

Genetic Testing Isn't What It Has Been Sold As



By *Jamie Wells, M.D.* — June 11, 2018



Credit: Steven Vacher, Flickr [1]

The use and concept of terms like “genetic testing” or “genome sequencing” as the key to future disease prevention is speculative at best. Besides the generic recommendations of healthy living through improved diet and routine exercise, there isn't much beyond entertainment value that the commercial products can offer along with an often newly found anxiety. Despite this reality, direct-to-consumer platforms and now health systems (e.g. [Geisinger Health System](#) [2], a health system and health insurance company) continue their marketing campaigns. Attempting to distinguish itself from the former which casts a wide net, [Geisinger promises](#) [2] to test in a targeted fashion with reporting of only actionable data inclusive of primary care involvement.

But, no one is addressing the downside and ethical complexities such mass, aimless screening causes. And even when there is fully informed consent in those already diagnosed with disease and being treated by a doctor, the process of genetic testing often raises more questions than answers. This will be discussed shortly.

The fallacy

It is no surprise that controlling your future by stopping the development of medical conditions draws a captive audience of willing participants, investors, and media attention. But, it is based on a fallacy.

The rhetoric describing DNA sequencing misleads what the technology is capable of today, and likely even in the near future. Few diseases have a single cause or purely genomic basis. At present, this technology is an unfocused screening tool creating angst and uncertainty regarding relative risks that may never come to fruition – patient's emotional energy is focused on probabilities. Screening of the general population, rather than select groups results in unnecessary

confirmatory testing, over diagnosis, and interventions due to the knee-jerk human reflex to do something instead of nothing.

Satisfying intellectual curiosity as an exercise rather than out of clinical concern can beget unnecessary actions, some that can do harm. Consider prenatal testing that provides probabilities, often false, that lead to irreversible interventions, even terminations. Of course, there is utility in actionable prevention when risk factors are known, but seeking the right testing is essential to promulgating useful data. To date, valuable precision medicine involves targeted therapies based on a patient's tumor tissue analysis and its genetic variants (for diagnoses that are already known). For otherwise healthy individuals, is knowing you might be at risk of heart disease or cancer and should, therefore, eat nutritiously and exercise progressing care? Only if you consider these recommendations a new revelation.

The wisdom that comes from perspective is priceless.

There is no better discussion of such testing and the concomitant personal ramifications it imposes than in a recent perspective piece for [The New England Journal of Medicine](#) [3] by Dr. Shekinah N.C. Elmore describing her own experience. She represents a case of a purposefully selected patient responsibly tested under a physician's care. Having survived a childhood cancer (rhabdomyosarcoma) and while deciding on treatment for dual breast and lung cancer diagnoses, [she underwent limited testing](#) [3] for a *p53* mutation known to be a culprit in those enduring multiple cancers.

Why is this particular specific test considered? In those with known *p53* mutations, a doctor will likely be more aggressive in cancer screenings of a variety of organ systems on a routine basis and the knowledge will guide him to consider seriously whether a radiation-associated cancer is worth the risk given the patient's predisposition. So, this awareness can inform therapies. But, in a young adult who has had three cancers it is highly likely anyway that a physician will take more precautionary measures with or without *p53* mutation identification. For the purposes of research and potential gene therapy discovery hopefully down the line, this understanding can ultimately be meaningful. But, trained health professionals are vital to shared patient decision-making as they help tease out the many questions that arise.

In her own words, Dr. Elmore eloquently [writes](#) [3]

“The modifications made to my medical care because of my mutation are surprisingly limited...The changes to my outlook, my psyche, have been much more profound. It's impossible to describe the unique panic that comes with imagining that any of your cells could decide to rebel at any moment... And then there are more practical questions: Should I wear sunscreen every day, or is it better just to stay inside?... A mutation like mine threatens to consume your whole imagination, especially with regard to the future... You start making crazy calculations, guessing the likelihood of being around for this or that event.”

She fittingly [articulates](#) ^[3] the spectrum of influence such information provides and underscores the damage when arbitrarily measured such testing imposes:

“So I wonder: Will knowledge about our personal genomes deliver us, or be our undoing? My knowledge has both empowered and broken me -- I don’t know which it’s done more. Flying between fatalism and denial, I eventually decided that I had to live, normally...But I doubt that knowing would be best for everyone.”

[Dr. Elmore believes](#) ^[3] the news is powerful if there can be action taken beyond the result. She speaks to her essential support system (e.g. oncologist, social worker, psychiatrist) in facilitating her coping with such uncertainty. In closing, when the choice is “doing less and doing more” she is “apt to wholly embrace the former” desiring “as many of these days as possible to be untethered” to medical procedures and studies.

In summary

Manufacturing worry for intellectual edification is a misguided approach. Being fully informed by health professionals who have a complete, comprehensive understanding of the risks and benefits for you, given your entire clinical picture and family history, is a more ideal path - however imperfect. Getting comfortable with ambiguity, in general, would serve us all well. Here’s hoping this notion reverses the current opposing cultural trend. When it comes to genetic testing, appreciating that the stakes are entirely different for those with a diagnosis as opposed to those without is an important, promising start.

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[2] <https://www.acsh.org/news/2018/05/25/precision-medicine-overselling-routine-dna-sequencing-primary-care-13014>

[3] <https://www.nejm.org/doi/full/10.1056/NEJMp1803542>