


PATRICK MALONE & ASSOCIATES, P.C.

From Tragedy To Justice - Attorneys For The Injured



We win exceptional verdicts and settlements for our clients in cases of brain injury, medical malpractice, wrongful death and other severe injuries.

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from Patrick Malone's book:
The Life You Save: Nine Steps to Finding the Best Medical Care -- and Avoiding the Worst

What Women (and Men) Need to Know about the BRCA Test for Cancer Risk

Dear Patrick,

Angelina Jolie's announcement that she had had both breasts removed because she tested positive for a mutation in the BRCA gene woke up a lot of people to the idea that cancer risk can lurk in our genes.

By coincidence, I had just finished a crash course in the ins and outs of cancer genetic testing, because a client of mine had been misdiagnosed as being mutation-free by a doctor who failed to order the right genetic test. My client got the correct test only after she had come down with a large tumor in one of her breasts and it was too late.

Here's a quick primer to help you figure out how to minimize worry and maximize benefit from genetic testing for cancer.

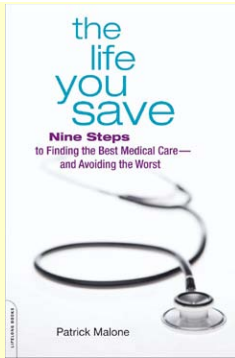
Who Needs to Worry and Why

Do you have a cancer pattern in your family? By that, I mean:

- * Has a sister, mother or grandmother been diagnosed with breast cancer before age 50?
- * Has someone in the family had cancer in both breasts?
- * Have multiple family members developed breast cancer?
- * Is there a history of both breast cancer and ovarian cancer in your family?

And even if no cancer pattern has emerged in your family, one last question: Are you of Ashkenazi Jewish descent?

If you answer yes to one or more of those questions, you should



Learn More



Read our [Patient Safety Blog](#), which has news and practical advice from the frontlines of medicine for how to become a smarter, healthier patient.



read to the end of this newsletter and strongly consider testing. If not, go outside and play. Thanks to serendipity and your wise choice of parents, you need not worry or think about getting this type of genetic test.

Why is family history so important? If a mutated gene that makes a person high risk for cancer is being passed down from generation to generation, it stands to reason that that gene will make its presence known by striking family members with early and multiple cancers.

So family history provides a strong clue, but only a clue. Proof comes with testing, but the testing is not perfect, especially if the right test isn't ordered, as I learned in my recent genetic testing malpractice case.

What Is the BRCA Gene, and How Is Testing Done?

Everybody carries both the BRCA-1 and BRCA-2 genes. The gene name comes from BR for breast and CA for cancer. In fact, we all have two copies of each gene, one passed down from our mother, and the other from our father. The genes make proteins that help repair errors in our DNA that pop up from time to time when our cells divide and duplicate their genetic code. Mutations in either BRCA gene can disable the repair process and make both women and men carriers of the defect susceptible to certain kinds of cancer.

Because a woman can inherit a BRCA mutation from her father, it's important to know the cancer history among grandmothers on the dad's side of the family. You're not necessarily out of the woods just because your own mother is healthy.

By the way, men should care about this for two reasons: first, males can pass the defective gene to children, whether or not the man gets cancer. Second, while the BRCA mutation usually hits women -- with a five-fold higher risk of breast cancer and 10- to 30-times higher risk of ovarian cancer -- men too with a defective BRCA gene have higher risks of some cancers, particularly colon, pancreas and prostate.

The BRCA genes were patented by a laboratory in Utah named Myriad. (A challenge to Myriad's patent is now before the U.S. Supreme Court, which will soon decide if a natural substance like a gene is patentable, but that's another story.)

The BRCA-1 gene is about 80,000 characters long in the DNA alphabet. BRCA-2 is a little shorter. (You'll recall from our earlier [primer on DNA testing](#) that we each have only four letters in our alphabet: A, C, G and T -- but each of us has a total "genome" that is three billion characters long.)

The size of these genes gives you a hint to the potential problems in the test. Two kinds of errors can be tested for: transpositions or spelling errors in the DNA "words," and duplications or deletions, when a whole chunk of DNA "words" get dropped out or duplicated.

Here's where it gets tricky. Myriad's main testing for BRCA mutations is called "Comprehensive BRACAnalysis" -- but it's not really comprehensive, and so the name is a bit misleading. If you read the fine print in Myriad's description of the test, it does do a comprehensive "sequencing" for all spelling-type errors in both BRCA-1 and BRCA-2, but it only tests for a few of the most common mutations of the duplication/deletion type. So if you want a truly comprehensive test, you have to get one additional test, which Myriad calls [BART: for BRACA large Rearrangement Test](#). This test finds all the duplications and deletions anywhere in the lengthy BRCA genes.

But getting the right test is just the first chapter of the story. You need to then figure out what it means. There are hundreds of variants in the BRCA genes, and only some of them are known to be harmful.

Myriad keeps a database of all the variants it has found over the years, including the known and suspected harmful ones, which outside researchers are pressing to be made public.

In the meantime, there is one strategy for getting the best answer to your BRCA profile: targeted testing.

Targeted Testing Is the Best Path to Peace of Mind

The place to start genetic testing, if you can, is with the family member who has been unlucky enough to come down with a cancer that looks suspicious for the inherited type, because it's early or aggressive or comes on the heels of a mother's or grandmother's cancer. Test THAT person for the BRCA genes to see what defect they might have, then test other family members for the same defect.

That strategy would have saved my client from a misdiagnosis. She had a sister who had developed breast cancer in her 30s, then had a recurrence a decade later in the opposite breast. Before that unlucky sister could be tested, my client received the "Comprehensive BRACAnalysis" test and got an all-clear signal. As we've already discussed, that test isn't as "comprehensive" as the name implies. Her all-clear signal turned out to be wrong -- what doctors call a "false negative," because once her sister with cancer was tested, the sister showed a mutation that wasn't covered by the first test. And of course it turned out that when my client got the targeted test, she had the same mutation. Once a family member is shown to have a defective BRCA gene, siblings have a one-in-two chance of possessing the same defect.

The doctor's error -- and it was inexcusable, because this doctor was a geneticist who should have known better -- was in failing to come back to my client and have her tested promptly for the same defect found in her sister.

We learn from our mistakes.

More reading on BRCA gene testing:

Here are two sources I recommend: the [Wikipedia article on BRCA mutation](#), and the [U.S. Preventive Services Task Force new draft recommendations on BRCA testing](#).

Recent Health Care Blog Posts

Here are some recent posts on our patient safety blog that might interest you.

- This should be common sense, but with all the pushback on how bad lawsuits are for doctors and hospitals, it's gratifying to see a new study describing how [malpractice lawsuits help hospitals improve their patient safety programs](#).
- [Baby boomers need to be tested for Hepatitis C virus](#), and they may need a second test, which many don't get. It's important to know for the prevention of liver disease and cancer.

Past issues of this newsletter:

Here is a quick [index of past issues of our Better Health Care newsletter](#), most recent first.

To your continued health!

Sincerely,

A handwritten signature in black ink that reads "Patrick Malone". The signature is written in a cursive, flowing style.

Patrick Malone
Patrick Malone & Associates