Lec 8 pedigree and Pattern of inheritance Mendel's laws

1. A pedigree is a graphical representation of a family's genetic history used to study inheritance patterns of traits or diseases.

- Applications:

1.Identifying carriers of genetic diseases.
2.Predicting recurrence risks for genetic disorders.
3.Counseling families about inherited conditions.

- Importance of Pedigree Analysis

Traces phenotypic expression across multiple generations.
Identifies inheritance patterns (autosomal dominant, autosomal recessive, X-linked, mitochondrial).
Aids in diagnosing rare genetic disorders.

- Standard Pedigree Symbols

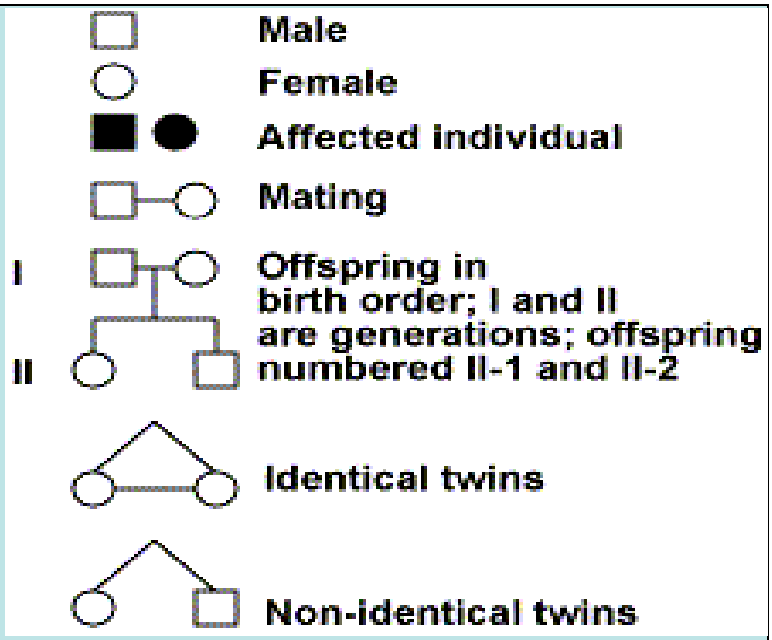
a. **Individuals**: Male: Square (□) Female: Circle (○)

Unknown Sex: Diamond (◇)

b. **Affected Status**:

- Affected: Shaded (■ or ●)
- Carrier: Half-shaded (◧ or ◑)
- Deceased: Slashed symbol (e.g., ⁄□)

c. **Relationships**:
   - Mating Line: Horizontal line connecting two individuals.
   - Offspring Line: Vertical line leading to descendants.
   - Twins: Two diagonal lines from a single point.



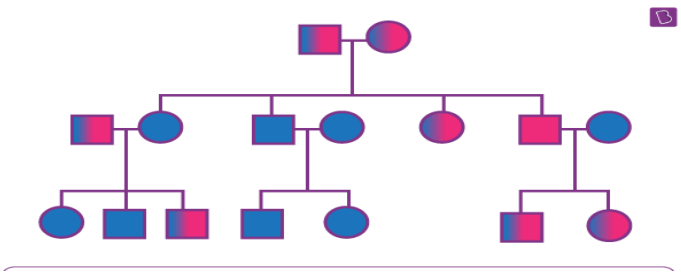


Figure (1) pedigree of family

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2. Mendel's Laws of Inheritance

Gregor Mendel, an Austrian monk, is known as the father of modern genetics. His experiments with pea plants laid the foundation for understanding heredity.

The reason for the selection of pea plants for the genetic experiments are

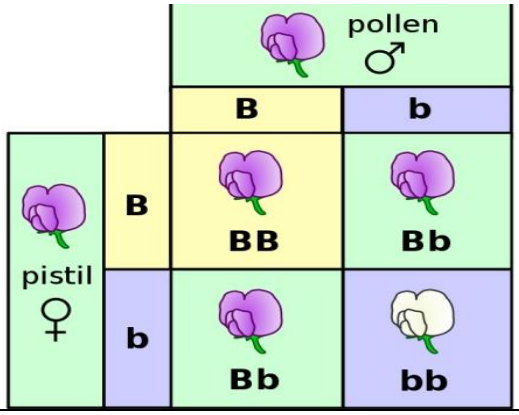
1. Easy to grow in the garden.
2. The flowers of pea plants are hermaphrodite.
3. Easy to obtain pure breed plant through self-fertilization
4. The generation time of pea plants is less.
5. They have excellent disease resistance and have an optimal rate ofsurvival

2. Punnett Square: is a tool used to predict the genotype and phenotype probabilities in offspring based on parental genotypes.

Types of cross

1. Monohybrid Cross: Examining inheritance of a single trait (e.g., pea plant color).

- **Example**: Cross between heterozygous purple-flowered plants (Bb) results in a 1:2:1 genotype ratio and a 3:1 phenotype ratio (purple to white). Purple-flowered plants (Bb) x purple-flowered plants (Bb)



BB: Bb: Bb : bb , phenotype ratio (purple to white) 3:1.

1. Dihybrid Cross: Examining inheritance of two traits (e.g., pea plant color and seed shape). **Example**: Cross between heterozygous purple, round-seeded plants (PpRr) results in a 9:3:3:1 phenotype ratio.

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(PpRr) x PpRr) 9:3:3:1 phenotype ratio

x	PR	Pr	pR	pr
PR	PRPR	PRPr	PRpR	PRpr
Pr	PrPR	PrPr	PrpR	Prpr
pR	pRPR	pR Pr	pRpR	pRpr
pr	prPR	Prpr	pRpr	prpr

NOTE :

Gene made up from 2 alleles, these alleles may be **Homogenous (either AA or aa) heterogenous (Aa)**

Dominant trait: is either AA or Aa
Recessive trait is only aa

Mendel's Law

A. Mendel's first law (Dominance): In a heterogonous trait, the dominant allele masks the expression of the recessive allele.

look at figure (2) if a cross occur between a homozygous dominant (AA) and a homozygous recessive (aa) will always express the dominant phenotype (Aa), while still having a heterozygous genotype

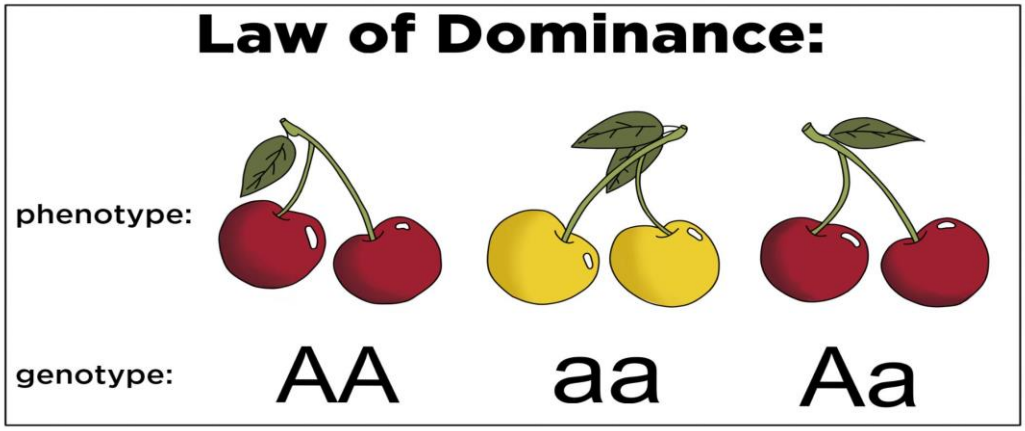

Figure (2) show dominance (**Aa is** heterogenous **trait** (A) masks the expression of a)

Q: In pea plants, the allele for purple flowers (P) is dominant over the allele for white flowers (p).make crossing between them by choose equal cross type and show their phenotypes and genotypes

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- The recessive allele requires two copies to be expressed in the phenotype.:

Example 1: In pea plants, heterogenous allele for purple flowers (Pp) is dominant over the recessive allele for white flowers (pp). Therefore, the genotype Pp and pp results in purple flowers and white flower 1:1

Question: If two pea plants with the recessive trait for white flowers (denoted by "p") are crossed, what will be the genotypes of their offspring?

Recessive homozygotes are produce d by three kinds of mating, although the first of these is by far the most common	
1- Two heterozygotes: Aa x Aa → AA: Aa: Aa: aa 3:1	risk = 25%.
2. Recessive homozygote and heterozygote: aa x Aa → Aa: aa 1:1	risk= 50%.
3. Two recessive homozygotes: aa x aa → aa	risk = 100%

B. (Law of Segregation): that state the two alleles of a pair segregate (or separate) during gamete formation (or meiosis cell division) such that only one allele will be present in each gamete. **law of segregation can be deduced from monohybrid cross**

Example: If a parent has the genotype Aa (where A is dominant and a is recessive), the gametes will carry either A or a.

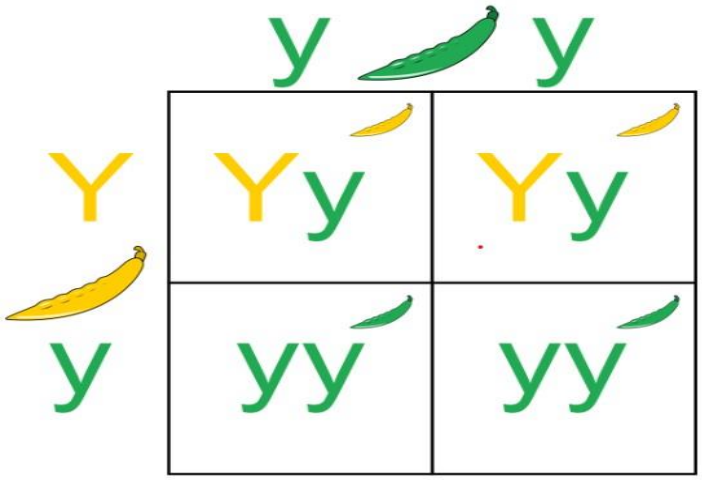

Figure (3a) show cross between homozygous recessive green pea (yy) with heterozygous dominant yellow pea (Yy)

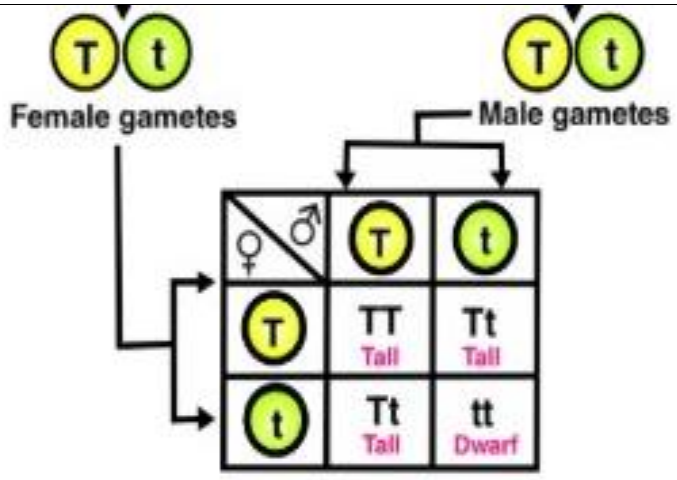

Figure (3 b) show cross between male has heterozygous dominant (Tt) tall with female has same genotype(Tt)

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Question: In pea plants, if a heterozygous purple flower plant (Pp) is crossed with a white flower plant (pp), by choose exact crossing type what will be the genotypes and phenotypes of the offspring, and in what ratio?

C. Mendel's Second Law(Law of Independent Assortment) : Genes for different traits (different pairs of alleles) assort independently of one another during gamete formation, provided the genes are located on different chromosomes.

This law applies only to genes located on different chromosomes or far apart on the same chromosome. This law **can be deduced from dihybrid cross**

Example: If a pea plant's genotype is AaBb, where A and B represent two different genes, the alleles A and B will assort independently into gametes.

AB: Ab: AB: ba

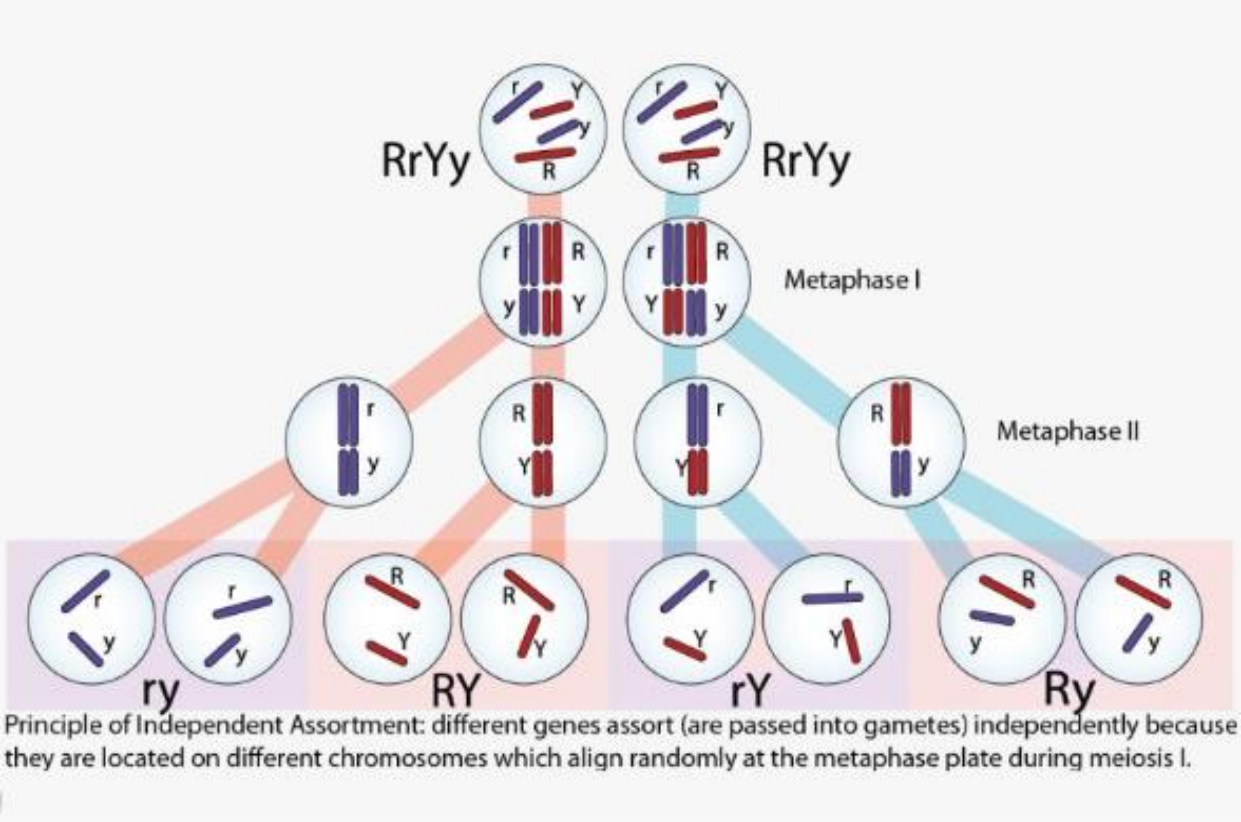

Figure (4) explain Mendel's Second Law(Law of Independent Assortment

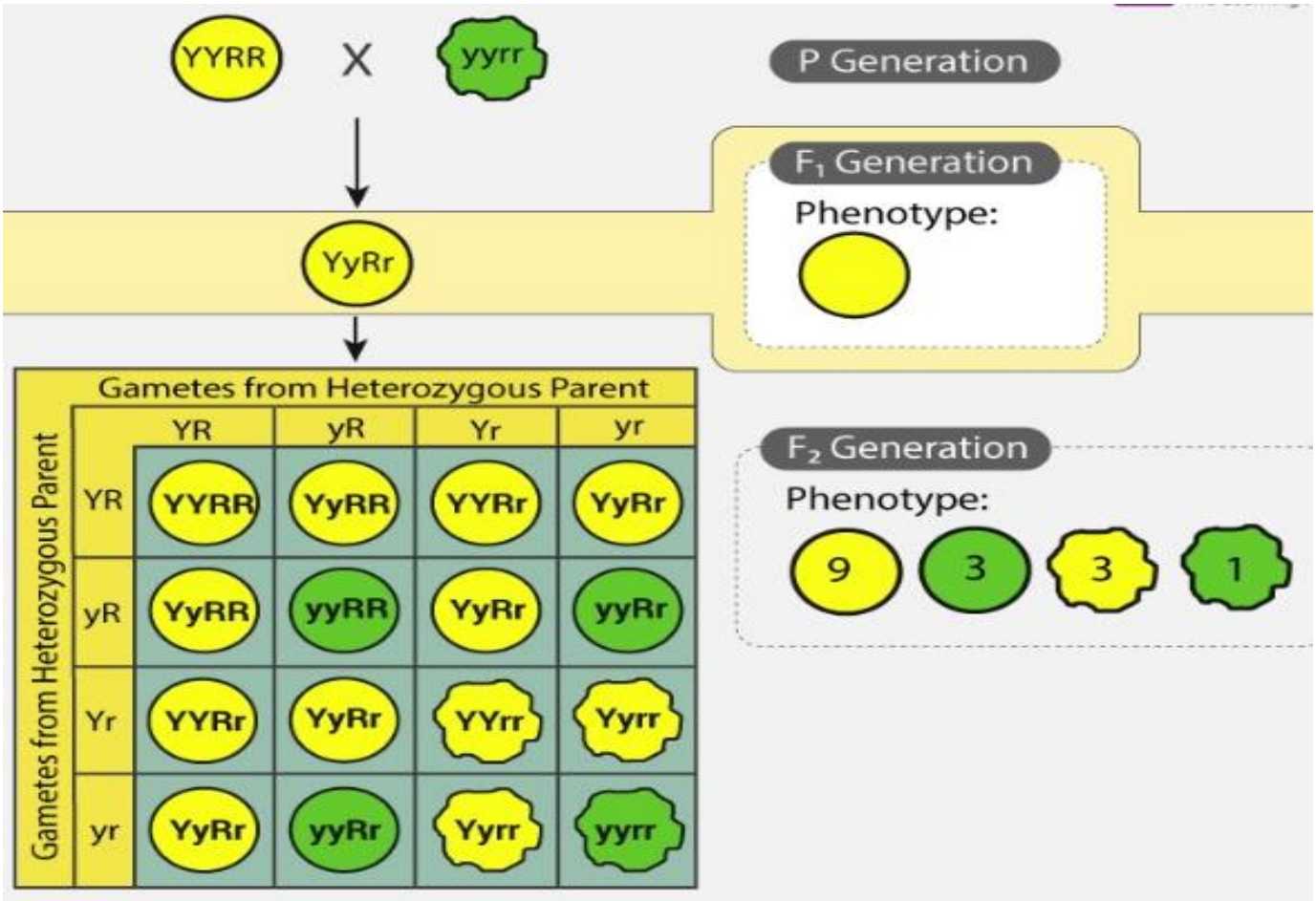

Figure (5) cross between homozygous dominant yellow peas and rounded seeded (YYRR) with homozygous recessive green pea and wrinkled seeded (yyrr)

Question: What will be the genotypes and phenotypes of the offspring when a homozygous dominant soft pea plant is crossed with a homozygous recessive rough pea plant with wrinkled seeds? If you know soft denoted by (S) and rounded seed denoted by (R)

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3.Patterns of Inheritance in Human Genetics

While Mendel's laws apply to many organisms, human inheritance can sometimes exhibit more complex patterns. The most common inheritance patterns include:

a. Autosomal Inheritance (Dominant & recessive): means the gene is located on a non-sex chromosome (not X or Y).
b. Sex linked inheritance(dominant & recessive):mean the transmission of traits located on genes found on the sex chromosomes, primarily the X chromosome
c. Mitochondrial Inheritance

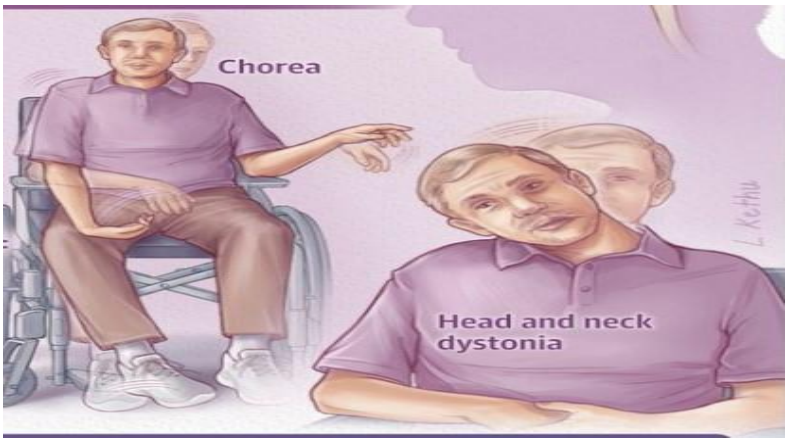
a. Autosomal Inheritance

Autosomal dominant Inheritance

- Characteristics:
- 1. One copy of the dominant allele causes the trait or disease.
- 2. An affected parent can pass the defective gene to their offspring.
- 3. Males and females are affected equally.
- 4. The trait or disorder appears in **every generation**.

Examples: Huntington's disease, Marfan syndrome.

✓ Huntington Disease (HD): A rare brain disorder causing uncontrolled movements (chorea), mental health issues, and memory loss (dementia). Symptoms usually appear between 30-50 years old. In some cases (Juvenile HD), symptoms start before 20 years with school difficulties and behavior problems. Cause: A defective gene with CAG repeats on chromosome 4, More than 36 repeats causes the disease; longer repeats mean earlier onset. Diagnosed Based on symptoms and family history. And Confirmed with a DNA test



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Autosomal Recessive Inheritance

- Characteristics:
- **Recessive disorders** occur when an individual is **homozygous** for a recessive allele (e.g., **a/a**).
- Unaffected individuals have at least one dominant allele (e.g., **A/a** or **A/A**) and show a normal phenotype.
- Affected children are typically born to **unaffected carrier parents**. Examples:

Example: Albinism, Cystic fibrosis, sickle cell anemia. B-Thalassemia and Phenylketonuria

Albinism is an autosomal recessive disorder observed in humans. Caused by mutations in genes responsible for melanin production, with both copies of the gene (alleles) lead to a defect in **tyrosinase** enzyme which involved in melanin synthesis, the pigment responsible for black and brown coloration. Melanin is produced by specialized cells called **melanocytes**, which are primarily found in the skin, hair follicles, and eyes. These **melanocytes** synthesize melanin from the amino acid **tyrosine** , by the enzyme **tyrosinase** which converting tyrosine into melanin.