HUMAN GENETICS

Lecture Three: MODE OF INHERITANCE

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Objectives:

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

- **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**

Father of Genetics

- Born in 1822
- Monk and teacher
- Discovered some of the basic laws of heredity
- Published his work, entitled Experiments on Plant Hybrids in1866 However, largely ignored
- He died in 1884 with his work still unnoticed
- His work rediscovered in 1900.
- His name is Gregor Mendel

Interpreting the outcomes of Mendel's breeding experiments:

- 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.

Genotypes

Homozygous: same alleles

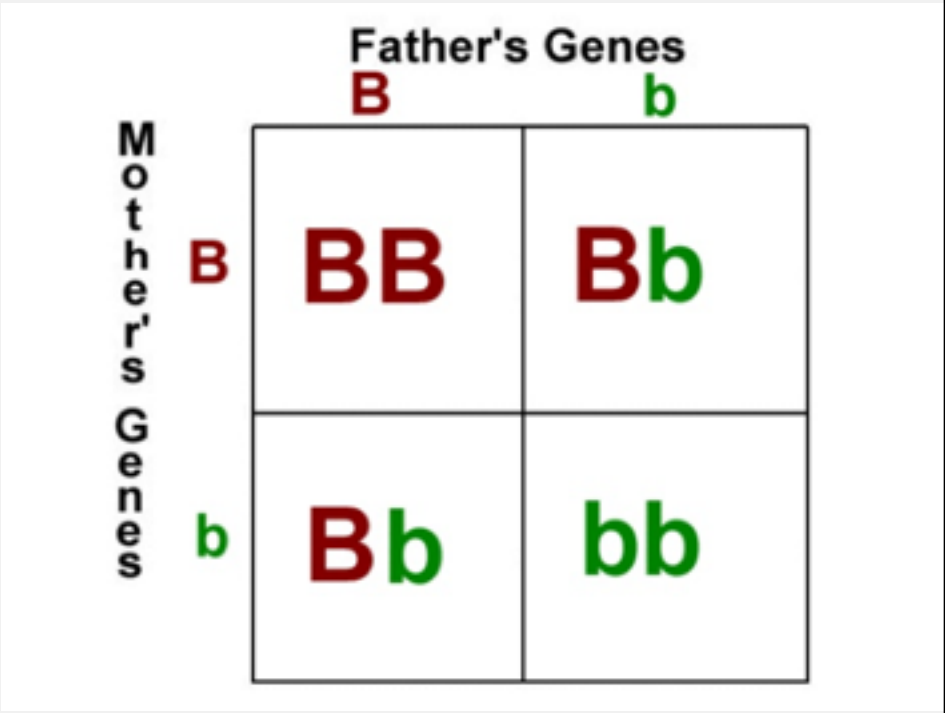
- Homozygous for dominant alleles: **AA**
- Homozygous for recessive alleles: **aa**

Heterozygous: different alleles

- Heterozygous is a combination of recessive and dominant alleles: **Aa**

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.



1. Law of Dominance or Uniformity

COMPLETE DOMINANCE: one allele is dominant to another allele

RECALL MENDEL'S 1st EXPERIMENT

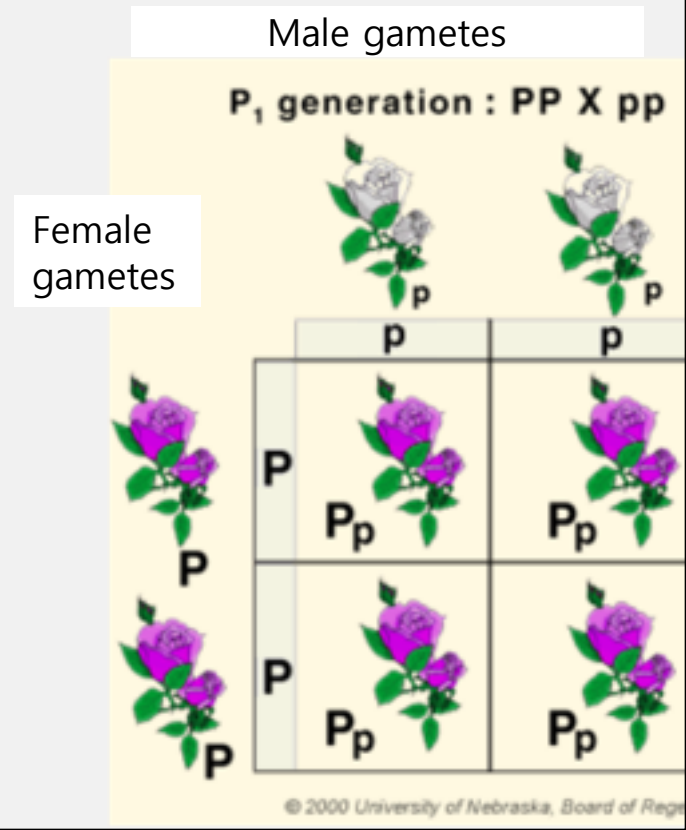
CROSS: Pure bred purple female(Dominant) **X** White male(Recessive)

P1 generation = PP x pp

F1 generation:

Genotype ratio: 1 Pp

Phenotype ratio: 1 purple



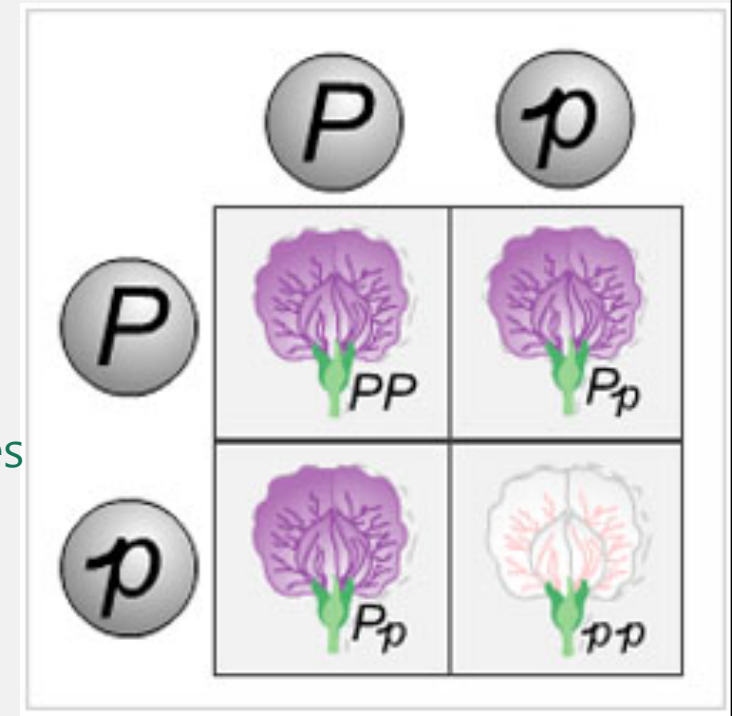
2. Law of Segregation

RECALL MENDEL'S 2nd EXPERIMENT

CROSS: Two F1 generation offspring with each other

P1 generation = Pp x Pp

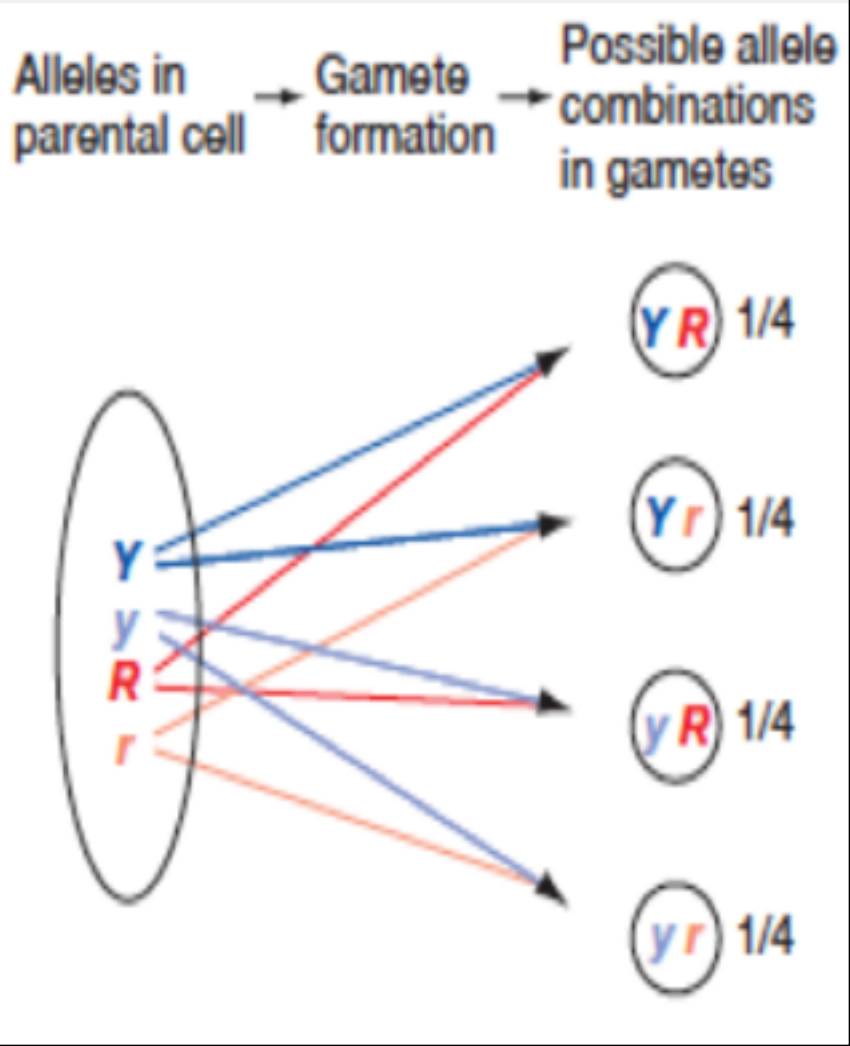
F1 generation:
- Genotype ratio = 1PP:2Pp:1pp
- Phenotype ratio = 3 purple : 1 white



3. Law of Independent Assortment

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.



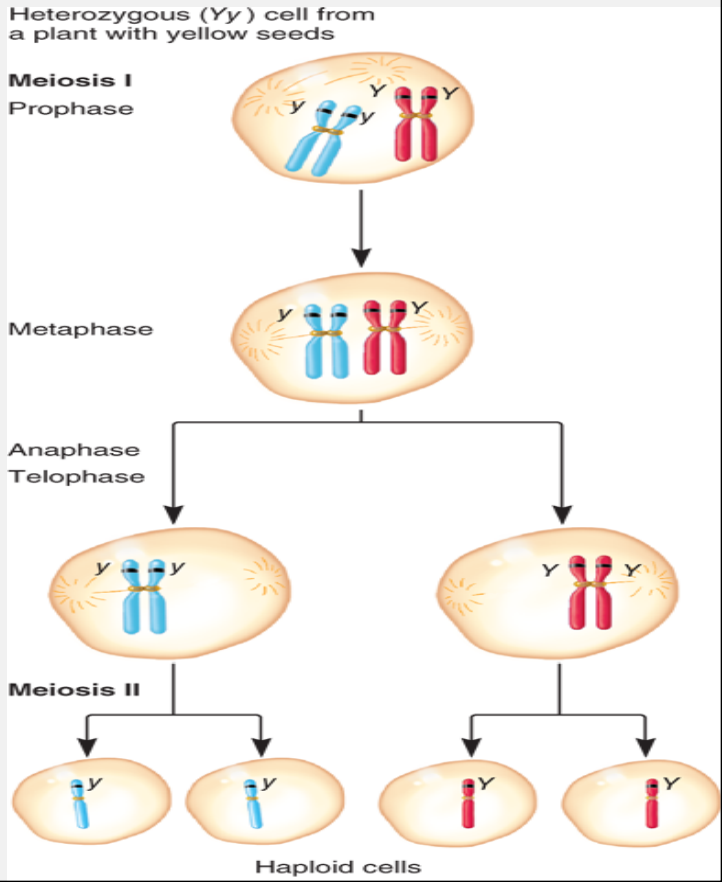
THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES

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

THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES

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

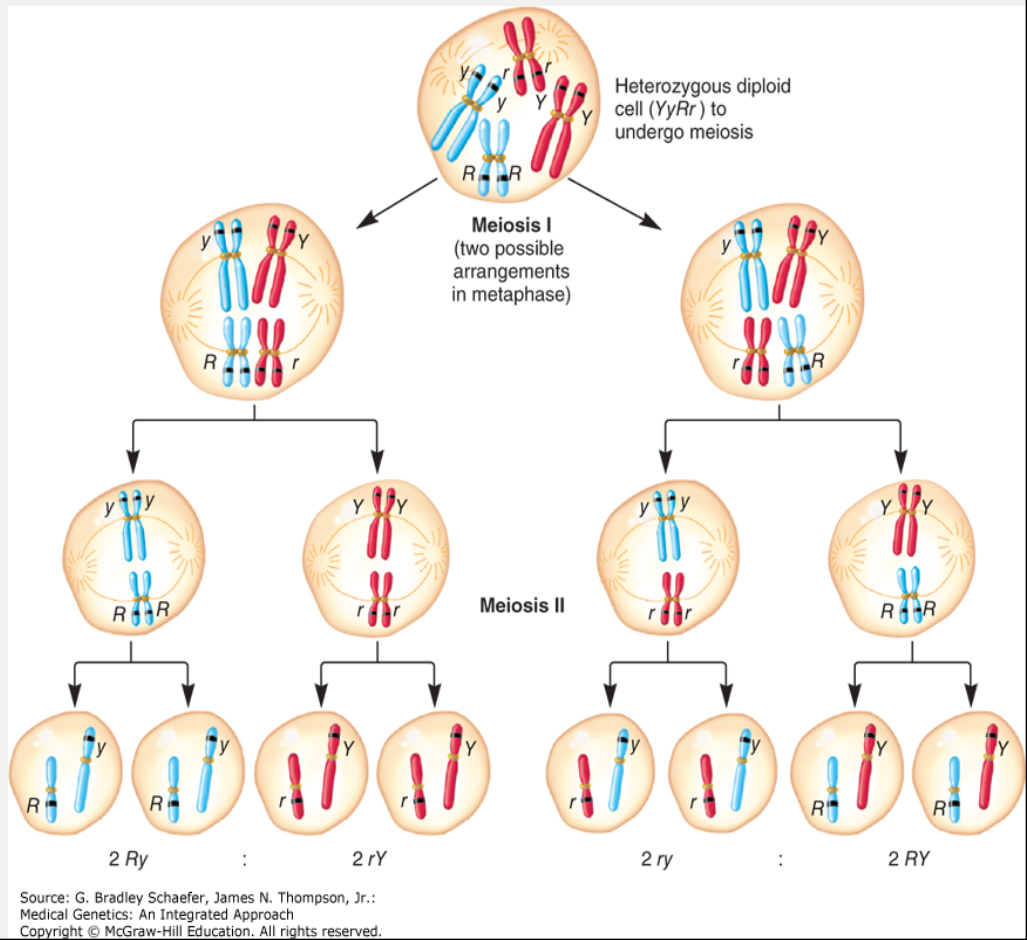
- The two copies of a gene are contained on homologous chromosomes.
- In this example using pea seed color, the two alleles are Y (red) and y (blue).
- During meiosis, the homologous chromosomes segregate from each other, leading to segregation of the two alleles into separate gametes.



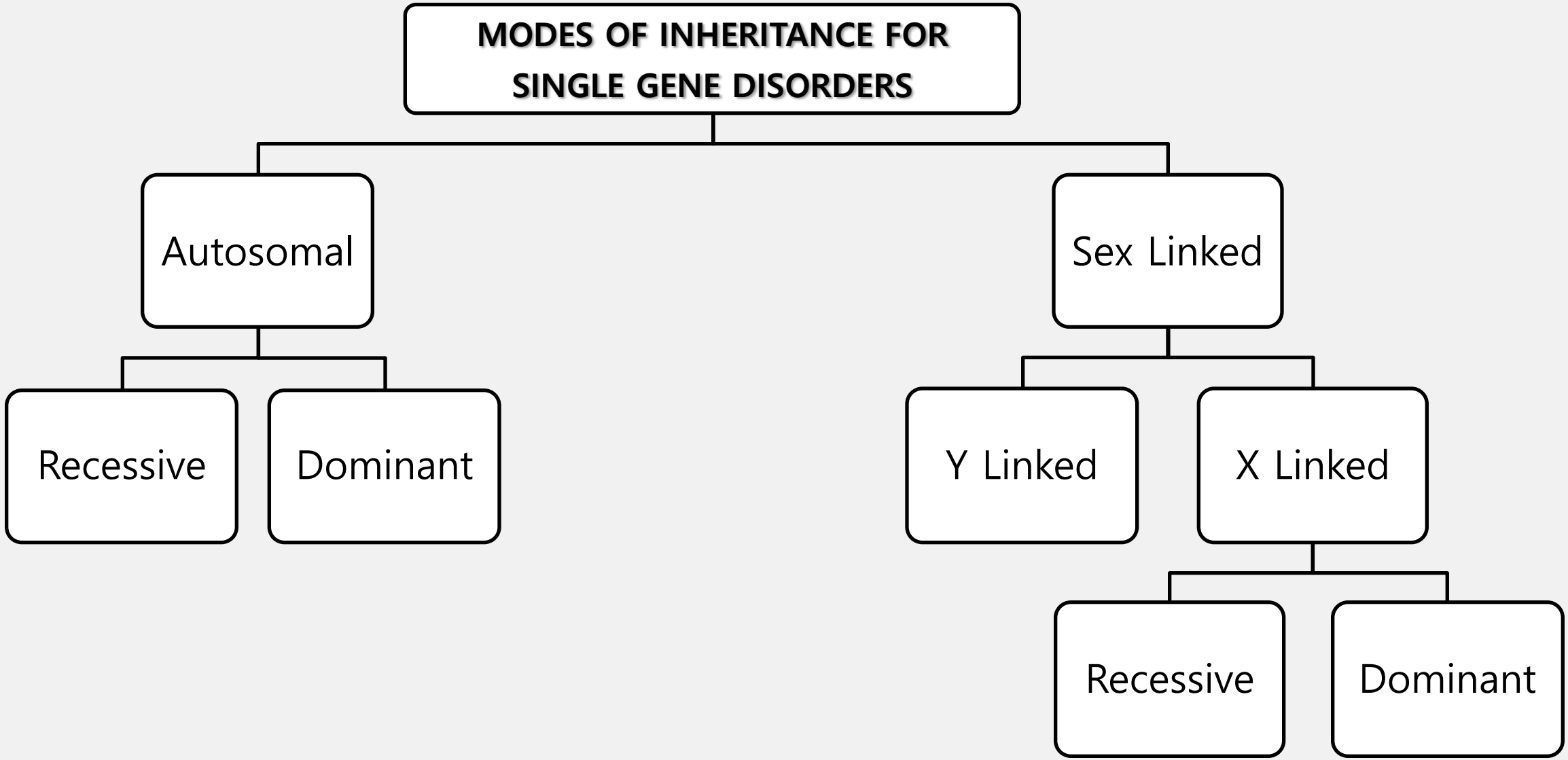
THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES

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

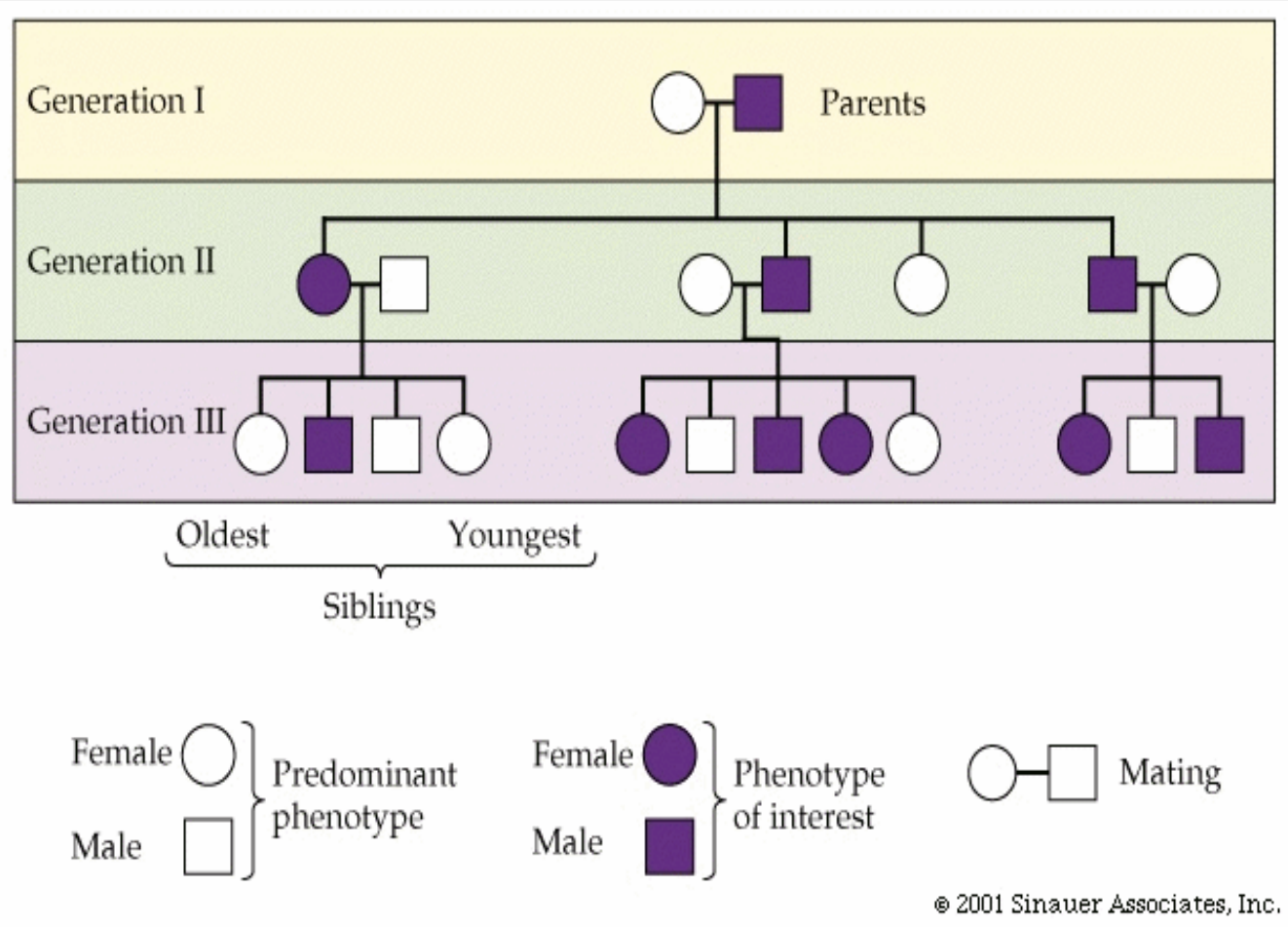
- This figure shows the assortment of two genes located on two different chromosomes, using pea seed color and shape as an example (YyRr).
- During metaphase of meiosis I, different possible arrangements of the homologs within bivalents can lead to different combinations of the alleles in the resulting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



THE CHROMOSOME THEORY OF INHERITANCE AND SEX CHROMOSOMES



A Pedigree Analysis



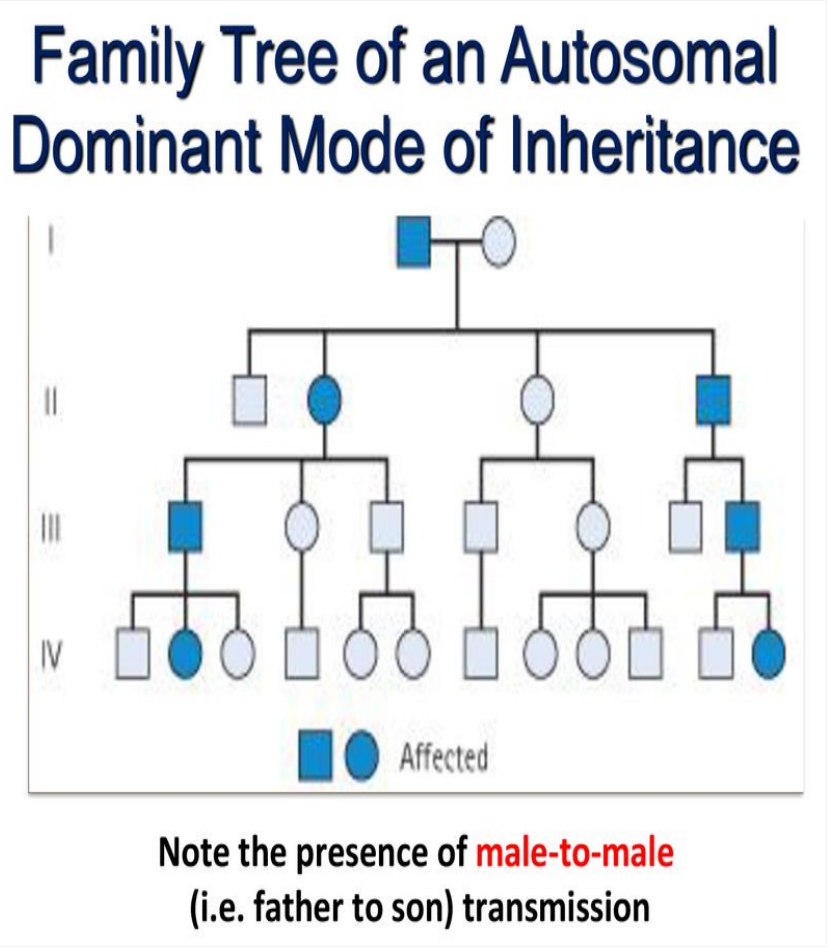
Autosomal Dominant

- 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..



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.

Autosomal Recessive

Punnett square showing autosomal recessive inheritance :

Affected | Carrier | Normal

(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:

- 50% normal but carrier "Heterozygous"
- 50% Normal

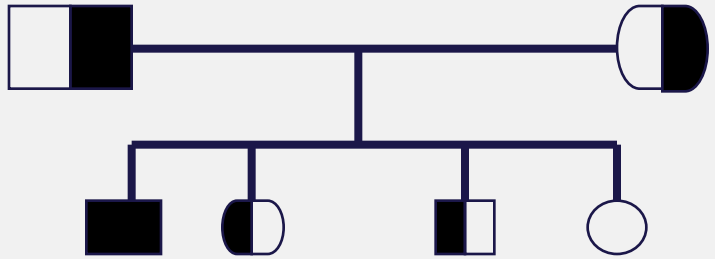
	A	a
A	AA	Aa
A	AA	Aa

(3) One Parent Homozygous:

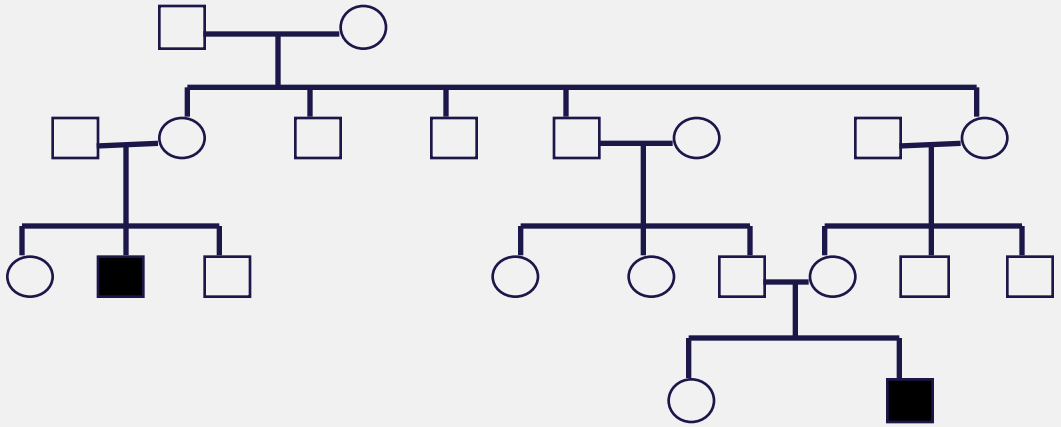
- 100% offspring carriers

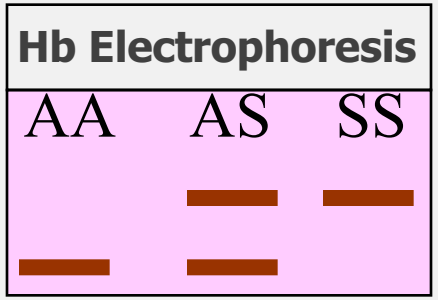
	A	A
a	Aa	Aa
a	Aa	Aa

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



A family with sickle cell disease -Phenotype

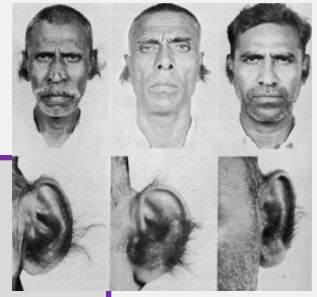




Sex – Linked Inheritance

- Is the inheritance of a gene present on the sex chromosomes (X,Y)
- Sex inheritance pattern (sex chromosomes) is different from autosomal inheritance pattern (autosomes 22 pairs)
- Inheritance differs in males from females

Y-Linked Inheritance



- The gene is on the Y chromosomes
- The gene is passed from fathers to sons only (Males)
- Daughters (Females) are not effected
- Males are Hemizygos, the condition exhibits itself whether dominant or recessive
- Hairy ears in India

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 homo zygous or heterozygous
- These disorders may be : recessive or dominant

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

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, Retinitis pigmentosa

(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	X*X	XX
Y	X*Y	XY

50% sons affected
50% daughters carriers "not affected"

(3) Homozygous female, normal male:

- All sons affected.
- All daughters carriers. "not 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

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% daughters are affected
50% sons

TAKE HOME MESSAGE :

- An accurate determination of the family pedigree is an important part of the work up 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 do minant or recessive

Quiz

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	D) Hemizygous
2-a person with these alleles (TT) is known to be:			
A) Homozygous recessive	B) Homozygous dominant	C) Heterozygous	D) Hemizygous
3-the physical expression of a character is called:			
A) Phenotype	B) Morphology	C) Genotype	D) dominant
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	D) Dominant inheitance
5-an individual inherits .... allele for each gene (character)			
A) 2	B) 3	C) 4	D) 5

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

Thank You

TEAM LEADERS:

- محمد المطيري
- جود العتيبي

GIRLS TEAM MEMBERS:

- طيف الشمري
- سفانا العمر
- ريم القرني
- ريناد الكنعان
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Special thanks to Team 437