked Inheritance	C-Autosomal Dominant	D-Autosomal Recessive
4 Affected male with X-linked Dominant disease and normal female, their sons will be:			
A-100% normal	B-50% normal 50% affected	C-50% normal 25% carrier 25% affected	D-100% Affected
5 Changes in the Y chromosome will lead to infected:			
A-Both female and male	B-Male only	C-Female only	D-None
6 Sex-linked disorders			
A- Higher in male	B-Higher in Female	C- Both have equal chance	D-Depends on the disease

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

Meet our team!

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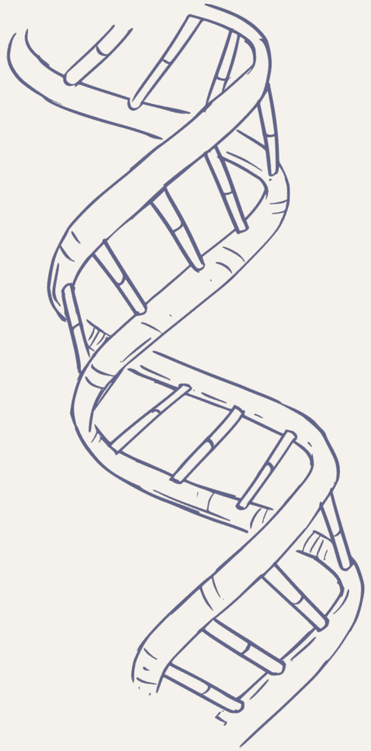
Rose Alsanie

Lama Alhayan

Marwah Fal

Najd Albawardi



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