KING SAUD UNIVERSITY
COLLEGE OF MEDICINE
FOUNDATION BLOCK

MODES OF INHERITANCE

HUMAN GENETICS 433

Objectives

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

Color index

Red: Important

Purple: Extra Information

Orange: Explanation

Mendel's breeding experiments: Interpretation of his results

- **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.**
- **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 and phenotype

Homozygous **Homo- =same**	**Dominant**	**AA**
	Recessive	aa
Heterozygous Hetero-=different		Aa

MENDELIAN LAW OF INHERITANCE

The traits, later called genes, normally occur in pairs in body cells and separates during the formation of sex cells. This happens in meiosis, the production of gametes. Of each pair of chromosomes, a gamete only gets one.

When two homozygotes with different alleles are crossed, all the offspring in the F1 generation are identical and heterozygous.

"The characteristics do not blend, as had been believed previously, and can reappear in later generations."

Punnett square

	Male gametes	
Female gametes	T	T
t	Tt	Tt
t	Tt	Tt

For example :
cross : purebred purple female (PP)
x White male(p*p*)

F1
Genotype / phenotype ratio= 1Pp/1 purple

	Male gametes	
Female gametes	P*	P*
P	Pp*	Pp*
P	Pp*	Pp*

P* =small p

Punnett Square

Each parent can only contribute one allele per gene
These genes are found on the chromosomes carried in the sex cells.
Offspring will inherit 2 alleles to express that gene

Female gametes	Male gametes: T	Male gametes: t
T	TT	Tt
t	Tt	tt

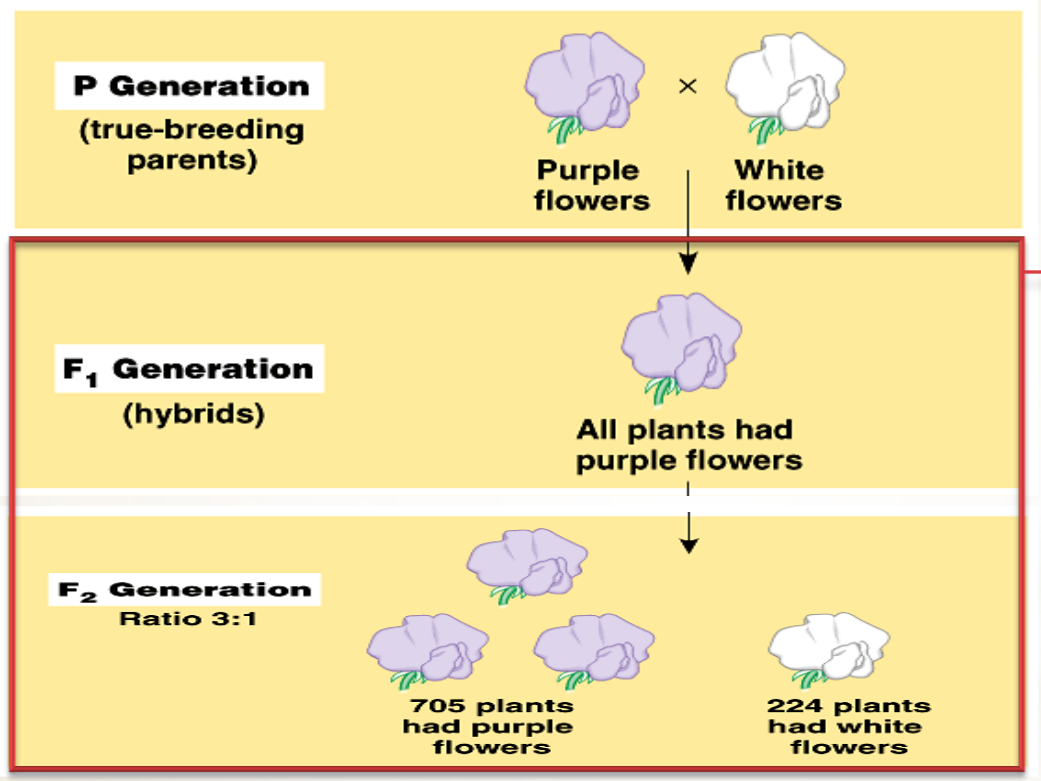
For example ;
two F1 generation offspring with each other
Pp X Pp

F2 generation
Genotypic / phenotypic ratio=
1PP:2Pp*:1p*p*/ 3 purple :1 white

Female gametes	Male gametes: P	Male gametes: P*
P	PP	Pp*
P*	Pp*	p*p*

Law of Dominance

In the monohybrid cross (mating of two organisms that differ in one character), one version disappeared



The first cross produce the F2 generation and the lost trait appeared with predictable ratio

F1 generation - Genotypic ratio = 1Pp / Phenotypic ratio = 1 purple

F2 generation - Genotypic ratio = 1PP:2Pp:1pp / Phenotypic ratio = 3 purple:1 white

MENDEL'S FIRST LAW OF SEGREGATION

The genes determine the organism's traits, and are inherited from its parents. As the pair of chromosomes separate, each gamete only receives one of each allele.

This Mendel called the Law of segregation. Mendel also noted that alleles of a gene could be either dominant or recessive

MENDEL'S SECOND LAW OF INDEPENDENT ASSORTMENT

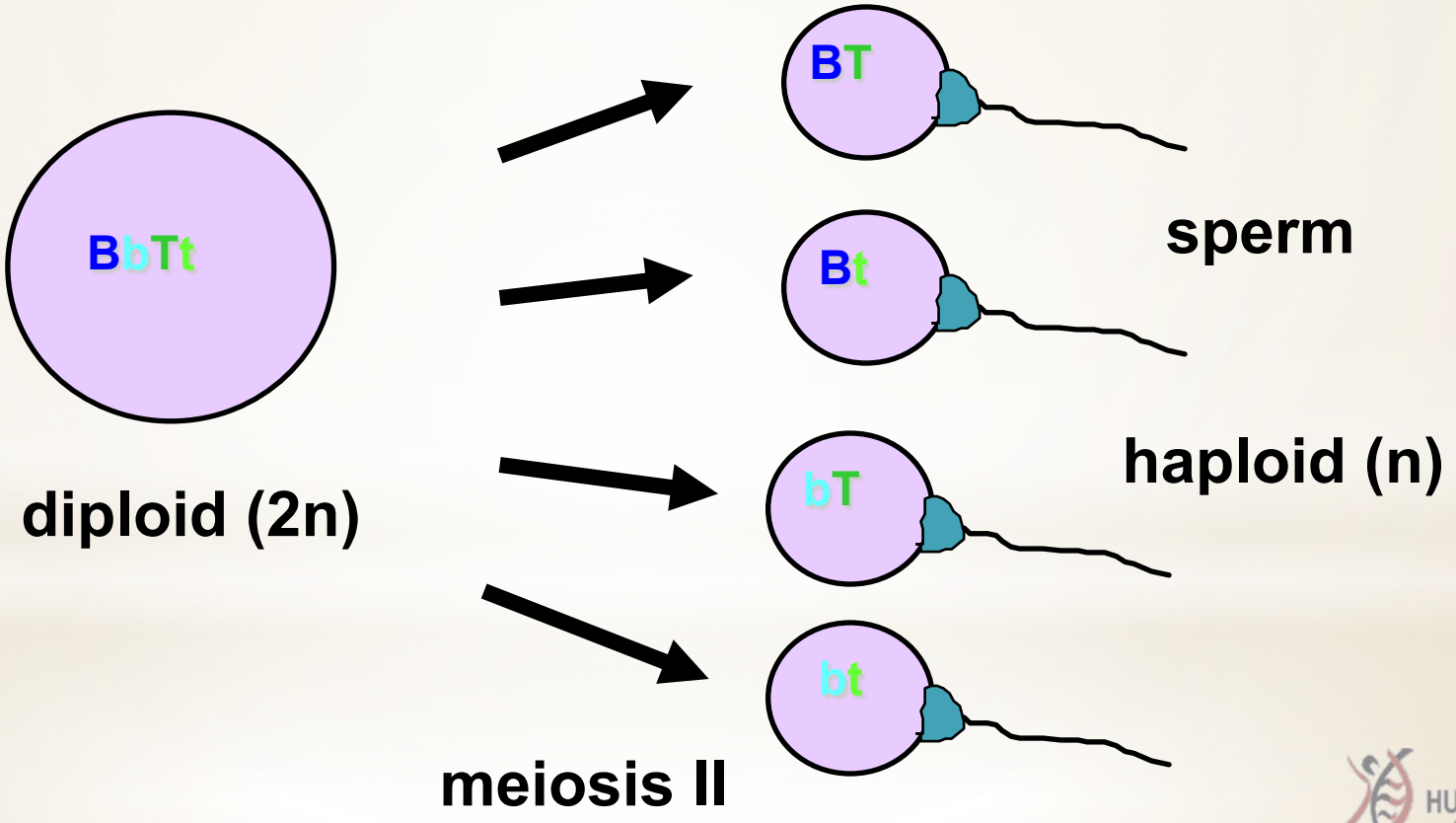
Alleles of different genes separate independently of one another when gametes are formed. So Mendel thought that different traits are inherited independently of one another.

The second law is only true if the genes are not on the same chromosome. If they are, then they are linked to each other

Principle of Independent Assortment

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.

B is an allele for the colour black and T is the allele for being tall



Principle of Independent Assortment

		BbTt			
		BT	Bt	bT	bt
BbTt	BT	BBTT	BBTt	BbTT	BbTt
	Bt	BBTt	BBtt	BbTt	Bbtt
	bT	BbTT	BbTt	bbTT	bbTt
	bt	BbTt	Bbtt	bbTt	bbtt

Phenotypic ratio: 9 Tall, Black: 3 Tall, White: 3 Short, Black: 1 White, Short → (9:3:3:1)

Genotypic ratio: 1 BBTT: 2 BBTt: 2 BbTT: 4 BbTt: 1 BBtt: 2 Bbtt: 2 bbTt: 1 bbTT: 1 bbtt

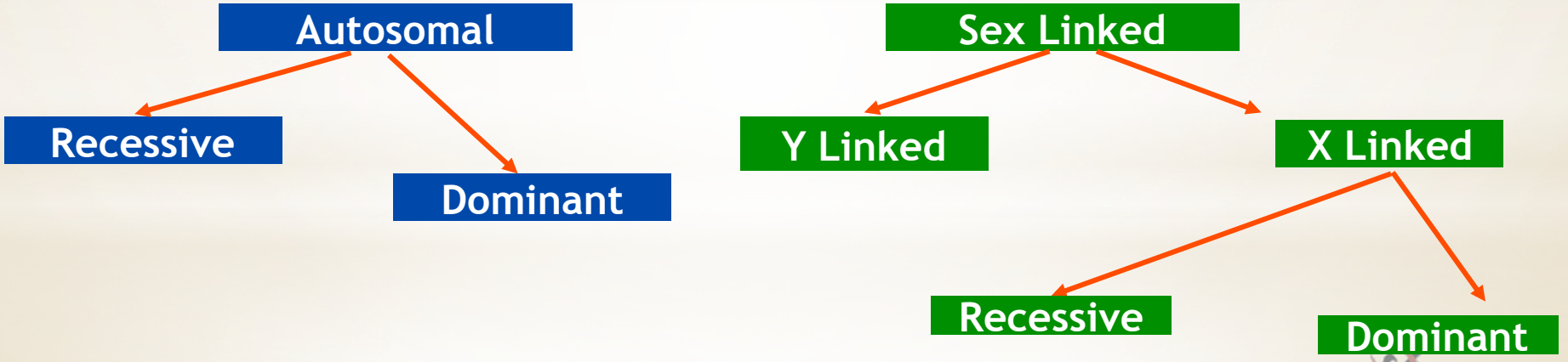
MENDELIAN INHERITANCE

Over 11,000 traits/disorders in humans exhibit single gene ***unifactorial*** or ***Mendelian*** *inheritance*.

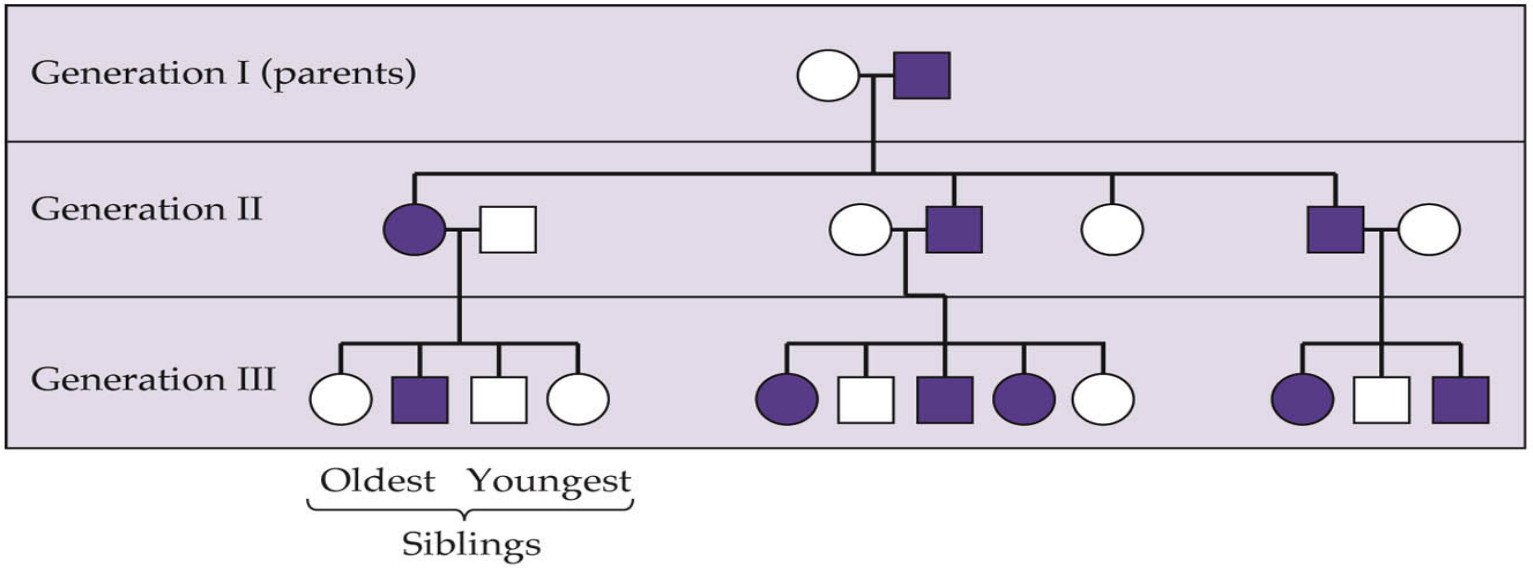
A trait or disorder that is determined by a gene on an ***autosome*** is said to show ***autosomal inheritance***.

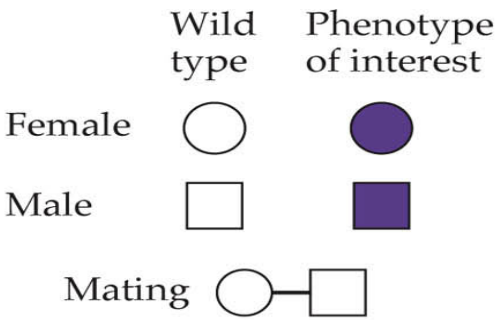
A trait or disorder determined by a gene on one of the ***sex*** chromosomes is said to show ***sex-linked inheritance***.

MODES OF INHERITANCE OF SINGLE GENE DISORDERS



A Pedigree Analysis for Disease





LIFE: THE SCIENCE OF BIOLOGY, Seventh Edition, Figure 10.10 Pedigree Analysis and Dominant Inheritance

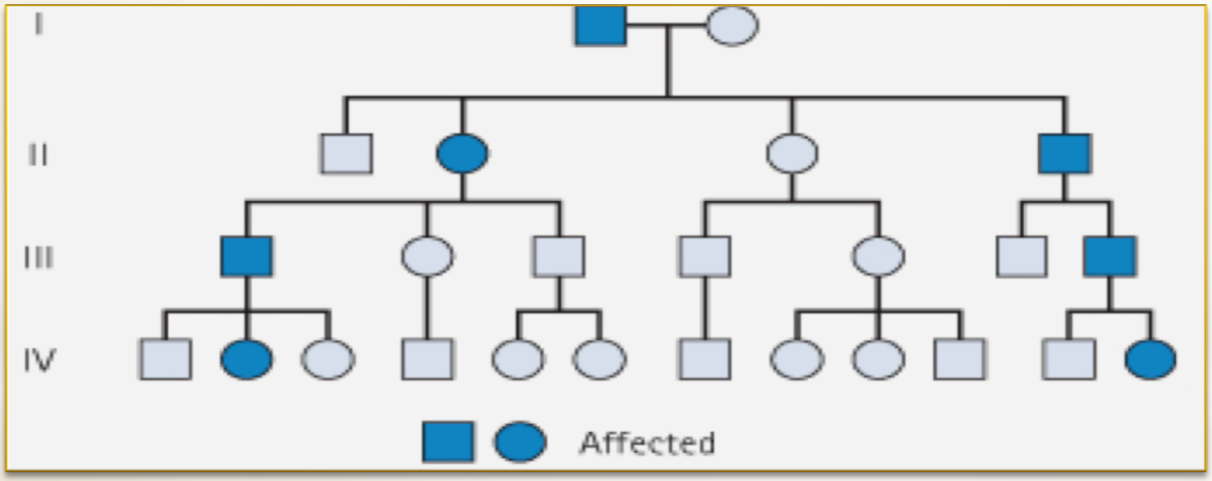
Autosomal Dominant[(1)] Mode of Inheritance

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 to son) transmission

(1) Autosomal dominant means penetrance of the mutation is very high , those who have a mutated copy of the gene will have the disease

Autosomal Recessive : Mode of Inheritance

- 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:

25% offspring affected Homozygous"

50% Trait "Heterozygous normal but carrier"

25% Normal

Mother

Father	A	a
A	AA	Aa
a	Aa	aa

(2) One Parent Heterozygous

50% normal but carrier "Heterozygous"

50% Normal

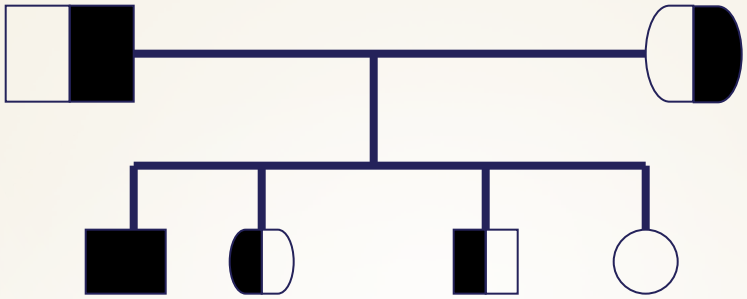
	A	a
A	AA	Aa
A	AA	Aa

(3) Both Parent Homozygous:

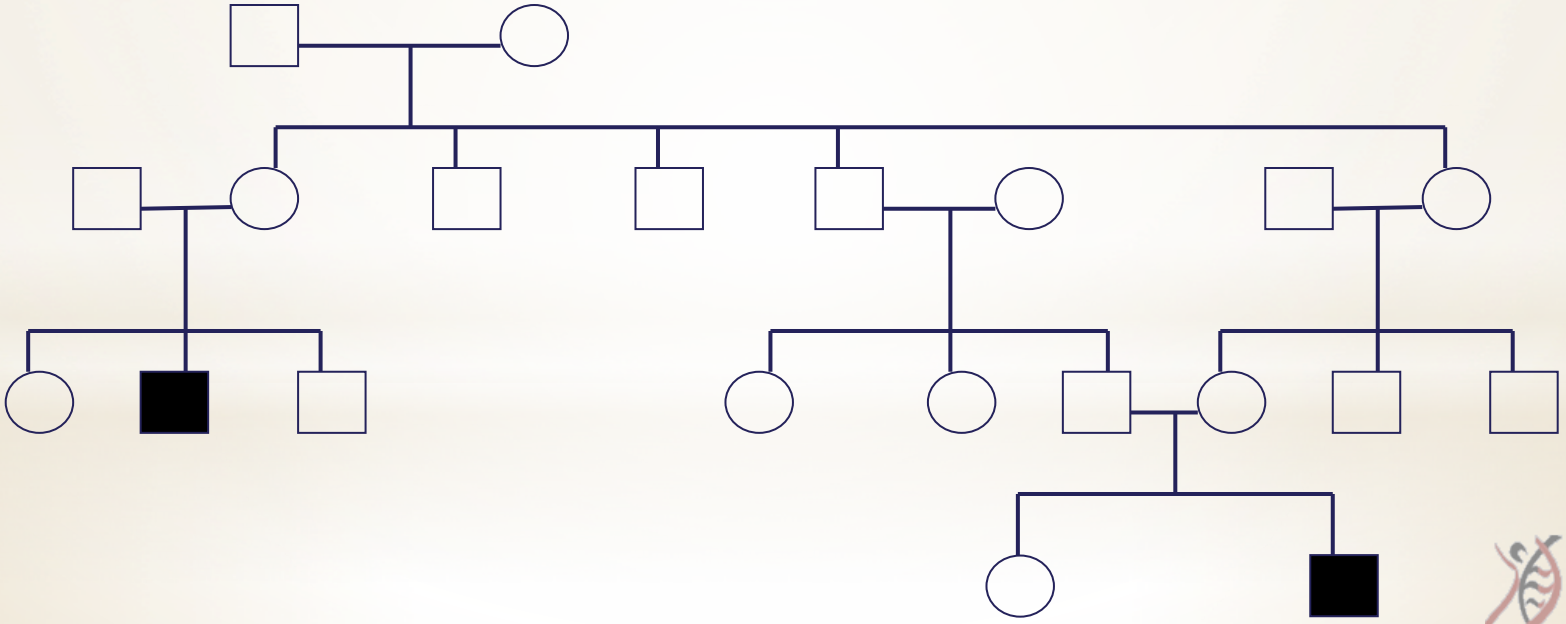
100% offsprings carriers.

	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

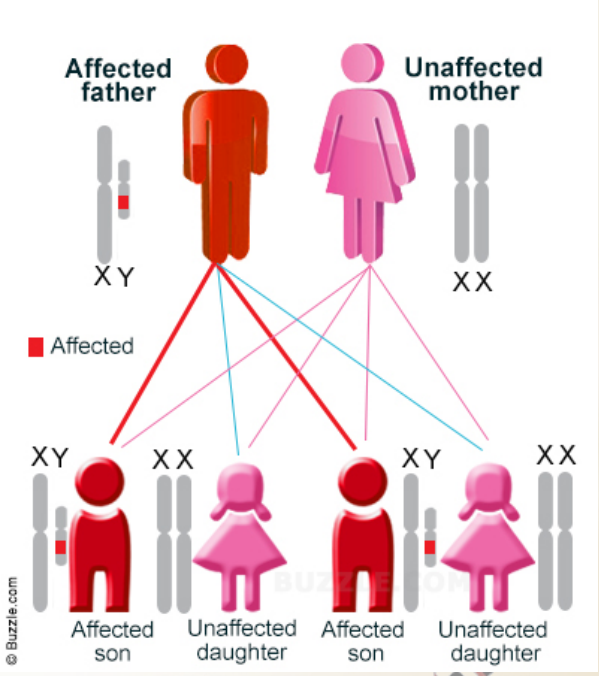


Sex – Linked Inheritance

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

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(1), the condition exhibits itself whether dominant or recessive



(1) Hemizygous : describe an individual who has only one member of chromosomes

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
- 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

Affected father x unaffected mother

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

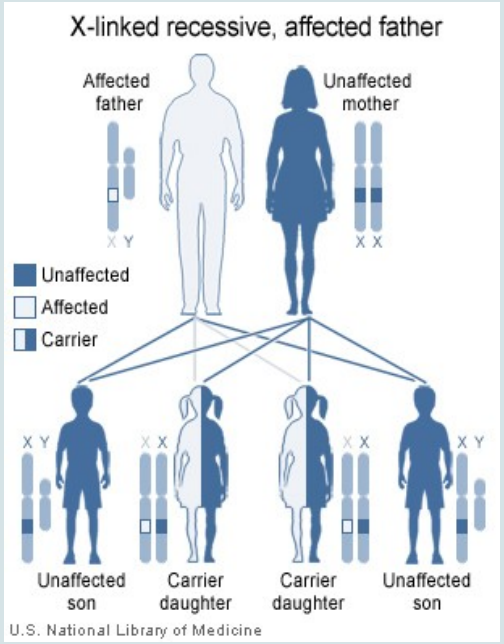
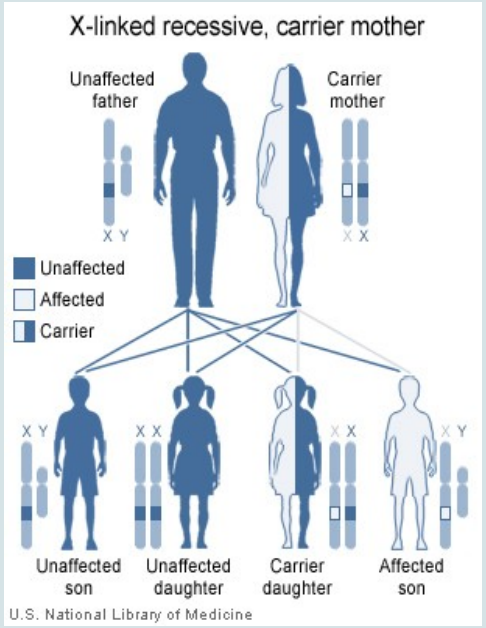
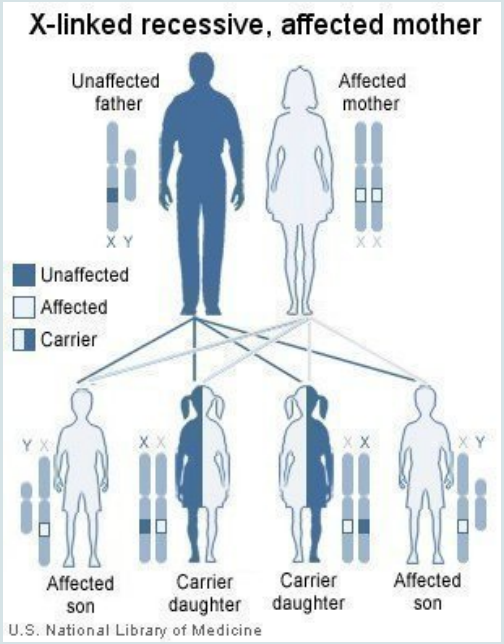
- All sons are Normal
- All daughters are carriers (not affected)

Unaffected father x unaffected carrier mother

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

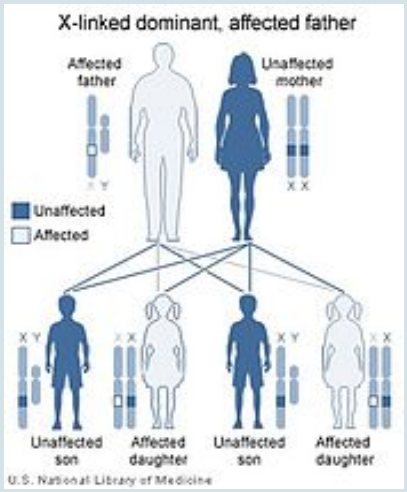
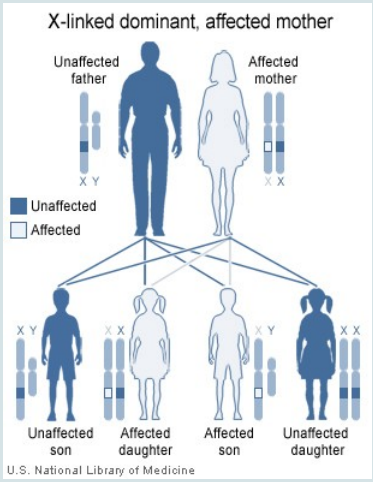
- 50% sons are affected
- 50% daughters are carriers

X – Linked Recessive Inheritance

Normal female, Affected male	Carrier female, Normal male	Homozygous affected female, Normal male
		
All sons are normal All daughters carriers "not affected	50% sons affected 50% daughters carriers	- All daughters carriers. - All sons affected.

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
- Hemizygous male and heterozygous females express the disease.

Normal female, Affected male	Homozygous female, Normal male
	
All daughters affected, all sons normal	50% sons & 50% daughters are affected

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MCQs

- Q1:Pure bred plants with identical genes are known as:

A:Hemizygous B:Heterozygous

C:Homozygous D:Azygous

- Q2:Which one of these is the Genotype of a homozygous recessive plant:

A:TT B:Tt

C:tt D:tT

- Q3: Alleles of different genes separate independently of one another when gametes are formed, which one of these fits this description:

A:Law of segregation B:Law of independent assortment

C:Law of isolation D:Law of forced division

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MCQs

- **Q4:The Phenotypic ratio in the Law of Independent Assortment for two traits is:**

A:3:1 **B:9:3:3:1**

C:4:3 **D:8:5:3:1**

- **Q5:In an autosomal recessive inheritance if heterozygous parents give birth to a child wh at is the percentige that the child is affected:**

A:100% **B:0%**

C:50% **D:25%**

- **Q6:In an autosomal recessive inheritance if homozygous recessive father and homozygous dominant mother give birth to a child what is the percentige that the child is affected:**

A:100% **B:0%**

C:50% **D:25%**

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MCQs

- **Q7:In Y-linked inheritance:**

  A:only males are affected **B:only females are affected**

  C:both are affected **D:none are affected**

- **Q8: In an X-linked dominant inheritance a normal father and affected mother wi ll have:**

  A:normal children **B:affected children**

  C:carrier children **D:affected Daughters and normal sons**

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