Human Genetics

LECTURE 3

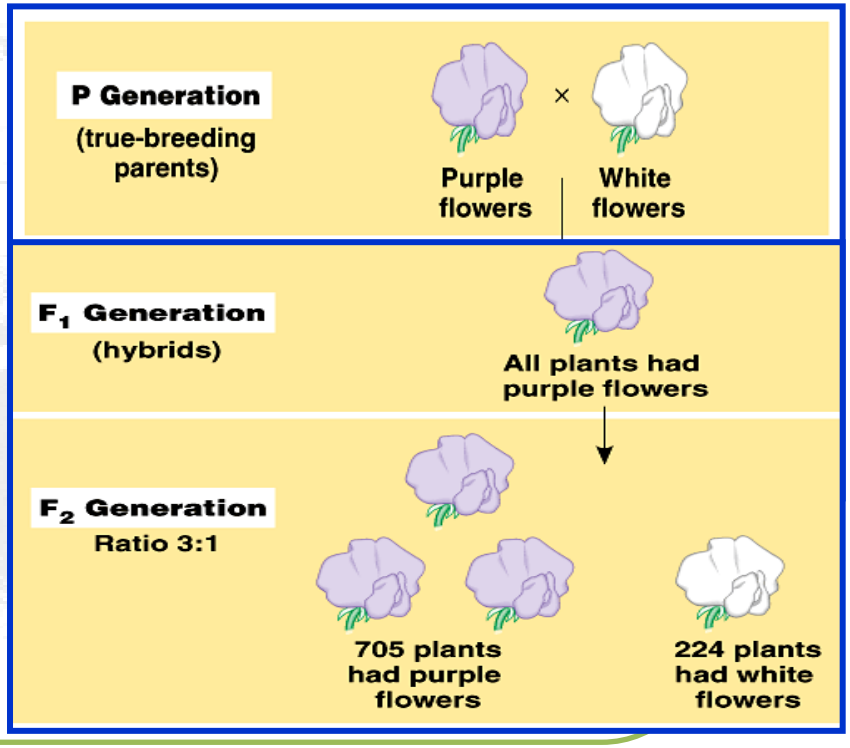
MODE OF INHERITANCE

Lecture Objectives

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

Father of Genetics

- Monk and teacher
- Discovered some of the basic laws of heredity
- Presentation to the Science Society in1866 went unnoticed
- He died in 1884 with his work still unnoticed
- His work rediscovered in 1900.



Mendel's Breeding Experiments : Interpretation of his results

1-The traits of the plant are controlled by a pair of **factors (alleles)** , each was inherited from each parent

2-The **pure-bred** plants, with **two identical genes**, used in the initial cross would now be referred to as **homozygous**.(**TT**)

3-The **hybrid F1** plants, each of which has one gene for tallness and one for shortness, would be referred to as **heterozygous**.(**Tt**)

4-Alleles means that there are 2 genes in the homologous chromosomes facing each other and they carry the same trait e.g. (color of the eyes)

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

Genotype And Phenotype

Homozygous Dominant (**TT**)
Homozygous Recessive (**tt**)
Heterozygous (**Tt**)

Note: If an individual has inherited 2 alleles from each parent(**Tt**) , and that trait was color of the eyes for example , If(**t**)was a blue color of the eye and (**T**) was a brown color of the eye .. That means his eyes would be BROWN , why ? Because brown color is dominant ((**T**))

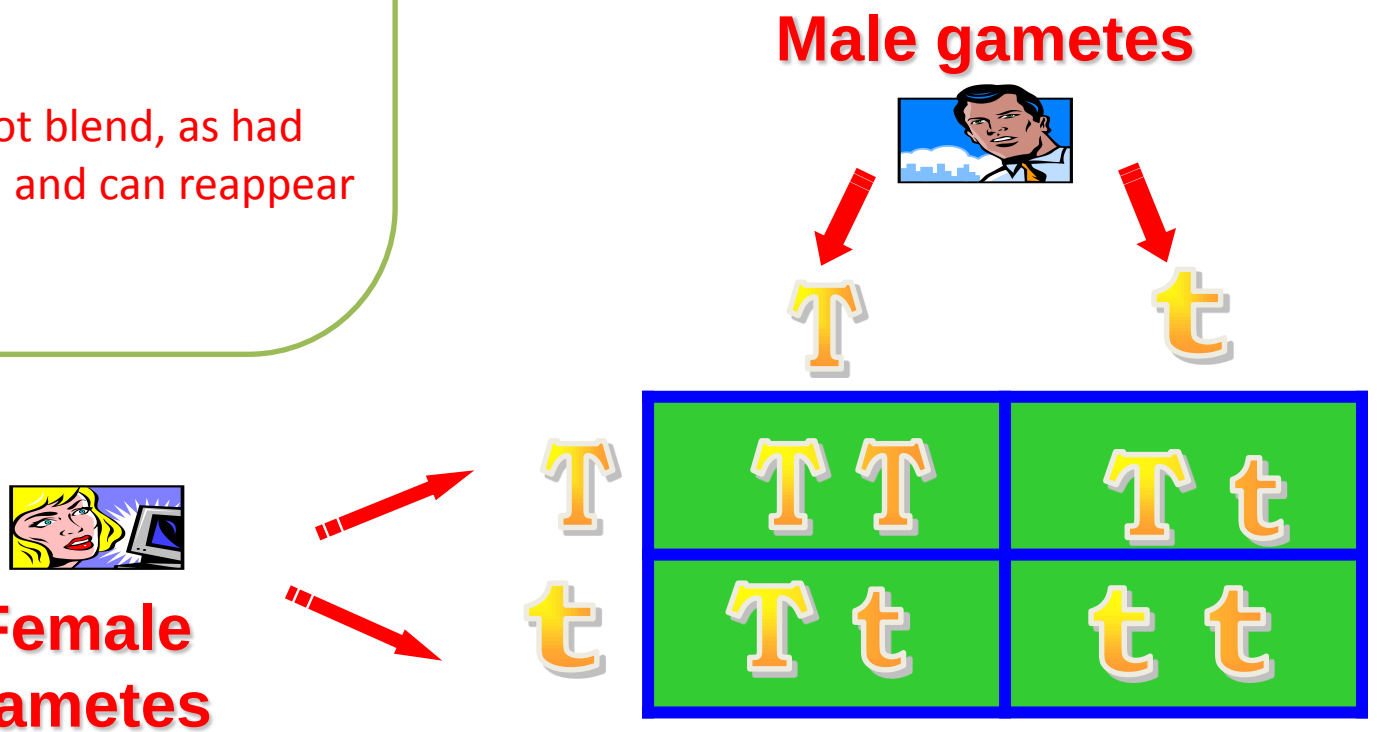
MENDELIAN LAW OF INHERITANCE

The traits, controlled by **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

- 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 gen.



MENDEL'S FIRST LAW OF SEGREGATION (the "First Law")

The genes determine the organism's traits .

A Sperm or egg carries only one allele for each inherited trait. When sperm and egg unite at fertilization,each contributes its allele , restoring the paired condition in the offspring .

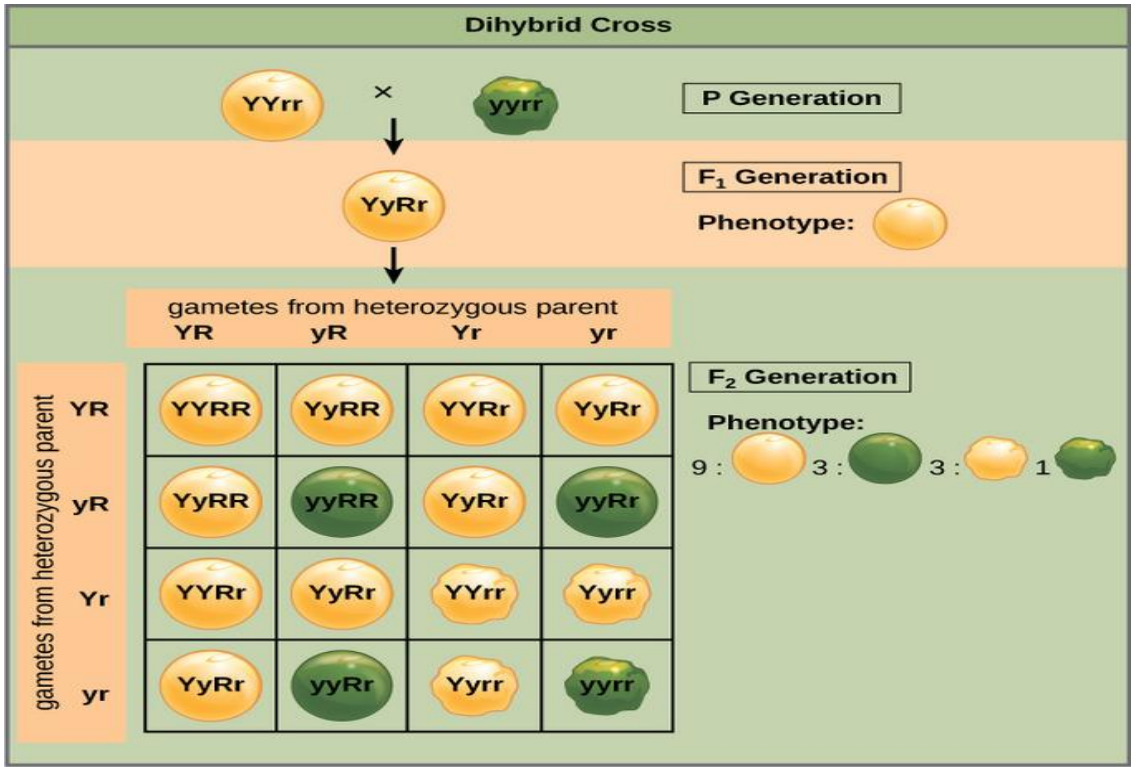
Note: Alleles could be either dominant or recessive.

Rare exceptions to this rule can occur when two allelic genes fail to separate because of chromosome non-disjunction at the first meiotic division.

MENDEL'S SECOND LAW OF INDEPENDENT ASSORTMENT (the "Second Law")

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 not always true if the genes are not on the same chromosome they tend to be inherited together . If they are, then they are linked to each other.

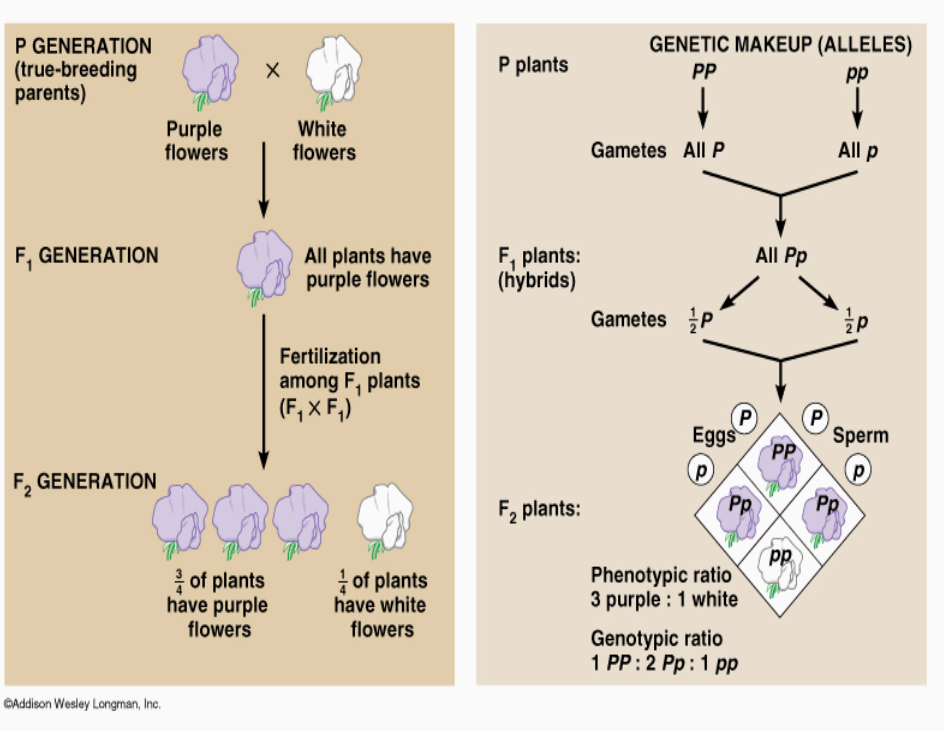


COMPLETE DOMINANCE - one allele is dominant to another allele.

Law of Dominance (3rd Law)

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

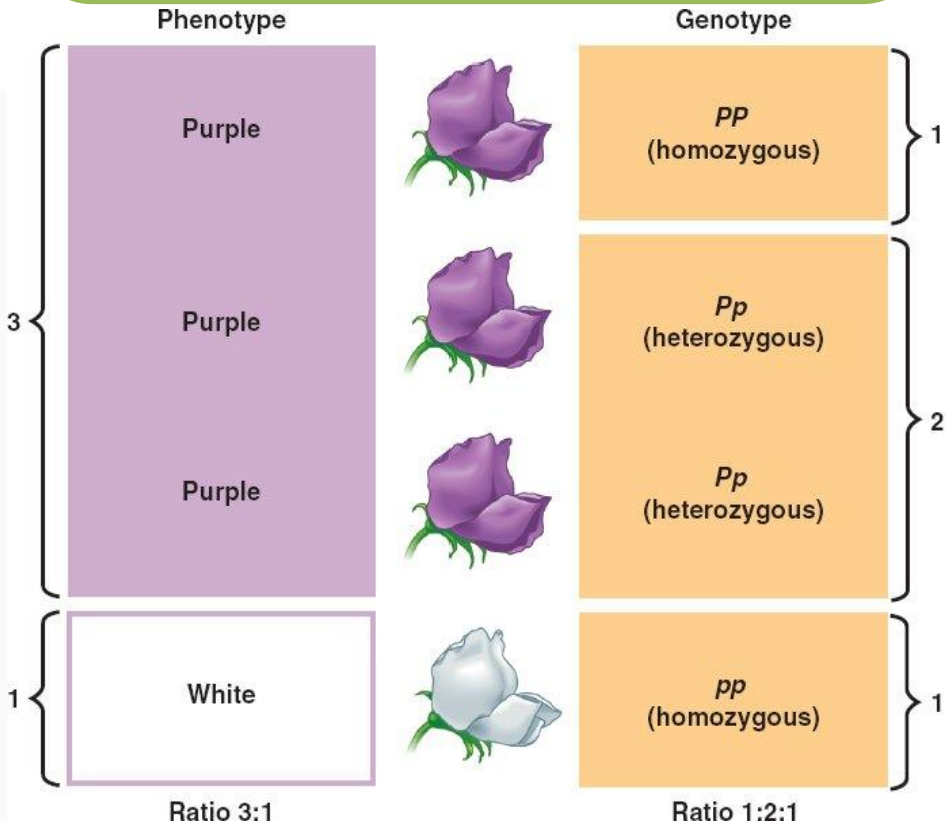
NOTE: Recessive alleles will always be masked by dominant alleles



Genotype versus phenotype

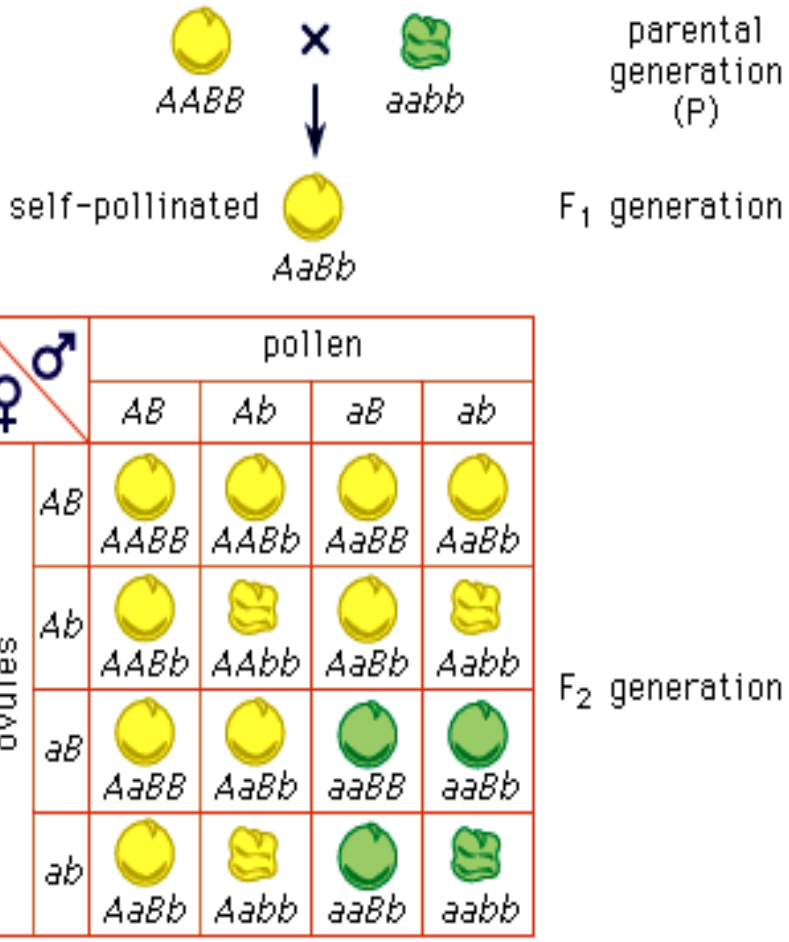
The F1 crossed produced the F2 generation and the lost trait appeared with predictable ratios.

This led to the formulation of the current model of inheritance.



Principle of Independent Assortment

"In dihybrid crosses you will see more combinations of the two genes"



MENDELIAN INHERITANCE

- in humans exhibit single gene ***unifactorial*** or ***Mendeli an*** *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

SINGLE GENE DISORDERS

Autosomal

Recessive

Examples: Cystic fibrosis, Phenyketonuria, Sickle cell anaemia, Thalassaemia etc.

Males and female are equally affected

Dominant

Examples: Huntington disease, Myotonic dystrophy, Neurofibromatosis type 1, Marfan syndrome etc.

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

Sex Linked

Y Linked

Examples: Hairy ears in India

The gene is passed from fathers to sons only Daughters are not affected

X Linked

Recessive

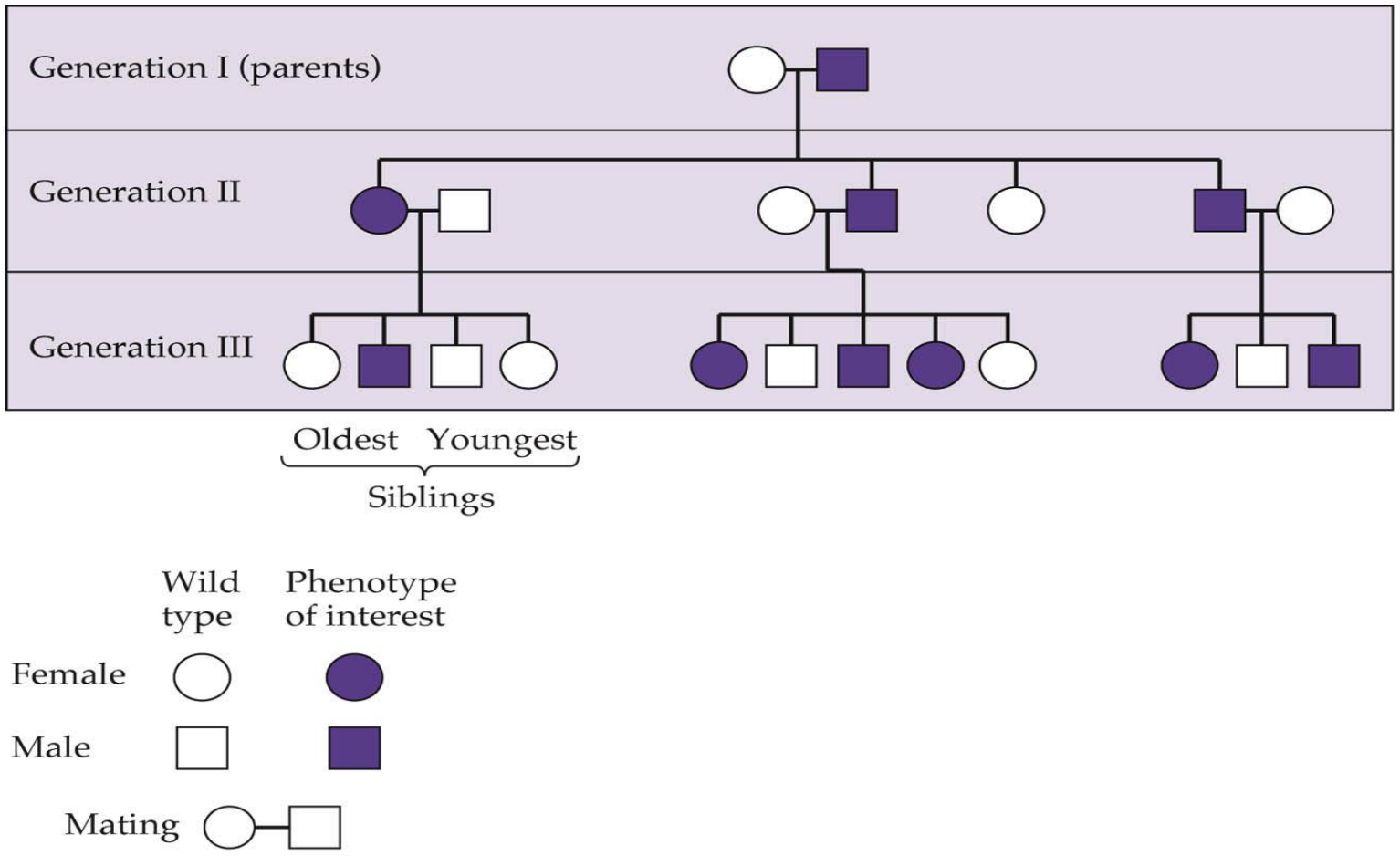
Examples: Albinism, Fragile X syndrome, Hemophilia, Muscular dystrophy, Retinitis pigmentosa

The incidence of the X-linked disease is higher in male than in female

Dominant

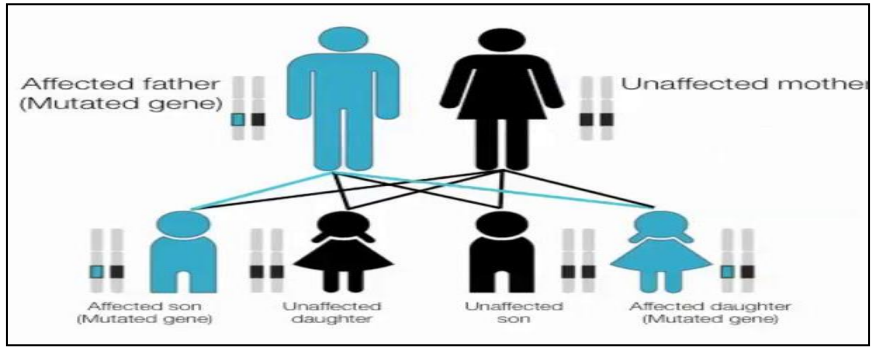
The trait occurs at the same frequency in both males and females

A Pedigree Analysis for Disease



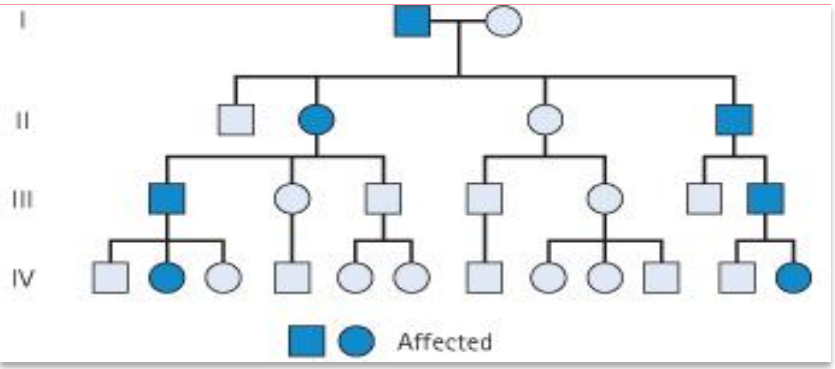
LIFE: THE SCIENCE OF BIOLOGY, Seventh Edition, **Figure 10.10 Pedigree Analysis and Dominant Inheritance**

Autosomal Dominant Mode of Inheritance



"Unaffected persons won't transmit the trait to their children."

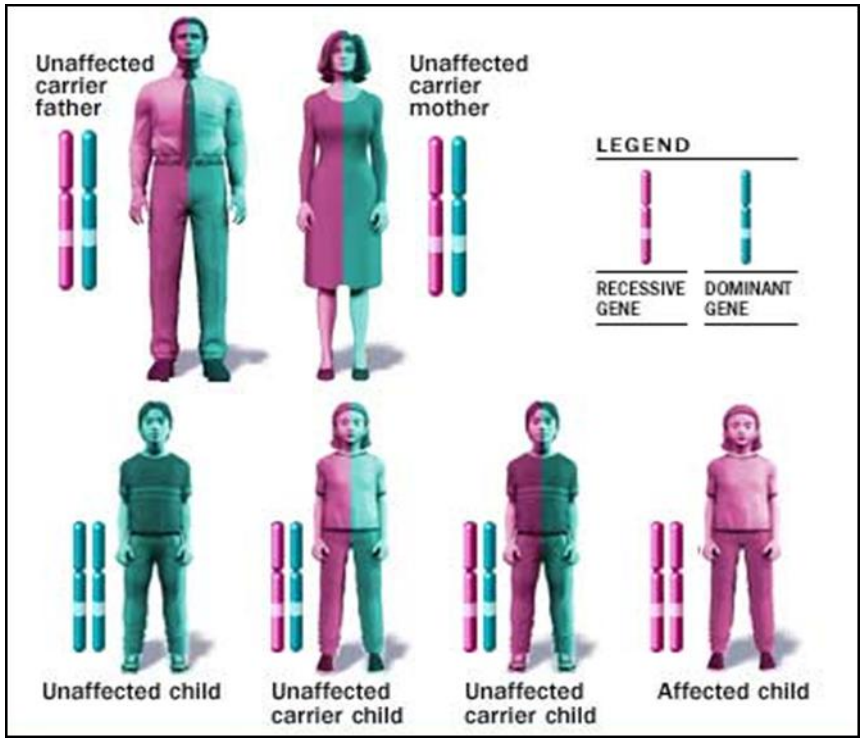
Family Tree of an Autosomal Dominant Mode of Inheritance



Note the presence of male-to-male (i.e. father to son) transmission
Both mother and father can transmit the disease.

Autosomal Recessive Mode of Inheritance

- ***The trait 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) (تربط الوالدين صلة قرابة)***



Punnett square showing autosomal recessive inheritance:

(1) Both Parents Heterozygous:

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

	A	a
A	AA	Aa
a	Aa	aa

2) One Parent Heterozygous

	A	a
A	AA	Aa
A	AA	Aa

50% normal but carrier "Heterozygous"

50% Normal

(3) Both Parent Homozygous:

	A	A
a	Aa	Aa
a	Aa	Aa

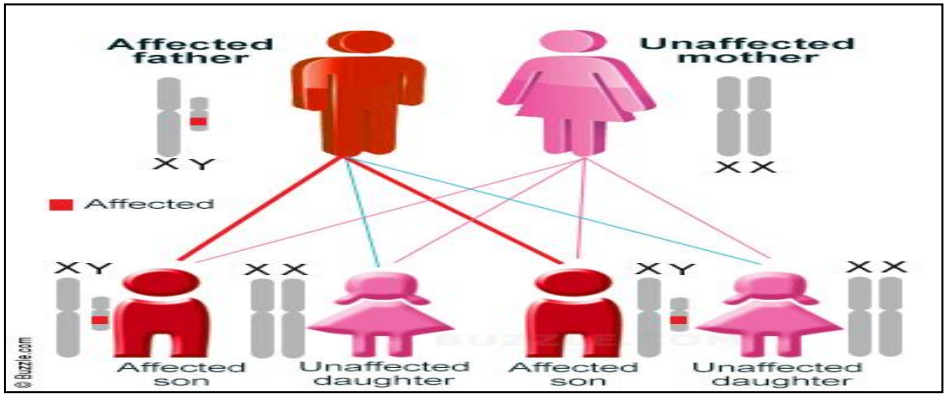
100% offsprings carriers.

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 is different in the males and females.

Y – Linked Inheritance

- The gene is on the Y chromosomes
- Male are Hemizygous, the condition exhibits itself whether dominant or recessive



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 in 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

(1) Normal female, affected male

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

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

(2) Carrier female, normal male

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

- 50% sons affected
- 50% daughters carriers

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.

showing X – linked dominant type of Inheritance

Affected female (heterozygous) and normal male

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

50% sons & 50% daughters are affected

Affected male and normal female

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

All daughters affected, all sons normal

How Mendel's Pea Plants Helped Us Understand Genetics
http://www.youtube.com/watch?v=Mehz7tCxjSE

Heredity
http://www.youtube.com/watch?v=CBezq1fFUEA

pedigrees, Patterns of Genetic Inheritance, Autosomal Dominant Recessive XLinked Mitocondrial
http://www.youtube.com/watch?v=BG6atAkSejc

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 the chromosomal location of the gene locus, which may be autosomal or sex chromosome-linked, and whether the phenotype is dominant or recessive
- Other atypical mode of inheritance will be discussed next lecture.

Quiz yourself!
https://www.examtime.com/en-US/p/1642859-Untitled-quizzes

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عمر العيسى

لا تسكن بلداً لا يكون فيه عالم يفتيك في دينك ولا طبيب ينبئك عن أمر بدنك..
الشافعي

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