

# GENETIC COUNSELING AND GENOMIC MEDICINE

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# 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.

# 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