Angelina's Story And Hereditary Cancers:
Is BRCA Genetic Testing Right For You?

By Hugo D. Ribot, Jr., M.D., F.A.C.O.G.
May 21, 2013

Most have heard about Angelina Jolie's decision to recently undergo a preventative double mastectomy after BRCA testing revealed she had an inherited BRCA1 gene mutation, which dramatically increases one's risk of developing breast and/or ovarian cancer. While Jolie's story rightly enlightened and empowered women about these hereditary cancers and their treatment options, it also raised questions and concerns among many about the test itself.

The BRCA gene test is a blood test that uses DNA analysis to identify harmful changes or mutations in either one of the two breast cancer susceptibility genes — BRCA1 and BRCA2. Women who have inherited mutations in these genes face a much higher risk of developing breast cancer (up to 87% higher) and ovarian cancer (up to 50% higher) compared with the general population. New research strongly suggests that these defective genes also greatly increase a woman's chance of developing cancer in her Fallopian tubes and peritoneum as well.

Women should know that inherited BRCA gene mutations are very uncommon. In addition, they are responsible for only a small fraction of all breast and ovarian cancers – about 5% and 10%-15% respectively. This is one reason why the BRCA test – which costs roughly $3,000 – is not routinely performed on women at average risk for those cancers. In general, the test is only prescribed for people who are likely to have an inherited mutation – such as Jolie, whose mother died at 56 of breast cancer.

This notwithstanding, Jolie's public announcement has, understandably, caused many women who, as she stated, "do not know that they might be living under the shadow of cancer" to question if they should have BRCA gene testing – even if their health insurance does not cover its high costs. Ovarian and other gynecological cancers are of special concern, as they are unarguably far more insidious and difficult to detect at an early, curable stage than breast malignancy. After all, aside from BRCA testing, there is currently no counterpart to the relatively easy mammogram for cancer screening of the ovaries and Fallopian tubes.

Your gynecologist can answer all of these questions and concerns, and is the first medical professional you should contact regarding these inherited female cancers. Based on your personal and family medical history, along with other criteria (see below), he or she can determine if you are a candidate for BRCA testing. Should that be recommended and you test positive for BRCA gene mutation, your gynecologist can provide an estimate of your personal risk of its associated cancers, which varies by individual. He or she also can provide you with information on the latest treatment options, and help you choose which is the right for you.

(more)

Criteria for BRCA Testing

Many medical, healthcare and medical-research organizations have established guideline criteria for BRCA testing. These criteria vary by organization.

According to the American Congress of Obstetricians and Gynecologists (ACOG) – the top standards-setting organization for the ob/gyn specialty – those who may be at increased risk of having a BRCA gene mutation and for whom genetic risk assessment *is recommended* are:

- Women with a personal history of both breast cancer and ovarian cancer*
- Women with ovarian cancer* and a close relative† with ovarian cancer or premenopausal breast cancer or both
- Women with ovarian cancer* who are of Ashkenazi Jewish ancestry
- Women with breast cancer at age 50 years or younger and a close relative† with ovarian cancer* or male breast cancer at any age
- Women of Ashkenazi Jewish ancestry in whom breast cancer was diagnosed at age 40 years or younger
- Women with a close relative † with a known BRCA1 or BRCA2 mutation

According to ACOG, those for whom genetic risk assessment *may be helpful* are:

- Women with breast cancer at age 40 years or younger
- Women with ovarian cancer, primary peritoneal cancer, or fallopian tube cancer of high grade, serous histology at any age
- Women with bilateral breast cancer (particularly if the first case of breast cancer was diagnosed at age 50 years or younger)
- Women with breast cancer at age 50 years or younger and a close relative† with breast cancer at age 50 years or younger
- Women of Ashkenazi Jewish ancestry with breast cancer at age 50 years or younger
- Women with breast cancer at any age and two or more close relatives† with breast cancer at any age (particularly if at least one case of breast cancer was diagnosed at age 50 years or younger)
- Unaffected women with a close relative† that meets one of the previous criteria

* Cancer of the peritoneum and fallopian tubes should be considered a part of the spectrum of the hereditary breast and ovarian cancer syndrome.

† Close relative is defined as a first-degree relative (mother, sister, daughter) or second-degree relative (grandmother, granddaughter, aunt, niece).

These criteria can, understandably, be confusing and daunting to many. Again, your ob/gyn is the first and best professional to contact if you have any questions about whether or not BRAC testing is right for you.

# # #

Hugo Ribot, Jr., M.D., is the founder and medical director of the Georgia Advanced Surgery Center for Women, and the managing partner of Cartersville Ob/Gyn Associates.