Epistasis

The phenotypic expression at one locus depends on the genotype at a different locus.

An Epistasis Problem

The ABO blood group is one set of blood antigens. There is a separate locus (H) that controls the production of these antigens. Individuals who are homozygous, hh, cannot produce antigens of the ABO type (and appear to be O-type). Consider the following cross:

P_0: AOHh $\times$ BOHh.

What are the frequencies of the apparent blood groups among the offspring?

ABO locus	Antigen Production	Blood Type
¼ A	¾ H-	A
	¼ hh No Production	O
¼ B	¾ H-	B
	¼ hh No Production	O
¼ AB	¾ H-	AB
	¼ hh No Production	O
¼ O	¾ H-	O
	¼ hh No Production	O

Summary:

$\frac{3}{16}$ A

$\frac{3}{16}$ B

$\frac{3}{16}$ AB

$\frac{7}{16}$ O

Modified dihybrid Mendelian ratios

Type of gene interaction	A-B-	A-bb	aaB-	aabb
No Epistasis	9	3	3	1
Duplicate Recessive	9	7		
Single Recessive	9	3	4	
Duplicate Dominant	15			1
Single Dominant	12		3	1
Dominant × Recessive	9	6		1

1. Duplicate Recessive (9:7)

- Recesssive at one either locus masks the expression of the dominant phenotype at the other locus.

2. Single Recessive (9:3:4)

- Recessive Trait at one locus masks the effect of the second locus.

3. Duplicate Dominant (15:1)

- The dominant trait at either locus will expression one phenotype, the other phenotype is homozygous at **both** loci.

4. Single Dominant (12:3:1)

- Dominant trait at one locus masks the expression of the second locus.

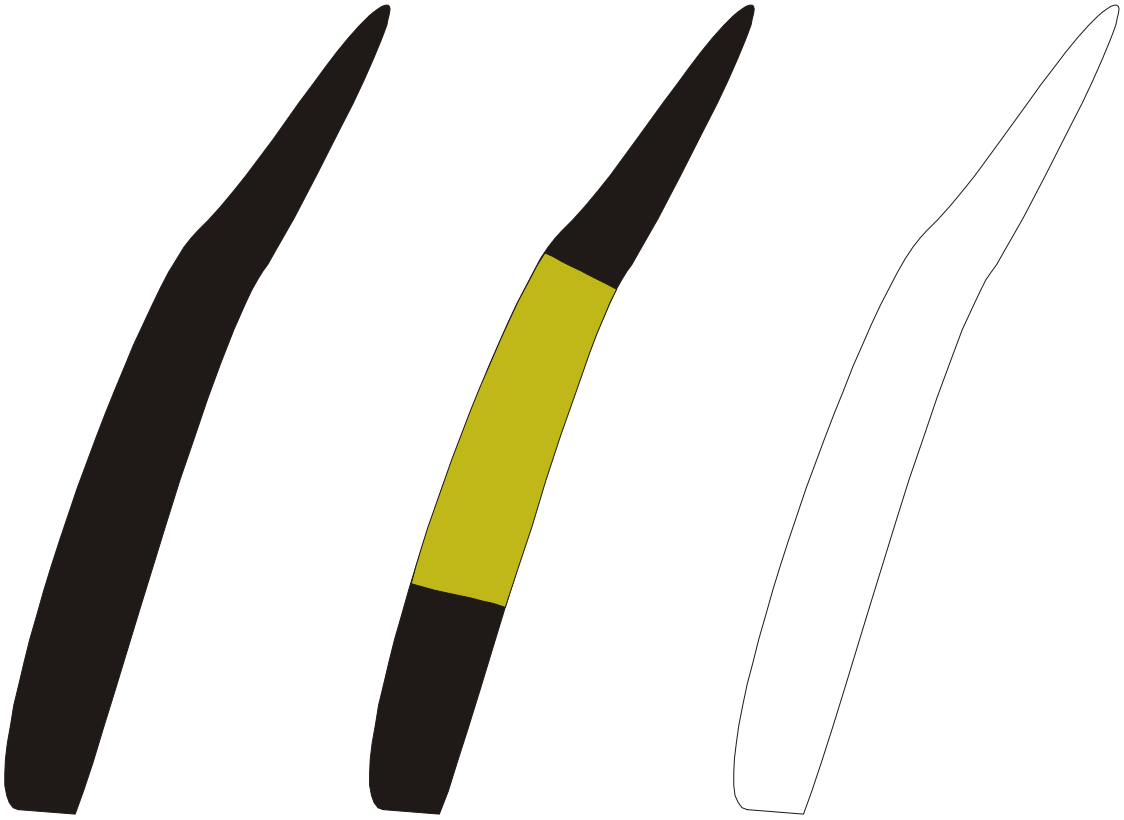
5. Dominant × Recessive (9:6:1)

- Duplicate effects from the two loci. The phenotypes are: Two dominant, One dominant, and None dominant.

Another Example

Agouti Coloring in rodents

Normal hair color in wild rodents (and other mammals) is agouti & controlled by two independent loci.



Black Agouti Albino

Locus 1: Black *vs* Black with Yellow Band (Agouti)
Locus 2: Albino

Agouti Example

The Cross:

Pure Breeding Black Crossed with an albino animal from a strain of Pure Breeding Agouti.

P_0: bbCC × BBcc

F_1: BbCc

F_2:

¾ Banded	¾ Colored	9/16 Agouti
	¼ Albino	3/16 Albino

¼ Black	¾ Colored	3/16 Black
	¼ Albino	1/16 Albino

Summary: 9/16 Agouti: 3/16 Black: 4/16 Albino

Single Recessive Epistasis

Lethal Alleles

Definition

A Lethal Gene (Allele) is a gene where the one of the expressed traits is lethal to the individual **before they are observed**.

Example: Yellow Gene in Mice

Genotype	Phenotype
YY	Normal Color (Agouti)
Yy	Yellow Color
yy	Lethal

Cross:

Yellow × Yellow

	½ Y	½ y
½ Y	¼ YY	¼ Yy
½ y	¼ Yy	¼ yy

Results:

1/3 YY + 2/3 Yy or 1/3 Agouti + 2/3 Yellow

Characteristics of Lethal Genes

- Cannot maintain pure breeding strains of one of the traits
- Families where both parents carry the gene have more miscarriages, smaller litter sizes, or smaller family sizes.