One gene can mask another

Epistasis is an interaction between different loci: one genotype masks or modifies the phenotypic effect of another. Dominance instead describes alleles at the same locus.

1. A TWO-ENZYME MODEL



Illustrative pathway: A_ makes enzyme A; B_ makes enzyme B. aa blocks step 1; bb blocks step 2. With aa, B cannot affect colour: aa is epistatic to the B locus (the masked, or hypostatic, locus). An underscore means either allele.

2. CROSS AaBb × AaBb: UNLINKED LOCI

Each parent produces AB, Ab, aB and ab gametes, each with probability 1/4. Assume complete dominance at each locus and equal survival.

Gametes	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

3. GROUP BY WHAT THE PATHWAY CAN MAKE

9 A_B_: black. 3 A_bb: brown. 3 aaB_ + 1 aabb: colourless.
Result: 9 black : 3 brown : 4 colourless. Derive the ratio from the gene effects; different interactions give different ratios. Masking does not remove the other gene or stop its inheritance.

Epistasis principles: OpenStax, Biology 2e 12.3.
Cambridge 16.2.3: interpret interactions; memorising epistasis ratios is not required.

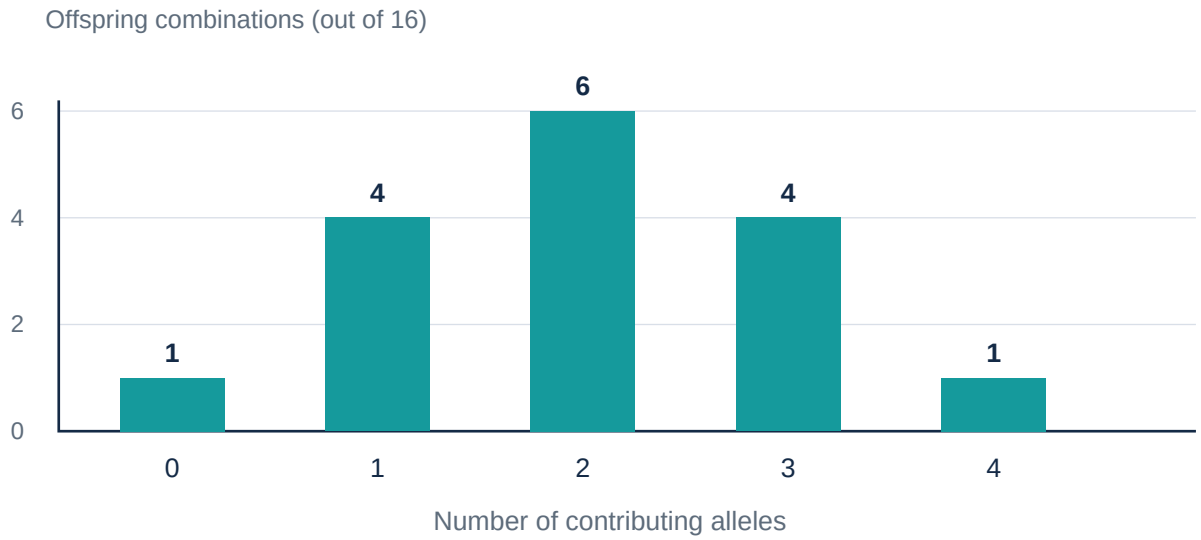
Scope: Cambridge 9700, 16.2.3; variation links to 17.1.1-3, pp. 41, 43.

Many genes, one characteristic

Polygenic inheritance means that two or more genes influence one characteristic. Small effects can combine, producing many possible phenotypic values.

1. AN ADDITIVE MODEL: COUNT CONTRIBUTING ALLELES

For AaBb × AaBb, assume unlinked loci and equal additive effects: each A or B adds one unit; a and b add none. There is no dominance in this model. Environment is held constant.



2. MORE COMBINATIONS OCCUR NEAR THE MIDDLE

AABB has four contributing alleles; aabb has zero. AAbb, AaBb and aaBB all have two, despite different genotypes. In this simplified cross, the five classes occur in 1 : 4 : 6 : 4 : 1. This is a teaching model, not a universal ratio or an exact model of human height.

3. FROM DISCRETE CLASSES TO A CONTINUOUS RANGE

Many loci with varied effects create many possible values. Environmental differences can further blur boundaries. Traits such as height therefore often show continuous variation. Polygenic does not mean that an individual has many alleles at one locus: several loci are involved.

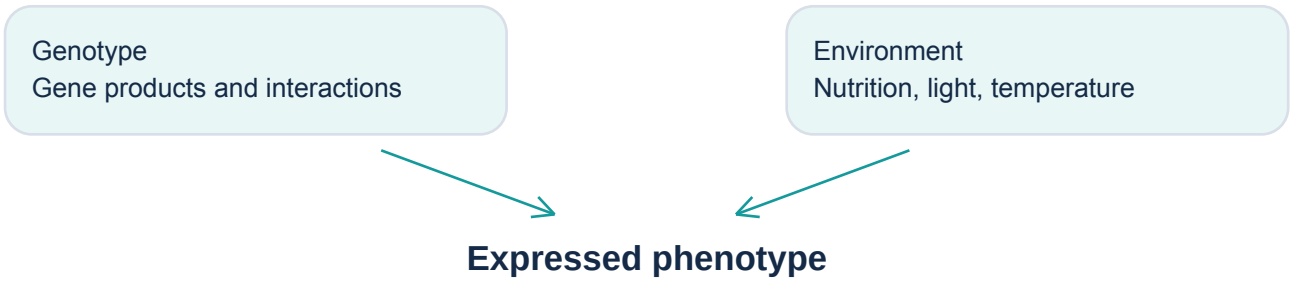
Polygenic and multifactorial traits: NHGRI, Polygenic Trait.
Continuous and discontinuous variation: Cambridge 9700, 17.1.1-3.

Scope: Cambridge 9700, 16.2.3; variation links to 17.1.1-3, pp. 41, 43.

Genotype is not the whole story

Phenotype develops through gene activity, gene products and environmental conditions. A changed phenotype does not necessarily mean a changed DNA sequence.

1. GENES AND ENVIRONMENT BOTH MATTER



2. EXAMPLE: HUMAN HEIGHT

Height involves many genetic variants, but childhood nutrition and health also influence growth. Relatives need not reach the same height. Sharing a genotype does not guarantee identical phenotypes under different conditions. This is multifactorial inheritance: genetic and environmental influences together.

3. CONTINUOUS OR DISCONTINUOUS?

Continuous: measured values form a range with intermediates, such as height. Often many genes plus environmental effects.

Discontinuous: distinct categories, such as ABO blood groups. Often one or a few genes with major effects. Several genes can still produce categories, as in the epistasis example.

4. SEPARATE CAUSES WITH A FAIR COMPARISON

Test an environmental effect using genetically matched plants, one changed condition, replication and other factors controlled. Compare genotypes in the same conditions. A continuous pattern or family resemblance alone does not prove its cause.

Phenotype and environmental influences: NHGRI, Phenotype.
Height, genes, nutrition and health: MedlinePlus Genetics.

Scope: Cambridge 9700, 16.2.3; variation links to 17.1.1-3, pp. 41, 43.