

The Use of DTC genetic testing for common disease risk assessment: update and current status
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There has been new activity in the DTC area recently, with the announcement by Walgreens Drug store chain that they were introducing an 'off the shelf' genetic testing kit, which the public could buy, take home, collect a specimen, and mail to a testing laboratory for a genetic risk assessment for common diseases including heart disease and diabetes. Several weeks ago, the FDA and CLIA raised concerns about this kit, and in addition, the state of New York has sent a letter to the company providing these kits, reminding them that no kits will be sold within the New York state physical boundary (DTC testing and patient-ordered testing for either diagnosis OR risk assessment is against NY state statute).

The number of internet-based DTC genetic testing companies has decreased in the past several years, due somewhat to the work of CLIA and the FDA, but there are still over 100 companies working and promoting various types of genetic testing. Some of these companies sell testing they describe strictly as 'recreational', i.e. ancestry, testing. Other companies are still promoting disease risk testing, which is the case for the kit being sold in Walgreens.

There has been some interesting data published online recently in the British Medical Journal (BMJ 2010:340:b4838 doi:10.1136/bmj.b4838). This study, which was performed over a period of 10 years, looked at the use of genetic and non-genetic risk factors in predicting who would ultimately develop type II diabetes. The study followed and assessed over 5,000 people in Great Britain, all of whom worked for the same government agency. Of the 5535 initially healthy individuals, 302 developed diabetes over the 10 year period. They compared the use of non-genetic risk variables (age, gender, drug treatment status, family history of type II diabetes, body mass index, HDL, cholesterol, triglyceride levels, and fasting glucose levels) to 20 different single nucleotide polymorphisms (SNPs) that have been associated with risk for this disorder. These SNPs are those published as part of various genome-wide association studies and are likely included in the DTC genetic tests that are offered by various companies.

The study clearly showed that performing genetic SNP analysis provided little additional improvement to risk calculation for type II diabetes, when compared to phenotype-based risk models. The study also showed that the parameters listed above resulted in better prediction of who would ultimately develop type II diabetes. The phenotype-associated type of assessment is what physicians have historically and can currently easily provide to their patient, in concordance with the appropriate laboratory testing. This paper is so important, as it shows very clearly that SNPs that contribute to the common disease of type II diabetes have only small additive effects and that personal lifestyle choices can absolutely influence an individual's risk for that disease. Since looking at someone's genotype for these disease-associated SNPs is a surrogate for having family history information, taking a good family history during a physical exam will be much cheaper for us as a health care system, to do.

You're also invited to read the following article, recently published in the New England Journal of Medicine: <http://content.nejm.org/cgi/content/full/358/2/105>.