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Motivations for Personalized Genetic Testing Include Explaining — Not Just Predicting



By Robert C. Green, MD, MPH

Last week, I invited the venerable [Lee Hood](#) to speak to us at Harvard Medical School, and I was reminded again of his prescience in describing and advocating for “[P4 Medicine](#)” — or predictive, preventive, personalized and participatory medicine — over 10 years ago, in 2004!

And while Lee’s vision of P4 Medicine embraces a model of systems biology that is broader than just the genome, the democratization of genetic testing over the past decade falls squarely within the powerful narrative prediction and prevention. Indeed, much of the controversy surrounding direct-to-consumer (DTC) genetic testing when it launched in 2007, and even today, has revolved around the degree to which genetic markers can, or cannot, meaningfully add to risk stratification, and do or do not result in health value.

Our work on the NIH-funded [Impact of Personal Genomics \(PGen\) Study](#) has been measuring exactly why people want DTC genetic testing, how they understand their results and what

changes they make in their lifestyle or health care.

Through our surveys and interviews, we kept hearing that DTC customers were not just interested in prediction, but that they were also interested in understanding conditions that they or their family members already had. So, in our most recent paper, we decided to ask: if a customer has already been diagnosed with a health condition, how does that affect their interest in learning about their genetic risk factors for that condition?

You see, if they were getting the test because they really wanted to know their future health risks, they would probably be *less* interested in their results for a condition they already have. After all, if you have been diagnosed with rheumatoid arthritis and you get genetic test results saying “you have a heightened risk of rheumatoid arthritis,” we might expect a response along the lines of: “Thanks, Captain Obvious.”

In a [PGen analysis led by visiting scholar Susie Meisel](#) of University College London, we examined survey responses from PGen Study participants after they had ordered their DTC tests, but before they had received personal genomic test results. We evaluated their level of interest in their results for eleven different conditions, and stratified those responses by whether they had a family history or had been personally diagnosed with any of those conditions.

Not surprisingly, we found that participants with a family history of a particular condition were more interested in their genetic risk results for that condition (compared to other conditions for which they did not have a known family history). This backs up similar findings in previous studies of people with family histories of [breast cancer](#) and [heart disease](#).

But here’s the really interesting part: Participants were consistently most interested in their genetic risk factors related to the condition that they already have. From the perspective of “predictive” genomics, this makes no sense whatsoever. If you

have already been diagnosed with a condition, why would you be especially interested in your risk of getting it?

One reason participants might be so interested in learning about the genetics of a disease they already had might be concerns about passing that condition on to their children. Genetic testing related to reproductive risks is already common; prospective parents undergo genetic testing to determine their carrier status for conditions like [Tay-Sachs](#), [cystic fibrosis](#) and [sickle cell anemia](#). But conditions like these usually have a straightforward, easy to understand inheritance pattern that is based on both parents having a defective version of a single gene—think of the [Punnett squares](#) that introduced us to genetics.

But the results in this study are a bit more complicated than Punnett squares. The participants in question were asked about — and had previously been diagnosed with — conditions that are inherited in more complex, harder-to-predict ways, a combination of multiple genetic and environmental factors. These were conditions like heart disease, obesity, high cholesterol, asthma, and skin cancer — all of which have genetic components, but none of which are directly inherited. Being born with a lower genetic risk of heart disease, for example, does not give you a free pass; an unhealthy lifestyle can still increase your overall risk dramatically. Similarly, having a higher genetic risk of heart disease is not a guarantee that you will develop the disease; a healthy lifestyle can still be protective.

So if it wasn't for their children, then why were the participants more interested in the results for conditions that they already had? Our study suggested that the motivations for this interest go beyond simply predicting risk... and that for these participants, their fascination with genetics was an attempt to understand the condition that they had.

If you live for years with a condition like multiple sclerosis or heart disease or diabetes, you may think of the condition as more than a diagnosis. It may be an integral part of how you see

yourself, a part of who you are. So naturally, you might want to understand it better.

And if a genetic test could shed light on the sticky question of “how did this happen to me,” well that might well justify the cost of a DTC genetic test.

Previous studies have found that some people take solace in knowing that a diagnosis wasn't totally their fault — that some risk factors were out of their control. In [one study that Susie conducted](#) before ours, overweight participants who learned about their increased genetic susceptibility for obesity said they felt less guilt and self-blame. Other studies have found that for people diagnosed with mental illness, learning of their genetic risk for the condition relieves internal speculation about its causes.


However, if you are looking for somewhere convenient to place the blame, a genetic test won't always help you with that. You may end up with results that suggest genetics didn't play a major role in your diagnosis, which, if you're inclined to see it this way, could reinforce notions of self-blame. On the other hand, you could see those results as a sign that you have some control over your own fate, that you aren't “doomed” by bad genes. Or you could go a different (probably more constructive) route: Whatever your results, you could simply use them to help you understand your condition and make more informed decisions about how to manage it going forward.

Before this study, we assumed that predicting future health risks is the main benefit most people derive from personal genome testing. For many people, that may be true. But gaining clarity and peace of mind through a better understanding of a condition you already have sounds like a benefit as well, and one that has infused DTC genetic testing with little recognition until now.

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