Quadrant II – Transcript and Related Materials

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Module Name: Epistasis and Hypostasis-II

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NOTES:

Epistasis is a non-mendelian inheritance pattern. It is an interlocus, inter-genic interaction. The term epistasis was first used in 1909 by Bateson to describe a masking effect whereby a variant or allele at one locus prevents the variant at another locus from manifesting its effect. This was seen as an extension of the concept of dominance for alleles within the same allelomorphic pair i.e. at a single locus.

TYPES OF EPISTATIC INTERACTIONS

1. Duplicate genes with cumulative effect (additive or duplicate gene action)

Duplicate genes are two identical pairs of alleles expressing the same phenotype, but localized in different chromosomes. Duplicate genes are generally given the same symbols, but with different suffixes or subscripts. A₁ is dominant over a₁ and A₂ over a₂. A₁ is identical with A₂ and a₁ with a₂. Since A₁ and A₂ have the same phenotypic expression, the combinations A₁a₂ and A₂a₁ are phenotypically equal and different from a₁a₂.

e.g: Coat colour in swine (pigs): in the Durac-Jersey breed of swine, two pairs of interacting genes are responsible for coat colour. There are three coat colours in the swine- red, sandy and white. When the dominant allele is present at both gene pairs (A,B), the phenotypic result is red coat colour. Either one factor alone yields the partial colour- sandy. The sandy coat colour can be caused by two different gene pairs, when either one is homozygous recessive and the other dominant (A-bb or aaB-). When both gene pairs are homozygous recessive (aa bb), no pigment formation takes place and the coat colour is white. A breed with genotype AA BB (red) was crossed with the genotype aa bb (white). The F1 generation individuals had a uniform Aa Bb genotype, giving red coat colour. A cross between the F1 heterozygous, produced F2 swine with the phenotypic ratio of **9 red: 6 sandy: 1 white**.

2. Dominant and recessive epistasis (inhibitory action)

There is complete dominance of both gene pairs, but one gene when dominant is epistatic to the first. Here a dominant gene is a suppressor of a dominant, (A epistatic to B) and a recessive gene is also a suppressor of a dominant gene, (bb epistatic to A). Therefore this type of epistasis is known as dominant and recessive epistasis. The genotype at one locus (A) and the recessive genotype at the other locus (bb) produce the same phenotype. Because of this, only two F2 phenotypes are produced. The A B-, A bb and the aa bb genotypes produce one phenotype, while the aa B genotype produces the other phenotype.

e.g – **Feather colour in fowl**: in the White Leghorn breed of fowl, white plumage is completely dominant over colour plumage (black, barred etc). In the

White Wyandotte and the White Plymouth Rock breeds, white plumage is recessive to colour plumage. White Leghorns are white because they carry a dominant colour-inhibiting gene I. White Wyandottes and White Plymouth Rocks are recessive white breeds. The genotype of white leghorns is II CC, i.e. they are double dominants. The genotype contains dominant colour genes, but since the inhibitory genes are epistatic to the colour genes, the plumage is white. The genotype of white wyandottes is double recessive (ii cc). Here the white colour is not due to the action of the inhibitor gene, but due to the absence of a dominant colour gene at the colour locus. When the white leghorns are crossed with white wyandottes, the F1 generation fowl are white with flecks. Inbreeding of the white – flecked F1 fowl results in the F2 generation with phenotypic ratio 13 white/flecked white: 3 coloured.

3. Duplicate recessive epistasis (complementary action)

There is complete dominance at both gene pairs, but either gene when dominant is epistatic to the other. The nature of duplicate genes has already been described in type III, duplicate genes with cumulative effect. In this type there is a cumulative or additive effect of the dominant alleles. In type VI, however, the dominant alleles of both loci each produce the same phenotype without cumulative effect. The F2 segregation ratio of 15:1 produced, provided that the dominant alleles of the duplicate genes do not act additively or cumulatively. The full phenotypic effect is produced by either of the dominant genes. The genotypes A- B-, aa- B- and A- bb all produce the identical phenotype. Only the double recessive aa bb produces the other phenotype.

An example of duplicate recessive interaction in humans is the inheritance of deafness. Children who are born deaf are also mute, unless special techniques are employed to teach them to speak. Persons with genotype A- and B- are normal for hearing and speech. Persons with the A-bb, aa B- or aa bb genotypes are deaf mutes. The homozygous aa is epistatic to the B gene and the homozygous bb is epistatic to the A gene for normal hearing. Since either of the homozygous gene pairs aa and bb is epistatic to the dominant allele of the other, this type of inheritance is called duplicate recessive epistasis.

4. Duplicate dominant epistasis (duplicate gene action)

There is dominance at both genes pairs, but either gene when dominant is epistatic to the other. The dominant alleles of both loci each produce the same phenotype without cumulative effect. The F2 segregation ratio of 15:1 is produced, provided that the dominant alleles of the duplicate genes do not act additively or cumulatively. The full phenotypic effect is produced by either of the dominant genes. The genotypes A-B-, aa B- and A-bb all produce the identical phenotype. Only the double recessive aa bb produces the other phenotype. In the fowl, the Black Langshan breed has feathered shanks, while the Buff Rock breed has non-feathered shanks. The presence of feathers on the shank of the fowl is due to two pairs of dominant alleles, AA and BB, each pair having a separate locus. when the homozygous double dominant (feathered shanks) is crossed with the homozygous double recessive (non-feathered shanks), the F1 progeny all have feathered shanks. Inbreeding of F1 generation fowl gives rise to the F2 generation which has the phenotypic ratio of 15 feathered shanks: 1 non-feathered shanks fowl.