Psychosocial and clinical factors affecting interest in genome sequencing results among young breast cancer patients

Kimberly A. Kaphingst, ScD
Department of Communication
Cancer Control and Population Sciences, Huntsman Cancer Institute
April 1, 2016
Genome sequencing results

- Genome sequencing is increasingly important in care of cancer patients
- Sequencing can generate different types of individual findings

McGuire, 2008; Roberts, 2010; Sharp, 2011
Genome sequencing results

- ACMG recommendations
  - Minimum list of 56 genes
  - Returned to clinician
  - Based on penetrance, pathogenicity, clinical actionability

- Limited data
  - Patient preferences
  - Cancer patients

Green, 2013
Communication preferences for genome sequencing results

Aim 1. Investigate communication preferences for whole genome sequencing results among young breast cancer patients
   • What, when, who, how

Aim 2. Examine factors influencing communication preferences
Why young breast cancer patients?

- Young patients more likely to carry mutations in cancer susceptibility genes
- Sequencing being done for research
- Clinical sequencing increasingly important in care
Young Women’s Breast Cancer Program cohort

Women diagnosed with breast cancer ≤ 40 (n=2200)

BRCA1/2 testing

No mutation

- Strong family history
- Moderate family history
- No family history
- Unknown family history

Mutation found

Have not received BRCA1/2 testing

Cohort:
- 91% Caucasian
- Mean age at diagnosis = 35 yrs
- Mean time since diagnosis = 7 yrs
Methods

• Completed survey of Young Women’s Breast Cancer Program cohort in December 2014

• N=1080 (60% response rate)

• Survey could be completed online, by mail, or by phone
  • 91% completed online version
Survey domains

- Communication preferences
  - Content, delivery
- Genome sequencing knowledge (Kaphingst et al. 2012)
- Worry (genetic, cancer) (Biesecker and colleagues; Gotay and Pagano, 2007)
- Present/future orientation (Strathman et al., 1994)
- Health information orientation (Dutta-Bergman, 2003)
- Health information seeking (National Cancer Institute, HINTS)
- Numeracy (Fagerlin et al., 2007)
Analysis

• Built six multivariable models with outcome of “very interested” in each of six types of results

• Tested for entry into the model
  • Clinical factors
    • BRCA1/2 mutation status
    • Prior genetic testing
    • Family history of breast cancer
  • Psychosocial factors
    • Genome sequencing knowledge
    • Worry about cancer and genetic risk
    • Future orientation
    • Health information orientation
• Controlling for education, marital status, biological children, age, time since diagnosis
Respondent characteristics (n=1080)

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>BRCA1/2 mutation carrier</td>
<td>118</td>
<td>11%</td>
</tr>
<tr>
<td>Prior genetic testing</td>
<td>899</td>
<td>83%</td>
</tr>
<tr>
<td>Strong family history of breast cancer</td>
<td>303</td>
<td>28%</td>
</tr>
<tr>
<td>Have biological children</td>
<td>740</td>
<td>69%</td>
</tr>
<tr>
<td>Current age</td>
<td>46</td>
<td>9.2</td>
</tr>
<tr>
<td>Time since diagnosis</td>
<td>10</td>
<td>7.9</td>
</tr>
</tbody>
</table>
# Content preferences (n=1080)

<table>
<thead>
<tr>
<th>Type of variant</th>
<th>Very interested</th>
</tr>
</thead>
<tbody>
<tr>
<td>Affects risk of preventable disease</td>
<td>77%</td>
</tr>
<tr>
<td>Medication response</td>
<td>74%</td>
</tr>
<tr>
<td>Carrier status</td>
<td>71%</td>
</tr>
<tr>
<td>Affects risk of unpreventable disease</td>
<td>28%</td>
</tr>
<tr>
<td>Ancestry/physical traits</td>
<td>20%</td>
</tr>
<tr>
<td>Uncertain meaning</td>
<td>16%</td>
</tr>
</tbody>
</table>
## Factors affecting content preferences

<table>
<thead>
<tr>
<th>Factor</th>
<th>Prev OR</th>
<th>Non-prev OR</th>
<th>Tx OR</th>
<th>Carrier OR</th>
<th>VUS OR</th>
<th>Non-health OR</th>
</tr>
</thead>
<tbody>
<tr>
<td>Knowledge about limits</td>
<td>1.1</td>
<td></td>
<td>1.1</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Knowledge about benefits</td>
<td>1.2</td>
<td>1.2</td>
<td>1.3</td>
<td>1.1</td>
<td>1.2</td>
<td>1.2</td>
</tr>
<tr>
<td>Genetic worry</td>
<td>1.2</td>
<td>1.1</td>
<td>1.3</td>
<td>1.2</td>
<td>1.2</td>
<td>1.2</td>
</tr>
<tr>
<td>Cancer worry</td>
<td>1.2</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Future orientation</td>
<td>1.0</td>
<td></td>
<td>1.0</td>
<td>1.1</td>
<td></td>
<td>1.1</td>
</tr>
<tr>
<td>Health information orientation</td>
<td>1.5</td>
<td>1.8</td>
<td>1.6</td>
<td>1.4</td>
<td>1.8</td>
<td>1.4</td>
</tr>
<tr>
<td>More than college</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>0.7</td>
<td></td>
</tr>
<tr>
<td>Prior genetic testing</td>
<td></td>
<td></td>
<td></td>
<td>1.4</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Age</td>
<td></td>
<td></td>
<td></td>
<td>1.0</td>
<td></td>
<td></td>
</tr>
<tr>
<td>BRCA1/2 mutation carrier</td>
<td></td>
<td></td>
<td></td>
<td>1.0</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Have kids</td>
<td></td>
<td></td>
<td></td>
<td>2.8</td>
<td>0.7</td>
<td></td>
</tr>
<tr>
<td>Never married</td>
<td></td>
<td></td>
<td></td>
<td>2.0</td>
<td>1.5</td>
<td></td>
</tr>
</tbody>
</table>
Discussion

• Primary predictors of strong interest in results
  • Higher genetics-related knowledge
  • Higher worry about genetic risks, but not worry about cancer recurrence
  • Higher importance of health information

• Clinical factors not significantly associated

• Suggests that lack of knowledge may not be driving interest in secondary findings

• Shared decision-making approaches may support choices about returning results
Acknowledgements

Investigators
• Melody Goodman
• Barbara Biesecker
• Jennifer Ivanovich
• Paul Goodfellow
• Lynn Dressler
• Rebecca Dresser

Research team
• Mackenzie Ray
• Sarah Lyons
• Goldie Komaie
• Joann Seo
• Keri Walton
• Ashley Elrick

Funding: R01 CA168608, National Cancer Institute, NIH; Intramural Research Program, NHGRI
Kimberly Kaphingst, ScD
Professor, Department of Communication
Director, Cancer Communication Research, HCI
The University of Utah
(801) 213-5724

kim.kaphingst@hci.utah.edu