The Role for Genetic Counseling and Genetic Testing in Survivorship

Health Care Disparities In Genetic Testing

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Background

• Hereditary cancer is rare and all healthcare providers have a critical role in identification
  – Approximately 5-10% of breast, colon, ovarian cancer is inherited
• Individuals with mutations have significantly higher risks for cancer than the general population
• Those with higher risk can benefit from different management
  – They have a higher risk for multifocal disease
  – They have (on average) an earlier age of onset
  – They may have a higher risk for additional malignancies
  – Their relatives are at risk
• And those who are not carriers in a family with inherited cancer can have average surveillance
Objectives- Part 1 Survivorship

• Provide a resource for family health history
• Describe the timeline for genetic testing in cancer survivors
• Engage patients in follow-up offering different opportunities and options for genetic

Objectives- Part 2 Health Disparities and Cultural Issues in Genetic Counseling and Testing

• Illustrate, using BRCA1/2 testing an example, studies describing disparities
• Present teaching resources (Toolkit) for genetic counselors to address workforce development for health disparities and cultural competencies
Objectives- Part 1 Survivorship

• Provide a resource for family health history
• Describe the timeline for genetic testing in cancer survivors
• Follow up - different opportunities and options for genetic
NCI Definition of Survivor

• from the time of diagnosis through the rest of his or her life.

• NCI definition of cancer terms
Cancer Survivorship—Genetic Susceptibility and Second Primary Cancers: Research Strategies and Recommendations

Lois B. Travis, Charles S. Rabkin, Linda Morris Brown, James M. Allan, Blanche P. Alter, Christine B. Ambrosone, Collins B. Begg, Nell Caporaso, Stephen Chanock, Angela DeMichela, William Douglas Higg, Mary K. Gospodarowicz, Eric J. Hall, Michie Hisada, Peter Inskip, Ruth Kleinerman, John B. Little, David Malkin, Andrea K. Ng, Kenneth Offit, Ching-Hon Pui, Leslie L. Robison, Nathaniel Rothman, Peter G. Shields, Louise Strong, Toshiyasu Taniguchi, Margaret A. Tucker and Mark H. Greene

Author Affiliations

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TABLE 3

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Prevalence</th>
<th>Associated malignancies (lifetime cancer risk%)</th>
<th>Causative genes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hereditary breast and ovarian cancer syndrome</td>
<td>1:400-1:800 (1:40 in the Ashkenazi Jewish population)</td>
<td>Female breast (56%-87%), Ovarian (27%-44%), Male breast (7%-9%), Prostate (20%), Pancreatic (up to 7%), Melanoma, Colon</td>
<td>BRCA1, BRCA2</td>
</tr>
<tr>
<td>Li-Fraumeni syndrome</td>
<td>Unknown, but rare</td>
<td>Lifetime risk for the following cancers 100% in females and 73% in males: female breast, sarcoma, leukemia, lymphoma, melanoma, colorectal, pancreas, adrenal cortex, brain</td>
<td>TP53</td>
</tr>
<tr>
<td>PTENhamartoma tumor syndrome</td>
<td>1:250,000</td>
<td>Female breast (85%), Thyroid (35%), Renal (34%), Endometrial (28%), Colorectal (9%), Melanoma (6%)</td>
<td>PTEN</td>
</tr>
<tr>
<td>Hereditary diffuse gastric cancer</td>
<td>Unknown, but rare</td>
<td>Diffuse gastric (&gt; 80%), Female breast, lobular (60%)</td>
<td>CDH1</td>
</tr>
<tr>
<td>Peutz-Jeghers syndrome</td>
<td>Unknown (estimates range from 1:25,000 to 1:280,000)</td>
<td>Small-bowel, colorectal, or gastric (52%), Female breast (45%), Ovarian, uterine, or cervical (18%), Pancreatic (11%)</td>
<td>STK11</td>
</tr>
</tbody>
</table>
Timing of genetic evaluation

- Along with the initial diagnosis and early treatment discussions
- During adjuvant chemotherapy
- During follow up and increased surveillance
  - Months or years later
    - Individuals question the role of genetics
    - Individuals are prepared for genetic counseling and possible genetic testing
    - Family history has changed and more evidence exists for the likelihood of an inherited cancer syndrome
    - As providers and patients inquire about “updates” to testing
      » Additions genes have been discovered
      » Additional technology has emerged
  - As other family members become concerned about their risks
Timing Until Genetic Testing


Continuity of Care

- Continuing contact with patients, inquiring about changes in family history
- Asking individuals who are carriers about other family members considering genetic counseling
- Discuss new and emerging genetic testing options
- Continue to monitor the status for changes in variants of uncertain significance
Objectives- Part 2 Health Disparities and Cultural Issues in Genetic Counseling and Testing

- Illustrate, using BRCA1/2 testing as an example, studies describing disparities
- Present teaching resources (Toolkit) for genetic counselors to address workforce development for health disparities and cultural competencies

Genetic Testing and Health Disparities
Genetic Testing and Disparities

- Disparities in testing
- Disparities in rates of telling family members about test results
- Disparities in understanding the implications of the test results from a scientific basis
- Estimation of population prevalence
Studies of Utilization of BRCA 1/2 Testing
Levy, et. al.

- Identified 1474 women age 20-40 with breast cancer from 14.4 million commercially insured patients
- Black and Hispanic women were significantly less likely to undergo testing than (non Jewish) white women
- Those with HMOs less likely than those with POS plans
- Testing rates were higher in 2007 than in 2004
Disparities in BRCA testing: when insurance coverage is not a barrier

W. Olaya, M.D.1,2,3,4, Pamela Engels, M.D.4,5, Jan Y. Wong, M.D.6,7, John W. Negri, Dr.P.H.1,7, Adam Freedberg, M.P.H.1,2, M.D.1,7, Shreela Nay-Dewellbury, M.B.B.S.8,9, Sheree S. Lam, M.D.1,7

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W. Olaya et al., BRCA testing

Table 2: Multivariable analysis of predictors of BRCA testing

<table>
<thead>
<tr>
<th>Predictor</th>
<th>Odds ratio (95% confidence interval)</th>
<th>P value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Personal history</td>
<td></td>
<td></td>
</tr>
<tr>
<td>No cancer</td>
<td>.31 (1.16–8.69)</td>
<td>.005</td>
</tr>
<tr>
<td>Cancer</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>Education</td>
<td></td>
<td></td>
</tr>
<tr>
<td>High school or less</td>
<td>.35 (1.13–9.51)</td>
<td>.03</td>
</tr>
<tr>
<td>Some college or more</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>SES</td>
<td></td>
<td></td>
</tr>
<tr>
<td>1 (lowest)</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>1.06 (0.56–1.98)</td>
<td>.28</td>
</tr>
<tr>
<td>3</td>
<td>.60 (0.18–2.14)</td>
<td>.45</td>
</tr>
<tr>
<td>4</td>
<td>.77 (0.30–2.04)</td>
<td>.70</td>
</tr>
<tr>
<td>5 (highest)</td>
<td>.84 (0.20–3.46)</td>
<td>.81</td>
</tr>
</tbody>
</table>

A GENETIC COUNSELING CULTURAL COMPETENCE TOOLKIT

http://www.geneticcounselingtoolkit.com/

The Genetic Counseling Cultural and Linguistic Competence Toolkit (GCCLCT) is the final product of the 2009 Jane Engelsberg Memorial Fellowship (JEMF) award of the National Society of Genetic Counselors. This award facilitated professional development opportunities for one member of the genetic counseling profession. Nancy Stemberg Warren, MS, CGC in diversity, cultural competence, and pedagogy. The resulting project outcome is this website, which is designed as a flexible, online resource portal to benefit the entire genetic counseling profession.
References- Health Disparities