Personal Genomics – Why 23&Me Doesn't Work

by **Bradley Miller** on **November 5, 2009**
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… It's certainly an important idea – scanning our individual genetic make-up to discern health risks and prevent them. Who wouldn't want to understand what preventable diseases they're prone to? I certainly would (well, to an extent)….

For the uninitiated, 23&Me is a personalized genomics company that will take a couple drops of your saliva, extract your DNA and screen it for hundreds if not thousands of genetic disease markers. The company name is derived from the 23 chromosomes humans contain – 23 from mom and 23 matching from dad.

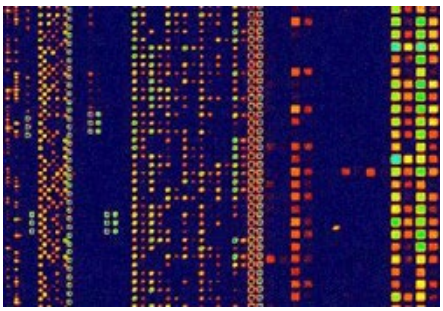
A gene chip by the company Affymetrix. That little square on the chip can yield information on 500,000 different genetic variations.

But the company over promises and under delivers. At the end of the day, the fact is that biomedical science isn't advanced enough yet for us to make meaningful predictions off of the information screened by 23&Me.

Let's dive right to the core of the issue – the biomedical science behind 23&Me. 23&Me use "gene chip" technology , which can screen thousands of genes at once and tell you where you have variation (mutations) that are known to be *correlated* to disease. In other words, if the gene chip picks up that you have a variation in a gene that has been correlated to a heart illness, 23&Me argues that you have a higher chance of developing heart disease. While that certainly seems logical – "I have a gene that shows a higher risk of heart disease, I better do something about it" – it remains somewhat misleading.

We have to dive a little deeper here to understand why the findings from these tests don't correlate to *real* disease risk. When researchers do genetic studies (the type of studies 23&Me base their tests on), most of the time they find correlations between gene variations and a disease. And I want to stress – these are correlations – and are not purely 100% causative like the genetic testing companies would like you to believe.

Put another way, these genes are found in these diseases, however they are not the *root cause* of the disease. Most diseases are due to multiple genetic mutations, which means the underlying causes for these diseases are much more complicated than just one genetic mutation.



A closer look at that square on the gene chip - this is what the chip looks like under magnification when it's read by a computer. The different colors indicate different gene results.

We now know of 20 genes that correlate to height, but they only explain 3% of variation in height between people. Only 3% of the difference in height between these two men! What about the other 97% of difference? More research!

For example, a study came out in the *New England Journal of Medicine* that detailed just how little we know about how our genes and how they become translated in to real world physical traits and disease. This review study illustrated that we know of 20 genes that correlate to the differences in height between people. While that sounds impressive, turns out that those 20 variations explain only about 3% of the true variations in height. I'm 6'5″ – those 20 genes explain only about **1/3 of an inch** of the variation in height between my 5'6″ wife and I! 20 genes!! Why then do we then think that 1 gene will detail risk for heart disease or cancer? The bottom line is that before we can accurately correlate and make meaningful disease predictions based on genomics, much much more research needs to be done.

So, let's come back up to the surface. I've detailed why genetic research to date isn't as complete as these companies would have you believe. The personal genetic variations they uncover, while using the most advanced technology and knowledge we have, isn't sufficient to fully explain disease risks.

The companies are selling a service based on scientific misconceptions – people are accepting 23&Me's marketing, rather than good science. And that lack of scientific and clinical substance is why the medical community hasn't embraced these tests.

I'm going to get in trouble with these companies because they don't directly make these claims – but I think it's implied based on their marketing and how they discuss their product. Doing these tests even just for curiosity's sake is even a waste of money – they don't truly tell you anything useful.

Don't get me wrong, these types of products and services are the future of medicine. Maybe not in this direct way, but we will be screening people for disease risk. No, not for insurance reasons, but rather to attempt to prevent diseases before they take hold. It's just too early for this type of genomic analysis to be accurate enough to truly act upon. Although I'd like to be an optimist, the technology isn't there, medical practice hasn't accepted these tests (and they shouldn't). All of this to say, as a consumer patient hold on to your money for now, the scientific community has a long way to go before we really have the information necessary to make strong clinical correlations and to make valid disease predictions.

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