The long arm of your chromosomes and the law

A great debate is currently raging in the wider genetics community directly relevant to day-to-day clinical medicine. In the last three years the advent of genome-wide association studies (GWAS) has facilitated the discovery of greater than 180 markers for risk of a growing list of common chronic diseases, including cancers, diabetes, coronary heart disease and Alzheimer's. In the last 6-9 months, a number of companies have moved to make these markers directly available to consumers in the form of genome-wide scans that can be obtained over the internet for about $1000-$2500 dollars – and several are seeking to lower that price point drastically. The companies make efforts to qualify that all test results provided to the consumer are preliminary in nature and that their products represent information, rather than medical advice. However, looking at the sites one could conclude that the companies - implicitly or explicitly - suggest to consumers that they might use the results to improve their health. Currently there is no direct evidence that providing patients with genetic risk information from genome-wide association studies improves health outcomes – though, importantly, this is very likely to change in the next few years. Yet, there are reports – many provided by the testing companies themselves - that patients are bringing their results to health care providers with the expectation that some form of action be taken to mitigate their newly discovered disease risk. However, beyond selected anecdotes, we know little about what providers are doing with the information patients are bringing them.

Though direct to consumer (DTC) testing for traditional genetic conditions (think hereditary breast cancer and ovarian cancer syndrome) has been around for a number of years, the sophistication, scale and potential reach of this new crop of offerings has raised the interest of both state and federal regulatory bodies. Not unexpectedly, these companies have also been subject to intense criticism from both the scientific and medical communities. The central theme of those voicing concerns is that the health care implications of this embryonic realm of genetic testing is unknown at this time – and that potential harms could result from either over-, under- or mis-interpretation of test results.

In the last few months, the intensity of the debate has ratcheted up: the state of California sent cease-and-desist letters to 13 concerns offering DTC genetic services to California residents http://ww2.cdph.ca.gov/HealthInfo/news/Pages/LabTestLandingPg.aspx . The letter stipulated, among other things, that the companies need to offer their tests through Medicare approved labs (CLIA certified) and that a licensed physician needs to be involved in ordering the test. At the federal level, there is ongoing Congressional scrutiny of the topic, evidenced by a June 12, 2008 roundtable held by Senator Gordon Smith of the U.S. Senate Special Committee on Aging http://aging.senate.gov/minority/index.cfm?Fuseaction=Hearings.Detail&HearingID=0f1d28bb-a9fb-403a-a462-8485b60b9d1f . On July 7 and 8, 2008 a committee that advises the U.S. Secretary of the Department of Health and Human Services on issues surrounding genetics/genomics examined this issue in some depth http://www4.od.nih.gov/oba/SACGHS/meetings/2008Jul/SACGHJul2008premeeting.htm . From these proceedings it is clear that there are widely divergent opinions on the topic

of DTC availability of genome-wide scans. Interestingly, this scrutiny has brought an unexpected windfall to those of us in primary care. Individuals from the most technology-driven reaches of medicine are discussing the need for increased research on determinants of health behaviors and a re-evaluation of how our current system values preventive interventions.

The core questions confronting DTC genetic testing are not new to medicine, nor even genetics/genomics: first, when is a new technology ready for clinical use; and second, how much regulation is appropriate to ensure its safe and effective application while fostering innovation and minimizing risk of disparities? One side of this debate argues strongly that consumers should be empowered with every bit of information about their health possible, and that to deny them direct access to their genetic makeup through overly strict regulation is old-fashioned and paternalistic. The other side argues that this type of genome-wide scanning is still a research tool. Consequently, offering it DTC at this point in time in a loosely regulated manner may substantially mislead the public and health care providers, incurring costs both in terms of morbidity and scarce health care resources. Both sides have valid points. The American Medical Association and the American College of Medical Genetics have taken note of the new DTC movement and have developed official positions critical of DTC genetic testing. It is unclear what effect these statements will have on the entities offering this type of testing. What is clear is that much hinges on consumer demand and opinion – and to some extent the ability to shape that demand rests in the hands of health care