I. Alleles Alter Phenotypes in Different Ways

A. A phenotype is under the control of one or more genes located at specific loci on one or more pairs of homologous chromosomes

B. Alleles are alternative forms of the same gene

1. Wild-type allele: occurs most frequently in a population (often, but not always dominant)

2. For an allele to be recognized, it must cause a change in the phenotype

3. Mutant alleles contain modified genetic information

a) loss of function mutation: causes an enzyme to change shape in such a way that it reduces or eliminates its affinity for the substrate (null allele)

b) gain of function mutation: increases the quantity of the gene product by affecting the regulation of transcription of the gene (dominant)

Oneutral mutations: can not be detected by changes in the phenotype

II. Geneticists Use a Variety of Symbols for Alleles

A. A denotes the dominant allele while a denotes the recessive allele

B. All letters are lowercase, but the wild-type allele is denoted by a superscript (+) a^+/a

1. Further abbreviation is possible such that $a^+/a = +/a$

C. When no dominance exists both letters are uppercase with superscripts to differentiate between the alleles: I^A and I^B

D. Abbreviations are usually chosen to reflect the function of the gene or a disorder caused by a mutant gene

III. Neither Allele Is Dominant in Incomplete or Partial Dominance

A. Incomplete (Partial) Dominance: When a cross between contrasting phenotypes generates an intermediate phenotype (red × white → pink)

1. If the phenotype is under the control of a single gene with two alleles where neither is dominant the results of F_2 can be predicted (1:2:1)

a. Tay-Sachs disease: homozygous recessive individuals have almost no activity of the enzyme hexosaminidase whereas heterozygous individuals have 50% of the normal levels

IV. In Codominance the Influence of Both Alleles in a Heterozygote is Clearly Evident

A. Codominance: the joint expression of both alleles in a heterozygote

B. Characterized by distinct expression of the gene products of both alleles

V. Multiple Alleles of a Gene May Exist in a Population

A. Multiple alleles can only be studied in populations because each individual can only have two alleles

B. The ABO Blood Group: three alternative alleles for blood type

1. four phenotypes: A antigen, B antigen, both antigens, or neither

2. Designated by I^A, I^B, or I^o

3. I^A & I^B are both dominant to I^o, but are codominant to one another

C. The Bombay Phenotype

1. A & B antigens are carbohydrate groups bound to lipid molecules protruding from the membrane of the RBC

2. The terminal sugar differentiates the A & B antigens

3. Both antigens are derived from a precursor molecule called the H substance to which one or two terminal sugars are added

4. Bombay Phenotype (Blood Type O) results from the incomplete formation of the H substrate due to a rare mutation at a locus separate from that controlling the A & B antigens

D. The White Locus in Drosophilia

1. Over 100 alleles for eye color in Drosophilia

2. White (w): complete absense of pigment

3. White-Satsuma (w^{sat}): deep ruby color

4. White-apricot (w^a): orange color

5. White-buff (w^{bf}): buff color

VI. Lethal Alleles Represent Essential Genes

A. Mutations resulting in the synthesis of a nonfunctional gene product can often be tolerated in the heterozygous state, but are lethal recessive alleles (phenotypic ratios - 2/3 Aa to 1/3 AA)

B. Dominant lethal alleles can also occur causing only homozygous recessive individuals to survive (Huntington's Disease)

VII. Combinations of Two Gene Pairs with Two Modes of Inheritance Modify the 9:3:3:1 Ratio

A. Independent assortment occurs so long as the genes controlling

Each character are not linked on the same chromosome, which is known as genetic linkage

B. Can be solved by the forked line method using the controlling character as the starting point

VIII. Phenotypes Are Often Affected by More Than One Gene

A. Genetic influence on the phenotype is often much more complex than Mendel had envisioned

B. Gene interaction: several genes influence a particular characteristic

C. Epigenesis: each step of development increases the complexity of this sensory organ and is under the control of one or more genes

1. Hereditary deafness: heterogeneous trait reflecting the involvement of over 50 genes

D. Epistasis: the expression of one gene/gene pair masks or modifies the expression of another gene/gene pair (antagonistic or cooperative)

E. When studying a single character, a ratio that is expressed in 16 parts suggests that two gene pairs are interacting during the expression of the phenotype under consideration

F. Discontinuous Variation: phenotypic categories are discrete and qualitatively different from one another

G. Novel Phenotypes: gene interactions yield completely new phenotypes in F_2

IX. Complementation Analysis Can Determine If Two Mutations Causing a Similar Phenotype Are Alleles of the Same Gene

A. Complementation analysis: by crossing two mutant strains and observing the phenotype of the F_1 generation we can determine if two mutations are alleles of the same gene

1. $m^1 \times m^2 \rightarrow$ all F_1 are normal/wild-type meaning that the mutations are NOT alleles of the same gene (complementation occurs)

2. $m^1 \times m^2 \rightarrow$ all F_1 are mutated meaning that the mutations are alleles of the same gene

B. Complementation group: all mutations determined to be present in a gene

X. Expression of a Single Gene May have Multiple Effects

A. Pleiotropy: when a single gene results in multiple phenotypic effects

1. Marfan Syndrome & porphyria variegata