

Direct-to-consumer genetic testing for health-related purposes in the European Union: the view from EASAC and FEAM

The rapid pace of advance in DNA analysis has led to increasing interest in the development of genetic tests for determining susceptibility to common, complex disorders. Such tests are increasingly being offered by companies through the internet. These direct-to-consumer genetic testing (DTC GT) services raise scientific, regulatory and ethical questions.

The issues were examined in a project initiated by the European Academies Science Advisory Council (EASAC) and the Federation of European Academies of Medicine (FEAM), which aimed to review the scientific evidence already available, to assess the regulatory developments underway and to ascertain the principles that should underpin the options for action by public policy-makers.

We note that there is controversy about whether using a nucleic-acid-based test is fundamentally different to using other types of biomarker as the predictor of risk, and whether concerns expressed about genetic testing are primarily related to the use of nucleic acids as the analyte or to the more general use of predictive risk information. In our view, efforts to devise recommendations relating specifically to genetic testing should be regarded as part of longer-term efforts to address all medical testing.

The scientific literature on potential benefits and harms of DTC GT is rather limited and, because it is drawn from consumers who can be regarded as 'early adopters', it may not be entirely relevant to the broader population. It is necessary to collect more evidence for the impact of testing on health outcomes and to share good practice in understanding, handling and communicating information about risk.

Based on an EASAC–FEAM Working Group discussion, it seems that all kinds of genetic testing require an appropriate and relevant level of professional advice. On the whole, DTC GT has little clinical value at present and, on occasion, has potential to be harmful. We would not wish to encourage European Union citizens to use DTC GT at present. We suggest special caution about DTC GT in several specific respects, for example relating to testing for high penetrance, serious disorders, prenatal screening, and nutrigenomic and pharmacogenetic testing. In developing general principles for the management of consumer genetic services, we emphasise that regulation must be on the basis that claims about the link between genetic markers and disease are scientifically valid. Other key issues to address include quality assurance, transparent supply of accurate information, consideration of the implications for established health services and clarification of consent procedures, including any use of data for research purposes.

These principles have consequences—for European Union policy-makers, for informed consideration of the regulatory alternatives; for the research community in developing an accessible evidence base; and for health professionals in translating research into practice:

- In Vitro Diagnostic Medical Devices Directive. The scope should be clarified to ensure that it covers all genetic information that is used to make medical claims. The European Commission will need to explore the options for introducing independent review of the claims made for a test, based on some form of risk stratification but independent of the nature of the analyte. The evidence base for all information provided must be accessible and verifiable.
- Professional and technical competences. Whatever can be achieved by reform of the In Vitro Diagnostic Medical Devices Directive to require demonstration of scientific validity of claims will need to be accompanied by appropriate mechanisms for ensuring professional and clinical good governance according to standard procedures.
- Industry code of practice. While awaiting public policy development, it would be highly desirable for DTC GT companies to work together to develop and implement industry-wide quality standards.
- Public databases of information. An international registry of information on the availability, validity and usefulness of genetic tests would help physicians and consumers to judge for themselves whether to avail of a particular test or service. The European Commission should consider what is needed to collect and validate the evidence on gene-disease associations.
- Professional education. It is vital for Europe to do better in educating medical and other health professionals about genetics, for example to improve the confidence of primary care physicians to interpret and explain risk and benefit.
- Public engagement. It is also critically important to address common public misconceptions about what genetic tests can offer in terms of medically relevant information so as to inform and empower consumers to decide for themselves when faced with offers of testing.
- Whole-genome sequencing. Very soon, it will be easier and cheaper to sequence an entire genome than to genotype a series of known mutations. The

challenges of consenting, communicating and acting on data will be accentuated by whole-genome sequencing, which has considerable potential to reveal incidental information that was not anticipated or requested by the consumer. Regulatory authorities and other policy makers need to prepare for the translation of this technology from the research setting to routine testing.

- Global implications. EU reform of Medical Devices legislation must be well integrated with global harmonisation efforts and this requires further work to develop shared understanding of test clinical performance. The situation is complicated by differences in the relevance of genetic information for different populations and it is important to build global databases

containing the clinical information on DNA variants of specific genes.

In conclusion, although some of these issues are controversial, there are opportunities to improve the regulatory and innovation framework for genetic testing in the EU. However, legislative reform will take time and can only be successful if there is also action to improve clinical governance and professional and public education; to facilitate translation of the available evidence base into practice and to support research to collect new evidence; and to ensure the widespread availability of accurate information.

The full report is available from the EASAC and FEAM websites: www.easac.eu and www.feam.eu.com.

EASAC

EASAC – the European Academies Science Advisory Council – is formed by the national science academies of the EU Member States to enable them to collaborate with each other in providing advice to European policy-makers. It thus provides a means for the collective voice of European science to be heard.

Its mission reflects the view of academies that science is central to many aspects of modern life and that an appreciation of the scientific dimension is a pre-requisite to wise policy-making. This view already underpins the work of many academies at national level. With the growing importance of the European Union as an arena for policy, academies recognise that the scope of their advisory functions needs to extend beyond the national to cover also the European level. Here it is often the case that a trans-European grouping can be more effective than a body from a single country. The academies of Europe have therefore formed EASAC so that they can speak with a common voice with the goal of building science into policy at EU level.

Through EASAC, the academies work together to provide independent, expert, evidence-based advice about the scientific aspects of public policy to those who make or influence policy within the European institutions. Drawing on the memberships and networks of the academies, EASAC accesses the best of European science in carrying out its work. Its views are vigorously independent of commercial or political bias, and it is open and transparent in its processes. EASAC aims to deliver advice that is comprehensible, relevant and timely.

For more information about EASAC and for copies of all our previous publications, please visit our website www.easac.eu.

FEAM

FEAM was founded in 1993 in Brussels with the objective of promoting cooperation between the national Academies of Medicine and of extending to the political and administrative authorities of the European Union the advisory role that the Academies exercise in their own countries on matters concerning medical sciences and public health. Since 31 March 1995, FEAM has enjoyed the civil status of an international association with a scientific objective. As an umbrella organization, it brings together national Academies of fourteen European member states (Austria, Belgium, Czech Republic, France, Germany, Greece, Hungary, Ireland, Italy, Portugal, the Netherlands, Romania, Spain and the United Kingdom) and aims to reflect the European diversity by seeking the involvement of additional Academies and experts in its scientific activities and by collaborating with other networks on scientific matters of common interest.

To find out more about FEAM, visit the website – www.feam.eu.com – or contact the FEAM secretariat at info@feam.eu.com.

EASAC Secretariat
Deutsche Akademie der Naturforscher Leopoldina
German National Academy of Sciences
Jägerberg 1, D-06108 Halle (Saale), Germany
Tel: +49 (0)345 4723 9833 fax: +49 (0)345 4723 9839
email: secretariat@easac.eu

EASAC Brussels Office
Royal Academies for Science and the Arts of Belgium (RASAB)
Hertogsstraat 1 Rue Ducale, B-1000-Brussels, Belgium
Tel: +32 (2) 550 23 32; fax: +32 (2) 550 22 05
email: brusseloffice@easac.eu
web: www.easac.eu

FEAM
Palais des Académies
Rue Ducale 1 Hertogstraat, B-1000-Brussels, Belgium
Tel: +32 (0)2 550 22 68; fax: +32 (0)2 550 22 65
email: info@feam.eu.com
web: www.feam.eu.com

