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The New England Journal of Medicine Gives Direct-to-Consumer Genome Scans Thumbs Down

by Dr. Hsien-Hsien Lei
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The New England Journal of Medicine (NEJM) is telling doctors what they should advise their patients when it comes to genome scans. And the advice is basically - **Don't Do It!**

Here are the concerns about genome scans as raised by the NEJM article (much of them overlap with those raised by the American Society of Human Genetics):

Analytic Validity - Quality control and oversight are unclear. Genetic tests may be highly accurate and reliable but even low error rates may mean large numbers of mistaken genotypes because each genome scan involves half a million to a million SNPs.

2. **Clinical Validity** - Sensitivity, specificity, and positive and negative predictive value for each test are unclear. Complex diseases, such as diabetes and heart disease, are caused by more than one gene variant as well as gene and environment interactions.

3. **Clinical Utility** - The desired effect on a patient's clinical care is unclear especially when most health interventions would apply even without genetic information, e.g. smoking cessation and weight loss. Also, co-author Dr. Muin Khoury says in the podcast below that family history is still more important clinically than results from genome scans.

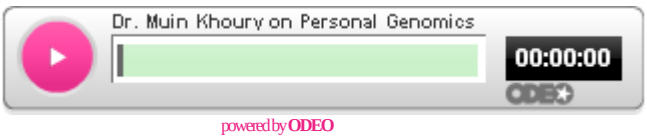
The authors conclude that genome scans have limited utility.

> **So what advice should a physician offer patients?** For the patient who appears with a genome map and printouts of risk estimates in hand, a general statement about the poor sensitivity and positive predictive value of such results is appropriate, but a detailed consumer report may be beyond most physicians' skill sets. For the patient asking whether these services provide information that is useful for disease avoidance, **the prudent answer is "Not now — ask again in a few years."** More information is needed on the clinical utility of this information in the light of existing disease-specific opportunities for prevention or early detection and the potential value that genomic profiles can add to that of

> simpler tools, such as the family health history. Finally, given the risk of commercial exploitation, if patients are determined to proceed, perhaps because they are simply curious, are genetic hobbyists, or are "early adopters" of new technology, it would make sense to **encourage them to enroll in formal scientific studies**. [emphasis added]

The idea of enrolling in formal scientific studies may be more prudent or altruistic but the reality is that when a customer pays for a genome scan, they have the right to keep their data private for their own use. Also, I doubt that scientific studies will come with the bells and whistles promised by the next-gen personal genome companies, including family tree building, social networking, and genome comparisons. For those interested in contributing to science and having their genomes scanned and/or sequenced, I would recommend checking out the the Personal Genome Project.

Co-author Dr. David Hunter gives more reasons for "why not to buy a scan of your genome" in US News & World Report. Below is the NEJM podcast interview of Dr. Muin Khoury.



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