MODE OF INHERITANCE

HUMAN GENETICS

Red: important.
Pink: F-slides
Blue: M-slides
Green: doctor's Notes
Gray: extra

Revised & Reviewed by:
Abdulaziz & Bahammam
Faye Wael Sendi

HUMAN GENETICS 441

MED441
KING SAUD UNIVERSITY

Objectives

01 Assess Mendel's laws of inheritance.

02 Understand the bases of Mendelian inheritance.

03 Define various patterns of single gene inheritance using family pedigree and Punnett square.

Father of genetics

Dr's note

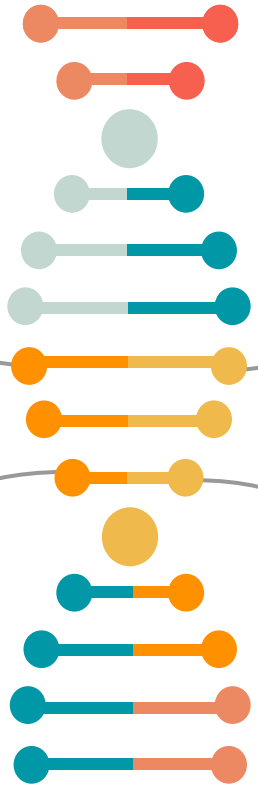
You don't need to remember his birth & death date.

1. Born in 1822
2. Monk (راهب) and teacher
3. Discovered some of the basic laws of heredity
4. His published his work, entitled Experiments on Plant Hybrids in1866. However, largely ignored
5. He died in 1884 with his work still unnoticed
6. His work rediscovered in 1900

Interpreting the outcomes of Mendel's breeding experiments

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**.

The plant characteristics being studied were each controlled by a 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.

Genotype

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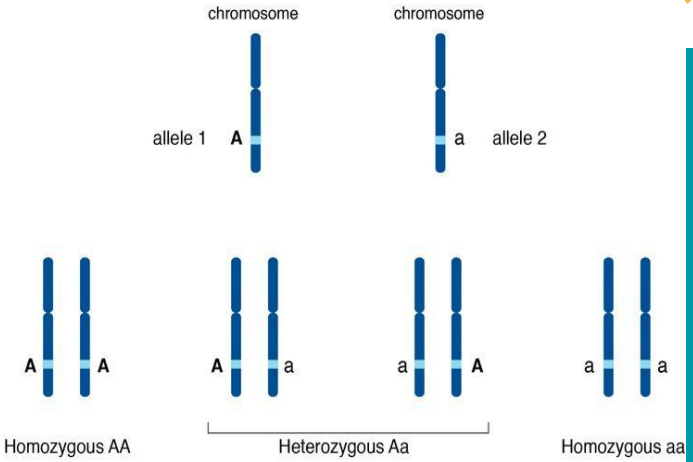
Homozygous (same alleles)

- for dominant alleles e.g. AA.
- for recessive alleles e.g. aa.

Heterozygous (differ alleles)

- It's a combination of recessive and dominant alleles e.g. Aa.



Punnett Square

(It is named after Reginald C. Punnett, who devised the approach)

USEFUL VIDEO

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We advise you to watch this video before moving to the next slide

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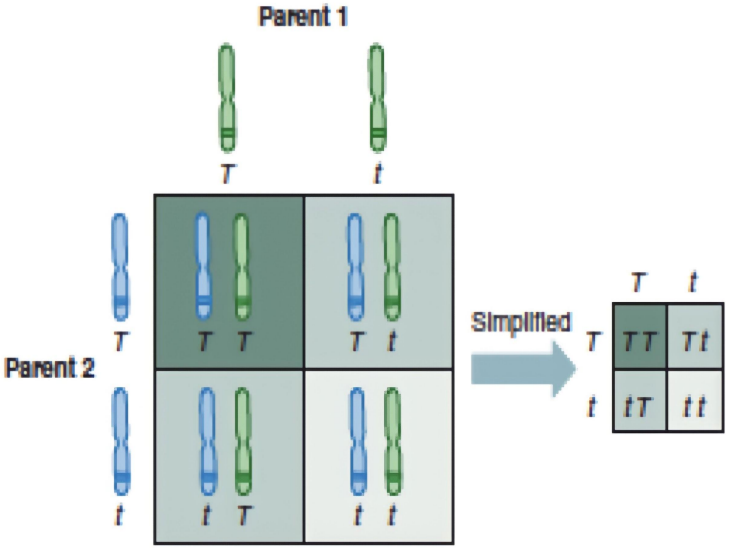
1. Each parent can only contribute **one allele** per gene.
2. These genes are found on the chromosomes of gametes of parents.
3. **Offspring** (ذرية) will inherit 2 alleles to express that gene.



Law of Dominance or Uniformity

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- 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 pp .

Female gametes

Male gametes

	P	P
p	Pp	Pp
p	Pp	Pp

F1 generation

Genotype ratio =1Pp

Phenotype ratio =1 purple

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.

Law of segregation

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Definition:
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 .

Male gametes

Female gametes

	P	p
P	PP	Pp
p	Pp	pp

Team 437:
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

F1 generation

Genotype ratio = 1PP:2Pp:1pp

Phenotype ratio = 3 purple:1 white

Law of independent assortment

USEFUL VIDEO

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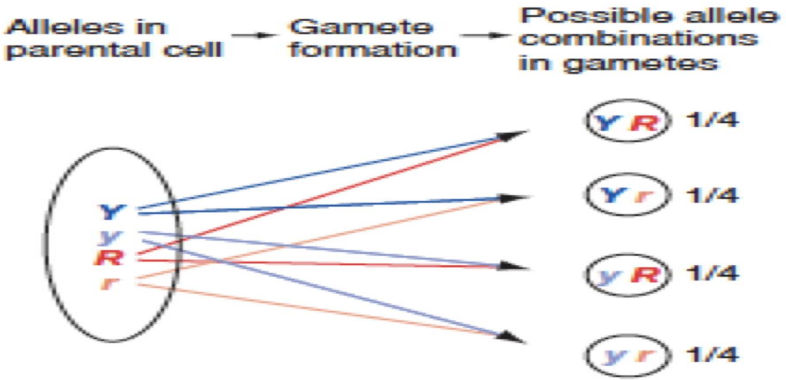
Definition:
During gamete formation, different pairs of alleles segregate independently of each other.

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.

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.

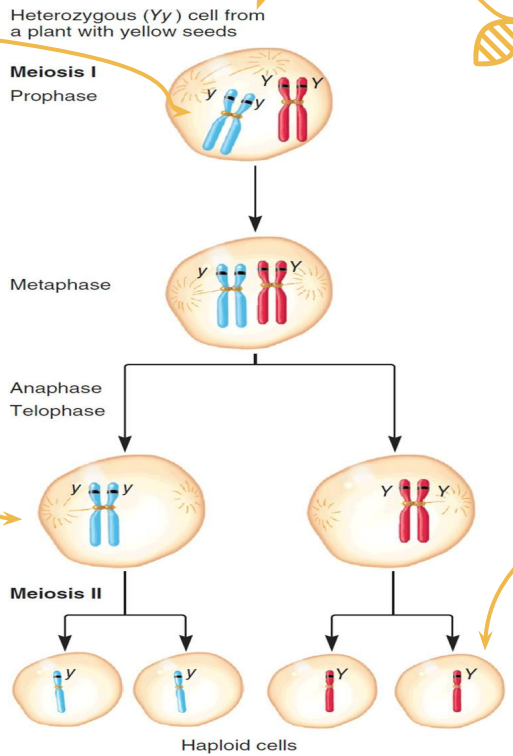


THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES

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.



In this example using pea seed color, the two alleles are Y (yellow) and y (green).

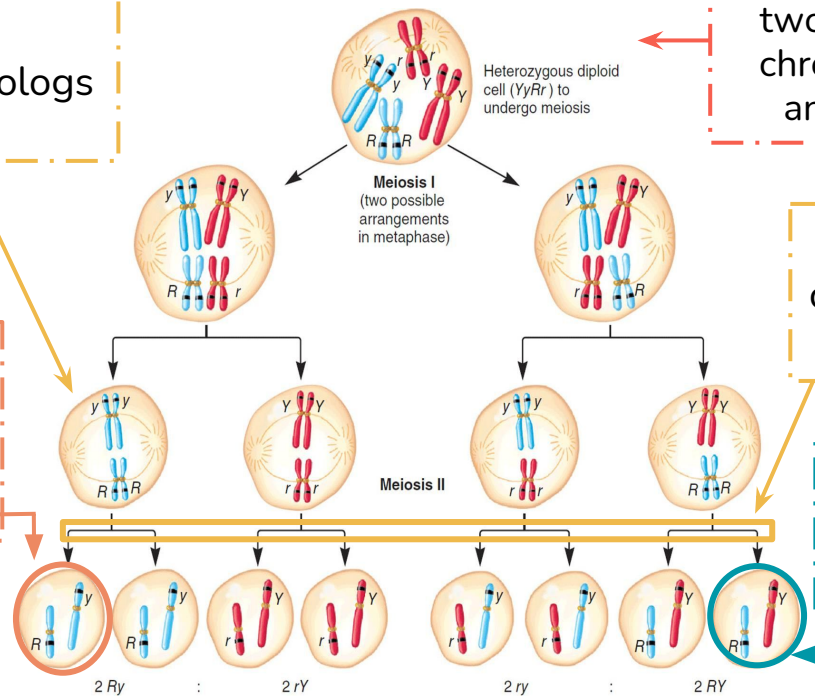
During meiosis, the homologous chromosomes segregate from each other

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

During **metaphase of meiosis I**, different possible arrangements of the homologs within bivalents .

This figure shows the assortment of two genes located on two **different** chromosomes, using pea seed color and shape as an example (**YyRr**).



On the left, the **dominant R** allele has sorted with the **recessive y** allele

can lead to **different** combinations of the alleles in the resulting gametes.

On the right, the **dominant R** allele has sorted with the **dominant Y** allele.

Dr's note

Bivalents mean pair of chromosomes.

MODES OF INHERITANCE FOR SINGLE GENE DISORDERS

Autosomal

Recessive

Dominant

Sex Linked

X Linked

Recessive

Dominant

Y Linked

Dr's note

A person needs 2 copies of the gene to be **affected (aa)**.

Autosomal Recessive

1. The trait (character, disease) is recessive.
2. The trait expresses itself only in homozygous state.
3. The parents of the affected child may be related (consanguineous).
4. Males and female are equally affected.
5. Examples:
   Cystic fibrosis, Phenyketonuria, Sickle cell anaemia, Thalassaemia

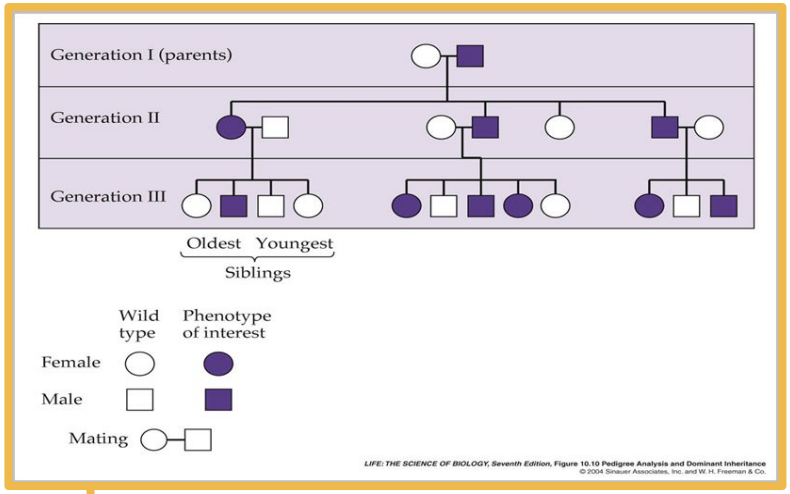
Autosomal Dominant

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

Dr's note

Which case that the trait will appear in **every generation**?
Autosomal Dominant.

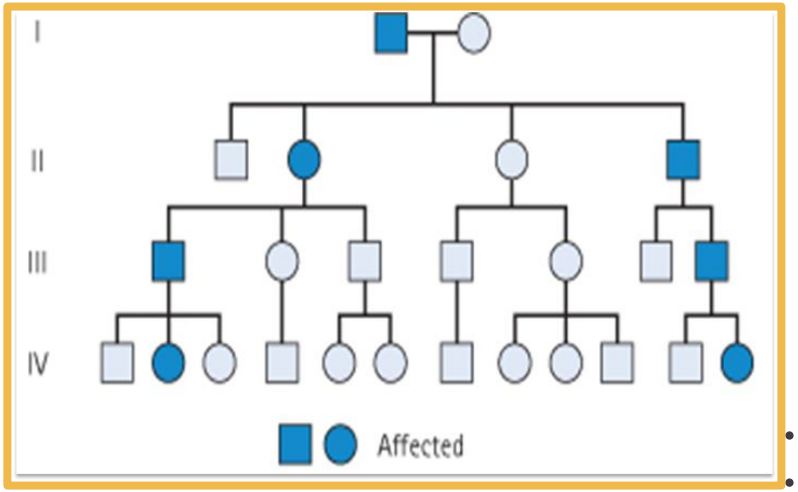
A Pedigree Analysis



The dr's note

- The pedigree is essential part of **history taking** in genetics.
- The probability of **each** child being affected is **25%** , but **not for all children**,**25%**.

Family Tree of an Autosomal Dominant Mode of Inheritance

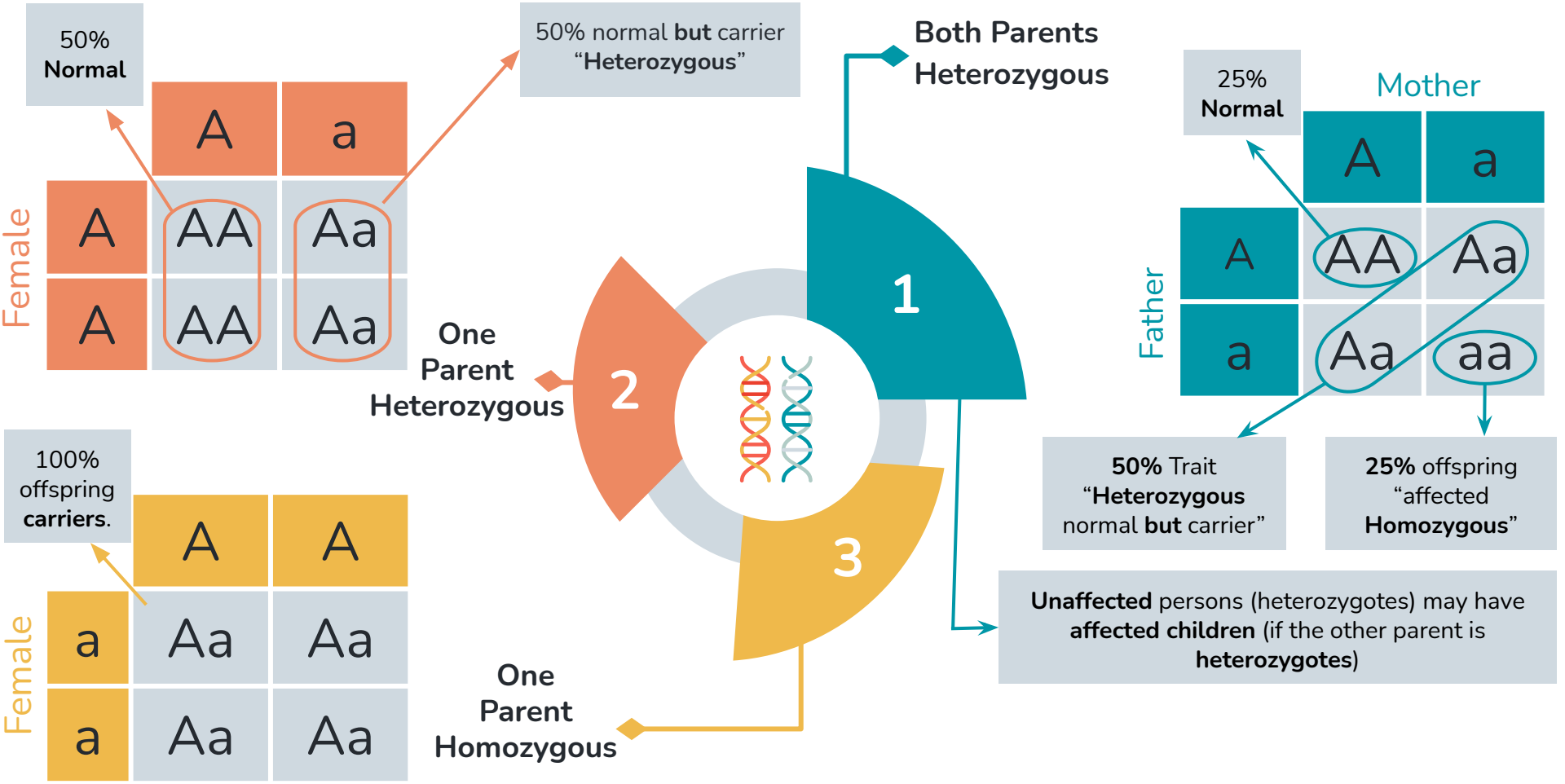


Note the presence of male-to-male (i.e. father to son) transmission.

The dr's note

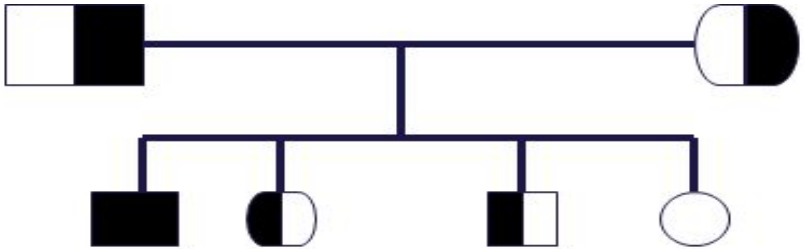
A person only needs 1 copy of the gene to be affected (**AA**,**Aa**).

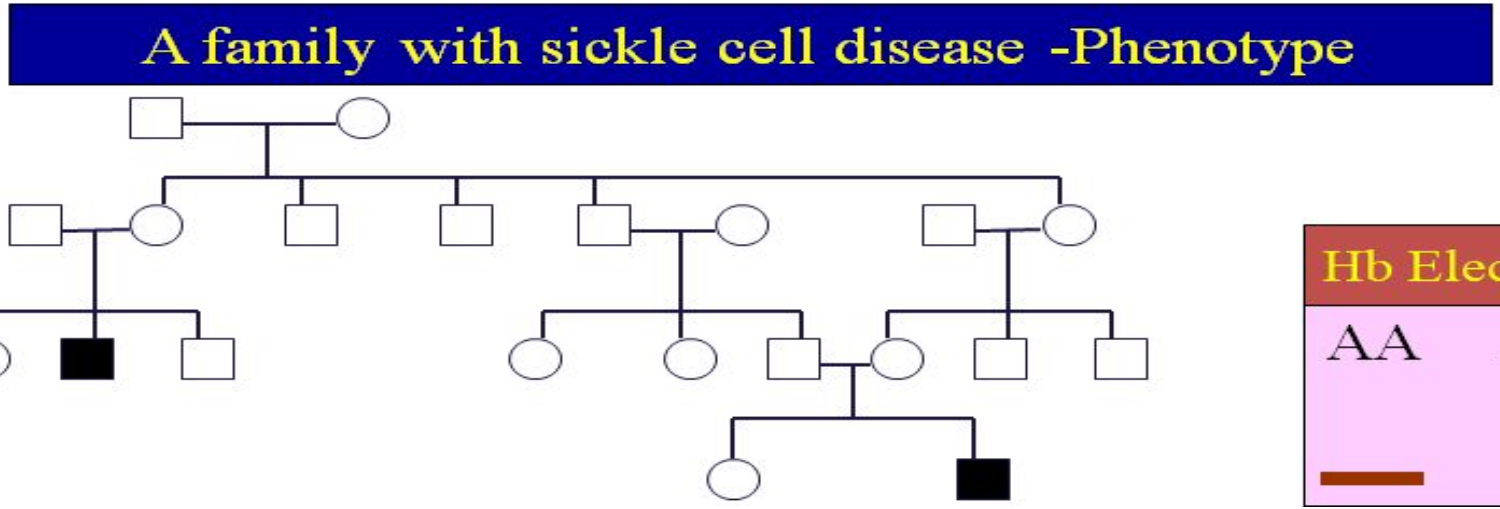
Punnett square showing autosomal recessive inheritance

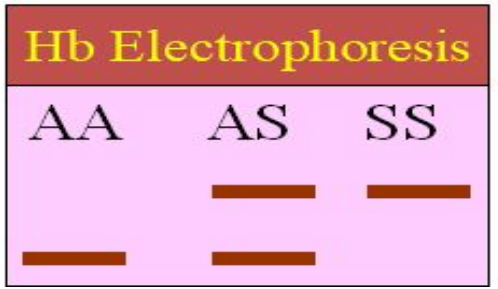


Family tree of an Autosomal recessive disorder Sickle cell disease (SS)

Team 439:
Skip one generation.







Sex – Linked Inheritance

Definition:
The inheritance of a gene present on the **sex chromosomes**.

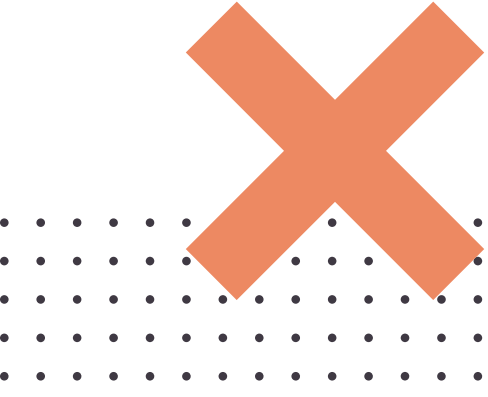
The Inheritance Pattern is **different** from the autosomal inheritance.

Inheritance differs in **males** from **females**.

Two types
- **X**-Linked
- **Y**-Linked

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**

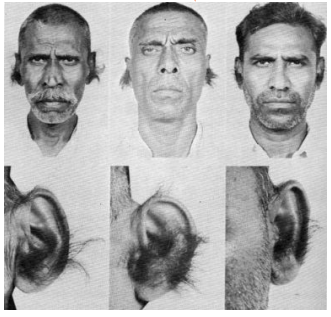


Y-Linked inheritance

- The gene is present 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*

Hairy ears in India

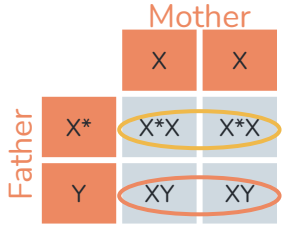


X – Linked Recessive Inheritance

- 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,

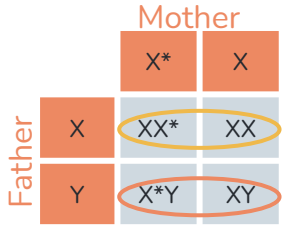
1 Normal female, affected male

1. All sons are normal
2. All daughters carriers “not affected”



2 Carrier female, normal male

1. 50% sons affected
2. 50% daughters carriers



3 Homozygous female, normal male

1. All daughters carriers.
2. All sons affected.

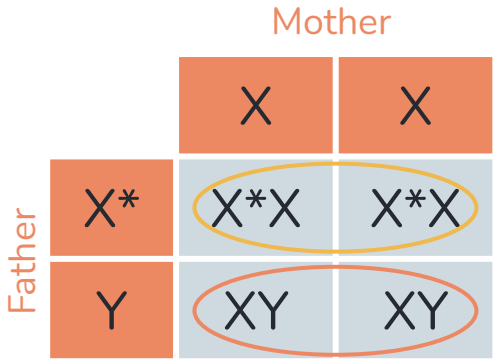
X – Linked Dominant Disorders

- 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

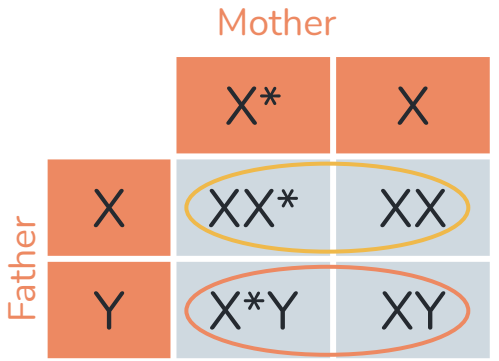
1 Affected male and normal female

All daughters affected
all sons normal



2 Affected female (heterozygous) and normal male

50% sons & 50% daughters are affected



TAKE HOME MESSAGES

- An accurate determination of the family pedigree is an important part of the work up of every patient
- 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
- Pedigrees for single-gene disorders may demonstrate a straightforward, typical mendelian inheritance pattern

MCQs

Q1: A trait or disorder determined by a gene on one of the sex chromosomes is said to be:			
A.autosomal inheritance	B.sex-linked inheritance	C.recessive inheritance	D.Dominant inheritance
Q2:Changes in the Y chromosome will occur in:			
A.Both female and male	B.Male only	C. Female only	D.Aliens
Q3:Transmission of genes occurs:			
A.Vertically	B.Horizontally	C.Between Siblings	D.Diagonally
Q4: The physical expression of a character is called:			
A. Morphology	B.Genotype	C.Phenotype	D.dominant

Answer Key: 1B - 2B - 3A - 4C

GIRLS TEAM

WAREEF ALMOUSA
AISHA ALHAMED
RAAOUM JOBOR
ALANOUD ALHAIDER
HAYA ALSHALOOB
LAMA ALEYADHY

BOYS TEAM

ABDULAZIZ ALMAJED
YAZAN ABUHOZA
ABDULLTAIF ALTALHAH
SAAD ALHANAYA
ABDULRAHMAN ALHOUMAILY
ABDULRAHMAN ALMUTAIRI

TEAM LEADERS

MARAM ALDEEJ

FAISAL ALHOGAIL

MADE BY

REVISED BY

HGMED441@GMAIL.COM

Editing file

“You are the designer of your destiny; you are the author of your story.”

—LISA NICHOLS