

Additional material submitted by Brian Whitley, Managing Director of GeneticHealth Ltd.

Referring to Question Q555 from the Chairman / Lord Patel.

Question matter:

1. In a recent newspaper article an individual was reported to have received different risk profiles of developing a particular common disease when tested by three different companies. This suggests that the information on risk prediction is not currently reliable. How can different companies reach different conclusions about an individual's disease risk profile? What can be done to ensure accuracy and consistency in the interpretation of genetic tests?

I feel that the article referred to was misleading and showed a poor understanding of the underlying science of genetic screening, and in particular, risk assessment. The author was aware that he was not comparing apples with apples as this was specifically explained in relation to heart disease (the main topic he led with and used for his pictorial illustration). He also did not seem broadly inline with "The sense about Science" approach.

[\(http://www.senseaboutscience.org.uk/index.php/site/about/6/\)](http://www.senseaboutscience.org.uk/index.php/site/about/6/)

Heart disease results

23andMe reported him to be at low risk and GeneticHealth reported a low to moderate risk (not moderate risk as printed). Decode looked at a completely different set of genes related to risk of heart attack. The author knew that these tests were not the same, they tested different polymorphisms, and this should have been pointed this out to the reader.

Alzheimer's results

In the case of Alzheimer's disease, GeneticHealth assessed that the risk was a four fold increase in developing Alzheimer's by his late eighties. DeCode placed a percentage on this of 74per cent above average. It is likely that both results are in agreement but the risk is being described in two different ways. It is with certainty that the tests are correct.

The article then finishes with a statement made by GeneWatch that "handing over your genetic information increases the likelihood that insurers will discriminate against you in the future".

This is an enormous and controversial subject in its own right, but I would propose that it would have been more responsible to point out that the Association of British Insurers has extended their moratorium on the use of genetic test results for insurance purposes from 2011 until 2014 and that the Human Genetics Commission have called for comprehensive legal protection against "discrimination on any genetic grounds".

In conclusion on this question;

It is not so much a case of the tests not being accurate. The technology platform that the companies use is very accurate. However the consistency of interpretation of the meaning of the results is where there is a need for more work and collaboration. In common with all medical advances, the field of personal genomics is rapidly evolving, and the clinical validity of new SNPs will have to reflect this.

I would agree that the specific gene polymorphisms that have been tested to assess a specific risk should always be made clear to the client before they take the test. Also I would agree that the clinical basis for any statement about risk should be provided. In our particular case, this is carefully explained in our detailed post test consultation with the client and in the documentation supplied to the client.

Referring to Question Q560 from Lord Taverne
Question matter: IVDD Directive.

Further to my previous comments highlighting our desire for a more cohesive, robust regulatory regime (ref: response to Q574) I would also like to suggest that a traffic light system is developed for genetic screening and each level be covered by increasing regulatory standards and procedures.

In the lowest category “green” would be tests such as ancestry, where no disease prediction or gene defect measurements are made.

In the medium category “amber” would be genetic tests which define such traits as hair colour, eye colour or tests for paternity.

And finally “red” where the test is of clinical significance. For example, cystic fibrosis or predicting disease risk. I would recommend that such tests should only be offered when combined with a consultation from suitably qualified health professionals (as has always been the case with GeneticHealth)

On the frontiers of science the opportunities for innovation always bring some risks, but genetic screening can also bring some major benefits if handled responsibly.

We want a framework in place that adequately informs and protects the client. That in the case of genetic screening for disease risk assessment, there should always a suitable trained doctor involved in a consultation process.

And finally media reporting should be responsibly researched and accurately presented.