What Can We Learn From Genetic and Genomic Testing

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HEALTHQUEST
GENETICS vs GENOMICS

- Genetics is the study of inherited traits and genes
- Genomics is the study of how a set of genes behave
Some basic genetics
Some basic genetics

Chromosomes
23 pairs of chromosomes

Hemoglobin

BRCA1

HD
Genes and Disease

Infections
Injuries
Accidents

Heart disease
Cancer
Psychiatric disease
Alzheimers
Diabetes
Autoimmune disease

Down syndrome
Cystic Fibrosis
Achondroplasia
Huntingtons
ALL cancer is genetic …

Complex, multifactorial etiology
BREAST CANCER GENETICS

- ALL cancer is genetic ... **BUT**

- **GENETIC** is not the same as **INHERITED**
  - Only a small portion of cancer is inherited
Hereditary Breast Cancer

Complex genetics and environmental factors

90-95%
Sporadic/Familial

5-10%
Hereditary

The BRCA1 and BRCA2 genes are associated with most inherited breast cancer

BRCA1 (37%)
BRCA2 (35%)
Other genes (28%)

Hereditary Breast Cancer

J Clin Onc 2002;20:1480-1490
J Clin Onc 2007, 25; 6-7
### BRCA mutation cancer risks

<table>
<thead>
<tr>
<th></th>
<th>General Population</th>
<th>BRCA1/BRCA2 mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Breast</td>
<td>12%</td>
<td>40-87%</td>
</tr>
<tr>
<td>Ovarian</td>
<td>1-2%</td>
<td>11-40%</td>
</tr>
<tr>
<td>Male Breast</td>
<td>0.1%</td>
<td>1-10%</td>
</tr>
<tr>
<td>Prostate</td>
<td>15-18%</td>
<td>&lt;30-39%</td>
</tr>
<tr>
<td>Pancreatic</td>
<td>0.5%</td>
<td>1-7%</td>
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Increased risk for a second primary breast cancer

Reports of increased risk for melanoma
What can we learn from genetic testing?

For a woman diagnosed with breast cancer -
- Can affect surgical decisions and risk management
- If a BRCA mutation is identified, other family members can be tested

If BRCA testing is negative...
What can we learn from genetic testing?

For women AT HIGH RISK for breast cancer

- Can affect risk management
  - Increased screening
  - Chemoprevention
  - Preventative surgery

If BRCA testing is negative...
GENETIC vs. GENOMIC