

## **ESHG Position Statement on the Inclusion of an Article on Genetic Testing in the Proposed Regulation on In Vitro Diagnostic Devices**

The European Society of Human Genetics is a non-profit organization. Its aims are to promote research in basic and applied human and medical genetics, to ensure high standards in clinical practice and to facilitate contacts between all persons who share these aims, particularly those working in Europe. The Society encourages and seeks to integrate research and its translation into clinical benefits and professional and public education in all areas of human genetics.

The European Society of Human Genetics fully supports the aims and objectives of the Council of Europe's Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes and the OECD Guidelines on Quality Assurance for Genetic Testing.

ESHG welcomes the European Commission's proposal for a Regulation on in-vitro Diagnostic Devices (IVDs). This Regulation will improve the quality, safety and oversight of IVDs marketed and used in the European Union. ESHG particularly welcomes the introduction of mandatory accreditation for health institution laboratories which manufacture their own tests. This will bring a new level of quality assurance to many genetic tests, especially tests for rare diseases.

ESHG also welcomes the proposal that all genetic testing should be carried out under individualised medical supervision, reflecting the dangers posed by "Direct to consumer" genetic testing for medical purposes. We believe that this can be achieved within the proposed Regulation by amendment of Article 1(5) as proposed by EuroGentest, which mandates that devices for human genetic testing be made available on prescription only.

ESHG also supports the Rapporteur's proposed Amendment 6 to the recitals, which emphasises the importance of the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes.

However, ESHG has serious reservations about the proposal to introduce provisions on genetic counselling into the Regulation on in vitro Diagnostic Devices (Rapporteur's proposed Amendment 6). While the Society supports any legislation that will bring clear benefits to patients, we do not support the use of the Regulation on IVDs for the purpose of regulating clinical practice in genetics. The purpose of the Regulation on IVDs is to ensure that IVDs are designed and manufactured to a high quality and that their use in medicine is based on sound scientific evidence. The Regulation does not govern the behaviour of the users of IVDs, except to oblige them to report adverse incidents involving devices. The proposed article attempts to extend the reach of the regulation beyond the user of the device to the clinician who sees the patient. This creates a burden on the users of the device (i.e. clinical laboratory staff) to determine, in some unspecified way, that the requirements of this article have been met. ESHG views this proposal as unworkable in the daily practice of genetic medicine.

Genetic testing is evolving rapidly, from a model based on tests for single-gene disorders with highly predictive outcomes to a mix of multiplex and even genome-wide tests, pharmacogenetic tests and companion diagnostics. The requirements for pre- and post-test genetic counselling differ greatly between different categories of test.

ESHG believes that the existing OECD Guidelines and the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes already provide a framework to guide those involved in genetic testing. The inclusion of the proposed new article as written would place a severe burden on genetics services which operate with limited resources, inevitably meaning that fewer, not more, patients would benefit from genetic testing.

The ESHG would strongly support Dr Liese if he were to propose a stand-alone Regulation or Directive on genetic testing, within the limits of EU competence in healthcare, as we believe that it is only by this means that our shared objectives can be achieved.

ESHG Executive  
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