British Society for Human Genetics

European Commission
Health and Consumers Directorate-General (DG SANCO)
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1st September 2010

Dear Sir/Madam

Revision of Directive 98/79/EC of the European Parliament and of the Council of 27 October 1998 on in vitro diagnostic medical devices. Public consultation (closure date 15th September 2010)

The British Society for Human Genetics (BSHG) represents 2,000 health professionals working in specialised genetic services in the UK National Health Service and scientists and health professionals in medical research. (www.bshg.org.uk). The Society is grateful for the opportunity to respond to the consultation on the revision of the IVDD directive.

Our main response concerns questions 7, 8, 9 and 10 in which we endorse the official position of the EuroGentest Network of Excellence in Genetic Testing. We will not repeat the points here but summarise that it is in the best interests of patients with genetic conditions for the exemption in article 1(5) for in house tests to be retained but be restricted to laboratories that have had their competence to design and manufacture, validate and deliver in house tests audited and recognised through accreditation to ISO15189 or equivalent.

We supplement the points made by EuroGentest by summarising the direction of travel of genetic testing. DNA sequencing technologies have been developed to serve the needs of the Human Genome Project and, as research findings have flowed from the project, the technologies have been adapted and exploited in healthcare diagnostics for inherited disorders, acquired disease (especially oncology), immunogenetics, infectious disease science and pharmacogenetics. New whole genome sequencing technologies readily allow scanning for previously unrecognised mutations in the cellular DNA of patients with symptoms of disease and in tumours. This challenges the paradigm of the IVDD which when applied to genetic analysis assumes that the test is designed to detect the presence or absence of a pre-determined set of genetic variants. Such tests are valuable and will continue to have a place in genetic analysis. We recognise that whole genome scanning techniques raise challenges in the interpretation of newly discovered variants or unexpected findings. However this situation is not new and karyotyping as the earliest comprehensive genetic analysis represents a whole genome scan for structural re-arrangements that has been part of medical practice since the 1960s. Interpretation of karyotyping or the new mutation scanning techniques then as now requires a close liaison between clinical doctors as phenotypers and scientists. A new feature is that interpretation will require the development and informed application of new bioinformatic tools which are suitable for healthcare use.

Although the focus of this response is on genomic technologies and their use in clinical practice highly parallel technologies for the detection and characterisation of proteins (proteomics) and small molecules (metabolomics) will follow during the lifetime of the proposed revision of the IVDD and present the same challenges.

In summary medical diagnostics is shifting from tests for a limited set of specific molecules and genetic variants to generic scanning techniques which will generate many thousands of data points; within five years most genetic testing will be synonymous with sequencing. In the view of the BSHG the best response is for regulators to shift their focus towards assuring the competence of the health institutions, enterprises and personnel responsible for delivering medical tests and for interpreting their results to patients and their families.

In this regard we draw the attention of the Commission to the OECD Guidelines for Quality Assurance in Molecular Genetic testing (2007) which make some relevant points concerning testing being delivered in a health-care framework (A2), the responsibility of laboratories to make information available on the validity of their tests (Aii) (Bvi), the value of accreditation as a mark of competence (B1), the value of collaboration and inter-laboratory comparisons (B8, C1) and the need for education and training to ensure the competence of personnel (E1).

Thanking you again for the opportunity to respond to this consultation

Yours Sincerely

Dr Christine Patch

Chair British Society for Human Genetics

Dr Rob Elles

Past Chair British Society for Human Genetics

Copy: Trevor Cole Joint Committee on Medical Genetics