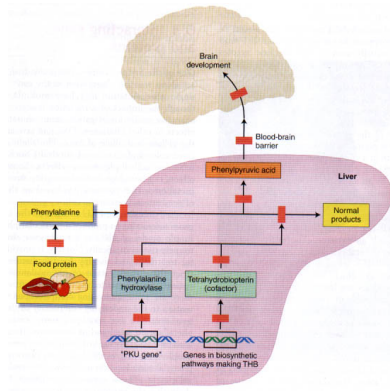


Extensions of Mendel II: *Biochemical Pathways and Epistasis*



*CF and PKU are Multifactorial Diseases
Examples of Epistasis*



Important Pre-Reading Concepts

Text Reading

- Chapter 5: pp 105-112 (Sections 5.5)
- Note:** We will **not** be covering pp113-122 (Coat Color in Dogs through section 5.7)

Important Pre-Lecture Concepts (quiz material)

- No quiz today

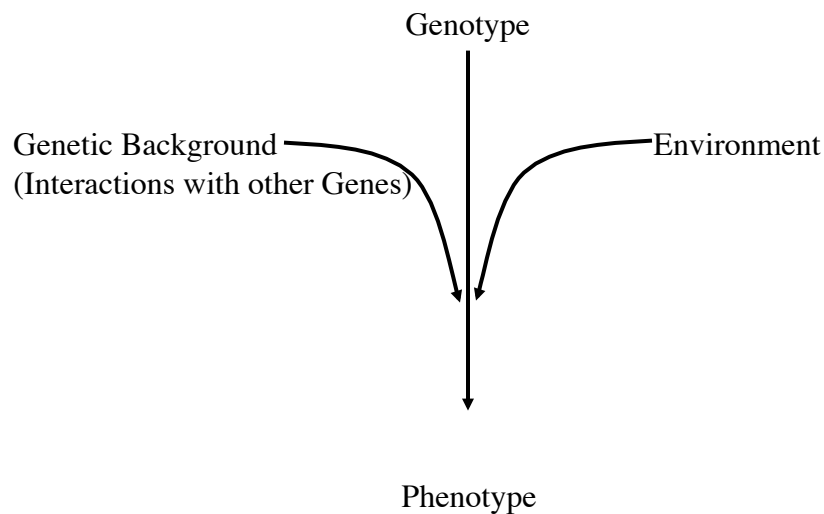


Important Class Discussion Concepts

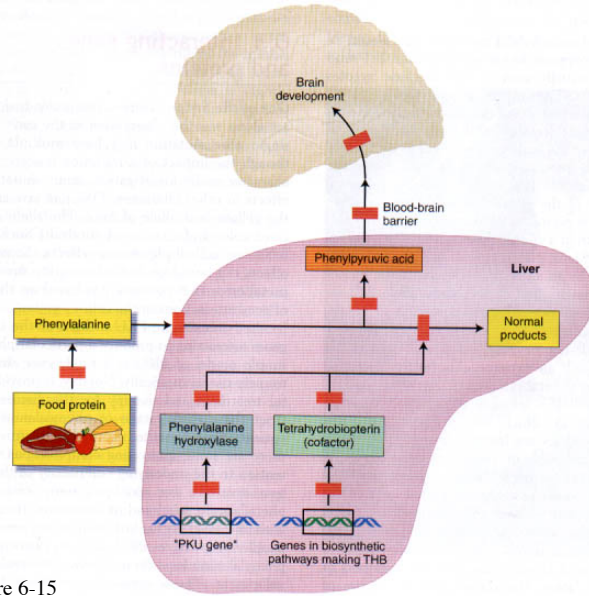
- *How do other genes, genetic background, and the environment affect the the phenotypic expression of a particular genotype?*
- *The main idea today will be examining biochemical pathways and using that to determine how mutations in proteins at different points in the pathway alter the resulting phenotypes.*

Think about these topics as we go through class and come back to them as you go over the material and practice problem solving.

From Gene to Phenotype : Phenotypes Do Not Form in a Vacuum

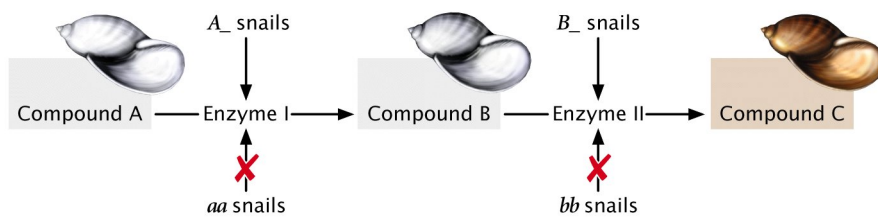


PKU is a Multi-factorial Disease: Biochemical Pathways and Epistasis



Griffiths et al. Figure 6-15

Interactions Between Alleles of Different Genes: Epistasis and Snail Pigmentation



1. Molecular Basis of Complementation
2. Epistasis: Duplicative Recessive

Recessive Epistasis: Genetics of Labrador Retriever Coat Color



Griffiths et al. Figure 6-21

Locus 1: Pigment Enzyme (Brown to Black Pigment)

B = Wildtype Allele, Produces Black

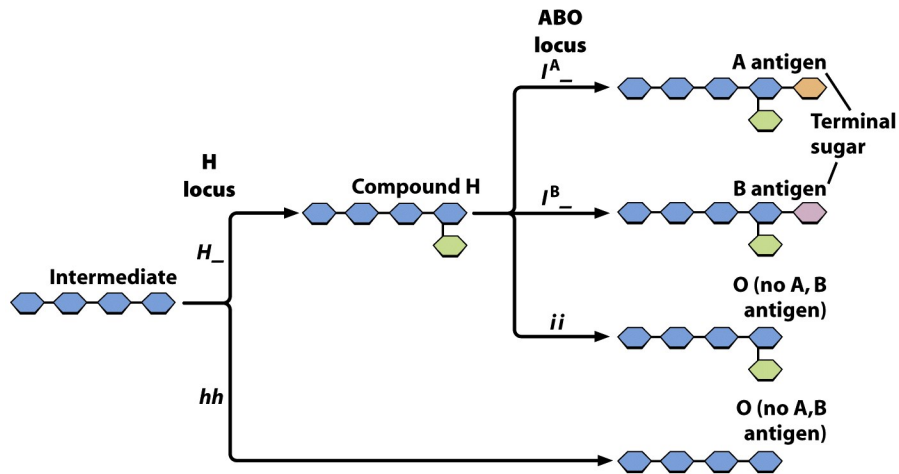
b = Loss-of-Function, Stays Brown

Locus 2: Pigment Deposition Protein

E = Wildtype Allele, Deposits Pigment (Brown or Black)

e = Loss-of-Function, No Deposition (yellow)

Recessive Epistasis: ABO blood type revisited



Conclusion: Genotypes at both the H locus and the ABO locus determine the ABO blood type.

Epistatic Interaction Summary

Table 5.2 Modified dihybrid—phenotypic ratios due to gene interaction

Ratio	Genotype				Type of Interaction	Example
	A_B_	A_bb	aaB_	aabb		
9:3:3:1	9	3	3	1	None	Seed shape and endosperm color in peas
9:3:4	9	3	4		Recessive epistasis	Coat color in Labrador retrievers
12:3:1	12		3	1	Dominant epistasis	Color in squash
9:7	9	7			Duplicate recessive epistasis	Albinism in snails
9:6:1	9	6		1	Duplicate interaction	—
15:1	15			1	Duplicate dominant epistasis	—
13:3	13		3		Dominant and recessive epistasis	—

*Each ratio is produced by a dihybrid cross ($AaBb \times AaBb$). Shaded bars represent combinations of genotypes that give the same phenotype.

Note-Review Pepper and Chicken Comb Interactions

A Problem to Try

Eye Color of the Oriental fruit fly is determined by several genes. You do the following crosses:

P: Wildtype eyes x yellow eyes

F1: all wildtype eyes (interbred for F2)

F2 Ratio:

9 wildtype

3 amethyst color (bright blue)

4 yellow eyes

What are the genotypes for P, F1 and F2

Is this an example of epistasis?

Take home: think about how a biochemical pathway could be constructed to explain this