



A Great Time for Guidance

Genetic testing for breast cancer opens up opportunities and emotions, says **Lillie Shockney, R.N.**, administrative director of the Johns Hopkins Breast Center and Cancer Survivorship Programs and a two-time breast cancer survivor. Having genetic counseling first is essential to prepare for whatever the outcome of testing might be. Having someone to support you as you make decisions can be just as important.

The genetic consultation and testing program at Johns Hopkins includes volunteers who have tested positive for the breast cancer gene. “Women who are considering genetic testing often like to talk to someone else who’s traveled that road,” Shockney says. “We have the ability to connect them with someone who would be a mirror image of themselves. That provides a lot of comfort. You don’t feel like you’re doing this on your own.”

An Ounce of Prevention

Angelina Jolie’s double mastectomy has people thinking about genetic testing

When Angelina Jolie told the world that she’d had a preventive double mastectomy after learning she was at high risk for developing breast cancer, the public reacted with surprise, compassion—and, for many women, wonder.

According to **Lillie Shockney, R.N.**, administrative director of the Johns Hopkins Breast Center and Cancer Survivorship Programs, only about 5 percent of breast cancers are thought to be caused by the BRCA1 or BRCA2 gene, though women like Jolie who carry BRCA1 have a 60 to 80 percent chance of developing breast cancer, as well as a 40 percent risk of ovarian cancer.

Before women rush out to get tested, however, Shockney strongly advises they first speak to a genetic counselor, who can help them determine what the results might mean for them and their families.

“If a woman tests positive for the gene, is she psychologically ready to take proactive measures?” she says. “There are other issues, too, because it impacts the entire family.” Will her children and siblings, who also might carry the gene, be ready for her news?

On the other hand, a negative test doesn’t mean a woman will never develop breast or ovarian cancer. “If your BRCA gene test is negative, but your family history highly suggests you might develop cancer, extensive testing of other genes may be

required,” says **David Euhus, M.D.**, chief of breast surgery and director of the Breast Center.

Men who have similar family histories should consider genetic testing, too. Their risk of developing breast cancer is low (6 percent for those carrying a gene), but they could pass the gene on to their daughters.

Some women who carry the BRCA1 gene choose to be proactive and have a double mastectomy. Others opt instead to screen for breast cancer more diligently and then take surgical measures if necessary. The latter is acceptable for breast cancer, “but not for ovarian cancer,” Shockney says. “We really have no good way of testing for ovarian cancer.”

She is an advocate for women with the BRCA1 gene having their ovaries and fallopian tubes removed as a precaution. Most important, Shockney urges women to have a thoughtful, serious discussion with a genetics expert before testing and after, if they test positive.

“Women need to determine how aggressive they want to be about prevention,” she says, “and understand every option available to them.” ■



WHO SHOULD GET GENETIC TESTING?

Watch a video of **Lillie Shockney, R.N.**, discussing genetic testing at bit.ly/hopkinsgeneticstesting. For more information, appointments or consultations, call **+1-410-614-4561**.



LOOKING FOR SIGNS

Amy McDougall was fortunate to get a quick stroke diagnosis from **Rafael Llinás, M.D.**, a neurologist at Johns Hopkins.

Some of the most common signs of stroke are numbness or weakness, especially on one side of the body; vision changes; a severe headache with no known cause; sudden confusion; and speech problems. McDougall's only symptoms were a "whoosh" sound in her head—a form of *tinnitus* caused by a change in blood flow—and a speech abnormality, which might be why her condition went undiagnosed as long as it did.

Another factor in the delayed diagnosis could have been McDougall's age. At 47, she was an example of a troubling trend: One in five strokes now occurs in people ages 20 to 55. The increased rate of strokes in younger adults is attributable to risk factors such as high blood pressure, smoking and hormonal birth control, but also greater awareness and better diagnostics.

Llinás is quick to point out that listening to a patient is just as important as looking at symptoms. To him, McDougall's family history of blood clots was a red flag. "Clots can form in the brain as easily as anywhere else," Llinás says. "The imaging is great, but it's all about what the patient tells you about what's going on. Nothing beats that."

It's important to tell your doctor about a personal or family history of conditions, including high blood pressure levels, high cholesterol levels or blood clotting disorders.

Young and at Risk

At age 47, Amy McDougall had no reason to think she was having a stroke. But that's exactly what happened

I've always had migraine headaches, but in April 2012 I had this intense pain in the back of my head, along with a bad earache and toothache. And I'd shrugged off some fleeting numbness I'd had a few months earlier.

It was allergy season, so I thought maybe it was just sinus pressure, and when I saw a doctor he put me on antibiotics.

A couple of days later, I started hearing this extremely loud "whoosh" sound in my head. I was hot and flushed and sat down because I couldn't figure out what I was supposed to be doing with a piece of paper I had in my hand.

My husband and daughter took me to the emergency room. They ran some tests, recommended I see a neurologist in the next two weeks and sent me home.

Later that night, in bed, the "whoosh" sound came back, and I told my husband. That's the last thing I remember until I woke up in the same hospital. This time the doctor identified a blood clot within my brain and jugular vein, but without a neurologist available he suggested we go to Johns Hopkins.

There, they confirmed a blood clot in a vein on the left side of my brain and down the back of my head. I didn't have any risk factors for stroke, other than a family history of clotting.

The team at Johns Hopkins put me on blood thinners, which I'll probably be on for the rest of my life. But I'm doing great now; the pressure in my head is gone, and I'm so grateful I was able to walk out without any lasting problems. ■



To watch a video of Amy McDougall telling her story, visit hopkinsmedicine.org/mystory. For more information, appointments or consultations, call +1-410-614-4561.