Barriers to Genetic Testing in Newly Diagnosed Breast Cancer Patients: Do Surgeons Limit Testing?

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Background
• Many genes are associated with hereditary breast cancer (BRCA1, BRCA2, PTEN, TP53, STK11, CHEK2, etc.).
• Genetic testing results in breast cancer patients influences surgical planning, modifies screening recommendations, and guides risk reduction strategies for family members.
• The National Comprehensive Cancer Network (NCCN) Clinical Guidelines recommend that breast cancer patients meeting specific criteria be referred to cancer genetics professionals.

Objectives
• Examine the utilization of genetic counseling and testing throughout our hospital system.
• Identify barriers to genetic counseling referral and appointment completion in newly diagnosed breast cancer patients.

Methods
• Analysis of prospectively collected, multi-site database of newly diagnosed breast cancer patients meeting NCCN criteria for genetics referral over 18 month period.
• The 4 hospital sites included 3 with accreditation by the National Accreditation Program for Breast Centers (NAPBC) and CoC (Commission on Cancer). One site was accredited by CoC only.
• Data: Demographics, pathology, imaging, genetic testing results.
• To identify barriers to appointment completion, a phone call was made to patients that were referred, but did not complete a genetic counseling appointment.
• Comparisons were done using two-sample t-tests, Wilcoxon rank sum tests, and chi-square tests. Statistical significance was defined as p<0.05. Institutional review board approved.

Figure 1: Genetic Testing Outcomes of Patients Meeting NCCN Criteria for Genetics Referral

Table 1: Results of Genetic Testing

<table>
<thead>
<tr>
<th>Genetic Testing Results</th>
<th>Number of patients (Total Number Tested: 2022)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathogenic Mutation (BRCA1, BRCA2, CHEK2, ATM, PALB2, MSH2, MTHFR, NBN, RAD51C)</td>
<td>32</td>
</tr>
<tr>
<td>Variant, Uncertain Significance</td>
<td>10</td>
</tr>
<tr>
<td>Variant, Likely Benign</td>
<td>23</td>
</tr>
<tr>
<td>Variant, Likely Pathologic</td>
<td>2</td>
</tr>
<tr>
<td>Negative</td>
<td>264</td>
</tr>
</tbody>
</table>

* The total number of tests is greater than the number of patients as some individuals had testing completed in a reflexive manner (i.e. BRCA1/2 with reflex to panel)
** Variant of unknown clinical significance

Figure 2: Patient Reasoning for Not Completing Genetic Counseling Appointment

Results
• 59% of breast cancer patients meeting NCCN criteria completed a genetic counseling appointment and 55% completed genetic testing (Figure 1).
• Age was only statistically significant difference between patients who were referred to genetics versus those that were not referred (50.9 years versus 60.6 years, p<0.001).
• The most common breast cancer in was low grade Stage 1 Invasive Ductal Carcinoma.
• 32 patients (11% of tested) had pathogenic mutations (Table 1).
• Once referred, there was a 78% appointment completion rate.
• The majority of patients who were referred but did not complete their appointment were contacted (65 of 89, 73%). 13 patients (13 of 89, 14%) had their care transferred to another health system. These patients had various reasons for not completing a counseling appointment (Figure 2).

Conclusions
• Even at NAPBC and CoC accredited sites, only 55% of breast cancer patients at risk for genetic mutation underwent genetic testing.
• The largest barrier to genetic testing in newly diagnosed breast cancer patients is lack of physician referral.
• We expect the main reasons for lack of referrals is a combination of provider education deficiency, limited genetic counselor availability, and potentially patient choice to defer to a later date.

Barriers to Genetic Testing and How to Improve

Figure 3: Barriers and Provider Referral

- Lack of Provider Referral
- Make counseling appointments convenient
- Provide resources on FAQ’s
- Proactive discussion

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American Society of Breast Surgeons Conference 2016, Poster #571

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Lack of Provider Referral

Provider Education

Make counseling appointments convenient

Provide resources on FAQ’s

Proactive discussion