Statewide Collection of Genetic Counseling and Testing of Hereditary Breast and Ovarian Cancer Syndrome (HBOC)

The Utah Department of Health is adding six new required data items for breast and ovarian cancer case reports to the Utah Cancer Registry from all facilities effective January 1st, 2019. Collecting these data items will allow for statewide surveillance of genetic counseling and testing as related to Hereditary Breast and Ovarian Cancer (HBOC). There are many public health and research uses for these data. The Utah Department of Health specifically proposed these items to fill surveillance gaps regarding cancer genetic services in order to inform statewide programmatic public health efforts aimed at reducing the burden of morbidity and mortality related to HBOC. The addition of these data items has been approved by the Utah Cancer Registry’s Advisory Research Committee.

These six state-specific data items are effective for Utah cases diagnosed January 1, 2019 and forward.

- HBOC – GENETIC COUNSELING REFERRAL
- HBOC - GERMLINE BRCA TESTING
- HBOC – GERMLINE BRCA1 RESULTS
- HBOC – GERMLINE BRCA2 RESULTS
- HBOC – GERMLINE BRCA NOS RESULTS
- HBOC – GERMLINE OTHER GENE RESULTS, NOT BRCA

Eligible cases include those meeting the following criteria:

- Female breast cancer cases diagnosed at age 60 or younger (invasive and ductal carcinoma in situ breast cancer)
- All male breast cancer cases diagnosed at any age (invasive and ductal carcinoma in situ breast cancer)
- All invasive ovarian/fallopian tube/primary peritoneal cancer cases diagnosed at any age

| Site and Histology Eligibility Criteria for HBOC Data Items |
|---------------------------------|-------------------------------|------------------|------------------|------------------|
| Cancer Diagnosis Description    | Site Codes (ICD-03)            | Behavior | Included Histology Codes (ICD-03) | Excluded Histology Codes (ICD-03) | Sex, Age |
| Breast, ductal carcinoma in situ | C50.0–C50.9                   | 2        | 8035, 8201, 8230, 8401, 8453, 8500, 8501, 8502, 8503, 8504, 8507, 8508, 8509, 8514, 8521, 8522, 8523, 8540, 8543, 8552 | all, unless listed as “included” | Female: age 60 or less  
Male: any age |
| Breast, invasive                | C50.0–C50.9                   | 3        | all, unless listed as "excluded" | 9050-9055, 9140, or 9590-9992 | Female: age 60 or less  
Male: any age |
| Ovarian, invasive epithelial    | C56.9                         | 3        | all except those listed as "excluded" | 9050-9055, 9140, or 9590-9992 | Any age |
| Primary Peritoneal              | C48.1-C48.2                   | 3        | all except those listed as "excluded" | 9050-9055, 9140, or 9590-9992 | Any age |
| Fallopian Tube                  | C57.0                         | 3        | all except those listed as "excluded" | 9050-9055, 9140, or 9590-9992 | Any age |

Exclusions: Cases identified only from a death certificate or autopsy
<table>
<thead>
<tr>
<th>State added data item name</th>
<th>Codes and description</th>
</tr>
</thead>
</table>
| HBOC – GENETIC COUNSELING REFERRAL | 0 Referred to genetic counseling, but declined  
1 Referred to genetic counseling  
6 Referral not recommended  
9 Unknown or no information |
| HBOC – GERMLINE BRCA TESTING | 0 Genetic testing recommended, but patient declined  
1 BRCA test was done  
6 BRCA testing not recommended  
7 BRCA test was ordered, but results not in medical record  
9 Unknown or no information |
| HBOC - GERMLINE BRCA1 RESULTS | 0 Negative or normal  
1 Positive for a pathogenic mutation  
3 Variant of unknown significance (VUS)  
9 Unknown, no information, or no BRCA 1 testing done |
| HBOC - GERMLINE BRCA2 RESULTS | 0 Negative or normal  
1 Positive for a pathogenic mutation  
3 Variant of unknown significance (VUS)  
9 Unknown, no information, or no BRCA 2 testing done |
| HBOC - GERMLINE BRCA NOS RESULTS | 0 Negative or normal  
1 Positive for a pathogenic mutation  
3 Variant of unknown significance (VUS)  
8 Not applicable  
9 Unknown or no information |
| HBOC – GERMLINE OTHER GENE RESULTS, NOT BRCA | 0 No positive or pathogenic mutations  
1 Positive for pathogenic mutation(s)  
9 Unknown, no information, or no other gene results |
HBOC – GENETIC COUNSELING REFERRAL

Genetic Counseling Referral records whether the patient was referred to genetic counseling or other comprehensive cancer risk assessment consultation for Hereditary Breast and Ovarian Cancer Syndrome (HBOC). The referral may be stated by any health care provider in the medical record.

This data item is effective for Utah cases diagnosed January 1, 2019 and forward. This data item is required only for female breast cancer cases diagnosed at age 60 or less (invasive and ductal carcinoma in situ breast cancer), all male breast cancer cases diagnosed at any age (invasive and ductal carcinoma in situ breast cancer), and all invasive ovarian/fallopian tube/primary peritoneal cancer cases diagnosed at any age. Cases identified only from a death certificate or autopsy or with histologies: 9050-9055, 9140, 9590-9991 will be excluded.

<table>
<thead>
<tr>
<th>Code</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>0</td>
<td>Referred to genetic counseling, but declined</td>
</tr>
<tr>
<td>1</td>
<td>Referred to genetic counseling</td>
</tr>
<tr>
<td>6</td>
<td>Referral not recommended</td>
</tr>
<tr>
<td>9</td>
<td>Unknown or no information</td>
</tr>
</tbody>
</table>

Coding Instructions

1. Assign code 0 when the medical record indicates the patient was referred to genetic counseling, but declined.

Example: Patient referred to genetic counseling by medical oncologist and appointment was scheduled. After being informed insurance will not cover counseling, patient cancelled appointment. Code Genetic Counseling Referral to 0 (Referred to genetic counseling, but declined)

2. Assign code 1 when a referral to a genetic counselor is documented in the medical record. The referral may be stated by any health care provider in the medical record. The medical record does not have to confirm that the patient received genetic counseling.

Example: Referral to genetic counseling is documented in the medical record, but it is unknown if the patient made an appointment or met with a genetic counselor. Code Genetic Counseling Referral to 1 (Referred to genetic counseling)

3. Assign code 6 when the medical record mentions a referral to genetic counseling was not recommended or indicated.

Example: Chart note from managing physician states the patient is unlikely to benefit from genetic testing due to lack of family cancer history. Physician determined that genetic counseling was not warranted. Code Genetic Counseling Referral to 6 (Referral not recommended)

4. Assign code 9 when there is no mention of a referral to genetic counseling in the medical record, it is unknown if a referral was made, or no information can be inferred.
**Note:** If a patient has an in home genetic kit test done and just does a self-referral for genetic counseling code that to 9 Unknown or no information. The referral must be made by a health care provider and stated in the medical record.

**Example:** Genetic counselor report states patient participated in a family conference with four sisters to discuss the finding of a BRCA2 mutation in the family and options for genetic testing. The patient has no history of cancer. No mention of a referral. Unknown if patient was referred by a health care provider. Code Genetic Counseling Referral to 9 (Unknown or no information):

5. Leave blank for cases diagnosed prior to January 1, 2019 and those who do not meet case criteria.
Germline BRCA Testing identifies patients at risk for Hereditary Breast and Ovarian Cancer Syndrome (HBOC) who received germline testing. DNA (usually from blood, saliva, or cheek swab sample) is needed for a genetic test.

This data item is effective for Utah cases diagnosed January 1, 2019 and forward. This data item is required only for female breast cancer cases diagnosed at age 60 or less (invasive and ductal carcinoma in situ breast cancer), all male breast cancer cases diagnosed at any age (invasive and ductal carcinoma in situ breast cancer), and all invasive ovarian/fallopian tube/primary peritoneal cancer cases diagnosed at any age. Cases identified only from a death certificate or autopsy or with histologies: 9050-9055, 9140, 9590-9991 will be excluded.

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<tr>
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<tbody>
<tr>
<td>0</td>
<td>Genetic testing recommended, but patient declined</td>
</tr>
<tr>
<td>1</td>
<td>BRCA testing was done</td>
</tr>
<tr>
<td>6</td>
<td>BRCA testing not recommended</td>
</tr>
<tr>
<td>7</td>
<td>BRCA testing was ordered, but the results not in the medical record</td>
</tr>
<tr>
<td>9</td>
<td>Unknown or no information</td>
</tr>
</tbody>
</table>

**Coding Instructions**

1. Assign code **0** when the medical record indicates germline genetic testing was recommended to the patient, but the patient declined.

   **Example:** Patient does not respond to attempts to set up an appointment for genetic testing or cancels the appointment. Code Germline BRCA Testing to 0 (Genetic testing recommended, but patient declined)

2. Assign code **1** when the medical record mentions BRCA testing was done.

   **Note 1:** Testing may be for a BRCA mutation only (single gene) or performed as part of multi-gene panel testing for several gene mutations.
   **Note 2:** This type of genetic testing for a germline mutation is usually done from a specimen collection. For example, blood, saliva or cheek (buccal) swab.
   **Note 3:** Color (Color Genomics, Inc.) is a recognized genomics lab test that is ordered by a physician – either the patient’s own physician or an independent physician.

   **Example:** Note in medical record states genetic testing was done in 2015, showing BRCA1 and BRCA2 were negative. Code Germline BRCA Testing to 1 (BRCA testing was done)

3. Assign code **6** when the medical record mentions BRCA genetic testing was not recommended or indicated.

   **Example:** Chart note from managing physician states the patient is unlikely to benefit from genetic testing due to lack of family cancer history or another family member testing negative for BRCA. Code Germline BRCA Testing to 6 (BRCA testing not recommended)

4. Assign code **7** when the medical record mentions BRCA testing was ordered, but the results are not in the medical record.

5. Assign code **9** when there is no mention of BRCA testing in the medical record or it is unknown if testing was ordered.
**Note 1:** When results for a 'home kit,' e.g. 23andMe, are noted in the medical record, code 9 Unknown or no information. There are no home genetic test kits, except Color (Color Genomics, Inc.), that do complete testing for BRCA mutations.

**Example 1:** Note in medical record mentions patient is moving out of state to live with a relative during treatment and will need to undergo genetic testing. No further information available. Code Germline BRCA Testing to 9 (Unknown or no information)

**Example 2:** Given the patient’s early onset ovarian cancer, the genetic counselor discussed with patient that she may benefit from BRCA testing, pending financial assistance. No indication that testing was performed. Code Germline BRCA Testing to 9 (Unknown or no information)

6. Leave blank for cases diagnosed prior to January 1, 2019 and those who do not meet case criteria.
Germline BRCA1 identifies test results for the germline \textit{BRCA} 1 mutation. Code results from the genetic test report. If the genetic test report is not available, the results may be stated by any health care provider or genetic counselor in the medical record.

This data item is effective for Utah cases diagnosed January 1, 2019 and forward.

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<tbody>
<tr>
<td>0</td>
<td>Negative or normal</td>
</tr>
<tr>
<td>1</td>
<td>Positive for a pathogenic mutation</td>
</tr>
<tr>
<td>3</td>
<td>Variant of unknown significance (VUS)</td>
</tr>
<tr>
<td>9</td>
<td>Unknown, no information, or no \textit{BRCA} 1 testing done</td>
</tr>
</tbody>
</table>

**Coding Instructions**

1. Assign code 0 when the medical record indicates the germline mutation testing results were negative for \textit{BRCA}1 mutation (normal \textit{BRCA}1 alleles).

2. Assign code 1 when the medical record indicates the germline mutation test results were positive for a pathogenic \textit{BRCA}1 mutation. This includes the wording, ‘deleterious mutation or positive for deleterious mutation’ or ‘heterozygous for a pathogenic mutation.’

3. Assign code 3 when the medical record indicates the germline mutation test results found no positive or pathogenic mutation, but a variant of unknown significance (VUS) is found for \textit{BRCA}1.

4. Assign code 9 when \textit{BRCA}1 results are unknown, there is no information in the medical record, or no \textit{BRCA}1 testing done.

5. Leave blank for cases diagnosed prior to January 1, 2019 and those who do not meet case criteria.
HBOC – GERMLINE BRCA2 RESULTS

Item Length: 1

Germline BRCA1 Results identifies test results for the germline \textit{BRCA} 2 mutation. Code results from the genetic test report. If the genetic test report is not available, the results may be stated by any health care provider or genetic counselor in the medical record.

This data item is effective for Utah cases diagnosed January 1, 2019 and forward.

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<tr>
<td>0</td>
<td>Negative or normal</td>
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<tr>
<td>1</td>
<td>Positive for a pathogenic mutation</td>
</tr>
<tr>
<td>3</td>
<td>Variant of unknown significance (VUS)</td>
</tr>
<tr>
<td>9</td>
<td>Unknown, no information, or no \textit{BRCA} 2 testing done</td>
</tr>
</tbody>
</table>

**Coding Instructions**

1. Assign code 0 when the medical record indicates the germline mutation testing results were negative for \textit{BRCA} 2 mutation (normal \textit{BRCA}2 alleles).

2. Assign code 1 when the medical record indicates the germline mutation test results were positive for a pathogenic \textit{BRCA} 2 mutation. This includes the wording, ‘deleterious mutation or positive for deleterious mutation’ or ‘heterozygous for a pathogenic mutation.’

3. Assign code 3 when the medical record indicates the germline mutation test results found no positive or pathogenic mutation, but a variant of unknown significance (VUS) is found for \textit{BRCA} 2.

4. Assign code 9 when \textit{BRCA} 2 results are unknown, there is no information in the medical record, or no \textit{BRCA} 2 testing done.

5. Leave blank for cases diagnosed prior to January 1, 2019 and those who do not meet case criteria.
HBOC – GERMLINE BRCA NOS RESULTS

Item Length: 1

Germline BRCA NOS Results identifies test results for the germline BRCA mutation, but it is unknown if the mutation was in the BRCA 1 or 2 gene. Code results from the genetic test report. If the genetic test report is not available, the results may be stated by any health care provider or genetic counselor in the medical record.

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<tr>
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</tr>
<tr>
<td>1</td>
<td>Positive for a pathogenic mutation</td>
</tr>
<tr>
<td>3</td>
<td>Variant of unknown significance (VUS)</td>
</tr>
<tr>
<td>8</td>
<td>Not applicable</td>
</tr>
<tr>
<td>9</td>
<td>Unknown or no information</td>
</tr>
</tbody>
</table>

Coding Instructions

1. Assign code 0 when the medical record indicates the germline mutation test results were negative for BRCA mutations (normal BRCA alleles), but neither BRCA 1 nor 2 is specified.

2. Assign code 1 when the medical record indicates the germline mutation test results were positive for a pathogenic mutation, but neither BRCA 1 nor 2 is specified. This includes the wording, ‘deleterious mutation or positive for deleterious mutation’ or ‘heterozygous for a pathogenic mutation.’

3. Assign code 3 when the medical record indicates the germline mutation test results found no positive or pathogenic mutation, but a variant of unknown significance (VUS) is found, but neither BRCA 1 nor 2 is specified.

4. Assign code 8 when BRCA 1 and/or BRCA 2 results are specified

5. Assign code 9 when BRCA results are unknown or there is no information in the medical record.

6. Leave blank for cases diagnosed prior to January 1, 2019 and those who do not meet case criteria.
Germline Other Gene Results, Not BRCA identifies other germline gene panel results when multi-gene panel testing is done. The multi-gene panel results will usually be part of the same genetic test report as BRCA. Some examples of other germline mutations associated with breast and ovarian cancer are Li-Fraumeni syndrome (TP53 gene mutation), Cowden syndrome (PTEN hamartoma tumor syndrome), Lynch syndrome (MSH2, MLH1, MSH6, PMS2, EPCAM gene mutations) and ATM carriers. Code any germline mutation results excluding BRCA in this data item. Code results from the genetic test report. If the genetic test report is not available, the results may be stated by any health care provider or genetic counselor in the medical record.

This data item is effective for Utah cases diagnosed January 1, 2019 and forward.

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<tbody>
<tr>
<td>0</td>
<td>Negative, normal or variants of unknown significance (VUS)</td>
</tr>
<tr>
<td>1</td>
<td>Positive for pathogenic mutation(s)</td>
</tr>
<tr>
<td>9</td>
<td>Unknown, no information, or no other gene results</td>
</tr>
</tbody>
</table>

**Coding Instructions**

1. Assign code 0 when the medical record indicates the other germline mutation test results were negative, normal or variant of unknown significance (VUS) for other mutations

2. Assign code 1 when the medical record indicates the other germline mutation test results were positive for other pathogenic mutation. This includes the wording, ‘deleterious mutation or positive for deleterious mutation’ or ‘heterozygous for a pathogenic mutation.’

   **Note:** When a multi-gene panel test reports a positive for pathogenic mutation and a variant of unknown significance (VUS) in genes other than BRCA, code the positive for pathogenic mutation(s).

   **Example:** 2/20/19 Myriad MyRisk report results include a positive for deleterious mutation in the CHEK2 gene and a variant of uncertain significance (VUS) in the BRIP1 gene. Code 1 (Positive for pathogenic mutation(s)).

3. Assign code 9 when other germline mutation test results are unknown, there is no information in the medical record, or no other gene testing done

4. Leave blank for cases diagnosed prior to January 1, 2019 and those who do not meet case criteria.