PROJECT MUSE®

The Impact of My Genetic Testing on My Father

Anonymous One

Narrative Inquiry in Bioethics, Volume 5, Number 3, Winter 2015, pp. 224-226 (Article)

Published by Johns Hopkins University Press

DOI: <https://doi.org/10.1353/nib.2015.0090>

➔ *For additional information about this article*

<https://muse.jhu.edu/article/701639>

very first words of Gottlob Frege's *Begriffsschrift*, "[i]n apprehending scientific truth[s]" the philosopher writes, "we pass, as a rule, through various degrees of certainty." While Frege speaks here of scientific truths, his insights are no less relevant to the process of apprehending information about personal health, disease and genetic risk. If there ever were a *Begriffsschrift* of Genetic Testing, perhaps its first words would read, "in apprehending our genes, we pass, as a rule, through various degrees and dimensions of certainty and uncertainty."

Two weeks later, an envelope containing the results arrived in my mailbox. Frightened by its heft, I opened it with haste.

While relieved to read that I was negative for the mutations tested, I soon began to question whether this sense of relief was in some sense undue or misplaced. My visceral response to the situation was akin to someone finding out about, say, biopsy results of a suspicious mass. But was this the right reactive attitude? Indeed, I had been living with this genome my whole life, and in just a few weeks, I called it into question, rendering it the subject of relentless questioning, and in some sense the axis around which much of my philosophical thinking turned.

As days, weeks, and months passed, and I continued to process and assimilate these experiences, I began reflecting on the ever-expanding role of genetics within our communities, especially as methods of testing grow increasingly efficient, accurate and affordable. I considered how undergoing genetic screening myself might inform and enrich my understanding of patient experiences as I transition from my role of medical student to practitioner.

These experiences, perhaps above all, impressed upon me the centrality of managing and mitigating uncertainty in patients' experiences. Uncertainty is a kind of great modulator, capable of shaping and reshaping bare earthly events and facts across a powerful assemblage of phenomenological frames. While the therapeutic and diagnostic yield of "getting into our genes" is tremendous, its impact on lived experiences of health and disease, on that essential dialectic of *das heimlich* und *das unheimliche*—the essentially familiar and

unfamiliar—remains underexplored. With some glimpse into this peculiar life-world, my sight was now cast on how to improve this process for others in the future.

Indeed, in our era of ever-expanding genomic knowledge, perhaps even more guidance will be needed on how to get out of our genes than on how to get into them.

The Impact of My Genetic Testing on My Father

Anonymous One

As a clinician and genetics researcher, I was excited to get my personalized genotyping and report from 23andMe. Silly as it seems, I didn't think twice about how these results would impact my family.

Prior to testing, I was concerned that I might have one of the genetic variants that cause breast cancer, because prostate cancer runs in my family and there are some genetic variants that contribute to both prostate cancer and breast cancer. Therefore, I was relieved to see that I am not a carrier of a high risk variant for breast or prostate cancer.

Surprisingly, I found out that I have an *APOE4* variant, one of the strongest genetic risk factors for Alzheimer's disease. This was particularly surprising because no one in my family has died of or with dementia. In addition, my paternal grandmother is 99 years old without dementia, and my maternal grandmother is 100 and only developed cognitive impairment from a stroke at age 95.

Outside of testing for specific genetic variants that cause relatively rare diseases or testing of tumor tissue to help guide cancer treatment, my "expert" opinion is that genetic testing for health risk is almost purely an intellectual exercise. Results can be used for medical care in a similar way to knowing family history, except these generalized genetic results are much less informative than even the most

cursory family histories. Specifically, knowing that my patient has a father with hypertension and had a heart attack at age 60 is more clinically useful than knowing that my patient had a 23andMe test that reported “elevated risk” of heart attack.

The relationship between Alzheimer’s Disease and *APOE4* is an exception to my opinion. Alzheimer’s Disease is not a rare disease, and *APOE4* increases your risk for Alzheimer’s Disease, but it is not deterministic. Strong medical research shows that, if you have a family history of Alzheimer’s Disease, you have a much higher risk for Alzheimer’s Disease, if you have the *APOE4* variant than if you don’t. In addition, there is evidence that *APOE4* increases your risk for Alzheimer’s Disease even without a family history. Basically, knowing that my patient has the *APOE4* risk allele is a clinically useful piece of information whether or not there is a family history of Alzheimer’s Disease.

Currently, we do not have treatments for Alzheimer’s Disease, although rapid advances are being made in understanding the biology behind Alzheimer’s Disease and I am confident that there will be effective treatments or preventions for Alzheimer’s Disease within the next 10 to 20 years. From my perspective, even though there is no treatment or clear prevention, knowing that I am at high risk for Alzheimer’s Disease is helpful for my own future planning.

This also significantly impacts my family. If I carry the *APOE4* variant, at least one of my parents does as well. Within an hour of receiving the results, I called my parents, who are in their late 60’s. Despite my medical experience and training in genetics, I was unprepared for the consequences of this phone call.

My father is a statistician in all senses of the word. He has a doctorate in statistics, and his entire career included statistical analyses of large databases. More unusual, he quantifies and analyzes most aspects of his life including (but not limited to) precise sleep hours, heart rate during exercise, dietary fat and cholesterol (starting in the 1970’s, well before it became popular). Therefore, he went through my complete family tree and determined that, although there has been no Alzheimer’s

Disease or dementia that we know of, because his relatives have died at younger ages than my mother’s relatives, the *APOE4* variant must come from him. He does not even speak of it as, “there’s a good chance that I carry the variant”, he states that definitively he carries the variant. At one point, I suggested that he get confirmatory testing, but he deemed that unnecessary because it is certain that he has the variant.

He changed his life immediately—he retired from his job, moved, and sought out investigational treatment for Alzheimer’s Disease. My decision to get genetic testing and communicate the results is responsible for these life changes.

Now, three years later, he shows no signs of Alzheimer’s, and he seems less driven by concern for his imminent cognitive decline. However, I cannot overstate the significance of my genetic testing on almost every decision he has made since I was tested.

My own feelings are highly conflicted. First and foremost, I am the cause this concern. But I feel strongly that it is not my fault. Although I had genetic testing, I am not responsible for my own results and I did the right thing by communicating these results to my parents. If he does have the variant (which is probable but not definitive), he made reasonable decisions to focus his life on moving forward with projects that are meaningful to him, and physically moving to be close to family in retirement. What is upsetting to me is that this experience has made him blind to the probabilities he so thoroughly understands: he does not acknowledge that he *may* not have the variant, and he does not acknowledge that, even if he does have the variant, he *may* not develop Alzheimer’s. I wonder how things would be different if I never had been tested. Would he have found something else health-related about which to be irrational?

Several years ago, the FDA effectively shut down the health-related component of 23andMe, stating that the health-related information given was acting as an unapproved medical device. Although you can pay \$100 to receive an extensive genotyping, the interpretation of these data is limited to a comprehensive ancestry report.

I firmly believe that we should be increasing access to genetic and other medical information rather than decreasing access, despite the fact that the medical implications most of the information is unclear. However, it is clear to me that my father's path, and by extension my family's path, has been dramatically altered by my decision to get myself tested.

A Sister, a Father and a Son: Autism, Genetic Testing, and Impossible Decisions

Anonymous Two

When I was in college, I had a friend with a brother that had fragile X syndrome. My older sister has autism, and didn't speak until she was 5 years old. We had long conversations about what it was like to grow up as the "typical" or "normal" sibling. We were each fiercely protective of our siblings, but we also knew what it had cost our families, particularly our mothers, to raise, protect, and advocate for them. Would we want to bear that burden? Even though we valued our siblings, and felt they enriched our lives, and taught other people a great deal, we worried about their futures once our parents were gone. We expected to serve as advocates for them for our entire lives.

But we also wondered, if we had children of our own, could we avoid the suffering that some aspects of their conditions caused? If they carried fragile X or autism, the challenges that they might face in education, employment, and independent living were very concrete for us. We had watched our siblings face their own abilities and limitations, and even more the inadequacies of our systems of health care, education and social welfare in supporting or failing those with disabilities.

Maya didn't know if she carried the fragile X gene—since her brother had it she had a 50 percent chance of carrying the gene on one of her X-chromosomes. And if she was a carrier, she had a 50 percent chance of passing it on with each pregnancy—if she

passed it to a girl, she would be a carrier; if a boy, he would have the condition. If she could not be tested, she said she wouldn't have children at all. We talked about whether she should be tested, but were worried about whether, if she were known to carry the gene, it would affect her own eligibility for insurance in the future.

Our conversations predated The Genetic Information Nondiscrimination Act (GINA) of 2008 and the Affordable Care Act (ACA) of 2010, which outlawed pre-existing condition exclusions, by more than a decade. But even now, it only protects against discrimination in health insurance and employment, not life, disability, and long-term care insurance.

We have the technology for preimplantation genetic diagnosis, so even if she carries the gene, Maya could have in vitro fertilization with testing to select an embryo without fragile X—if she has insurance that covers it, or in the ballpark of \$25,000 to pay out of pocket for each attempt.

I struggled with the same questions, but unlike Maya, I could not be tested—we still don't have a clear sense of what causes autism. So for me, having children would have to be an act of faith. Either I would be lucky and my children would not have autism, or I would do my best, as my parents had. The best data I could find suggested that as a sibling, I had somewhere between a 2 and 10 percent chance of having a child with severe autism spectrum disorder, but closer to a 50 percent chance of having a child with some autistic traits, even if he or she did not have the full syndrome.

After I finished school, and was secure in my first job, I became pregnant.

At seven weeks gestation, I met with obstetrics and had 26 tubes of blood sent for a dizzying array of tests. As part of that baseline testing for my pregnancy, I had screening tests for cystic fibrosis, spinal muscular atrophy, and fragile X (the only test directly relevant to the family history of autism), and a hemoglobin electrophoresis to look for sickle and other hemoglobinopathies.

Because my age, 40 at the time, placed my child at increased risk for chromosomal trisomies such as the one that causes Down's Syndrome, I was