Altered Cellular Tissue

Heredity

- DNA
  - Chromatin
  - Chromosome
  - Karyotype
- Protein Synthesis
  - Transcription
  - Translation

DNA Replication

Karyotype/Chromosomes

Protein Synthesis

Genetics

- GENES MAKE PROTEINS!!!!!!!!
- Gene
  - Autosomal
  - X-linked
  - Y-linked
- Inheritance
  - Mendelian
  - Non-Mendelian
    - Chromosomal Abnormalities
    - Polygenic
    - Multifactorial
Mendelian Genetic Disorders

- **Autosomal Dominant:**
  - Presence of the protein causes disease
- **Autosomal Recessive**
  - Lack of the protein causes disease
- **Sex linked**
  - X-linked
  - Y-Linked

Autosomal Dominant: Huntington’s Disease

Autosomal Recessive: Cystic Fibrosis, Sickle Cell Anemia, PKU

X-linked Recessive: Hemophilia, Color Blindness

- **Polygenic Inheritance**
  - Each allele produces larger amounts of the protein

Multi-Factorial: CAD

- Genetic
  - LDL receptors
  - Liver Function
  - Clotting
  - Inflammation
  - Disposition
  - Blood Pressure
- Environmental
  - Age
  - Smoking
  - Exercise
  - Diet
  - Blood Pressure
Inheritance

• Genotype vs phenotype
• Penetrance
• Variable expressivity
• Lethal allele

Environmental Congenital Disorders

• Teratogens
  – Alcohol
  – Cigarettes
  – Medications
  – Radiation
• Infections
  – Toxoplasmosis
  – Other: hepatitis B, syphilis, herpes zoster
  – Rubella
  – Cytomegalovirus (CMV)
  – Herpes simplex virus

Screening and Treatment

• Genetic Counseling
• Fetal Diagnostics
• Newborn screening
  – FL: PKU, hypothyroidism, galactosemia, hemoglobinopathies
• Management of Phenotype
• Dietary Modification
• Replacement
• Surveillance
• Surgery
• Gene therapy

Worldview Issues

• Health/Disease
• Genetic diseases