12.3F: Epistasis

Epistasis occurs when one gene masks or interferes with the expression of another.

Learning Objectives

• Explain the phenotypic outcomes of epistatic effects between genes

Key Points

• In many cases, several genes may contribute to a particular phenotype; when the actions of one gene masks the effects of another, this gene is said to be epistatic to the second.
• Epistasis can occur when a recessive genotype masks the actions of another gene, or when a dominant allele masks the effects of another gene.
• Epistasis can be reciprocal: either gene, when present in the dominant (or recessive) form, expresses the same phenotype.
• Any single characteristic that results in a phenotypic ratio that totals 16 (such as 12:3:1, 9:3:4, or others) is typical of a two-gene interaction.

Key Terms

• epistasis: the modification of the expression of a gene by another unrelated one
Epistasis

Mendel’s studies in pea plants implied that the sum of an individual’s phenotype was controlled by genes (or as he called them, unit factors): every characteristic was distinctly and completely controlled by a single gene. In fact, single observable characteristics are almost always under the influence of multiple genes (each with two or more alleles) acting in unison. For example, at least eight genes contribute to eye color in humans.

In some cases, several genes can contribute to aspects of a common phenotype without their gene products ever directly interacting. In the case of organ development, for instance, genes may be expressed sequentially, with each gene adding to the complexity and specificity of the organ. Genes may function in complementary or synergistic fashions: two or more genes need to be expressed simultaneously to affect a phenotype. Genes may also oppose each other with one gene modifying the expression of another.

In epistasis, the interaction between genes is antagonistic: one gene masks or interferes with the expression of another. “Epistasis” is a word composed of Greek roots that mean “standing upon.” The alleles that are being masked or silenced are said to be hypostatic to the epistatic alleles that are doing the masking. Often the biochemical basis of epistasis is a gene pathway in which the expression of one gene is dependent on the function of a gene that precedes or follows it in the pathway.

An example of epistasis is pigmentation in mice. The wild-type coat color, agouti (AA), is dominant to solid-colored fur (aa). However, a separate gene (C) is necessary for pigment production. A mouse with a recessive c allele at this locus is unable to produce pigment and is albino regardless of the allele present at locus A. Therefore, the genotypes AAcc, Aacc, and aacc all produce the same albino phenotype. A cross between heterozygotes for both genes (AaCc x AaCc) would generate offspring with a phenotypic ratio of 9 agouti:3 solid color:4 albino. In this case, the C gene is epistatic to the A gene.
Epistasis in mouse coat color: In mice, the mottled agouti coat color (A) is dominant to a solid coloration, such as black or gray. A gene at a separate locus (C) is responsible for pigment production. The recessive c allele does not produce pigment and a mouse with the homozygous recessive cc genotype is albino regardless of the allele present at the A locus. Thus, the C gene is epistatic to the A gene.

Epistasis can also occur when a dominant allele masks expression at a separate gene. Fruit color in summer squash is expressed in this way. Homozygous recessive expression of the W gene (ww) coupled with homozygous dominant or heterozygous expression of the Y gene (YY or Yy) generates yellow fruit, while the wwyy genotype produces green fruit. However, if a dominant copy of the W gene is present in the homozygous or heterozygous form, the summer squash will produce white fruit regardless of the Y alleles. A cross between white heterozygotes for both genes (WwYy × WwYy) would produce offspring with a phenotypic ratio of 12 white:3 yellow:1 green.

Finally, epistasis can be reciprocal: either gene, when present in the dominant (or recessive) form, expresses the same phenotype. In the shepherd’s purse plant (Capsella bursa-pastoris), the characteristic of seed shape is controlled by two genes in a dominant epistatic relationship. When the genes A and B are both homozygous recessive (aabb), the seeds are ovoid. If the dominant allele for either of these genes is present, the result is triangular seeds. That is, every possible genotype other than aabb results in triangular seeds; a cross between heterozygotes for both genes (AaBb x AaBb) would yield offspring with a phenotypic ratio of 15 triangular:1 ovoid.

Keep in mind that any single characteristic that results in a phenotypic ratio that totals 16 is typical of a two-gene interaction. Recall the phenotypic inheritance pattern for Mendel’s dihybrid cross, which considered two non-interacting genes: 9:3:3:1. Similarly, we would expect interacting gene pairs to also exhibit ratios expressed as 16 parts. Note that we are assuming the interacting genes are not linked; they are still assorting independently into gametes.

LICENSES AND ATTRIBUTIONS

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