

## **Direct-to-Consumer Genetic Testing for Health-Related Purposes in the European Union**

### **Summary of points from a meeting organised by EASAC, FEAM and STOA in the European Parliament 3 December 2012**

Until recently, human genetic testing was mainly confined to specialist medical genetic services, traditionally focussing on the relatively rare inherited disorders. However, the rapid pace of advance in DNA analysis has led to increasing interest in the development of genetic tests for determining susceptibility to the more common, complex disorders. Such tests are increasingly being offered by companies through the internet but provision of these services raises scientific, regulatory and ethical questions.

A recent report<sup>1</sup> published by the academies of science (European Academies Science Advisory Council, EASAC) and medicine (Federation of European Academies of Medicine, FEAM) reviewed the evidence and ascertained the principles that should underpin the regulatory options for managing Direct-to-Consumer Genetic Testing (DTC GT). A workshop in the European Parliament on 3 December 2012, organised by EASAC and FEAM together with Science and Technology Options Assessment (STOA), provided the opportunity for further discussion of emerging issues, some controversial, and for comparison of the current situation with that described in the STOA report in 2008<sup>2</sup>.

In presenting the main conclusions from the EASAC-FEAM report, Martina Cornel (VU University Medical Center, Amsterdam) referred to the growing realisation that the human molecular system is more complex than had been anticipated a decade previously when the draft of the human genome sequence was first published. In consequence, the impact of genomics on public health services has, so far, been relatively modest and the potential advantages of DTC GT services were contentious. The EASAC-FEAM report concluded that DTC GT had little clinical value at present and expressed especial concern in several specific respects, most notably with reference to testing for high penetrance serious disorders, prenatal screening, nutrigenomic and pharmacogenetic testing. In developing broad principles for the management of DTC GT, it was emphasised that claims must be regulated on the basis of the scientific validity of the link between genetic marker and disease. Other key criteria for assessing test provision include quality assurance, to cover both testing procedures and the

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<sup>1</sup> Available from <http://www.easac.eu> and <http://www.feam.eu.com>

<sup>2</sup> Available from [http://www.europarl.europa.eu/stoa/publications/studies/stoa32and39\\_en.pdf](http://www.europarl.europa.eu/stoa/publications/studies/stoa32and39_en.pdf)

professional interpretation of results, transparent supply of accurate information, and existence of additional consent procedures for any use of data for research purposes.

Many of the issues addressed by the EASAC-FEAM recommendations have been taken into account in the recent proposal by the European Commission for a Regulation<sup>3</sup> on in vitro diagnostic medical devices. This clarification of a preferred regulatory pathway also begins to answer some of the questions posed by the earlier STOA report. Nonetheless, Professor Cornel observed that there were several regulatory dimensions that would benefit from further consideration: to ensure robust, independent review of the technical and clinical evidence on claims for tests; to facilitate provision of accurate and meaningful information to the public; and to entertain the possibility of alternative legal frameworks, such as that used to regulate medicines' provision.

Leonhard Hennen (Karlsruhe Institute of Technology) summarised the conclusions from the 2008 STOA study, noting that the scope of DTC GT services had expanded significantly since that time (and prices had decreased). Among regulatory options proposed in 2008 were the restriction to prescription by a physician, industry self-regulation by a code of practice, and pre-market approval. The latter option was most similar to the Regulation route now proposed by the European Commission although, in agreement with the point made by Professor Cornel, a more stringent approach, emulating pharmaceutical regulation, might also warrant further consideration. Whichever approach is adopted, there is the additional challenge in ascertaining how Europe can regulate in a global, internet-based, market. Therefore, it is important both that the various regulatory authorities work together and that the scientific community raises awareness of the issues internationally. Thus, there may be a role for academies of science and medicine globally to build on the initiative of the EU academies.

Many of the points made by Professors Cornel and Hennen were developed further by Panel members and in general discussion.

Pascal Borry (University of Leuven) noted that the DTC GT sector was in a state of flux with new companies appearing, others disappearing and some increasingly involving physicians in their services. He also referred to weaknesses of the new proposed legislation on in vitro diagnostic medical devices in relation to the assessment of whether genetic tests are useful. The proposed Regulation focuses on safety and performance of the devices, leaving room for companies to bring new tests to market that conform to technical requirements but lack evidence for predictive value and

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<sup>3</sup> Available from [http://ec.europa.eu/health/medical-devices/documents/revision/index\\_en.htm](http://ec.europa.eu/health/medical-devices/documents/revision/index_en.htm)

utility. He emphasised that a European approach in dealing with these issues would be useful but current frameworks may leave too much responsibility to Member States. Some countries are seeking a more rigorous path in enforcing legislation that requires provision of genetic testing services through a physician or in conformity with national screening priorities and assessment. It is also vital that any approach to regulation is accompanied by improved public education and engagement to empower the consumer to make informed choices.

Anne Forus (Committee on Bioethics, Council of Europe) referred to the Council of Europe's protocol<sup>4</sup> on genetic testing for health purposes which, although not specific to DTC GT, identifies generic problems relating to appropriate provision of information to the individual before, during and after testing, data validity and confidentiality, and understanding of risk information. In support of the points from previous speakers on the importance of public engagement, the Council of Europe recently acted to publish information for the public on genetic testing, including DTC GT<sup>5</sup>, and this will be widely translated.

Sabine Lecrenier (DG for Health and Consumers, European Commission) reviewed some key features of the proposed Regulation, relating to points raised in the EASAC-FEAM report:

- For the purposes of the Regulation, it is now clear that the definition of in vitro diagnostic medical devices covers genetic testing and includes predisposition to medical conditions or disease.
- The Regulation also applies to services offered through the internet.
- Tests are classified according to risk, with all genetic tests included within a single risk category.
- In-house (laboratory-developed) tests are exempted from the Regulation if the health institute is accredited and providing that any safety issues are reported.
- Instructions for test use must include information on test limitations and advise that the consumer should not take medical decisions without consulting a medical professional.
- There is an enforced requirement for test scientific validity and clinical performance. Clinical utility will not be covered by the Regulation as this is perceived as a matter for Member States.
- An EU data bank on manufacturers and marketed tests will be established, with public access to summaries of test safety and performance.

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<sup>4</sup> Available from <http://conventions.coe.int/treaty/en/treatise/html/203.htm>

<sup>5</sup> Available from [http://www.coe.int/t/dg3/healthbioethic/Source/en\\_geneticTests\\_hd.pdf](http://www.coe.int/t/dg3/healthbioethic/Source/en_geneticTests_hd.pdf)

- The Regulation requires a qualified person within the company to be responsible for regulatory compliance.

Among the questions raised in general discussion were:

*Appraising genetic data* Are genetic data different in principle from other types of test data and should all genetic tests be considered to fall within the same category of risk?

*Strengthening pre-market approval* How will the weaker Member State Notified Bodies be strengthened to deliver uniform quality of technology assessment across the EU?

*Alternative regulatory approaches* If the FDA grants marketing authorisation to some DTC GT companies, what will be the impact on EU regulatory procedures? Would the EU consider asking the European Medicines Agency to act in a manner analogous to the FDA?

*Impact on public health systems* What else needs to be done in the EU to increase use of responsible testing and protect against unsound testing? In particular, what are the priorities for the continuing education of medical professionals, for clinical governance and for improvements in public medical genetic services in translating research advances into clinical practice? Might DTC GT ever be reimbursed by medical insurance?

*Sharing information* How will quality of information on gene-disease associations and their implications in the genetic testing data base (registry) be validated? How will it be made accessible to the general public?

There is significant work to be done to explore these and other issues. In his final remarks, Antonio Correia de Campos (STOA Chairman) concluded that the European Commission's proposal was an important step forward, addressing some of the issues raised in the EASAC-FEAM and STOA reports, but more would be needed. In order to respond to the growing opportunities and challenges, legislation would need to be complemented by increased commitment to professional medical training, public engagement, health services development and collective action for an international, standardised repository of information.

The impending legislation at the EU level is timely. EASAC and FEAM will continue to examine how best to inform the deliberations in the EU Institutions and, in addition, to stimulate discussion in Member States and at the global level on the options for managing genetic testing.