Supplementary Table 1. Distribution of mutation types in *BRCA1* and *BRCA2*

A. *BRCA1* mutation types by clinical significance (%)

<table>
<thead>
<tr>
<th>Mutation type</th>
<th>Significant</th>
<th>Not significant</th>
<th>Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td>3′UTR</td>
<td>0.0</td>
<td>0.5</td>
<td>0.2</td>
</tr>
<tr>
<td>5′UTR</td>
<td>0.0</td>
<td>0.0</td>
<td>0.9</td>
</tr>
<tr>
<td>Frameshift</td>
<td>58.3</td>
<td>0.0</td>
<td>2.7</td>
</tr>
<tr>
<td>In-frame deletion/insertion</td>
<td>0.2</td>
<td>1.1</td>
<td>3.8</td>
</tr>
<tr>
<td>Intervening sequence</td>
<td>10.9</td>
<td>13.0</td>
<td>25.2</td>
</tr>
<tr>
<td>Missense</td>
<td>9.9</td>
<td>78.4</td>
<td>60.2</td>
</tr>
<tr>
<td>Nonsense</td>
<td>20.5</td>
<td>0.0</td>
<td>1.6</td>
</tr>
<tr>
<td>Splice</td>
<td>0.0</td>
<td>0.0</td>
<td>0.4</td>
</tr>
<tr>
<td>Synonymous</td>
<td>0.3</td>
<td>7.0</td>
<td>5.2</td>
</tr>
<tr>
<td><strong>Total</strong>a</td>
<td><strong>903</strong></td>
<td><strong>185</strong></td>
<td><strong>560</strong></td>
</tr>
</tbody>
</table>

B. *BRCA2* mutation types by clinical significance (%)

<table>
<thead>
<tr>
<th>Mutation type</th>
<th>Significant</th>
<th>Not significant</th>
<th>Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td>3′UTR</td>
<td>0.0</td>
<td>0.0</td>
<td>0.0</td>
</tr>
<tr>
<td>5′UTR</td>
<td>0.0</td>
<td>2.5</td>
<td>0.8</td>
</tr>
<tr>
<td>Frameshift</td>
<td>67.4</td>
<td>0.8</td>
<td>0.3</td>
</tr>
<tr>
<td>In-frame deletion/insertion</td>
<td>0.1</td>
<td>0.0</td>
<td>2.7</td>
</tr>
<tr>
<td>Intervening sequence</td>
<td>5.7</td>
<td>10.0</td>
<td>12.3</td>
</tr>
<tr>
<td>Missense</td>
<td>2.7</td>
<td>76.7</td>
<td>78.4</td>
</tr>
<tr>
<td>Nonsense</td>
<td>23.4</td>
<td>1.7</td>
<td>0.1</td>
</tr>
<tr>
<td>Splice</td>
<td>0.3</td>
<td>0.0</td>
<td>0.1</td>
</tr>
<tr>
<td>Synonymous</td>
<td>0.4</td>
<td>8.3</td>
<td>5.3</td>
</tr>
<tr>
<td><strong>Total</strong>a</td>
<td><strong>774</strong></td>
<td><strong>120</strong></td>
<td><strong>924</strong></td>
</tr>
</tbody>
</table>

Data were obtained from the Breast Cancer Information Core (BIC) database and updated using recent classification of variants of unknown significance (VUS) as neutral or pathogenic according to the literature (see Supplementary Note).

*a*Number of mutations.
Supplementary Note

A. References used in Figure 2.


B. References used in Supplementary Table 1.