## Physicians as guardians of genetic knowledge

The genetic revolution in medicine, envisaged since the completion of the draft sequence of the human genome over a decade ago, is moving forward at a painfully slow pace. Although truly personalised care is, perhaps, still decades away, private companies have been quick to exploit the genetic information that has become available from the Human Genome Project. The past 5 years have seen a proliferation of personal genetic tests, which promise to predict risk for an array of complex conditions such as type 2 diabetes and Parkinson's disease, determine drug or food metabolism, or uncover carrier status for inherited diseases. And although the predictive power of some of these tests is questionable, they have been offered directly to consumers, who send off a swab of saliva or a blood sample (and US\$200-\$2000, depending on the test) in return for their genetic risk profile. Last week saw a new development in the regulation of these tests in the USA. An expert advisory panel of the US Food and Drug Administration (FDA) recommended that direct-to-consumer (DTC) genetic tests be subject to medical supervision; interpreted, and possibly also ordered by doctors, rather than by the lay public. The FDA has so far indicated that it might require physicians' involvement for some, but not all, types of genetic test.

The advice of the advisory panel is wise and will be welcomed by many health professionals concerned about the effect of such tests on consumers, as well as the dubious claims made by some manufacturers. Last year, an undercover investigation by the US Government Accountability Office (GAO) found examples of "deceptive marketing" by companies selling DTC tests, including claims made by four companies that consumers' DNA could be used to create personalised supplements to treat diseases. The clinical validity of some tests has also proven problematic. The GAO's investigation showed that disease risk predictions varied across companies for identical DNA samples. One DNA donor, for example, was told that he had below average, average, and above average risk for prostate cancer and hypertension by different companies. Whether doctors become the guardians of genetic tests or not, tighter federal regulation will be needed to ensure companies that do market products are making clinically valid claims. A registry for genetic tests that is being developed by the US National Institutes of Health should help towards this goal.

There are also question marks over the clinical utility of some DTC tests. For example, can anything useful be done with the information gleaned from disease susceptibility tests? After all, most of the interventions for reducing the risk of complex diseases are the same as those that physicians will recommend to all patients—eq, maintain a healthy weight, eat healthily, exercise, and refrain from drinking excessively and smoking. There is also some evidence to suggest that knowledge of disease risk might not hold much extra sway with patients in terms of leading a healthy lifestyle. A paper in the New England Journal of Medicine published in January showed that there were no short-term measurable changes in diet or exercise or use of screening tests in a selected group who underwent DTC genome-wide profiling. Some have argued, however, that it could take many years to show improved health outcomes for genetic tests intended to assist in the prevention and treatment of chronic diseases. In view of this situation, the American Heart Association has urged the FDA "to allow tests with clear clinical validity to be marketed even if their use has not been shown to result in improved clinical outcomes".

In the future, full genome sequencing might prove a valuable adjunct to clinical care, especially in tailoring drug treatment, as reported in an Article by Euan Ashley and colleagues published in *The Lancet* last year. Some companies already offer near full or full genome sequencing for consumers but at a prohibitively high price for the general public (\$10 000–40 000). However, the costs are expected to come down considerably in the next 5 years to around \$1000.

Are doctors prepared for the increasing use of genetics in clinical care? Some evidence suggests not. A 2009 survey of more than 10000 US physicians by the American Medical Association showed that only 26% had any type of education in the use of genetic testing to guide treatment decisions. And only 10% felt they had the necessary training and knowledge to put pharmacogenetic testing to good use when treating patients. Medical schools and professional organisations will have an important part to play in improving this situation. Doctors will have to become increasingly adept at not only using genetic tests in clinical care but also at explaining their results and, importantly, their limitations to patients. 

The Lancet



See Articles Lancet 2010;

For the **GAO report** see http:// www.gao.gov/new.items/ d10847t.pdf

For the **paper on the effects of DTC genome-wide profiling** see *N Engl J Med* 2011; **364:** 524–34