



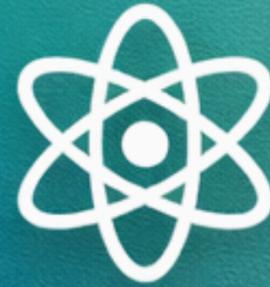
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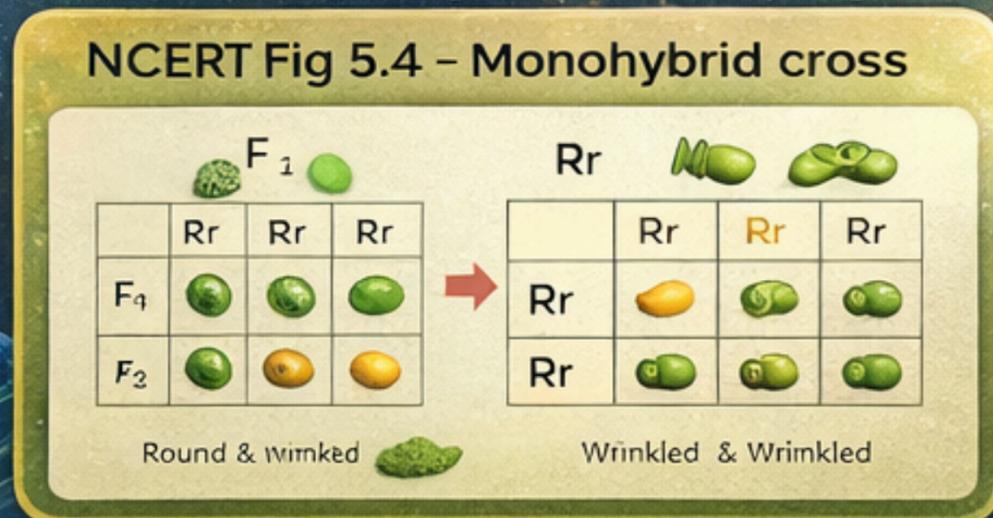
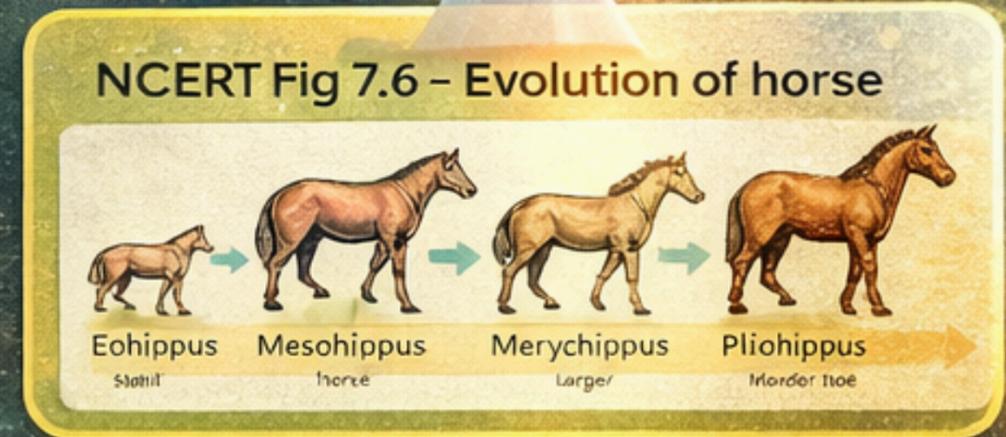
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CHAPTER 4



PRINCIPLES OF INHERITANCE AND VARIATION

- 4.1 Mendel's Laws of Inheritance
- 4.2 Inheritance of One Gene
- 4.3 Inheritance of Two Genes
- 4.4 Sex Determination
- 4.5 Mutation
- 4.6 Genetic Disorders

Have you ever wondered why an elephant always gives birth only to a baby elephant and not some other animal? Or why a mango seed forms only a mango plant and not any other plant?

Given that they do, are the offspring identical to their parents? Or do they show differences in some of their characteristics? Have you ever wondered why siblings sometimes look so similar to each other? Or sometimes even so different?

These and several related questions are dealt with, scientifically, in a branch of biology known as Genetics. This subject deals with the inheritance, as well as the variation of characters from parents to offspring. Inheritance is the process by which characters are passed on from parent to progeny; it is the basis of heredity. Variation is the degree by which progeny differ from their parents.

Humans knew from as early as 8000-1000 B.C. that one of the causes of variation was hidden in sexual reproduction. They exploited the variations that were naturally present in the wild populations of plants and animals to selectively breed and select for organisms that possessed desirable characters. For example, through artificial selection and domestication from ancestral

KEY CONCEPTS

- Genetics is the branch of biology dealing with inheritance and variation.
- Inheritance refers to transmission of characters from parents to offspring.
- Variation refers to differences among offspring.
- The study of heredity explains similarities and differences among siblings.

◆ IMPORTANT EXPLANATIONS

- Offspring resemble parents because of inheritance of traits.
- Differences among individuals occur due to variation.
- Humans recognised early (8000–1000 B.C.) that sexual reproduction causes variation.
- Early humans used selective breeding and domestication to obtain desirable traits.

◆ EXAMPLES (concept understanding)

- Elephant giving birth to an elephant – shows inheritance.
- Mango seed growing into mango plant – species-specific inheritance.
- Selective breeding of plants and animals – use of variation.

◆ DEFINITIONS / TERMS

- Genetics – study of heredity and variation.
- Inheritance (Heredity) – transmission of traits from parents to offspring.
- Variation – differences in traits among individuals.

FIGURES / DIAGRAMS

- No specific figure on this page (chapter introduction layout only).

◆ MEMORY TRIGGERS (quick revision keywords)

- Genetics → inheritance + variation
- Heredity → traits passed
- Variation → differences
- Selective breeding
- Sexual reproduction → variation

| Character | Dominant trait | Recessive trait |
|-----------------|---|--|
| Seed shape |  Round |  Wrinkled |
| Seed colour |  Yellow |  Green |
| Flower colour |  Violet |  White |
| Pod shape |  Full |  Constricted |
| Pod colour |  Green |  Yellow |
| Flower position |  Axial |  Terminal |
| Stem height |  Tall |  Dwarf |

Figure 4.1 Seven pairs of contrasting traits in pea plant studied by Mendel

wild cows, we have well-known Indian breeds, e.g., Sahiwal cows in Punjab. We must, however, recognise that though our ancestors knew about the inheritance of characters and variation, they had very little idea about the scientific basis of these phenomena.

4.1 MENDEL'S LAWS OF INHERITANCE

It was during the mid-nineteenth century that headway was made in the understanding of inheritance. Gregor Mendel, conducted hybridisation experiments on garden peas for seven years (1856-1863) and proposed the laws of inheritance in living organisms. During Mendel's investigations into inheritance patterns it was for the first time that statistical analysis and mathematical logic were applied to problems in biology. His experiments had a large sampling size, which gave greater credibility to the data that he collected. Also, the confirmation of his inferences from experiments on successive generations of his test plants, proved that his results pointed to general rules of inheritance rather than being unsubstantiated ideas. Mendel investigated characters in the garden pea plant that were manifested as two opposing traits, e.g., tall or dwarf plants, yellow or green seeds. This allowed him to set up a basic framework of rules governing inheritance, which was expanded on by later scientists to account for all the diverse natural observations and the complexity inherent in them.

Mendel conducted such artificial pollination/cross pollination experiments using several true-breeding pea lines. A true-breeding line is one that, having undergone

continuous self-pollination, shows the stable trait inheritance and expression for several generations. Mendel selected 14 true-breeding pea plant varieties, as pairs which were similar except for one character with contrasting traits. Some of the contrasting traits selected were smooth or wrinkled seeds, yellow or green seeds, inflated (full) or constricted green or yellow pods and tall or dwarf plants (Figure 4.1, Table 4.1).

KEY CONCEPTS

- Gregor Mendel proposed the laws of inheritance through experiments on garden pea plants.
- He used hybridisation experiments and applied statistical analysis.
- Mendel studied contrasting traits to understand inheritance patterns.
- Experiments used true-breeding pea lines for accuracy.

◆ IMPORTANT EXPLANATIONS

- Mendel's work (1856–1863) provided a scientific basis of heredity.
- Large sample size and successive generations increased reliability of results.
- True-breeding lines show stable traits after continuous self-pollination.
- Mendel selected plants similar except for one contrasting character to study inheritance.
- His findings formed a framework of general rules of inheritance later expanded by scientists.

◆ EXAMPLES (concept understanding)

- Smooth vs wrinkled seeds
- Yellow vs green seeds
- Violet vs white flowers
- Full vs constricted pods
- Green vs yellow pods
- Axial vs terminal flower position
- Tall vs dwarf plants

◆ DEFINITIONS / TERMS

- Hybridisation – crossing of plants with contrasting traits.
- True-breeding line – plants producing same traits over generations.
- Contrasting traits – two opposite forms of a character.

| Character | Dominant trait | Recessive trait |
|-----------------|---|--|
| Seed shape |  Round |  Wrinkled |
| Seed colour |  Yellow |  Green |
| Flower colour |  Violet |  White |
| Pod shape |  Full |  Constricted |
| Pod colour |  Green |  Yellow |
| Flower position |  Axial |  Terminal |
| Stem height |  Tall |  Dwarf |

Figure 4.1 Seven pairs of contrasting traits in pea plant studied by Mendel

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FIGURES / DIAGRAMS

• Fig 4.1 – shows seven pairs of contrasting traits in pea plant studied by Mendel.

◆ MEMORY TRIGGERS (quick revision keywords)

- Mendel → pea plant
- 1856–1863 experiments
- True-breeding lines
- Hybridisation
- Seven contrasting traits

Table 4.1: Contrasting Traits Studied by Mendel in Pea

| S.No. | Characters | Contrasting Traits |
|-------|-----------------|----------------------|
| 1. | Stem height | Tall/dwarf |
| 2. | Flower colour | Violet/white |
| 3. | Flower position | Axial/terminal |
| 4. | Pod shape | Inflated/constricted |
| 5. | Pod colour | Green/yellow |
| 6. | Seed shape | Round/wrinkled |
| 7. | Seed colour | Yellow/green |

4.2 INHERITANCE OF ONE GENE

Let us take the example of one such hybridisation experiment carried out by Mendel where he crossed tall and dwarf pea plants to study the inheritance of one gene (Figure 4.2). He collected the seeds produced as a result of this cross and grew them to generate plants of the first hybrid generation. This generation is also called the **Filial₁ progeny** or the **F₁**. Mendel observed that all the F₁ progeny plants were tall, like one of its parents; none were dwarf (Figure 4.3). He made similar observations for the other pairs of traits – he found that the F₁ always resembled either one of the parents, and that the trait of the other parent was not seen in them.

Mendel then self-pollinated the tall F₁ plants and to his surprise found that in the Filial₂ generation some of the offspring were 'dwarf'; the character that was not seen in the F₁ generation was now expressed. The proportion of plants that were dwarf were 1/4th of the F₂ plants while 3/4th of the F₂ plants were tall. The tall and dwarf traits were identical to their parental type and did not show any blending, that is all the offspring were either tall or dwarf, none were of in-between height (Figure 4.3).

Similar results were obtained with the other traits that he studied: only one of the parental traits was expressed in the F₁ generation while at the F₂ stage both the traits were expressed in the proportion 3:1. The contrasting traits did not show any blending at either F₁ or F₂ stage.

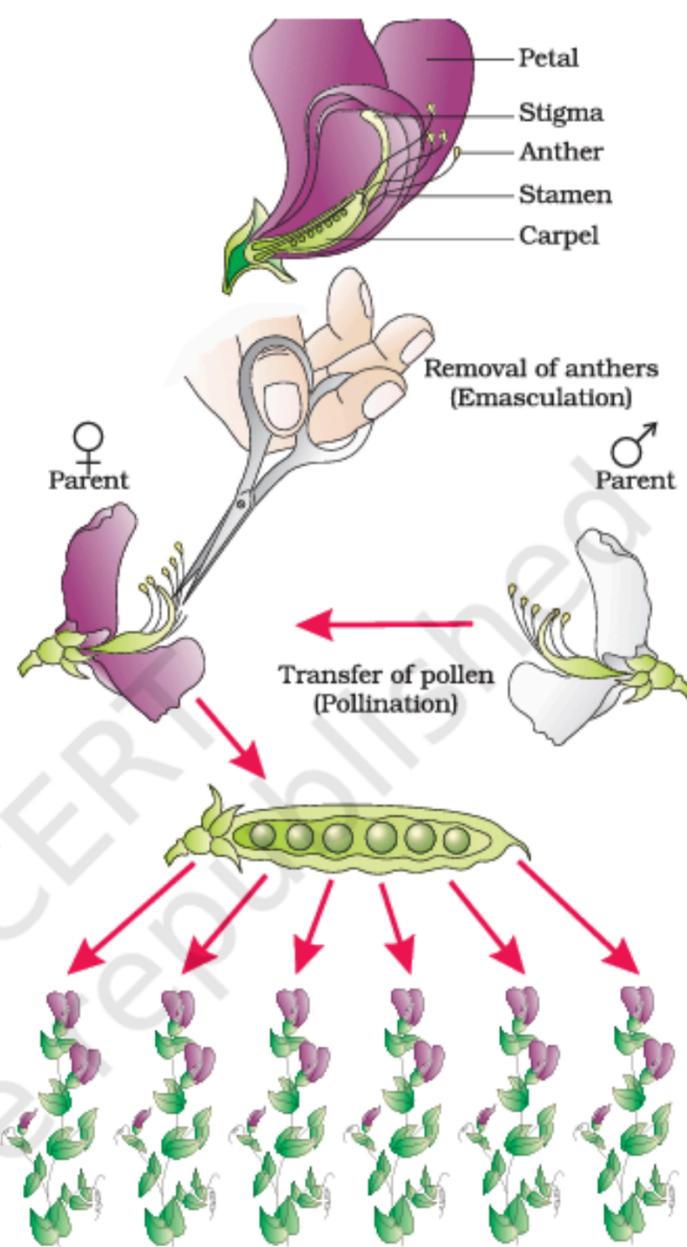


Figure 4.2 Steps in making a cross in pea

KEY CONCEPTS

- Mendel studied inheritance of one gene using tall × dwarf pea cross.
- First hybrid generation is called F₁ (Filial generation).
- F₁ plants all showed dominant trait (tall).
- In F₂ generation, both traits reappeared in 3:1 ratio.
- Traits showed no blending; offspring were either tall or dwarf.

◆ IMPORTANT EXPLANATIONS

- Mendel crossed true-breeding tall and dwarf plants.
- All F₁ progeny resembled one parent only (dominant trait).
- Self-pollination of F₁ plants produced F₂ generation.
- In F₂, recessive trait reappeared, proving traits are inherited as discrete units.
- Similar pattern observed for other contrasting traits.

◆ EXAMPLES (concept understanding)

- Tall × dwarf cross → F₁ all tall.
- F₂ ratio: 3 tall : 1 dwarf.
- Same pattern seen in other pea traits.

DEFINITIONS / TERMS

- F₁ generation – first filial generation produced from parental cross.
- F₂ generation – offspring produced by self-pollination of F₁.
- Dominant trait – trait expressed in F₁ generation.
- Recessive trait – trait masked in F₁ but reappears in F₂.

◆ FIGURES / DIAGRAMS

- Fig 4.2 – shows steps in making a cross in pea (emasculatation, pollination, offspring).
- Table 4.1 – lists seven contrasting traits studied by Mendel.

◆ MEMORY TRIGGERS (quick revision keywords)

- Monohybrid cross
- F₁ → all dominant
- F₂ → 3:1 ratio
- No blending
- Dominant vs recessive

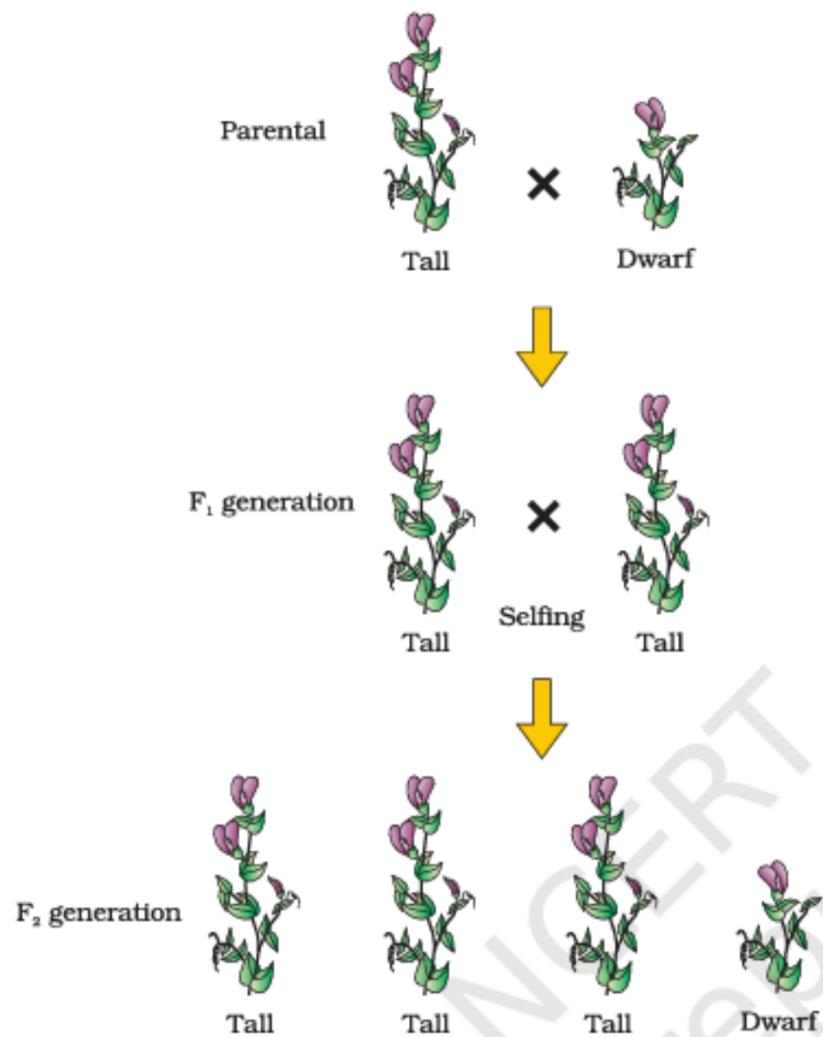


Figure 4.3 Diagrammatic representation of monohybrid cross

identical or **homozygous**, **TT** and **tt**, respectively. **TT** and **tt** are called the **genotype** of the plant while the descriptive terms **tall** and **dwarf** are the **phenotype**. What then would be the phenotype of a plant that had a genotype **Tt**?

As Mendel found the phenotype of the F₁ heterozygote **Tt** to be exactly like the **TT** parent in appearance, he proposed that in a pair of dissimilar factors, one dominates the other (as in the F₁) and hence is called the **dominant** factor while the other factor is **recessive**. In this case **T** (for tallness) is dominant over **t** (for dwarfness), that is recessive. He observed identical behaviour for all the other characters/trait-pairs that he studied.

It is convenient (and logical) to use the capital and lower case of an alphabetical symbol to remember this concept of dominance and recessiveness. (Do not use **T** for tall and **d** for dwarf because you will find it difficult to remember whether **T** and **d** are alleles of the same gene/character or not). Alleles can be similar as in the case of homozygotes **TT** and **tt** or can be dissimilar as in the case of the heterozygote **Tt**. Since

Based on these observations, Mendel proposed that something was being stably passed down, unchanged, from parent to offspring through the gametes, over successive generations. He called these things as 'factors'. Now we call them as **genes**. Genes, therefore, are the units of inheritance. They contain the information that is required to express a particular trait in an organism. Genes which code for a pair of contrasting traits are known as **alleles**, i.e., they are slightly different forms of the same gene.

If we use alphabetical symbols for each gene, then the capital letter is used for the trait expressed at the F₁ stage and the small alphabet for the other trait. For example, in case of the character of height, **T** is used for the Tall trait and **t** for the 'dwarf', and **T** and **t** are alleles of each other. Hence, in plants the pair of alleles for height would be **TT**, **Tt** or **tt**. Mendel also proposed that in a true breeding, tall or dwarf pea variety the allelic pair of genes for height are

KEY CONCEPTS

- Mendel proposed that traits are controlled by factors (genes) passed through gametes.
- Genes are the units of inheritance.
- Different forms of a gene are called alleles.
- Traits are expressed as genotype (genetic makeup) and phenotype (observable trait).
- Dominance explains why only one trait appears in F₁.

♦ IMPORTANT EXPLANATIONS

- Genes carry information required for expression of traits.
- Capital letters denote dominant allele and small letters denote recessive allele.
- For plant height: T (tall) and t (dwarf) are alleles.
- Possible allele combinations: TT, Tt, tt.
- Homozygous – identical alleles (TT or tt).
- Heterozygous – different alleles (Tt).
- Phenotype of Tt is tall because dominant allele masks recessive allele.

♦ EXAMPLES (concept understanding)

- TT or Tt → Tall phenotype.
- tt → Dwarf phenotype.
- F₁ heterozygote showing same appearance as dominant parent.

DEFINITIONS / TERMS

- Gene – unit of inheritance controlling a trait.
- Alleles – alternative forms of a gene.
- Genotype – genetic constitution of an organism.
- Phenotype – observable characteristics.
- Homozygous – identical allele pair.
- Heterozygous – different allele pair.
- Dominant – allele expressed in heterozygous condition.
- Recessive – allele masked in heterozygous condition.

♦ MEMORY TRIGGERS (quick revision keywords)

- Genes → factors
- Alleles → T & t
- Genotype vs phenotype
- Homozygous vs heterozygous
- Dominant masks recessive

the **Tt** plant is heterozygous for genes controlling one character (height), it is a **monohybrid** and the cross between **TT** and **tt** is a **monohybrid cross**.

From the observation that the recessive parental trait is expressed without any blending in the F_2 generation, we can infer that, when the tall and dwarf plant produce gametes, by the process of meiosis, the alleles of the parental pair separate or **segregate** from each other and only one allele is transmitted to a gamete. This segregation of alleles is a random process and so there is a 50 per cent chance of a gamete containing either allele, as has been verified by the results of the crossings. In this way the gametes of the tall **TT** plants have the allele **T** and the gametes of the dwarf **tt** plants have the allele **t**. During fertilisation the two alleles, **T** from one parent say, through the pollen, and **t** from the other parent, then through the egg, are united to produce zygotes that have one **T** allele and one **t** allele. In other words the hybrids have **Tt**. Since these hybrids contain alleles which express contrasting traits, the plants are **heterozygous**. The production of gametes by the parents, the formation of the zygotes, the F_1 and F_2 plants can be understood from a diagram called **Punnett Square** as shown in Figure 4.4. It was developed by a British geneticist, Reginald C. Punnett. It is a graphical representation to calculate the probability of all possible genotypes of offspring in a genetic cross. The possible gametes are written on two sides, usually the top row and left columns. All possible combinations are represented in boxes below in the squares, which generates a square output form.

The Punnett Square shows the parental tall **TT** (male) and dwarf **tt** (female) plants, the gametes produced by them and, the F_1 **Tt** progeny. The F_1 plants of genotype **Tt** are self-pollinated. The symbols ♀ and ♂ are used to denote the female (eggs) and male (pollen) of the F_1 generation, respectively. The F_1 plant of the genotype **Tt** when self-pollinated, produces gametes of the genotype **T** and **t** in equal proportion. When fertilisation takes place, the pollen grains of genotype **T** have a 50 per cent chance to pollinate eggs of the genotype **T**, as well as of genotype **t**. Also pollen grains of genotype **t** have a 50 per cent chance of pollinating eggs of genotype **T**, as well as of

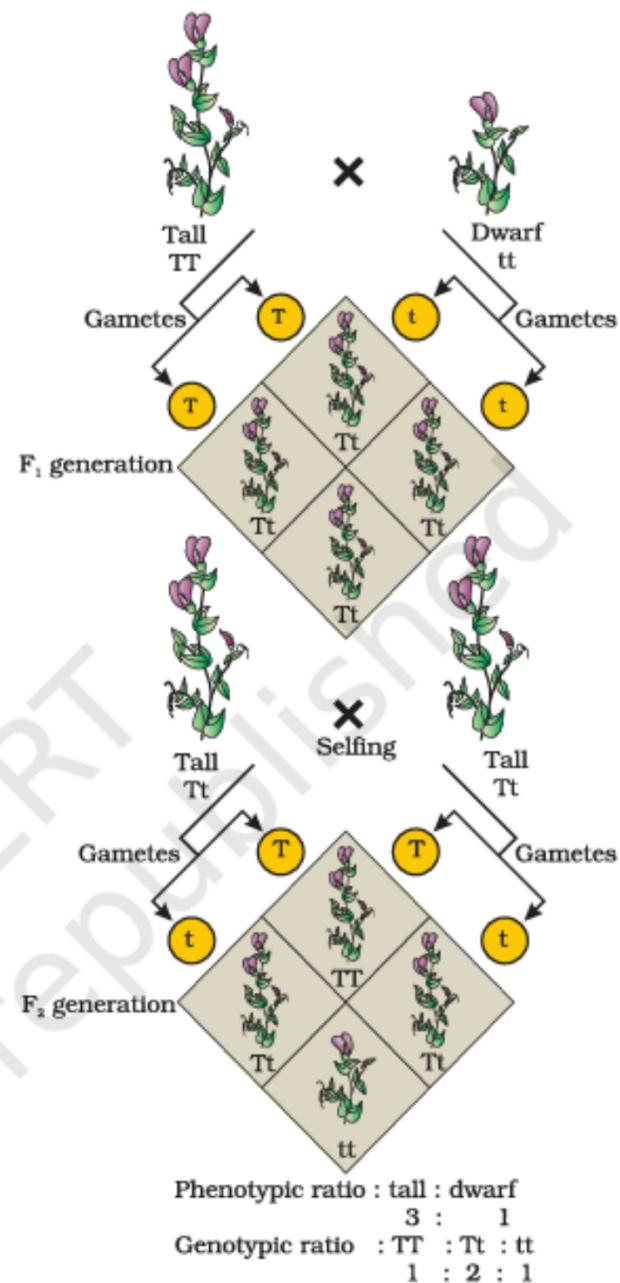


Figure 4.4 A Punnett square used to understand a typical monohybrid cross conducted by Mendel between true-breeding tall plants and true-breeding dwarf plants

KEY CONCEPTS

- Monohybrid cross involves inheritance of one character (e.g., height).
- During gamete formation, alleles segregate so each gamete carries only one allele.
- Segregation is random, giving 50% chance for each allele.
- Punnett square is used to predict genotypic and phenotypic ratios.

♦ IMPORTANT EXPLANATIONS

- In cross $TT \times tt$, parents produce gametes T and t respectively.
- Fertilisation results in Tt hybrids (heterozygous) in F_1 generation.
- Selfing of $Tt \times Tt$ produces F_2 generation.
- F_2 shows phenotypic ratio 3 tall : 1 dwarf.
- F_2 shows genotypic ratio 1 TT : 2 Tt : 1 tt.
- Punnett square gives a graphical representation of allele combinations.

♦ EXAMPLES (concept understanding)

- TT (tall) \times tt (dwarf) $\rightarrow F_1$ all Tt (tall).
- $Tt \times Tt \rightarrow F_2$ showing 3:1 phenotype ratio.

♦ DEFINITIONS / TERMS

- Monohybrid – organism heterozygous for one character.
- Monohybrid cross – cross involving one trait.
- Segregation – separation of alleles during gamete formation.
- Punnett square – diagram to calculate genetic probabilities.

MEMORY TRIGGERS (quick revision keywords)

- Law of segregation
- Gametes \rightarrow one allele
- Punnett square
- F_2 ratio \rightarrow 3:1
- Genotype \rightarrow 1:2:1

genotype **t**. As a result of random fertilisation, the resultant zygotes can be of the genotypes **TT**, **Tt** or **tt**.

From the Punnett square it is easily seen that $1/4^{\text{th}}$ of the random fertilisations lead to **TT**, $1/2$ lead to **Tt** and $1/4^{\text{th}}$ to **tt**. Though the F_1 have a genotype of **Tt**, but the phenotypic character seen is 'tall'. At F_2 , $3/4^{\text{th}}$ of the plants are tall, where some of them are **TT** while others are **Tt**. Externally it is not possible to distinguish between the plants with the genotypes **TT** and **Tt**. Hence, within the genotypic pair **Tt** only one character '**T**' tall is expressed. Hence the character **T** or 'tall' is said to dominate over the other allele **t** or 'dwarf' character. It is thus due to this dominance of one character over the other that all the F_1 are tall (though the genotype is **Tt**) and in the F_2 $3/4^{\text{th}}$ of the plants are tall (though genotypically $1/2$ are **Tt** and only $1/4^{\text{th}}$ are **TT**). This leads to a phenotypic ratio of $3/4^{\text{th}}$ tall : ($1/4$ **TT** + $1/2$ **Tt**) and $1/4^{\text{th}}$ **tt**, i.e., a 3:1 ratio, but a genotypic ratio of 1:2:1.

The $1/4 : 1/2 : 1/4$ ratio of **TT**: **Tt**: **tt** is mathematically condensable to the form of the binomial expression $(ax + by)^2$, that has the gametes bearing genes **T** or **t** in equal frequency of $1/2$. The expression is expanded as given below :

$$(1/2T + 1/2t)^2 = (1/2T + 1/2t) \times (1/2T + 1/2t) = 1/4 TT + 1/2Tt + 1/4 tt$$

Mendel self-pollinated the F_2 plants and found that dwarf F_2 plants continued to generate dwarf plants in F_3 and F_4 generations. He concluded that the genotype of the dwarfs was homozygous – **tt**. What do you think he would have got had he self-pollinated a tall F_2 plant?

From the preceding paragraphs it is clear that though the genotypic ratios can be calculated using mathematical probability, by simply looking at the phenotype of a dominant trait, it is not possible to know the genotypic composition. That is, for example, whether a tall plant from F_1 or F_2 has **TT** or **Tt** composition, cannot be predicted. Therefore, to determine the genotype of a tall plant at F_2 , Mendel crossed the tall plant from F_2 with a dwarf plant. This he called a **test cross**. In a typical test cross an organism (pea plants here) showing a dominant phenotype (and whose genotype is to be determined) is crossed with the recessive parent instead of self-crossing. The progenies of such a cross can easily be analysed to predict the genotype of the test organism. Figure 4.5 shows the results of typical test cross where violet colour flower (V) is dominant over white colour flower (v).

Using Punnett square, try to find out the nature of offspring of a test cross.

What ratio did you get?

Using the genotypes of this cross, can you give a general definition for a test cross?

KEY CONCEPTS

- In F_2 generation, genotypes occur in 1:2:1 ratio (TT:Tt:tt).
- Dominance causes Tt and TT plants to appear tall.
- Phenotypic ratio in F_2 is 3 tall : 1 dwarf.
- Test cross is used to determine the genotype of a dominant phenotype.

◆ IMPORTANT EXPLANATIONS

- Random fertilisation produces $1/4$ TT, $1/2$ Tt, $1/4$ tt.
- Though F_1 genotype is Tt, phenotype appears tall due to dominance.
- External appearance cannot distinguish TT from Tt.
- Mathematical probability explains ratios using binomial expansion.
- Mendel confirmed that dwarf plants are homozygous (tt) through repeated selfing.
- To find genotype of a tall plant, Mendel crossed it with a recessive parent (tt).
- In a test cross, progeny analysis reveals the genotype of the unknown parent.

◆ EXAMPLES (concept understanding)

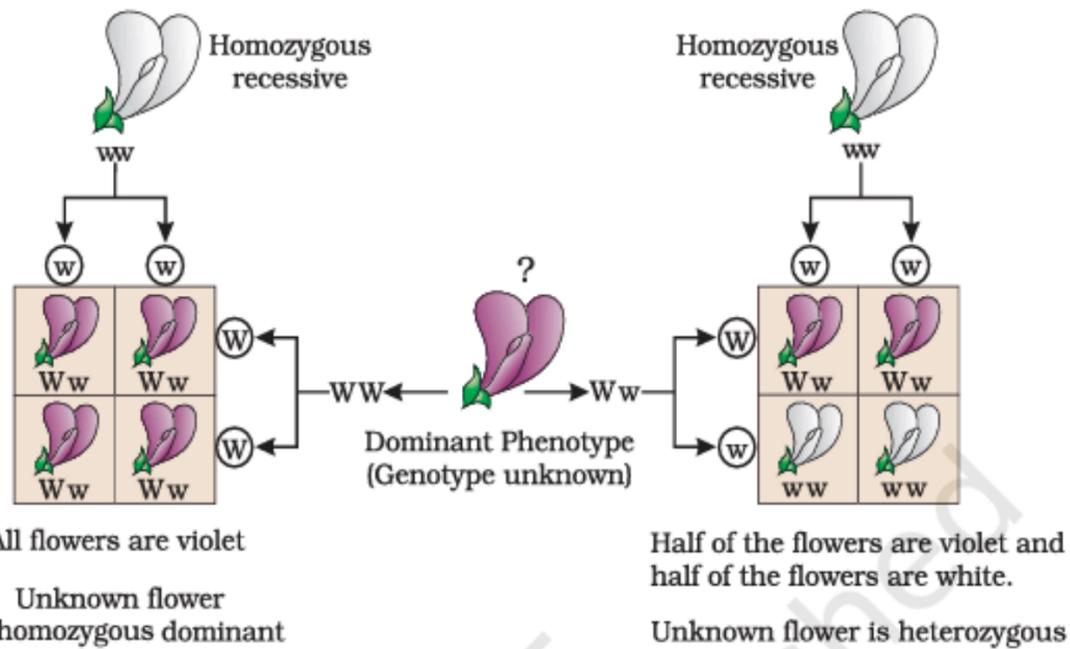
- Tall (TT or Tt) × dwarf (tt) — used as a test cross.
- Observation of offspring ratios helps identify genotype of tall plant.

◆ DEFINITIONS / TERMS

- Genotypic ratio — proportion of different genotypes in offspring.
- Phenotypic ratio — proportion of observable traits.
- Test cross — cross of dominant phenotype with recessive parent to determine genotype.

◆ MEMORY TRIGGERS (quick revision keywords)

- Genotype ratio → 1:2:1
- Phenotype ratio → 3:1
- TT vs Tt indistinguishable
- Test cross → with recessive
- Dominance



Result All flowers are violet

Interpretation Unknown flower is homozygous dominant

Half of the flowers are violet and half of the flowers are white.

Unknown flower is heterozygous

Figure 4.5 Diagrammatic representation of a test cross

Based on his observations on monohybrid crosses Mendel proposed two general rules to consolidate his understanding of inheritance in monohybrid crosses. Today these rules are called the **Principles or Laws of Inheritance**: the First Law or **Law of Dominance** and the Second Law or **Law of Segregation**.

4.2.1 Law of Dominance

- (i) Characters are controlled by discrete units called **factors**.
- (ii) Factors occur in pairs.
- (iii) In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

The law of dominance is used to explain the expression of only one of the parental characters in a monohybrid cross in the F_1 and the expression of both in the F_2 . It also explains the proportion of 3:1 obtained at the F_2 .

4.2.2 Law of Segregation

This law is based on the fact that the alleles do not show any blending and that both the characters are recovered as such in the F_2 generation though one of these is not seen at the F_1 stage. Though the parents contain two alleles during gamete formation, the factors or alleles of a pair segregate from each other such that a gamete receives only one of the two factors. Of course, a homozygous parent produces all gametes that are similar while a heterozygous one produces two kinds of gametes each having one allele with equal proportion.

KEY CONCEPTS

- Test cross results reveal whether dominant phenotype is homozygous or heterozygous.
- Mendel proposed two main laws: Law of Dominance and Law of Segregation.
- Traits are controlled by discrete units (factors/genes) occurring in pairs.

♦ IMPORTANT EXPLANATIONS

Test Cross Interpretation

- If all offspring show dominant trait, parent is homozygous dominant.
- If offspring show 1:1 ratio of dominant and recessive, parent is heterozygous.

Law of Dominance

- Factors (genes) occur in pairs.
- In a pair of dissimilar factors, one dominates (dominant) and the other is recessive.
- Explains why only one trait appears in F_1 and 3:1 ratio in F_2 .

Law of Segregation

- Alleles do not blend and separate during gamete formation.
- Each gamete receives only one allele.
- Homozygous parent produces one type of gamete, heterozygous produces two types.

♦ EXAMPLES (concept understanding)

- $WW \times ww \rightarrow$ all violet flowers \rightarrow parent homozygous dominant.
- $Ww \times ww \rightarrow$ 1 violet : 1 white \rightarrow parent heterozygous.

DEFINITIONS / TERMS

- Law of Dominance – dominant allele masks recessive allele in heterozygote.
- Law of Segregation – separation of allele pairs during gamete formation.
- Factors – Mendel's term for genes.

MEMORY TRIGGERS (quick revision keywords)

- Test cross \rightarrow identify genotype
- Dominance \rightarrow one trait expressed
- Segregation \rightarrow alleles separate
- Gamete \rightarrow one allele
- 1:1 ratio \rightarrow heterozygous

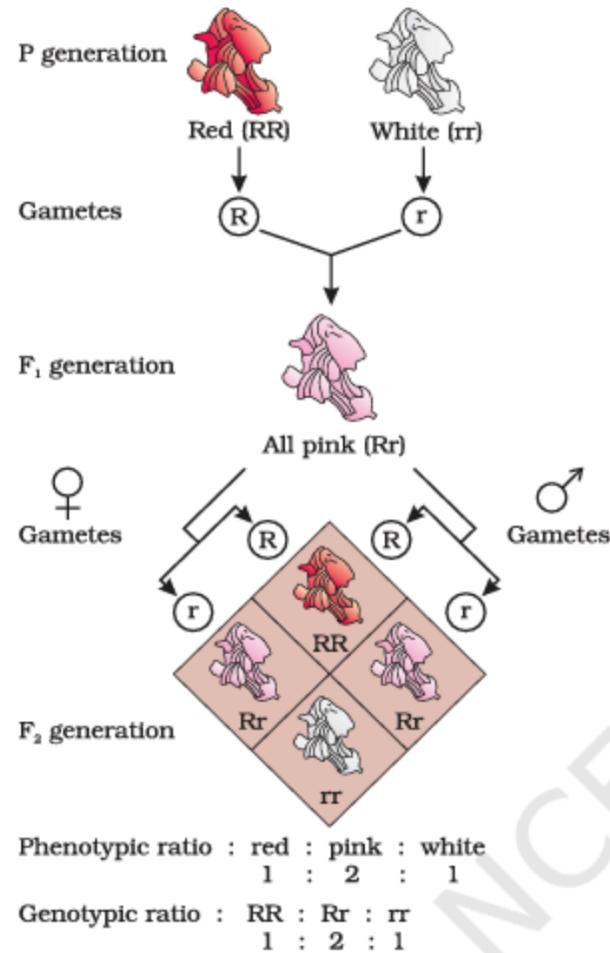


Figure 4.6 Results of monohybrid cross in the plant Snapdragon, where one allele is incompletely dominant over the other allele

4.2.2.1 Incomplete Dominance

When experiments on peas were repeated using other traits in other plants, it was found that sometimes the F₁ had a phenotype that did not resemble either of the two parents and was in between the two. The inheritance of flower colour in the dog flower (snapdragon or *Antirrhinum* sp.) is a good example to understand incomplete dominance. In a cross between true-breeding red-flowered (**RR**) and true-breeding white-flowered plants (**rr**), the F₁ (**Rr**) was pink (Figure 4.6). When the F₁ was self-pollinated the F₂ resulted in the following ratio 1 (**RR**) Red : 2 (**Rr**) Pink : 1 (**rr**) White. Here the genotype ratios were exactly as we would expect in any mendelian monohybrid cross, but the phenotype ratios had changed from the 3:1 dominant : recessive ratio. What happened was that **R** was not completely dominant over **r** and this made it possible to distinguish **Rr** as pink from **RR** (red) and **rr** (white).

Explanation of the concept of dominance:

What exactly is dominance? Why are some alleles dominant and some recessive? To tackle these questions, we must understand what a gene does. Every gene, as you know by now, contains the information to express a particular trait. In a diploid organism, there are two copies of each gene, i.e., as a pair of alleles. Now, these two alleles need not always be identical, as in a heterozygote. One of them may be different due to some changes that it has undergone (about which you will read further on, and in the next chapter) which modifies the information that particular allele contains.

Let's take an example of a gene that contains the information for producing an enzyme. Now there are two copies of this gene, the two allelic forms. Let us assume (as is more common) that the normal allele produces the normal enzyme that is needed for the transformation of a

substrate S. Theoretically, the modified allele could be responsible for production of –

- the normal/less efficient enzyme, or
- a non-functional enzyme, or
- no enzyme at all

KEY CONCEPTS

- Incomplete dominance occurs when F₁ phenotype is intermediate between parents.
- Neither allele is completely dominant over the other.
- Genotypic ratio remains 1:2:1, but phenotypic ratio changes to 1:2:1.

◆ IMPORTANT EXPLANATIONS

- In snapdragon (*Antirrhinum*), cross RR (red) × rr (white) gives F₁ pink (Rr).
- Selfing of F₁ produces F₂ with red, pink, and white flowers.
- Phenotypic ratio in F₂ is 1 red : 2 pink : 1 white.
- Incomplete dominance occurs because dominant allele does not fully suppress recessive allele.
- Genes control production of enzymes, and differences in enzyme activity lead to intermediate phenotype.

◆ EXAMPLES (concept understanding)

- Snapdragon flower colour – classic example of incomplete dominance.
- RR → red, rr → white, Rr → pink.

◆ DEFINITIONS / TERMS

- Incomplete dominance – condition where heterozygote shows intermediate phenotype.
- Intermediate phenotype – trait expression between two parental forms.

◆ MEMORY TRIGGERS (quick revision keywords)

- Incomplete dominance → intermediate trait
- F₁ → pink
- F₂ ratio → 1:2:1 (phenotype)
- RR red / Rr pink / rr white
- No complete dominance

In the first case, the modified allele is equivalent to the unmodified allele, i.e., it will produce the same phenotype/trait, i.e., result in the transformation of substrate S. Such equivalent allele pairs are very common. But, if the allele produces a non-functional enzyme or no enzyme, the phenotype may be affected. The phenotype/trait will only be dependent on the functioning of the unmodified allele. The unmodified (functioning) allele, which represents the original phenotype is the dominant allele and the modified allele is generally the recessive allele. Hence, in the example above the recessive trait is seen due to non-functional enzyme or because no enzyme is produced.

4.2.2.2 Co-dominance

Till now we were discussing crosses where the F_1 resembled either of the two parents (dominance) or was in-between (incomplete dominance). But, in the case of co-dominance the F_1 generation resembles both parents. A good example is different types of red blood cells that determine ABO blood grouping in human beings. ABO blood groups are controlled by the gene **I**. The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene. The gene (**I**) has three alleles I^A , I^B and i . The alleles I^A and I^B produce a slightly different form of the sugar while allele i does not produce any sugar. Because humans are diploid organisms, each person possesses any two of the three **I** gene alleles. I^A and I^B are completely dominant over i , in other words when I^A and i are present only I^A expresses (because i does not produce any sugar), and when I^B and i are present I^B expresses. But when I^A and I^B are present together they both express their own types of sugars: this is because of co-dominance. Hence red blood cells have both A and B types of sugars. Since there are three different alleles, there are six different combinations of these three alleles that are possible, and therefore, a total of six different genotypes of the human ABO blood types (Table 4.2). *How many phenotypes are possible?*

Table 4.2: Table Showing the Genetic Basis of Blood Groups in Human Population

| Allele from Parent 1 | Allele from Parent 2 | Genotype of offspring | Blood types of offspring |
|----------------------|----------------------|-----------------------|--------------------------|
| I^A | I^A | $I^A I^A$ | A |
| I^A | I^B | $I^A I^B$ | AB |
| I^A | i | $I^A i$ | A |
| I^B | I^A | $I^A I^B$ | AB |
| I^B | I^B | $I^B I^B$ | B |
| I^B | i | $I^B i$ | B |
| i | i | $i i$ | O |

KEY CONCEPTS

- Dominance depends on whether an allele produces a functional enzyme.
- Co-dominance occurs when both alleles express equally in F_1 .
- ABO blood group is a classic example of co-dominance and multiple alleles.

◆ IMPORTANT EXPLANATIONS

Dominance (enzyme concept)

- If an allele produces functional enzyme, trait is expressed normally.
- A non-functional allele results in recessive phenotype.
- Dominant allele represents functional form, recessive represents modified/non-functional form.

Co-dominance

- In co-dominance, F_1 resembles both parents simultaneously.
- In ABO system, gene **I** has three alleles: I^A , I^B , i .
- I^A and I^B are dominant over i .
- When I^A and I^B occur together, both express → blood group AB.
- Because of three alleles, there are six genotypes and four phenotypes.

◆ EXAMPLES (concept understanding)

- AB blood group — co-dominance (both A and B antigens expressed).
- ii genotype → blood group O (no antigen).

◆ DEFINITIONS / TERMS

- Co-dominance — condition where both alleles express equally in heterozygote.
- Multiple alleles — more than two forms of a gene in a population.
- ABO blood group system — classification based on antigens on RBCs.

◆ MEMORY TRIGGERS (quick revision keywords)

- Functional enzyme → dominant
- Co-dominance → both expressed
- I^A , I^B , i
- AB → both antigens
- Multiple alleles

Do you realise that the example of ABO blood grouping also provides a good example of **multiple alleles**? Here you can see that there are more than two, i.e., three alleles, governing the same character. Since in an individual only two alleles can be present, multiple alleles can be found only when population studies are made.

Occasionally, a single gene product may produce more than one effect. For example, starch synthesis in pea seeds is controlled by one gene. It has two alleles (**B** and **b**). Starch is synthesised effectively by **BB** homozygotes and therefore, large starch grains are produced. In contrast, **bb** homozygotes have lesser efficiency in starch synthesis and produce smaller starch grains. After maturation of the seeds, **BB** seeds are round and the **bb** seeds are wrinkled. Heterozygotes produce round seeds, and so **B** seems to be the dominant allele. But, the starch grains produced are of intermediate size in **Bb** seeds. So if starch grain size is considered as the phenotype, then from this angle, the alleles show incomplete dominance.

Therefore, dominance is not an autonomous feature of a gene or the product that it has information for. It depends as much on the gene product and the production of a particular phenotype from this product as it does on the particular phenotype that we choose to examine, in case more than one phenotype is influenced by the same gene.

4.3 INHERITANCE OF TWO GENES

Mendel also worked with and crossed pea plants that differed in two characters, as is seen in the cross between a pea plant that has seeds with yellow colour and round shape and one that had seeds of green colour and wrinkled shape (Figure 4.7). Mendel found that the seeds resulting from the crossing of the parents, had yellow coloured and round shaped seeds. *Here can you tell which of the characters in the pairs yellow/green colour and round/wrinkled shape was dominant?*

Thus, yellow colour was dominant over green and round shape dominant over wrinkled. These results were identical to those that he got when he made separate monohybrid crosses between yellow and green seeded plants and between round and wrinkled seeded plants.

Let us use the genotypic symbols **Y** for dominant yellow seed colour and **y** for recessive green seed colour, **R** for round shaped seeds and **r** for wrinkled seed shape. The genotype of the parents can then be written as **RRYY** and **rryy**. The cross between the two plants can be written down as in Figure 4.7 showing the genotypes of the parent plants. The gametes **RY** and **ry** unite on fertilisation to produce the F_1 hybrid **RrYy**. When Mendel self hybridised the F_1 plants he found that $3/4^{\text{th}}$ of F_2 plants had yellow seeds and $1/4^{\text{th}}$ had green. The yellow and green colour segregated in a 3:1 ratio. Round and wrinkled seed shape also segregated in a 3:1 ratio; just like in a monohybrid cross.

KEY CONCEPTS

- Multiple alleles exist when a gene has more than two forms in a population.
- A single gene may show different dominance patterns depending on the trait studied.
- Inheritance of two genes is studied using dihybrid crosses.

♦ IMPORTANT EXPLANATIONS

Multiple Alleles & Dominance

- In an individual only two alleles are present, even if more exist in population.
- Example: gene controlling starch synthesis in pea seeds (B and b).
- BB → large starch grains → round seeds.
- bb → small starch grains → wrinkled seeds.
- Bb → intermediate starch grain size, showing incomplete dominance if grain size is considered.
- Dominance depends on gene product and phenotype considered.

Inheritance of Two Genes

- Mendel crossed plants differing in seed colour and seed shape.
- Yellow colour (Y) dominant over green (y).
- Round shape (R) dominant over wrinkled (r).
- Parental cross: RRYY × rryy → F_1 RrYy.
- In F_2 , each trait shows 3:1 ratio, similar to monohybrid cross.

♦ EXAMPLES (concept understanding)

- Starch synthesis gene (B/b) affecting seed shape.
- Yellow round × green wrinkled pea cross.

DEFINITIONS / TERMS

- Multiple alleles – more than two alternative forms of a gene in population.
- Dihybrid cross – cross involving two characters.

♦ MEMORY TRIGGERS (quick revision keywords)

- Multiple alleles → >2 forms
- BB round / bb wrinkled
- Dominance depends on trait
- Dihybrid cross → two genes
- Y over y, R over r

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