Epistasis, Additivity, Pleiotropy

The gene interactions studied earlier are allelic or intragenic, i.e. different alleles of single gene interact and affect the expression of each other, and may also lead to the development of a new phenotype. However, there are interactions in which two or more genes located at different loci are responsible to produce a phenotype. The expression of these genes is influenced by each other and interactions among these genes may create new phenotypic combinations exhibiting modified Mendelian ratios. Such interactions are called non-allelic or inter-genic interactions. The different non-allelic interactions are complementary gene interaction, epistasis, supplementary gene interaction and duplicate genes.

Epistasis:

Epistasis is a kind of gene interaction **in which one gene masks the expression of other non-allelic gene.** The gene that shows the masking action is called **epistatic gene** while the one whose expression is masked is called a **hypostatic gene**.

On the basis of the effect exerted on another gene, epistasis can be of two types- dominant and recessive epistasis.

a) **Dominant Epistasis:** This type of gene interaction occurs when a dominant gene suppresses the expression of a gene at some otherlocus. For example, in *Cucurbita pepo* (summer squash), gene for white fruit colour is dominant and is designated as 'W' (Fig. 2.10). Another gene 'Y' controls the expression of yellow colour in fruit. The gene W exerts epistatic effect on the gene Y, therefore, the yellow colour is not expressed in fruits in presence of gene W. When both the genes (W and Y) are absent, green phenotype appears. The F2 phenotypic ratio in dominant epistasis is **12:3:1**.

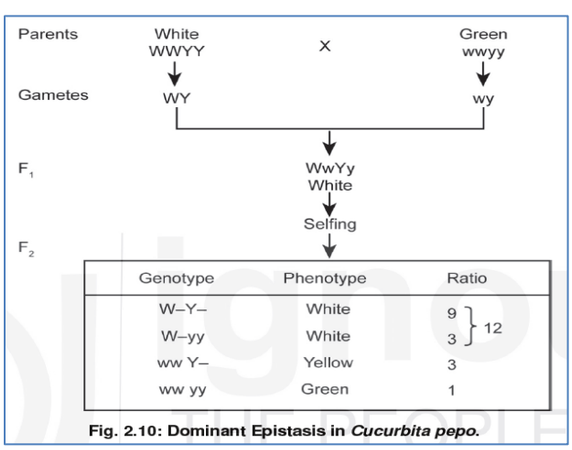
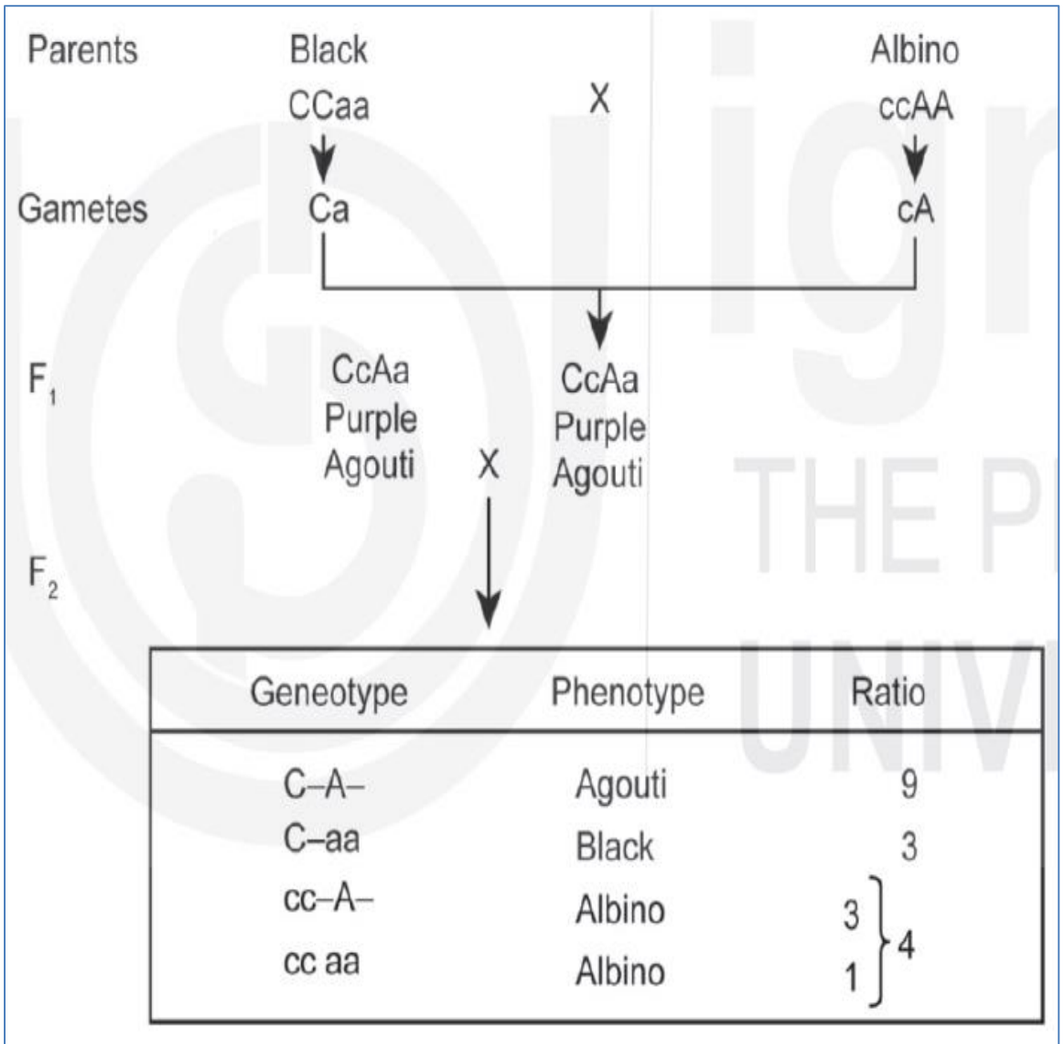


Fig. 2.10: Dominant Epistasis in *Cucurbita pepo*.

b) **Recessive Epistasis**: In this type of gene action, a recessive pair of alleles inhibits or masks the expression of a gene at another locus. The epistatic action is exerted by the homozygous recessive gene pair and its own expression by its dominant form occurs only in the presence of other dominant gene.

Coat colour in mice is a common example of recessive epistasis. The coat colour in mice can be black, agouti or albino, and the phenotype is controlled by two pairs of genes- 'A' and 'C', both of which are non-allelic. The agouti coat colour in mice is expressed by gene 'A' only in presence of another dominant gene 'C'. However, the recessive pair of alleles 'cc' masks the expression of the gene A, for both the genotypes AA and Aa. The black coat colour in mice is expressed by gene 'C' only in absence of dominant gene A. Recessive homozygous forms of both the gene pairs produce albino phenotype. The F2 phenotypic ratio in such a case is **9:3:4** (Fig. 2.11).



Geneotype	Phenotype	Ratio
C–A–	Agouti	9
C–aa	Black	3
cc–A–	Albino	3 } 4
cc aa	Albino	1 }

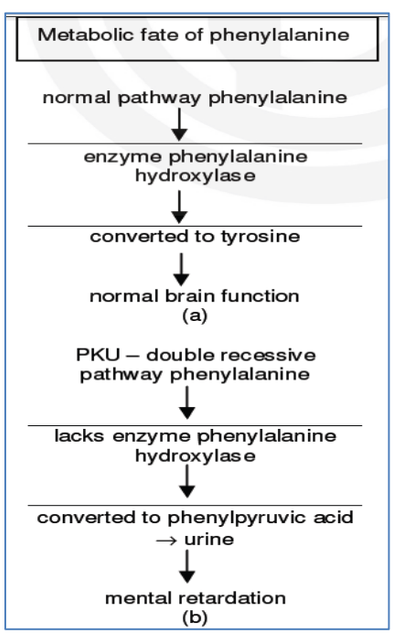
Fig. 2.11: Inheritance of coat colour in mice.

PLEIOTROPY:

The action of a gene at the cellular level is unitary, that is, one gene one action. Sometimes the presence of a gene results in a broad spectrum of phenotypic changes, so that it appears that the **gene has multiple actions.** This phenomenon is called **pleiotropy,** and is found primarily in higher organisms where complex and interrelated developmental events occur.

Many lethal alleles are pleiotropic. For example, the **yellow coat colour in mice,** as discussed, is an allele that affects more than one character, that is, it produces yellow colour of the coat in heterozygotes, and it also affects survival, causing lethality in homozygotes. Another example of multiple effects is the gene affecting **seed shape in garden peas**; this gene also affects starch grain morphology. In fact, many genes affect more than one trait. Mendel also noticed that genes causing the flower colours, like violet and white, also influenced seed colour and caused the presence or absence of coloured areas on the leaves. This is due to pleiotropy as a single gene affects more than one character.

Pleiotropic traits also occur in humans. One such disease is **phenylketonoria,** abbreviated as **PKU.** This occurs in individuals that are homozygous for a defective, recessive allele. The diseased people lack the enzyme necessary for the metabolism of the amino acid phenylalanine. When normal and PKU individuals are compared, the level of phenylalanine is much higher in diseased group. In addition to this basic biochemical difference, a number of other features are seen in the untreated PKU patients, such as lower IQ, similar head size and lighter hair.



Additivity:

Genes may be classified as additive genes or non-additive genes. Additive genes are those genes that code for the same trait and their effects work together on the phenotype. An example of a function of additive genes is on the eye colour. Several genes work together to determine the colour of the eye of an offspring. Additive genes are associated with epistasis since they contribute to epistatic effects. In contrast, non-additive genes are genes in which there is no summation of the effects of the genes. In a non-additive gene pair, for instance, not all members of the gene pair will be expressed. The gene in a gene pair that is expressed is described as dominant. That means the dominant gene will not allow the expression of the other gene in the pair. Additive genes affect the same trait. That is when the dominant forms of both genes are present together and produce double effect. Members of a gene pair may be equally expressed and therefore their expression is about the sum of individual effects of the genes in a pair.