

# Q&A: More Patients Experience Inadequate Genetic Testing, Advocacy Official Says

January 07, 2009

Newsletter: [Pharmacogenomics Reporter](#)



**Sue Friedman**  
*Founder and  
Executive Director  
Facing Our Risk of  
Cancer Empowered*

A growing number of patients are reporting instances of receiving either too little or the wrong information about their genetic risk for developing cancer, Sue Friedman, executive director of the patient advocacy group Facing Our Risk of Cancer Empowered, told the HHS Secretary's Advisory Committee on Genetics, Health, and Society last month.

"The wrong tests are being ordered, the wrong individuals within a family are being tested, and people are being given wrong information about what the results mean," Friedman told SACGHS. "A lot of this is based on the fact that the company that develops these tests is providing doctors and consumers with all the information that they are getting. There is no one else filling in the gap."

Regulating genetic tests has become a hotly debated topic in recent months as more genetic tests come to market and the public becomes increasingly aware of their availability both as clinical tests and as direct-to-consumer services.

Some industry observers have said that makers of such tests are doing consumers and patients a disservice by selling diagnostics for which little or no clinical utility has been established.

For its part, the genetic-testing industry has taken the general position that individuals want to know their genetic risk for diseases and they have a right to know this information.

This debate played out prominently when Myriad Genetics launched a television advertising campaign in September 2007 for its BRCAAnalysis test.

Greg Critchfield, president of Myriad Genetic Laboratories, told *Pharmacogenomics Reporter* at the time that it was “a very good time to launch the campaign” since “there’s currently a lot more awareness [among health-care professionals] of the importance of hereditary cancer risk in managing the health care of individuals who carry mutations.”

However, after Myriad’s DTC campaign launched, many doctors complained that their practices were not equipped to handle the resulting influx of patients who saw the ads and wanted to be tested for BRCA mutations.

Regardless of the lack of patient and physician education, diagnostic companies are increasing their marketing budgets and growing their sales teams. Companies like Decode Genetics, Genomic Health, and Laboratory Corporation of America combine direct-to-physician marketing with DTC promotions to get the word out about their tests.

Friedman spoke to *Pharmacogenomics Reporter* this week about how the current regulatory and marketing environment for genetic tests has affected patients represented by FORCE.

Below is an edited transcript of that conversation.

---

### **What is FORCE’s mission?**

FORCE’s mission is to improve the lives of people and families affected by hereditary breast and ovarian cancer. We focus on hereditary cancer syndromes

that include breast and ovarian cancer, such as BRCA and HNPCC, and a lot of the highly penetrant gene mutations that have been discovered. But our members also include people who have strong family history of breast cancer and no identifiable mutation. So, some of these newer, low-penetrant gene assays and genome-wide association studies that are coming out may be relevant to people in our community ... as they struggle to figure out what their risk is [for cancer] even though they are BRCA negative.

Genetics is a rapidly changing area of research information and there's a lot of new information coming out. It's really hard for some healthcare providers to sort through [the information], learn everything there is to know, and then take the time to provide consumers with all the information to make good decisions about their risk and what to do about it. There are options available [for managing cancer risk]. There are things that someone who may be high risk may pursue if they have a better sense of what their [personal cancer] risk is.

### **What do you think FDA's role should be in regulating genetic tests?**

[The regulation of tests] is very relevant to our community as more tests become available and as companies begin to promote them. It's not that I have strong feelings regarding who should be regulating [genetic tests], but I recognize that there is a huge regulatory gap. What I do believe is that some agency should be regulating testing. Consumers already think that these tests are being regulated. There is very little understanding on the part of the consumer that if a test is done at a CLIA-approved lab there is very little oversight over the actual test itself ...

People assume that once a test is made available to the public, and has been commercialized, and is being marketed to them, that there has been some regulations and government oversight over the test. Most people assume that it is the FDA regulating these tests, but so few people understand the regulatory landscape ... FORCE just wants to make sure that this is on the radar of the government and someone addresses this regulatory gap. In the mean time, our organization and others like us need some assistance in what information we can give to consumers that is evidence-based, credible, and isn't totally discounting

the science. People want answers. They know what technology is out there and they want to have access to it ...

I'm not anti-technology in any way. I feel I have personally benefited greatly from having [BRCA] genetic testing. Many in our community feel the same way. But, currently, it's very hard for healthcare providers to know what they do and don't know about genetic testing unless they are experts in genetics. There are some physicians and facilities that are doing a great job in informing consumers, [directing them] to genetic counseling, and telling them what a test will and will not do. Others aren't doing such a great job of it.

The other issue is that some of the biotech companies that are doing the genetic testing are discouraging a full genetic counseling process with experts who are qualified in genetics and facilitating less accomplished health care providers.

**Can you give an example of where patients have been discouraged from seeking out genetic counselors?**

Let me rephrase that. It's more that [the companies] are directing patients to providers who will order genetic testing without actually disclosing the qualifications, or lack thereof, of these providers. Physicians are being told they should be doing genetic testing in their office and being given access to genetic test kits without any real guidance about what information should be provided up front, how to do a full genetic counseling session, and what the limitations are of the genetic tests being promoted.

What we are seeing at FORCE are people who have had genetic testing, who either have no idea about what [their results] mean. Or, they are considering genetic testing because their doctor handed them a pamphlet and said, 'I have a kit here and I want to pull your blood,' and they said, 'Wait a minute, I want to find out more information.' Those are the people who we will refer to a genetics expert. I cannot tell you how many times people I have referred have said, 'I had no idea. My doctor had no idea.' There have been women who have been given their genetic test results while they were driving a car without any [advance] preparation or discussion about the impact the results may have. So, we're seeing

that. And that's concerning because there is an increase in it. I think it's doing harm to the community that our organization serves.

**Is there a strong marketing effort from genetic testing companies, where similar to pharmaceutical firms, they will go to doctors and urge them to incorporate tests into their practice?**

Absolutely. There is this kind of marketing of genetic testing; it's allowed. There is even access allowed to consumers where they can order certain tests online. It still has to go through a physician. I'm not trying to say that a doctor shouldn't be doing genetic testing, ... But I don't think a laboratory should be going around telling physicians that they are qualified [to order and interpret genetic tests] either. I think that's a conflict of interest.

So, I do believe it is happening for sure. We are seeing it.

**Can you say which companies are marketing in this way to doctors?**

Let's put it this way: There are no regulations that prevent [genetic testing] companies from [marketing to physicians]. When you look at the pharmaceutical industry, as of Jan. 1 it is not even allowed to put their name on a pen. There are a lot of regulations over what they can and cannot say to physicians. There are no similar regulations over many biotech companies.

I believe it's a shared responsibility. It's not only up to the labs. Physicians should understand the tests they are ordering. They should be doing their research. They should invest in resources to hire a genetic counselor if they want to do genetic testing or find out where they can refer patients, so patients interested in genetic testing can get all the information they need to make an informed decision on whether or not genetic testing will benefit them.

Genetic testing, depending on the tests, is not equally informative for every patient and [if the wrong test is ordered or the test is misinterpreted] it's a waste of money. I've heard the argument that it's patronizing to tell consumers what [technology] they should and should not have access to. I'm not trying to tell

people what they should have access to. But I also know it's also very difficult for a consumer to make an informed decision if they are not getting all the information. People trust that their doctors are giving them all the information that they need and that's not always happening.

**How does FORCE help the patients who come to the organization and are confused about genetic testing? And have you seen an increase in the number of patients coming to you for more information?**

We have seen an increase in the past year.

There are a few things we do to provide people with credible information. We have a whole area on our website with information about genetic testing and genetic counseling, and how people can find genetics experts in their area. There are a few databases that we turn to in order to find a genetics expert in someone's area. One is the National Society of Genetic Counselors website. They have a look-up tool [with which] you can find a genetic counselor. Often we'll direct people there or we will help them look up someone ... The National Cancer Institute also has a look up tool that is not just for genetic counselors, but lists physicians and nurses. This tool includes information about whether they are board certified ... The Genetic Nurse Credentialing Commission, they have credentialed genetics nurses. [FORCE] is in the process of trying to compile a database of genetics services and facilities that offer specific services to people with hereditary conditions, so we can help people find genetics experts in their area.

We know that there are some areas where there isn't equal access to genetic counselors. There is a company [called Informed Medical Decisions] that does genetic counseling by telephone with board-certified genetic counselors. They volunteer time to answer help-line calls for [FORCE]. We refer people who have questions to them. They work through the insurance company to make sure people have good access to credible information. They do not do direct-to-consumer marketing ...

We're also seeing an increase in people who were not told that there were genetic

counselors in their area, and they lived in big cities like Chicago and Washington, DC. And in some of these cases there was no emergency. For example, we are seeing young people, 18- and 19-year-olds, who didn't have a cancer diagnosis, who had genetic testing in large cities who were not referred to genetic counselors. It would be hard for us to say that was an access-to-care issue. That's a marketing issue. That is [the kind of scenario] where the patient was told about the test by their OB-GYN or their general care provider. The doctors have information or a test kit, and they tell the patient that they can test for a gene, and then the patient is not provided access to genetic counseling.

**Another example of direct-to-consumer marketing of genetic testing would be through recently launched consumer genomics companies like 23andMe and Navigenics. Within the FORCE community, have you seen an increase in people getting their genomic profile done?**

Not yet ... There's been a lot of media coverage on these tests, but I haven't seen a lot of direct-to-doctor marketing on it ... We deal with a specific segment of the population that is interested in the risk for cancer ... So, at this point, no we're not seeing a lot of that.

But, the processes are the same. If there is anyone concerned about their risk, I'm going to recommend they see someone who has expertise in risk assessment ... Sending them to a genetics expert is the best way I know that these patients are going to get a three-generation family pedigree if available, and genetics experts are going to be looking at what syndrome does this fit, not what test kit do I have on my desk that I can test my patient for.

I've gotten calls from people who have had breast cancer and thyroid cancer in their family, which can be Cowden syndrome and they were given BRCA testing. And I worry that those people, if they haven't been to a genetic counselor's office, that's going to be the end of the story. They are going to be told that there is no risk of a hereditary syndrome, and no further follow up is going to happen for them. That's false reassurance and they're missing the potential to get an answer. There are syndromes out there that are less common for which there aren't commercially-available tests. So, there's less interest in that from the testing

companies, so doctors get less information. The lack of information is not the same as a negative test result.

**Some doctors have expressed that as more genetic tests become available and consumers become aware of the availability of these tests, there may be an increase in lawsuits if doctors fail to do genetic testing.**

I don't think a physician is going to get sued for referring someone to a genetics expert. I find that really frustrating when I hear that argument. The doctor doesn't have to do the test to do the right thing. I would argue that they are putting themselves at more liability ordering a test they have no idea how to interpret.

So, you go to your primary care physician and he hears a heart murmur. Is he going to work you up or is he going to send you to a cardiologist? It's not any different. Where's the liability? Is the liability in him failing to work up your heart murmur or in failing to send you to an expert who could work up your heart murmur appropriately?

There is a field of genetics ... There are health care providers with advanced training in genetics, and who are credentialed in it. Doctors need to bother to refer people to these experts. It's the same responsibility. So, I think it's one of those comments you hear from people who promote the concept of genetic testing in the primary care setting. I think it is missing the boat. Physicians need to realize they may be putting themselves at equal liability for ordering a test they do not understand and might misinterpret.