

# Genetic Testing

- What is genetic testing?

# Multiple kinds

1. Single gene test
  - Cystic Fibrosis (CFTR)
  - Sickle-cell anemia (hemoglobin)
  - BRCA1/2 gene
2. Multiple gene/RNA test
  - MammaPrint for breast cancer treatment
  - Paternity tests
3. Genome/Exome Sequencing
  - Sequence (or genotype) extensively the genome or exome

# Rise of cheap sequencing/genotyping has changed everything

- One off genetic tests are FDA approved and require counseling with genetic counselor
- Genetic tests for disease diagnosis or treatment must be ordered by physician
- Then came 23andme!

# Direct to Consumer Genomics

- 23andMe, Ancestry.com, Mapmygenome, Helix
  - Going around physicians and counselors
  - Mostly doing genotyping
  - Focus on Population genetics
  - To give out health information still need FDA approval

# 23andMe vs the FDA

2013 – 23andMe started direct to consumer marketing of their test (~600,00 SNPs)  
Included information about SNPs that are health related

2013– After repeated requests for data, FDA finally banned 23andMe from providing health data

April 2017 – After long negotiations, FDA approved 23andMe to start providing health data again.

<https://www.nytimes.com/2017/04/06/health/fda-genetic-tests-23andme.html>