Extensions of Mendelian Genetics

Alleles Alter Phenotypes in Different Ways

- Alternative forms of a gene are called alleles.
- Mutation is the source of alleles.
- The wild-type allele is the one that occurs most frequently in nature and is usually, but not always, dominant.
- A mutation can cause the reduction or loss of the wild-type function. Such a case is called a loss of function mutation.
- If the loss is complete, the mutation has resulted in a null allele.
- If the mutation increases the function, it is called a gain of function mutation.

Lethal Alleles Represent Essential Genes

- A loss of function mutation can sometimes be tolerated in the heterozygous state but may behave as a recessive lethal allele in the homozygous state. Homozygous recessive individuals will not survive.
- It can also result in a distinctive mutant phenotype. Such an allele is behaving as a recessive lethal, but is dominant with respect to the phenotype.
- In some cases, a mutation can be a dominant lethal allele, in which case the heterozygote will not survive. Huntington disease is one example.
- For dominant lethal alleles to exist, the affected individual must reproduce before dying.

Multiple Alleles

- Multiple alleles is a MOI that has more than two alleles in the population for one trait.
- It can be studied only in populations, because any individual will have at most two alleles of the same gene.
- Example - ABO blood group.
  - –Each individual is A, B, AB, or O phenotype as a result of dominance of the I^A and I^B alleles to the I^O allele and codominance of the I^A and I^B alleles to each other.

In Incomplete Dominance, Neither Allele Is Dominant

- In incomplete dominance, neither trait is dominant and offspring from a cross between parents with contrasting traits have an intermediate phenotype.
- The phenotypic ratio is identical to the genotypic ratio in incomplete dominance.

In Codominance, the Influence of Both Alleles in a Heterozygote Is Clearly Evident

- Codominance occurs when both of the two alleles of a gene in a heterozygote results in phenotypic detection of both gene products. One example is the MN blood group.

Phenotypes Are Often Affected by More Than One Gene

- Epistasis occurs when one gene masks the effect of another gene.
  - Ex. - Bombay phenotype for ABO blood. The homozygous recessive condition at one locus masks the expression of a second locus.
- When studying a single characteristic, a ratio expressed in 16 parts (e.g., 3:6:3:4) suggests that epistasis is occurring.
- These include recessive epistasis (case 1), dominant epistasis (case 2), and complementary gene interaction (case 3).

Phenotypic Expression Is Not Always a Direct Reflection of the Genotype

- It may be influenced by environment as well as by genotype.
- The degree of expression depends on the penetrance (the percentage of individuals that show at least some degree of expression) of a mutant genotype and the expressivity (the range of expression) of the mutant genotype.
- Completely penetrant means that everyone that inherits it will have some symptoms.
- Incompletely penetrant means that some do not express the phenotype.
- Variably expressive is a phenotype that varies in intensity in different individuals.

Expression of a Single Gene May Have Multiple

- Pleiotropy occurs when expression of a single gene has multiple phenotypic effects, and it is quite common.
  - Examples - Marfan syndrome and porphyria variegata.

Besides the genes in the nucleus, there are also genes in the mitochondria.

- A father gives only one set of chromosomes to its offspring. The mother gives a set of chromosomes and all the organelles. The traits that come with the mitochondrial genes are called maternally inherited because only females transmit them.

Linkage

- Transmission of genes on the same chromosome
- No independent assortment
- Do not get normal ratios
- Linkage maps show the order and distance of genes on a chromosome

X-Linked Describes Genes on the X Chromosome

- Genes on the X chromosome exhibit unique patterns of inheritance due to the presence of only one X chromosome in males.
- *Drosophila* eye color was one of the first examples of X-linkage described.
- Lethal X-linked recessive disorders are observed only in males, since females can only be heterozygous carriers that do not develop the disorders.