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The Genetic Tsunami

By Jean Elliott

A Virginia Tech genomics expert discusses personalized medicine, science in the cinema, and the four questions people should ask before peering deeply into their own DNA.

Doris T. Zallen, a professor of science and technology in society in the College of Liberal Arts and Human Sciences, has navigated the curricular divide between science and the humanities. Trained as a geneticist, her research focuses on the ethical, social, and policy issues raised by genetic testing.

We hear a lot about “the new age of genomic medicine.” What does this mean?

Genomic medicine is a term that describes the practical application of genetic information in the health-care setting. This includes the use of genetic tests for establishing a diagnosis, for selecting treatments tailored to each individual’s genetic makeup, for predicting one’s risk of developing an illness in the future, and even for developing programs of disease prevention.

Genetics itself is not new but for a long time the applications in medicine didn’t occur because humans are hard organisms to study. Their generation time is too long. They seldom have enough children to sort out genetic issues, the way you can with fruit flies, or mice, or bacteria. Starting in the 1950s, this began to change as new tools were developed that could explore the genetic material, DNA, in considerable detail.

As years went on, scientists were able to develop tests that could determine the status of whole chromosomes and also determine the status of individual genes, tiny parts of chromosomes. Just over 10 years ago, with the conclusion of the Human Genome Project, the genetic code within all of the human chromosomes was identified. That knowledge has made it possible to identify specific genes that may, in concert with other genes and environmental factors, be responsible for particular disorders. So the code within the genes can help us determine if someone has a disorder or predict if someone is more apt to develop a disorder.

Invent the Future

What prompted you to write your books, “To Test or Not to Test: A Guide to Genetic Screening and Risk” and “Does It Run in the Family? A Consumer’s Guide to DNA Testing” (both Rutgers University Press)?

I was part of the early effort to develop tests for individual genes. When we came to offer them to families for whom those tests would be appropriate, I discovered that although many were happy to have the information that those tests would provide, many others were unhappy.

At first I was surprised, but then I began talking with them and I realized that they had excellent reasons for not wanting the information. Trying to understand why genetic information prompted these two different reactions propelled me across the curriculum into the field of bioethics. I wanted to pursue the reasons that this technology, which has great potential to do good in medicine, was also in some cases a cause of great concern for patients.

I interviewed many people on the front lines of genetic testing. This includes genetic professionals, doctors, genetic counselors, and researchers, but also I talked to the families, because they are the ones who ultimately have to make decisions about whether a particular genetic test is right for them.

These oral histories have remained the foundation of my work to this day and led me to write those books for a general audience, encouraging people to make decisions for themselves about genetic testing and not to do it because their friends were doing it or because they saw an article in the newspaper about it, but to think through carefully about whether genetic testing is right for them.

Based on your research, what are the questions that everyone considering genetic testing should ponder?

Making a decision is not easy. Based on my interviews, I’ve come up with four main questions that people should ask themselves:

1) Is there a family history?

Having a family history of colon cancer, Alzheimer’s disease, or cardiovascular disease could be an indicator of a genetic component. It is important to keep in mind that, since these disorders also have environmental components, there can be a family history without any genetic basis.

2) Will the test give me information that will be helpful?

Most medical tests give precise, definitive information. Some genetic tests do too. For the common health problems like breast cancer or Alzheimer’s disease, however, the genetic tests do not provide a definite answer. They provide an estimate of risk, a probability that your chances are higher or lower than the population average.

If one has the APOE4 gene, the risk for having late-onset Alzheimer's goes up, but many people with this genetic signature do not develop it and many people develop it even without the APOE4 gene. There are other genes involved, there are environmental factors, and all of these fit together in a very complex way that we still don't understand.

3) Is this the right time in your life to do this?

Genetic testing comes with a strong emotional component. So you shouldn't take on genetic testing when your life is complicated with other factors. If you're moving or if you're having problems at work or someone is sick and you have to be there for them – it's probably not the best time to take on genetic testing; it is best to have genetic testing at a relatively calm point in your life.

4) Do the advantages of getting this information outweigh the disadvantages?

Here each person has to weigh a number of factors that are unique to them. Are you an information seeker? Are you a worrier? If you find out that you are at a higher risk for Alzheimer's and then you forget where you put your keys, will you begin to wonder if today's the day that the disease is going to start? People are different and you have to think about how that genetic information is going to affect you.

What is the value to your family? Perhaps your family would benefit if you find that you have a higher risk for colon cancer. They might also have inherited the same gene. If so, they could take steps to have regular checkups, colonoscopies. But in some families, genetic information has caused divisiveness as people have to deal with information that is complicated and tough to take onboard.

Then there is the problem of discrimination. What happens if other people find out about your genetic predisposition? The Genetic Information Nondiscrimination Act protects people from having genetic information used against them in their health insurance and in the work place but that law does not apply to long-term care insurance, or life insurance, or disability insurance. What happens if other agencies or institutions find out?

Other people say, if I find out, I'm going to be more proactive. I'm going to take part in research and try to reduce my risk.

It's not a one-size-fits-all decision. For instance, a woman at risk for breast cancer because of family history was tested and found to have a risk-raising mutation in one of the breast-cancer genes. She was able to have more regular screenings, to take drugs to lower her risk. She felt that was a useful test to have. But when she considered the test for Alzheimer's disease, for which she felt there was nothing she could do, she decided it wasn't useful to have that information. We each have to determine what fits best for us.

What has been your biggest challenge?

Among the biggest challenges is keeping up with the pace of research. We're on the brink of having affordable whole-genome sequencing. Instead of testing for an individual gene, you can have your whole genome sequenced for your doctors to inform your health care. It's a challenge to anticipate what issues are going to arise with this tsunami of genetic information, how to deal with these issues, how to make the wisest health-care policies.

You recently received a grant from the Alzheimer's and Related Diseases Research fund. What are you working on now?

That grant is enabling us to evaluate an online educational tool we've developed here at Virginia Tech, a tool that provides guidance in how to make a genetic testing decision. It's called a decision aid. Decision aids are used in many places in medicine usually to allow people to decide which treatment they prefer, such as to have surgery or to use radiation. Our decision aid allows people to understand the four questions I mentioned as they think about having a genetic test for Alzheimer's disease.

As a consultant for the entertainment industry, what shows and what particular issues have you addressed?

I was a consultant for "Chicago Hope" when they featured a story about gene therapy. At the time, gene therapy was a very new field. I worked with them to explain how it worked, the prospects for success, and helped them flesh out a plot that was scientifically accurate and not misleading. When the science presented is not accurate, it can mislead people into thinking that certain options are available when they are not or that they're more successful than they really are.

In recent years I've been part of the Science and Entertainment Exchange of the National Academy of Sciences and asked to comment on particular lines of dialogue related to genomic medicine. One was for the program "House." I'm pleased that people in the field of entertainment really do want to get it right.

You've served in an advisory role for the National Institutes of Health (NIH) and the Centers for Disease Control and Prevention. How can science and the public collaborate?

There have to be ongoing opportunities set up for that collaboration. When I was serving on the Recombinant DNA Advisory Committee, we had a public audience able to contribute their thoughts and reactions, and we

learned an awful lot from them. The public audience also benefited from listening to the discussions about genes and the science behind what was being proposed.

Collaboration between scientists and the public in forging policies for genetic testing is essential. I've found that the people who have faced genetic testing have a great deal to add to decisions, policies, and programs. The collaboration helps both sides. We need more venues for people to get together and talk to each other. That remains part of our Choices and Challenges forum series at Virginia Tech, to provide space for people to get together, tackle hard issues, and to value each other's contributions.

How do human sciences and education help to inform today's scientists?

I think I'm the living example of this. Without drawing on insights and expertise from the humanities, I could not have addressed the issues that arose when people were telling me their concerns about the genetic tests I was developing as a researcher.

Understanding the history of genetic testing contributed to my ability to recognize and address concerns people have about genetic privacy or possible discrimination. The philosophical and the literary components of the humanities enriched my understanding of what I was doing in the lab, and how I should do it better.

I do miss the lab from time to time, but I have no regrets for crossing the curricular divide and immersing myself in the traditions and the approaches of the humanities.

I'm convinced that these approaches are essential to figuring out how to usher in the new age of genomic medicine — and other scientific fields as well — in a way that can accomplish the most good for patients and families.