Genomic Medicine: A Primer

Michelle Amaral, PhD
UAB Heflin Center
Study Navigator
Background

- Human Genome Project, 2003

- Uses *personal* genomic data to better predict, diagnose, and treat disease
Genomic Technologies

• Next generation sequencing
  – Massively parallel sequencing
Genomic Technologies

• Array-based assays
  – Gene expression profiling
  – Genotyping
Example

• 19 year old male collapsed & died during hockey game

• **Autopsy:** autosomal dominant arrhythmogenic right ventricular cardiomyopathy
  – DNA test detected deletion of gene encoding desmoglein 2

• **Could his life have been spared?** Microarray test would have detected deletion

• **Family testing?** Benefits from early detection

Buchanan JA et al. *Genome Medicine*, 2009
Study: Emphysema and COPD

B-cell receptor signaling pathway

TGFβ pathways and cellular structure, blood vessel morphogenesis, vascular endothelial growth factor, extracellular matrix production, integrin signaling

Campbell JD et al. Genome Medicine, 2012
Ethical Issues

• Consent forms
  – Research vs patient care

• Examples
  – Study: genetic changes in colon cancers
  – Family history of breast cancer
ENCODE Project

• Less than 2% of genome encodes for proteins
• Goal: determine function of the remaining 98%
  – About 80% is dedicated to regulation
    • Enhancers
    • Promoters
    • Non-protein encoding RNAs
• Variants correlated with disease lie within or very near non-coding functional DNA elements