Genetics Notes Chapter 6

6.1 Interactions between the Alleles of a Single Gene: Variations on Dominance

- The known mutant alleles of a gene and its wild-type allele are called **multiple alleles** or **allelic series**.
- The simplest type of dominance is full or complete dominance, which will be phenotypically expressed even when there is only one copy such as in a heterozygote.
- In full dominance the homozygous dominant is phenotypically indistinguishable from the heterozygote.
- Wild type full dominance occurs when one wild-type allele is haplosufficient.
- Mutant type full dominance occurs when the one wild-type allele is haploinsufficient.
- Another important type of dominant mutation is a **dominant negative.**
- In the case of a homodimeric protein, one mutated protein acts as a "spoiler" binding to the wildtype protein and somehow distorting or blocking its function. Same problem can occur in a hetero dimer (two proteins from two different genes)
- **Incomplete Dominance** occurs when the heterozygote displays an intermediate phenotype between the recessive and dominant phenotype. Ex: White flowers mixed with Red flowers make Pink flowers. Shown by C+/C+ C/C and C+/C
- On the molecular level, two copies produces the most genetic transcripts, produces the most protein, produces the most pigment resulting in the most red color.
- **Codominance** is when the heterozygotes express both alleles evenly.
- An example is blood type which has the allelic series i, I^A, and I^B. Resulting in 6 possible genotypes and four different phenotypes. A, B, AB, and O.
- I^A and I^B are fully dominant over i but when put together they are codominant.
- An example of the arbitrary nature of dominance is given by sickle cell anemia. In terms of having anemia the wild type is fully dominant over the mutant. In terms of cell shape there is incomplete dominance, in heterozygotes cells have a slightly sickled shape. In terms of hemoglobin production there is codominance, two types of hemoglobin are produced in heterozygotes.
- An allele capable of causing death is a **lethal allele.**
- This displays that the gene potentially controls something essential to the organism's operation.
- An allele that affects several properties of an organism are called **pleiotropic**.
- EXAMPLE: The effect of the yellow coat color allele on mice. Affects two characters in mice, coat color and survival. Lethal in the homozygous YY, non-lethal yellow coated mice Yy, wildtype brown mice yy.
- Sometimes alleles are only lethal in the homozygous state.
- Lethality can range, lethalness sometimes is sublethal it doesn't kill off all progeny.
- A gene can be tested for its essentialness to survival but seeing if a null allele of the gene is lethal.

6.2 Interaction of Genes in Pathways

- **One gene-one polypeptide theory**-Each gene at one locus controls the creation of one polypeptide.
- The majority of genes are converted to mRNA and then amino acid chains.

- A minority of genes however, are converted into **Functional RNAs** like tRNA or rRNA and not changed into proteins.
- So the one gene one polypeptide rule is not necessarily true as a gene may encode a functional RNA not a polypeptide.
- Genes encode for enzymes along a **biochemical pathway** that constitute a functionally interacting subset of the genome.
- To determine if two different mutations are either two different mutated genes or two mutant alleles of the same gene a **complementation test** is done.
- In this test two homozygous recessive organisms each with its own mutation are crossed. If the heterozygote progeny has the wild-type phenotype then the mutations are likely on different genes. If the progeny have the recessive mutations then they both must be mutated alleles of the same gene.
- When two independently derived recessive mutant alleles producing similar recessive phenotypes fail to complement they must be alleles of the same gene.
- If complementation is seen and the F1 generation is selfed the phenotypic ratio of the F2 generation will be **9:7** instead of the 9:3:3:1 this is because the genes interact through a biochemical pathway and if even one gene is mutated means the mutant phenotype will be produced.
- The **9:3:4 ratio** is diagnostic of an epistatic pattern. **Epistasis** is the interaction between the genes at two or more loci, so that the phenotype differs from what would be expected if the loci were expressed independently.
- The double mutant shows the phenotype of one mutation but not the other.
- The gene whose phenotype is expressed in the double mutant is said to be epistatic, while the phenotype altered or suppressed is said to be hypostatic.
- A **suppressor** is the mutant allele of one gene that reverses the effect of a mutation of another gene.
- Sometimes when mutants are exposed to genetic changing environmental effects (such as high energy radiation) their progeny are reverted back to the wild-type. These are **revertants.**
- Other time however, there are **psuedorevertants**, organisms that appear phenotypically and genotypically wild type but are not due to the presence of a mutated suppressor gene.
- Another type of mutation is the **modifier mutation**, this is a gene at a second locus that can modify the gene expression of the gene at the first locus resulting in four genotypes each varying in their expression of the phenotype.

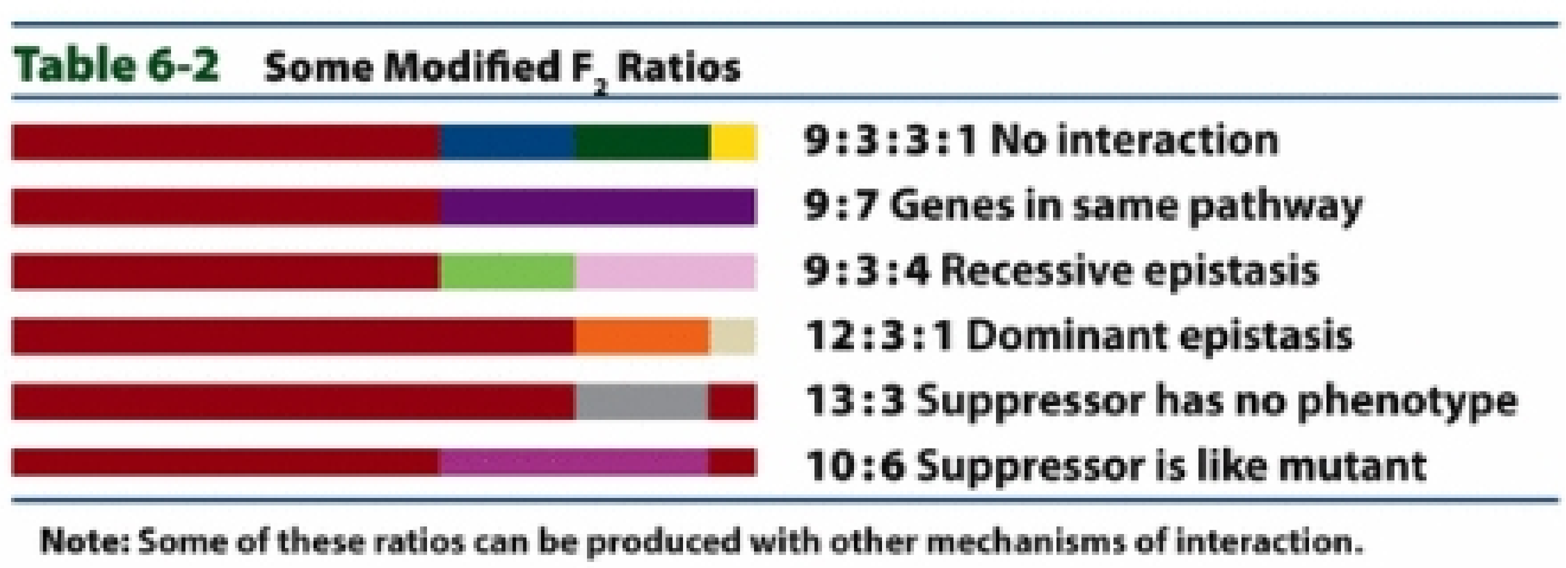
**Table 6-2** Some Modified $F_2$ Ratios

9:3:3:1 No interaction

9:7 Genes in same pathway

9:3:4 Recessive epistasis

12:3:1 Dominant epistasis

13:3 Suppressor has no phenotype

10:6 Suppressor is like mutant

Note: Some of these ratios can be produced with other mechanisms of interaction.

6.4-Expressivity and Penetrance

- **Penetrance** is defined as the percentage of individuals with a given allele that exhibit the phenotype associated with that allele.
- 3 reasons for why a phenotype may not be expressed:
- Environmental Influence
- Influence of gene interaction (modifiers, suppressors, epistatic genes)
- The mutant phenotype is too subtle to measure.
- **Expressivity** measures the degree or intensity to which the phenotype is expressed.