Growth of BRCA1/2 Genetic Testing in Michigan, 2008-2011

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Background

The general population likelihood of BRCA1/2 breast and ovarian cancer susceptibility gene mutation is estimated between 1/300 and 1/1,000. In the Ashkenazi Jewish population, it is as high as 1/40. Immediate relatives of any individual with a BRCA mutation have a 50% likelihood of mutation.

In 2005, the United States Preventive Services Task Force (USPSTF) issued a Grade B recommendation for genetic counseling and testing based on family history of breast and ovarian cancer and a Grade D recommendation against routine referral for those whose history is not associated with increased risk. In addition, Healthy People 2020 added a related goal:

Healthy People 2020 Goal G-1

Increase the proportion of women with a family history of breast and/or ovarian cancer who receive genetic counseling

Genetic counseling facilitates accurate risk assessment and appropriate genetic testing with applicable follow-up care and surveillance. This process evaluates patients for testing and determines which tests are most relevant. However, BRCA1/2 testing can be ordered by non-genetics providers; the frequency of testing by non-genetics providers was previously unknown.

In the United States, clinical testing for the BRCA1/2 breast and ovarian cancer susceptibility genes is performed by Myriad Genetics, Inc. In 2012 Myriad disclosed data on the number of BRCA tests performed yearly in Michigan by type (comprehensive sequencing, rearrangement, three site and single site testing).

Results

<table>
<thead>
<tr>
<th>BRCA Test Type</th>
<th>Clinical Indication5-6</th>
<th>Cost</th>
<th>Description of Test</th>
</tr>
</thead>
<tbody>
<tr>
<td>Comprehensive Sequencing</td>
<td>Significant family and/or personal history</td>
<td>$3,340</td>
<td>Full sequencing of BRCA1/2 and five specific large genomic rearrangements analyzed</td>
</tr>
<tr>
<td>Rearrangements</td>
<td>No mutation detected with Comprehensive, with significant family and/or personal history</td>
<td>$700</td>
<td>Large genomic rearrangements of BRCA1/2</td>
</tr>
<tr>
<td>Three Site</td>
<td>Ashkenazi Jewish ancestry with family and/or personal history</td>
<td>$575</td>
<td>Three specific mutations examined (accounts for &gt;90% of BRCA mutations in Ashkenazi Jewish population)</td>
</tr>
<tr>
<td>Single Site</td>
<td>Known deleterious mutation in the family</td>
<td>$475</td>
<td>Specific single known mutation analyzed</td>
</tr>
</tbody>
</table>

Methods

As part of a cooperative agreement with the Centers for Disease Control and Prevention, the Michigan Department of Community Health Cancer Genomics Program maintains a surveillance database with visit and testing information on all patients seen for BRCA genetic counseling by a board-certified genetics professional in Michigan, beginning with visits on October 1, 2007.

The data on BRCA tests ordered by board-certified genetics providers was compared to Myriad’s total test numbers in Michigan annually from 2008 through 2011. For each of the four test types, percent growth from 2008 through 2011 and the annual proportion of tests ordered by genetics providers were calculated.

Conclusion

BRCA testing by both genetics and other ordering providers is increasing in Michigan. This is primarily due to increases in comprehensive and rearrangement testing; single site and three site tests constitute a diminishing proportion of all BRCA tests and display little growth in recent periods. Because single site and three site tests may both be used for family members of individuals with identified mutations, we would expect to see these tests increase when high risk patients are correctly identified and tested.

While comprehensive and rearrangement testing are appropriate for those at increased risk of hereditary breast and ovarian cancer, single site and three site tests represent both appropriate clinical care and cost-savings in testing because they:

1) Apply to patients at the highest risk of BRCA mutation
2) Are lower cost compared to comprehensive and rearrangement testing
3) Are often used to identify known family mutations, which can impact recommended follow-up care.

Genetics providers disproportionately order single site and three site tests, and these providers represent a declining measure of all BRCA testing.

Policy and education efforts are needed to:

* Promote genetics referral and improve provider knowledge of available genetics services
* Educate providers about risk identification and appropriate testing options

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References


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