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The gap is widening on genetic testing, too

By Ricki Lewis

Yesterday, Ricki examined the [growing gap between science and the public on stem cells](#). Today, she turns her attention to genetic testing.

Marketing tests based on whole genome association (WGA) studies were perhaps inevitable, for the approach makes theoretical sense. Scan genomes of people who share a particular disorder for single nucleotide polymorphisms (SNPs, or single base differences) that they have but unaffected controls do not. Then translate the population odds ratios of the telltale DNA patterns into individual disease risk statements, stressing such words as “variant” and “wellness” and censoring use of “disease.”

Consumers submit DNA-bearing saliva, and algorithms based on published WGA studies spit back risk stats. The tests are exempt from FDA scrutiny because they aren't medical, according to the language. Load up the website with an intro genetics course, assemble an advisory board, and the FTC is satisfied, too.

But some people think it is too soon to hawk these WGA-based tests. The truth is, and the direct-to-consumer company websites actually say so in the fine print, we just don't know yet whether an association seen in a population can provide meaningful information for an individual. The many calculations of WGA studies produce some false positives, and associations have been known to dissipate as data accumulates. Consumers may not be aware of these limitations, nor realize that “link,” “marker,” and “association,” have precise scientific meanings. It is all too easy to see slick company websites and assume that tests based on results of association studies have been validated. They have not.

Several companies now offer services based on these WGA studies, not to be confused with the regulated tests for well-studied genes, such as BRCA1 or the cystic fibrosis gene. We geneticists have mixed feelings about the new generation of tests.

When Navigenics unveiled its “Health Compass” at a special session at the [American Society of Human Genetics](#) annual meeting last fall, many in the packed room were so disturbed that even grad students ignored the free food.

As soon as deCODE Genetics announced a similar service, I sold my stock. Yet some very notable notables have leant their names to the advisory boards of these companies. Perhaps they figure the proverbial cat is already out of the bag, so why not help provide the most accurate information?

When I caught a segment of the Today Show that featured yet a third company, 23andMe, I decided to investigate further.

The founders of, and the website for, 23andMe were very impressive. The company seems to be making the best of technology that is, I believe, not quite ready for prime time. Caveats and qualifiers abound. They use association studies published in the best journals, with the largest samples and most convincing statistics. Descriptions are clear, privacy protection paramount.

But, again, a gap glares for those in the know. Genetic counseling is mentioned only deep down in the “help” list on the 23andMe website. People receiving medical news from a doctor often fall into a deer-in-the-headlights trance. Won't reading about one's DNA on a website, without benefit of face-to-face professional interpretation, lead to at least some misunderstanding? Navigenics does offer in-house 24/7 genetic counseling, but each web page has the disclaimer “Navigenics does not provide medical advice, diagnosis or treatment.” Genetic counselors are medical professionals. It's confusing.

Like a mantra, the 23andMe website repeatedly states that their services are for “research and education use only,” are not licensed by any health care agency, and that the risk estimates have not been validated in the clinic. Yet the first image that pops up is a “Personal Genome Service” that promises to “unlock the secrets of your own DNA. Today.” Similarly, Navigenics' Health Compass reveals “what your genes have to say about the future of your health,” but also runs a disclaimer that they do “not provide medical advice, diagnosis or treatment.” Can they really have it both ways?

What, exactly, do these companies test for? The Today Show reporter learned his genetic associations to eye color, ability to taste bitter substances, and the consistency of his earwax, traits that could be more economically detected by looking in a mirror, eating broccoli, and sticking a finger in an ear. But that's not all these companies offer. Instead of affirming that I have hazel eyes, love broccoli, and have thin earwax, I might learn that I have an elevated risk of developing multiple sclerosis, heart disease, colorectal cancer, and diabetes. At Navigenics I'd learn my risk of developing obesity, lupus, and Alzheimer disease, and at deCODE Genetics, their under-\$1,000 deCODEme “introduction to your genome” panel estimates risks of developing atrial defibrillation, psoriasis, and age-related macular degeneration. Oddly, all three companies analyze restless legs syndrome – now who wouldn't want a heads-up on that?

What's a consumer to do with all this information? It beats me. But I'm not alone.

The same journals that publish WGA studies are beginning to also publish challenges to their uses. In the March 2008 issue of the **American Journal of Human Genetics**, for example, a team led by A. Cecile J. W. Janssens from Erasmus MC University Medical Center in the Netherlands did a meta analysis of meta analyses to determine that extrapolating from genomic profiles to assess health risks is premature. Kenneth Offit, of Memorial Sloan-Kettering Cancer Center, in the March 19 Journal of the American Medical Association contrasted the unregulated direct-to-consumer genome companies with the decade of prospective clinical trials that validated genetic testing for cancer predisposition.

To end on an up note, perhaps the information that direct-to-consumer companies provide may inspire and empower health care consumers to alter their behaviors in ways that counter what their genes may set into motion. But that, too, needs to be validated. And so, like the stem cell saga, a gap between scientific progress and health consumer expectation gapes. It isn't ethical to market DNA tests based on whole genome population-based studies without randomized, controlled clinical trials, replication, and validation.

Whether considering stem cells or DNA tests, that's simply the way that good medical science is done.