Chapter 18
Genes and Medical Genetics

Outline
- Genotype vs. Phenotype
- Dominant vs. Recessive Traits
- Punnett Squares
- Autosomal Recessive Disorders
- Autosomal Dominant Disorders
- Pedigree Charts
- Multiple Allelic Traits
- Incomplete Dominance
- Sex-Linked Traits

Genotype and Phenotype
- Genotype refers to an individual’s genes.
  - Alleles are alternate forms of a gene.
    » Dominant alleles are assigned uppercase letters, while recessive alleles are assigned lowercase letters.
    » Homozygous Dominant = EE.
    » Homozygous Recessive = ee.
    » Heterozygous = Ee.
- Phenotype refers to an individual’s physical appearance.

Genetic Inheritance

Dominant/Recessive Traits
- Forming the Genes.
  - Reduction of chromosome number occurs when pairs of chromosomes separate as meiosis occurs.

Gametogenesis

Figuring the Odds
- A Punnett square is used to determine the phenotypic ratio among the offspring when all possible sperm are given an equal chance to fertilize all possible eggs.
  - If both parents are heterozygous, each child has a 25% chance of exhibiting the recessive phenotype.

Heterozygous-Heterozygous Cross

Autosomal Recessive Disorders
- Recessive disorders can be passed on by parents who are unaffected.
  - Tay-Sachs Disease.
    » Allele located on chromosome 15.
  - Cystic Fibrosis.
- Allele located on chromosome 7.
  - Phenylketonuria.
- Allele located on chromosome 12.

10. Autosomal Dominant Disorders
   - Dominant disorders are passed on by a parent who has, or will develop, the disorder.
   - Neurofibromatosis.
     - Allele located on chromosome 17.
   - Huntington Disease.
     - Allele located on chromosome 4.

11. Pedigree Charts
   - A pedigree chart shows the pattern of inheritance for a particular disorder.
   - Males are designated by squares.
   - Females are designated by circles.
   - Shaded circles or squares are affected individuals.
   - Vertical line down represents a child, while an attached horizontal line across represents more children (siblings).

12. Autosomal Recessive Pedigree Chart

13. Autosomal Dominant Pedigree Chart

14. Beyond Simple Inheritance
   - Polygenic Inheritance.
     - One trait is governed by two or more sets of alleles.
       - Continuous variation of phenotypes.
         - Skin Color.

15. Polygenic Inheritance

16. Beyond Simple Inheritance
   - Multiple Allelic Traits.
     - Gene exists in several allelic forms, although an individual usually only has two of the possible alleles.
     - ABO Blood Types.
       - A - A antigen on red blood cells.
       - B - B antigen on red blood cells.
       - O - Neither A or B antigen on red blood cells.

17. Inheritance of Blood Type

18. Beyond Simple Inheritance
   - Incomplete Dominance.
     - Codominance occurs when alleles are equally expressed in a heterozygote.
     - Incomplete Dominance is exhibited when the heterozygote has an
intermediate phenotype between that of either heterozygote.
- Sickle Cell Disease.
- Heterozygotes protected from malaria.

19 Incomplete Dominance

20 Sex-Linked Traits
- Traits controlled by alleles on the sex chromosomes are said to be sex-linked.
  - X chromosome = X-linked.
  - Y chromosome = Y-linked.
- Most sex-linked alleles are on the X chromosome.
  - Red-Green Color Blindness.
  - Muscular Dystrophy.
  - Hemophilia.

21 Color-Blindness Cross

22 Royal Hemophilia Pedigree

23 Review
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