

Genetic testing

“Hey Doc, what are you going to do to help me? I’m at risk for obesity, gout, diabetes, gallstones, heart disease, and kidney disease.”

“Bob, despite the fact that obesity is already more of a fact than a risk for you, what are you talking about?”

“I did one of those home DNA tests, and my printout said I’m at higher risk, so I’m worried.”

“And you weren’t worried when I explained last month that 300 lbs on your 5’6” frame increased your chances of heart disease and other adverse conditions?”

We live in interesting times. Patients now have access to home genetic testing, and for less than \$200 these at-home kits allow individuals to research their ancestry. In addition, these companies provide genetic reports on disease predisposition and carrier states. One could argue that this is a wonderful technological advance because patients can now take control of their lives and mitigate risks. However, this information is being given without genetic counseling to people who might not be ready to hear it or understand what information they are actually getting. A genetic mutation doesn’t actually mean you will get the disease; nor does it explain the other complex factors that go into disease risk.

Probably the most well-known genetic test is for mutation of the BCRA1 and BCRA2 genes, which if positive significantly increase a woman’s risk for breast and ovarian cancer. But how can a patient make an informed decision about future treatments to mitigate risk without an expert guiding them through this maze of gene expression and penetrance? Also, these tests check for some mutations but not all, and if negative

might convey a false sense of security and lead individuals to stop potentially lifesaving screening.

I did some online research, and it appears that not all companies have the same level of credibility. Some make unsubstantiated claims that seem a little farfetched. One compa-

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ny will test for 31 conditions, 53 carrier states, 12 drug-response genes, 6 wellness traits, 11 other traits, and 11 addictions. Your genetics will even be matched to dietary and exercise advice along with supplements and skin products that the company conveniently sells. Some of the listed traits did make me smile, particularly ear-wax type (who cares), digit ratio (can’t you just look at them?), and newborn hair amount (didn’t anyone take baby photos?). The drug-response item also caused me to pause. Apparently, your genes reveal how you will metabolize proton pump inhibitors, respond to hepatitis C treatment, and how sensitive you will be to warfarin and clopidogrel (I admit this would be valuable information to have). As for addiction testing, if the result is negative, should an individual use alcohol, cocaine, and heroin without a care in the world?

Lastly, the wellness tests listed include alcohol flush, caffeine metabolism, depth of sleep, lactose intolerance, and muscle performance. If you are predisposed to being a black-coffee-swilling, red-faced alcoholic-insomniac with weak muscles, should you just give up?

As technology continues to advance, physicians will definitely face new challenges in processing the wealth of ever-expanding testing and knowledge. I believe my DNA is heavily sprinkled with the stalling-while-I-Google gene, so hopefully I will be okay. Good luck to the rest of you.

—DRR

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Who am I to judge?

One of my kids has picked up a hobby for which he sometimes appears on stage, in film, and on TV. Believe me, it is not something we even knew anything about until our family was suddenly in it, and it is both an interesting and occasionally ridiculous pastime for all of us. One of his brothers is a competitive diver, who has trained thousands of hours to get to a place where he can rip dives at provincial and national competitions, and he may even be presented with scholarship opportunities at some point.

They and we are very happy to have these opportunities, and hopefully enough talent to feel part of the game, but there is also a difficult side to these activities—subjective judgment alone determines how successful they are. Not a clock or a goal or a measuring stick—a human ultimately decides who deserves what. No matter how hard you work and how well you perform, there is no guarantee that everyone will see the performance the same way. And subjectivity is prone to create biased or single-point-of-view judgments, critiques that are

narrow and misdirected, and maybe even wrong sometimes!

Professionally, before we even recognize it, we can find ourselves falling onto the path of needing to be uncomfortably judgmental. For instance, when acting as part of the

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residency selection committee in our division for several years, having also helped choose which fellows we train, comparing and ranking submissions to academic meetings and publications to determine which ones will be accepted, winnowing out who we might hire as colleagues; we judge and judge and judge. And I'm often


left thinking that these people have done amazing work, even the ones we don't end up choosing, and because the majority of them are usually way better than me on paper, I am left thinking, who the heck am I to judge?

And that's just at work. The world has become so critical and often unapologetically mean, it's hard to even read what is published sometimes. Anonymity means that online critique, tweets, and blogs can be simply brutal. For example, your doctor rating can be submarined by a solitary unhappy patient, and there is no burden of truth or even discussion allowed. A restaurant can be maligned with a review that never has to be proven. Children affected by gun violence are dragged over the coals publicly for daring to try to create legal change. The same film or play your kid is in can be described as both "must see" and "the worst of the year" in sequential tweets, with the more unsavory one usually appearing higher in the search engine algorithm. Your daughter may find herself being scored in the less-happy column of an uninvited, immature "hot or not" site. Blogger after blogger creates and promotes self-made critique sites, and tries to one-up their competition with edginess. And who the heck are these people to judge?

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When we absolutely do have to make a call or criticism, we should be reminded that we should keep things real and fair and transparent. "Is it true? Is it necessary? Is it kind?" No one really knows who first said that about how we speak our mind, but it would sure be a nicer world if people took it to heart.

—CV



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