Researchers call for better regulation of genetic tests

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Better regulation is urgently needed for genetic tests, particularly those marketed directly to the public, argue researchers in this week's BMJ.

In the past 18 months, studies have identified dozens of inherited DNA variations associated with common conditions such as heart attacks, diabetes and asthma, write Professor David Melzer and colleagues.

In many cases, these findings provide insight on the cause of the disease, but clinical applications are still mostly unclear. Much work is now needed to identify and evaluate each potential clinical application. Yet, although the work of translating discovery into evidence based practice is just beginning, several companies have already marketed tests, many directly to the public.

Using tests without proper evaluation, they warn, could trigger erroneous treatment and involve major hazards. For example, direct marketing of the BRCA1 and 2 familial breast cancer tests to women at low risk was criticised for causing unfounded anxiety and unnecessary preventive surgery.

False reassurance from tests for common diseases could also result in effective prevention measures, such as controlling weight and exercising, being ignored. Problems with insurance or implications for other family members may also arise, they add.

So what needs to change to ensure that tests are used appropriately?

The authors call for harmonisation of regulatory standards internationally and for more transparency regarding the clinical evidence base for new tests.

For example, a scientifically independent system for identifying and checking higher risk genetic tests is needed in Europe. Currently, genetic tests in Europe are marketed without pre-market scrutiny by regulators, and companies can keep secret the clinical evidence on their tests. Similarly, few genetic tests developed 'in-house' by laboratories in the USA are currently reviewed by regulators.

Both consumers and professionals should push for a regulatory system that encourages clinical evaluation and makes the results (or lack of them) easily available to all, write the authors. Professional bodies and health care providers should also remind professionals that using tests in routine practice without evidence of utility is incompatible with good clinical practice.

These improvements in the clinical evaluation of tests may prove as important as the discoveries themselves in realising the promise of genomics to improve health, they conclude.

An accompanying editorial supports these views, saying that international collaboration to set standards and methods is urgently needed.

Source: British Medical Journal