

On-Line Personal Genetic Testing: Recreational Genetics



“Medicine’s ability to prognosticate always exceeds its ability to intervene—and genetics only widens this gap”

Were you aware that if you order a genetic test from certain on-line commercial genetic testing companies, you can find out your probability of having blue, versus brown eyes? Is it too obvious to point out that it is quicker and cheaper to look in the mirror? Did you know that you can “share” your DNA sequence and SNP (single nucleotide polymorphisms) genotype with others on-line, and even get matched up for on-line dating based on your genotype? There are a number of other test results provided by on-line “personal genomics” that will also provide you with information you may already know: the consistency of your ear wax, for example, and whether you are overweight. However, for this \$750 to \$2500 (cash or credit card accepted), you can also gain information on your risk for things like prostate and colon cancer, heart disease or Alzheimer’s diseasewill people make lifestyle changes to the extent that their risks can be altered? Who knows, in many diseases, what that would take?

There has been a large number of genome-wide association studies published in the past decade. These commercial companies wade through and compile the published data and produce computer generated risks based on that data. However, it is currently known that the predispositional risk associated with many of the genes and alleles examined in these reports result in an odds ratio between 1 and 2²—and we do not really understand the inter-play between genes or between the genes and environment, in determination of an individual’s risk. Additionally, it has been reported that there is a high incidence of false positive results stemming from the study designs used in these genome-wide association studies, and that some risks will not be validated.³ So, we have medical information based on weak validation and poorly characterized genetic variants being provided to consumers. Never mind the fact that many of the studies have not taken into account ethnicity of the study population, something that cannot be extrapolated to the average on-line U.S. consumer ordering these tests.

This is truly a situation of “buyer-beware”; even though these companies have been under scrutiny by the FDA for several years, they are still operating and taking people’s money. There is an “uncritical enthusiasm”¹ driving this use of new genotyping and sequence technology, while the technology does, truly, hold promise to improve health and medicine. However, at the current time, there is no good published data supporting use of these test results for patient care—for anything other than expensive genetic recreation.

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References

1. Evans, J.P. Genetics in Medicine 10:709, '08.
2. Hunter, D.J., et al. NEJM 358:105, '08.
3. Pearson, T.A. and Manolio, T.A. JAMA 299:1335, '08.

At Home Genetic Tests: A Healthy Dose of Skepticism
<http://www.ftc.gov/bcp/edu/pubs/consumer/health/hea02.pdf>