EPISTASIS

the Expression and Interaction of Genes

We have already discussed the Mendelian laws of (i) segregation and (ii) independent or random assortment of genes in the previous chapter. To properly understand the basis of these two laws, we must acquire a clear concept of the process of *meiosis*. Reduction of chromosome number, as also of the genes carried on them, takes place during meiosis. Segregation of genes means the separation of the alleles of a gene pair through the separation of the homologous chromosomes, while independent assortment of genes refers to segregation of the members of one pair of genes regardless of the members of the other gene pairs.

The phenotypic F_2 ratio is 3:1, when the individuals crossed differ in only one gene pair (monohybrid) and one of the two character forms is dominant. The F_2 phenotypic ratio is 9:3:3:1, when the difference between the two crossing individuals is in the nature of two gene pairs (dihybrid) and dominance is involved in both the characters. This phenotypic ratio of 9:3:3:1 implies that a total of 16 possible combinations resulting from the random union of 4 types of male and female gametes produce only four phenotypes; 9 of them show the double dominant phenotype (A—B—), 3 show one dominant and one recessive character (A—bb); 3 show the other dominant character and the recessive (aaB—), whereas only one has a phenotype representing both the recessive characters (aabb). It must be borne in mind that 9:3:3:1 ratio is dependent upon the following facts:

(i) The difference between the two parents is of two independent gene pairs which determine mostly two different characters.

(ii) Dominance is involved in both the characters.

(iii) The assortment of the genes controlling the two characters is independent of each other.

(iv) The union of the gametes is random.

If these conditions get disturbed, the usual F$_2$ phenotypic ratio of 9:3:3:1 will also be upset. For instance, if the two gene pairs involved in a dihybrid cross control expression of only one character instead of two the two gene pairs must be present to produce the given phenotype, the absence of the one or the other results in the non-expression of that phenotype and the 9:3:3:1 ratio will change.

So far, we have emphasised that a character is controlled by one pair of allele, and the crosses involving contrasting characters show segregation into definite Mendelian ratios. It has been, however, observed that in some cases more than one gene pair may be essential to the expression of a particular character and the same is also true inversely, i.e., a single pair may affect more than one character. In the development of a character, there may be many genes involved, interacting in a complex manner. The phenomenon of interaction of non-allelic genes modifies the dihybrid Mendelian ratio. In the usual dihybrid ratio of 9:3:3:1, some of classes may add up together on the basis of their phenotypic similarity but the number of total combinations remains the same, i.e., 16. The change occurs only in the number of the phenotypic classes.

I. Modification of the 9:3:3:1 ratio by the lack of dominance

In snapdragons, a pair of alleles controls the expression of flower colour, i.e. RR produces red colour, Rr produces pink and rr produces white. Similarly, there is another pair of alleles affecting leaf shape, so that BB produces broad leaves, Bb intermediate and bb narrow leaves. When RRBB is crossed with rrbb, the F$_1$ will consist of pink flowered intermediate leaved plants and in the F$_2$ will be obtained this ratio: $(1:2:1)^2$ i.e., 1:2:2:4:1:2:1:2:1. This is a modification of 9:3:3:1 ratio when dominance is absent.

II. Modification of the 9:3:3:1 by epistasis

Sometimes, it so happens that the genes which are non-allelic, but affect the same characters of an organism, show interaction in much the same way as the effect of the recessive genes is masked by its dominant allele. When a non-allelic gene exerts a dominant influence over another the dominating gene is said to be *epistatic* and the gene whose effect is suppressed is regarded as *hypostatic*. This phenomenon is called *epistasis*. It may be noted that dominance involves allelic genes, while epistasis involves non-allelic genes. The interaction may take the form of a complementary gene action, supplementary gene action or an inhibiting action on the part of one of the genes. Several modifications of the 9:3:3:1 ratio representing the various types of gene interaction are briefly discussed hereunder:

(Supplementary Gene action)

(1) 9:3:4 ratio (*recessive epistasis*). To produce a black rat two dominant genes must be present R.C; *cc* produces albino and is epistatic to R. Gene interaction in which recessive allele at one locus can mask the expression of both the alleles at other locus.

Parents (Black) RRCC × rrcc (albino)
Gametes (RC) (rc)
F_1 RrCc (All black)
RrCc × RrCc (Cross F_1 amongst themselves)
F_2 9 R–C— black
3 R–cc albino
3 rrC– cream
1 rrcc albino
16

The ratio is 9:3:4 and is a case of recessive epistasis.

(2) 12:3:1 ratio (*dominant epistasis*). In summer squash to produce yellow coloured fruit, Y gene is essential. In some white fruit varieties W gene is epistatic to Y gene. The epistatic W gene does not allow the hypostatic gene Y to produce yellow colour. Yellow colour appears when W is absent and Y gene alone is present. Both white and yellow fruit colours ~~and~~ are dominant over green.

Parents (White) WWYY × wwyy (green)
Gametes WY wy
F_1 WwYy All white
F_2 9 W–Y– } White
3 W–yy }
3 ww Y– Yellow
1 ww yy Green
16

The F_2 ratio is modified to 12:3:1 due to dominant epistasis.

(3) 9:7 *ratio* (*duplicate recessive epistasis*). In daisy, there are two yellow-flowered varieties which are genetically different; the gene producing yellow colour e.g., P in one is different from the gene producing yellow colour, R, in the other. The recessive alleles are epistatic. When both these independent dominant genes are brought together by crossing they produce purple colour:

Parents	(Yellow)	PPrr × ppRR	(Yellow)
Gametes		Pr pR	
F_1		PpRr	Purple
F_2		9 P–R–	Purple
		3 P–rr 3 ppR– 1 pprr	Yellow
		16	

Gene Interaction in which recessive alleles at either of two loci can mask The expression dominant alleles at The two loci called Complementary epistasis.

The F_2 ratio is modified to 9:7 and the genes involved here are complementary. (Genes which are similar in phenotypic effect when present in different individuals, but produce a different expression by interaction on coming together are called complementary genes.

(4) 15:1 *ratio* (*duplicate dominant epistasis*). When two factors affect the same character and the dominant allele of each acts as epistatic. This case may be illustrated by an example from the poultry.

Parents	(Feathered shanks)	FFSS × ffss	(Unfeathered)
Gametes		FS fs	
F_1		FfSs	
F_2			(Feathered shanks)
		9 F–S– 3 F–ss 3 ffS–	(Feathered shanks)
		1 ffss	(Unfeathered shanks)
		16	

The F_2 ratio is changed to 15:1;

Gene interaction in which Two dominant alleles at either of The loci Can mask The expressin

Two identical pairs of genes with the same expression present in one individual are called *duplicate genes.*

(5) 13:3 *ratio* (*dominant and recessive epistasis*). An inhibiting gene is generally assumed to have no effect of its own but only inhibits the function of another gene. In poultry, White Leghorn birds are genetically coloured birds but they are unable to develop colour because alongwith the colour gene C they also possess an inhibitor gene I.

Gene Interaction in which dominant allele at one locus can mask the expression of both dominant & recessive allele at second locus.

The white plumage in White Plymouth Rocks is recessive to coloured plumage.

Parents	(White Leghorn)	IICC × iicc	(White Plymouth Rocks)
Gametes		IC ic	
F_1		IiCc	White
F_2		9 I–C–	White
		3 I–cc	White
		3 iiC–	Coloured
		1 iicc	White
		16	

I gene is epistatic (inhibitor) to C gene which develops colour only where I is absent. The F_2 ratio is changed to 13:3.

(6) 9:6:1 *ratio* (*factor interaction*). In summer squash A and B genes are essential to the production of disc-shaped fruit. When A or B gene is separately present in different individuals each produces round-shaped fruit. Their recessive alleles *a* and *b* interact to develop elongated fruit shape.

Parents	(Round fruit)	AAbb × aaBB	(Round fruit)
Gametes		Ab aB	
F_1		AaBb	Disc-shaped fruit
F_2		9 A–B–	Disc
		3 A–bb 3 aaB–	Round
		1 aabb	Elongated
		16	

Polymeric Gene action

Gene action in which two dominand alleles have similar effects when they are separate but produced enhanced effect when come together

III. 9:3:3:1 ratio resulting from factor interaction

The walnut comb shape in poultry birds results from the interaction between the two independent dominant genes R and P, which, when present in different individuals, produce rose and pea combs, respectively.

Parents	Rose	RRpp × rrPP	(Pea)
Gametes		Rp rP	
F_1		RrPp	Walnut (interaction between dominant alleles)
F_2		9 R–P–	Walnut
		3 R–pp	Rose
		3 rrP–	Pea
		1 rrpp	Single (Interaction between recessive alleles)
		16	

Two things may be noted: (i) the F_1 resembles neither parent and (ii) 1/16 F_2, birds have a new comb shape (single) not present in the parents and the F_1. These are the products of factor interaction.

PROBLEMS

Note: In poultry, gene R alone produces rose comb, while gene P is responsible for the pea comb. R and P together produce walnut comb and their recessive alleles pprr single comb.

✓ 1. What phenotypic ratios will be obtained from the following crosses?

(a) PrPP × rrpp
(b) RrPp × Rrpp
(c) RrPp × RrPP
(d) RrPp × rrPP