Genetic Counseling

- **Genetic Counseling** is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.

- This process integrates:
  - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
  - **Education** about inheritance, testing, management, prevention, resources and research.
  - Counseling to promote **informed choices** and adaptation to the risk or condition.

*National Society of Genetic Counselors*
Genetic Counseling of the Genome

Genome Cost

Interpretation Cost
Types of Genomic Testing

- Clinical testing
  - Pharmacogenetic
  - Known genetic syndromes
  - Family history of genetic disease

- Research testing
  - Undiagnosed diseases
  - Genetic modifiers of disease

- Direct to Consumer Genetic Testing
  - Initiated by the patient outside of the medical setting
  - Integration into the patient’s medical record
Informed Consent Challenges

- Communicating the limitations of the data

- Incidental findings and obligation to disclose
  - Other pathogenic mutations not known to the family
  - False paternity
  - Findings that alter medical management

- Plan for ongoing evaluation of data

- Variants of unknown significance and reclassification process
Variants of Unknown Significance

- Most people expect a “positive” or “negative” test

- Risk of misinterpretation by the individual or healthcare provider that reviews the results

- Reclassification of variants as benign or pathogenic

- Duty to re-contact individuals when variants are reclassified
Genetic Discrimination Concerns

• Genetic Information Nondiscrimination Act

• Patient Protection and Affordable Care Act
Direct to Consumer Genetic Testing

• Points to consider:
  • Clinical utility:
    • Pharmacogenetic testing
    • Genetic carrier testing
    • Multifactorial disease risk assessment
  • Non-genetics professionals’ knowledge and interpretation of results
  • Inclusion in the medical record
  • Incorporation of family and medical history with DTC sample
  • Consumers expectation of primary care practitioners to be knowledgeable about DTC results
  • Consumer access to genetics professionals