Genetic Testing for Hereditary Breast/Ovarian Cancer

In 2005 the United States Preventive Services Task Force (USPSTF) issued a recommendation stating that “women whose family history is associated with an increased risk for deleterious mutations in \textit{BRCA1} or \textit{BRCA2} genes be referred for genetic counseling and evaluation for \textit{BRCA} testing.” The purpose of this fact sheet is to help you understand if genetic counseling and testing for hereditary breast/ovarian cancer may be helpful to you or your family.

How are breast and ovarian cancer inherited?

- Most (70-75%) breast and ovarian cancers are \textit{SPORADIC} - due to age and non-inherited factors.
  - \textit{Example}: Grandmother with breast cancer diagnosed at age 75.
  - \textit{Cancer Risk}: Same as general population.

- About 15-20% of breast and ovarian cancers are \textit{FAMILIAL} – where multiple \textit{minor} genetic factors interact with environment to increase risk. \textit{Genetic testing is unlikely to be helpful for this type of family}.
  - \textit{Example}: Mother breast cancer at 65 and sister with breast cancer at 58.
  - \textit{Cancer Risk}: Somewhat higher than general population, but most women in this type of situation will not get breast or ovarian cancer.

- Approximately 5-10% of breast and ovarian cancers are \textit{HEREDITARY} – due to inheritance of a mutation (mistake) in a single \textit{major} cancer susceptibility gene such as \textit{BRCA1} or \textit{BRCA2}.
  - \textit{Genetic testing for mutations in BRCA1/2 may be helpful to individuals with certain family history characteristics (see below)}.
  - \textit{Breast and ovarian cancer risks are greatly increased above general population, and age of diagnosis is earlier}.

The chart below shows how risk for breast and ovarian cancer will differ depending on the level of genetic risk:

\textbf{Chance for a 20 year-old woman to develop cancer by age 60}

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<tr>
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<th>Sporadic (General Population)</th>
<th>Familial (Minor Genetic Risk)</th>
<th>Hereditary (BRCA1/2 mutations)</th>
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<tbody>
<tr>
<td>Breast cancer</td>
<td>4-5% or 1 out of every 20-25 women</td>
<td>10-15% or 1 out of every 10-12 women</td>
<td>30-50% or 1 out of every 2-3 women</td>
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<tr>
<td>Ovarian cancer</td>
<td>0.5% or 1 out of every 200 women</td>
<td>1-2% or 1 out of every 50-100 women</td>
<td>10-30% or 1 out of every 3-10 women</td>
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When should I find out more about genetic testing for hereditary breast/ovarian cancer?

If any of the statements below are true of your family, you may want to talk to your health care provider about a genetic counseling appointment to learn more about BRCA1/2 genetic testing.

Family History characteristics related to BRCA1 or 2 mutations:

- Multiple (2 or more) cases of breast and/or ovarian cancer on the same side of the family
- Breast cancer diagnosed at a young age (under 50 years old)
- Cancer diagnosed in both breasts (bilateral breast cancer)
- Both breast and ovarian cancer diagnosed in the same person
- Breast cancer in a man
- Ashkenazi (Eastern European) Jewish Ancestry and any family history of breast or ovarian cancer

How can I find out if BRCA1/2 genetic testing is right for me?

The best way to learn if BRCA1/2 genetic testing will be helpful to you, is to have an appointment with someone trained to provide Genetic Counseling for cancer.

The process of cancer genetic counseling begins with the collection of a detailed family and medical history, followed by a discussion to answer the following questions:

- Am I at increased risk for breast or ovarian cancer?
- What are the BRCA1/2 genes and how are they related to cancer?
- What is the chance that I might have a BRCA1/2 mutation?
- How accurate is genetic testing for BRCA1/2 mutations?
- What are the possible test results and what do they mean?
- How will knowing if I carry a BRCA1 or 2 mutation help me to lower my risks for cancer?
- Are there risks and limitations of BRCA1/2 genetic testing?
- How will the results of BRCA1/2 genetic testing affect my family members?

Answering these and other questions are important so that you can make an informed choice about genetic testing.
Why should I consider genetic counseling and possible BRCA1/2 genetic testing?

There is now good scientific evidence that women who carry BRCA1/2 mutations can take effective steps to prevent cancer and/or catch it at early, more curable stages. These screening and prevention strategies are very different from those offered to women in the general population and include:

- Screening with breast magnetic resonance imaging (MRI) and mammogram starting at age 20-25
- Screening more often (every six months instead of once per year)
- Taking medications that can help reduce the chance of getting breast cancer
- Undergoing surgery to remove the ovaries and/or breasts to prevent cancer

It is also important to understand that on average, only half the people in a hereditary breast/ovarian cancer family will inherit the BRCA1 or 2 mutation. Thus, family members can learn they are NOT at high risk and avoid unnecessary screening, surgery, and worry.

Where can I go for more information about Genetic Counseling/Testing for Hereditary Breast/Ovarian Cancer?

The following websites provide information on cancer genetics services and hereditary breast/ovarian cancer.

- [http://facingourrisk.org](http://facingourrisk.org) – excellent resource for individuals at potential hereditary risk for breast/ovarian cancer

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