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My experience with direct to consumer genetic testing

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As a genetics researcher interested in learning more about direct to consumer genetic testing, I thought the best way to start studying this topic was to get myself tested by the various companies. At the time, there were four different companies that offered this service (23andMe, deCODE me, Navigenics and Pathway Genomics), so I sent a sample to each company to both experience the process and see how my results differed across companies.

The first results that came back were from Pathway Genomics. I was shocked by the results that told me I have a very high genetic risk for macular degeneration, the most common cause of age related blindness. None of my family members has or had macular degeneration, so these results surprised me. Coincidentally, at the time I got the results, I was at a meeting with an ophthalmologist who collected and manages one of the largest genetic databases for macular degeneration. I was anxious about my results, concerned that I was almost definitely going to go blind, so I approached her to talk about my results. When I told her my concern, she looked at me blankly because there really are no genetic determinants that increase your risk for macular degeneration to the point where you will definitively get the disease. She did not understand what the company could have tested that would justify my concern. That weekend, I started taking expensive eye vitamins.

Over time, I received my results from 23andMe, Navigenics and deCODEme. The results were strikingly different. Although I had an elevated risk for macular degeneration across the companies, my risk of macular degeneration was different for each company. Pathway Genomics reported that I should “be proactive” regarding my risk for macular degeneration. This is the highest risk option; the other two levels were “learn more” and “live a healthy lifestyle” (more recently, Pathway Genomics changed their risk levels to “average risk”, “above-average risk”, and “increased risk”). In contrast to the vague risk levels given by Pathway Genomics, the other three companies gave me probabilities for my estimated lifetime risk of macular degeneration: Navigenics gave me a 61% lifetime risk, 23andMe gave me a 38.1% lifetime risk, and deCODEme gave me a 90% risk.

Because I’m a medical researcher, I then investigated how it can be that these risk estimates are so wildly different from one another. I first looked to confirm that they genotyped the same genetic markers. This was the easiest thing to do because the specific genotypes were in all of my reports. Then I looked to see if they used the same medical studies to compute the risk. These were also essentially the same. It turned out that the difference is the methods they used for calculation of risk. I had assumed that was the easy part because it’s just a matter of multiplying numbers together including baseline risk of macular degeneration, the joint impact of the genetic variants on this risk. It turns out that the problem is that we actually don’t know the baseline risk of macular degeneration for people

who don't have the genetic variants, and we don't know how the genetic variants interact with each other and the environment to lead to the development of macular degeneration. So probably the most accurate result I received was "be proactive".

Prior to engaging in this exercise, I didn't realize how difficult and imprecise it is to calculate risk of disease that includes genetic information. It's interesting that although the science has come a long way towards identifying genetic risk factors of disease, we cannot translate these findings to everyday medicine until we know how these risk factors contribute to the overall risk of disease at a personal level.

Many medical professionals and genetic researchers are against direct-to-consumer genetic testing because of the difficulty with interpretation and potential for unnecessary medical procedures that may result from this information. How is it helpful, they ask, to have a test that should be interpreted as "be proactive"?

In general, we don't have precise probabilities for most things in medicine. Although medical studies result in risks that would give probabilities like 38.1%, the studies themselves are often in specific populations and under highly regulated conditions that don't easily apply to the general population. Many diagnoses are based on somewhat arbitrary cut points on a scale (for example high blood pressure, where there is a distinct range for normal blood pressure, a distinct range for high blood pressure, and a large grey area in between), or on a somewhat arbitrary number of related symptoms (for example dementia where a certain amount of forgetfulness is in the range of normal, and severe confusion under the right circumstances is clearly dementia, but again there is a large grey zone). Ideally, however, medical tests should help inform management of a patient. Does "be proactive" help us know who to screen more often? It may not.

While I was debating the utility of genetic testing, Navigenics and deCODEme closed, and the FDA shut down the 23andMe medical reports deeming that the report was acting as an unapproved medical device. Pathway Genomics still offers testing, although it is conducted through your physician (and has always been conducted this way; I served as my own physician). This regulation feels paternalistic to me, but it is a way of protecting consumers from over-interpreting the risk estimates in the 23andMe health reports.

The best word I have for describing my journey into direct-to-consumer testing is "confusing". I am confused about the risk interpretations; I am confused about the relevance of this for my health; and I am confused about whether limiting direct access to genetic testing is overly paternalistic. In the end, I just stopped taking my eye vitamins.