Evaluating surgeon-driven preoperative genetic testing in newly diagnosed breast cancer patients

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Introduction

- Preoperative genetic testing may influence decisions for breast conservation and contralateral prophylactic mastectomy (CPM)
- In 2017, our breast cancer team developed a protocol for breast surgeons to initiate limited breast cancer-specific gene panel testing for surgical decision without a preoperative consultation with a certified genetic counselor (CGC)
- We sought to identify the consequences of surgeon-driven testing, including completion of a formal CGC consultation, larger panel testing, and CPM outcomes

Objectives

- To evaluate the outcomes of surgeon-driven preoperative genetic testing in an academic women’s oncology program
  - Barriers to larger panel testing or CGC consultation
  - Influence on patient receipt of CPM

Methods

- Using our prospective genetic testing database, women who were diagnosed with breast cancer (in-situ or invasive) and underwent genetic testing between July 1, 2017 and September 30, 2019 were identified
- Retrospective chart review was performed to verify age, genetic testing status, presence of deleterious genetic mutation or variant of uncertain significance (VUS), dates of genetic testing, surgical consultation, genetics consultation, and resultant surgical decision making process

Results

### Demographics, n=139

<table>
<thead>
<tr>
<th>Age</th>
<th>Race</th>
<th>Cancer Type</th>
<th>Stage</th>
<th>Grade</th>
<th>Receptor Status</th>
</tr>
</thead>
<tbody>
<tr>
<td>47.1 mean</td>
<td>White (85.6%)</td>
<td>Ductal (80%)</td>
<td>1 (65.5%)</td>
<td>2 (53%)</td>
<td>ER/PR-pos (73%)</td>
</tr>
<tr>
<td></td>
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<td></td>
<td></td>
<td></td>
<td>HER2/neu-neg (74%)</td>
</tr>
</tbody>
</table>

### Genetic Test Results

- VUS: 39 (35.7%)
- Negative: 65 (59.6%)
- Positive: 10 (7.2%)

### Results Delivered By

- CGC Consultation: Yes 82%
- Surgeon: 18%

### Larger Genetic Panel Performed

- Yes: 118 (85%)
- No: 21 (15%)

### Documented Reason for CPM, n(%) (Total CPM: 118)

<table>
<thead>
<tr>
<th>Reason</th>
<th>n</th>
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</thead>
<tbody>
<tr>
<td>Strong family history</td>
<td>5</td>
</tr>
<tr>
<td>Desire to avoid further screening</td>
<td>4</td>
</tr>
<tr>
<td>Improved cosmesis/symmetry</td>
<td>10</td>
</tr>
<tr>
<td>Anxiety about 2nd cancer</td>
<td>7</td>
</tr>
</tbody>
</table>

### Additional Unique Genes with Mutation on Larger Panel

- BRCA2 (4)
- CHEK2 (2)
- PALB2 (2)
- BRCA1 (1)
- ATM (1)
- CDH1 (1)
- TP53 (1)

### Additional Unique Genes with VUS on Larger Panel

- BRCA2 (5)
- ATM (5)
- CHEK2 (2)
- PALB2 (2)
- BRCA1 (1)
- PALB2 (2)
- ATM (1)
- CDH1 (1)
- TP53 (1)

Conclusions

- The majority of patients (92%) completed a consultation with a CGC and 85% proceeded to panel testing
- Of those patients that selected CPM, 53% had a negative genetic test result, highlighting that desire for improved symmetry (63%) and anxiety about a contralateral breast cancer (44%) were also drivers to CPM
- Implementing surgeon-driven preoperative genetic testing was not a significant barrier for patients to complete a formal consultation with a CGC, nor did it present additional barriers to completing larger panel testing

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