EuroGentest Committee

https://www.eshg.org/egt

Presentation NHGS Meeting – Milano 2025

EuroGentest Committee

Harmonization and standardization in genetic services
- quality in the laboratory
- quality for genetic counselling

Identify gaps in quality issues within diagnostic and clinical genetic testing services
Promote harmonization with EQA providers to reduce poor performance
Identify the specific needs for professional guidelines
Promote the uniform translation of technologies into diagnostic practice
Address policy issues on behalf of a genetic community

Started as a Network of Excellence (EC) in 2005, fully integrated in ESHG since 2015

Harmonization in genetic counselling

Working group members: Rebecka Pestoff (Sweden)
Christi van Asperen (the Netherlands)
Christophe Cordier (Switzerland)
Donna Darmanin (Malta)
Katrin Õunap(Estonia)

Harmonization necessary to provide quality assurance and patient safety.

A review of published state-of-the art in Europe is being prepared (updating 2008 survey):

- describing the current evidence base
- presenting identified issues and
- proposing potential solutions.

In collaboration with the European Board of Medical Genetics (EBMG)

eurogentest@eshg.org

IVDR Task force

To inform diagnostic laboratories and raise awareness within the genetics community and towards regulatory authorities about key concerns related to the implementation of the IVDR.
To highlight concerns regarding the loss of innovation and the availability of IVD devices for orphan diagnostics.

Formal representation
ESHG representative in Biomed Alliance TF IVDR
Member of MDCG IVDR workgroup

> opportunity to provide examples of “orphan diagnostic devices” that remain unaddressed in the IVDR and its associated MDCG documents.

IVDR survey of European laboratories
Responded to the European Commission’s “Public consultation and call for evidence for the targeted evaluation of the EU Regulations on medical devices”

ESHG / EuroGentest IVDR Task force

Els Dequeker (Belgium)
Milan Macek Jr (Czech Republic)
Gunnar Houge (Norway)
Thomas Liehr (Germany)
Gert Matthijs (Belgium)
Joris Vermeesch (Belgium)

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IVDR Survey

In preparation

Call for evidence

Public consultation

Feedback and consultation period

12 December 2024 - 21 March 2025

Feedback: Closed

Upcoming

Commission adoption

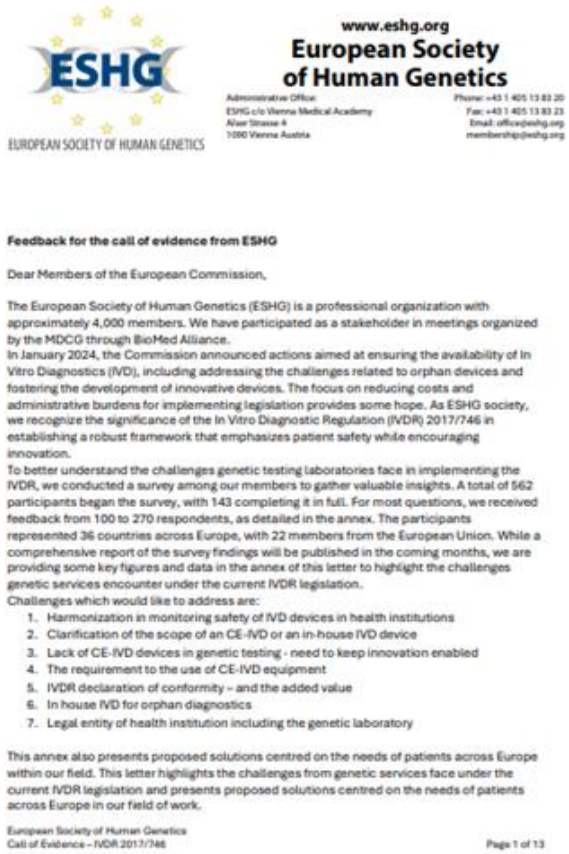
Planned for

Fourth quarter 2025

Survey was distributed to all ESHG members: 143 complete response (out of 562 participants)

Report submitted March 21st, 2025 (supported by EURORDIS-Rare Disease Europe)

Allowed to reinforce 7 key points to highlight the challenges genetic services face under the current IVDR framework.



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European Society of Human Genetics

Feedback for the call of evidence from ESHG

Dear Members of the European Commission,

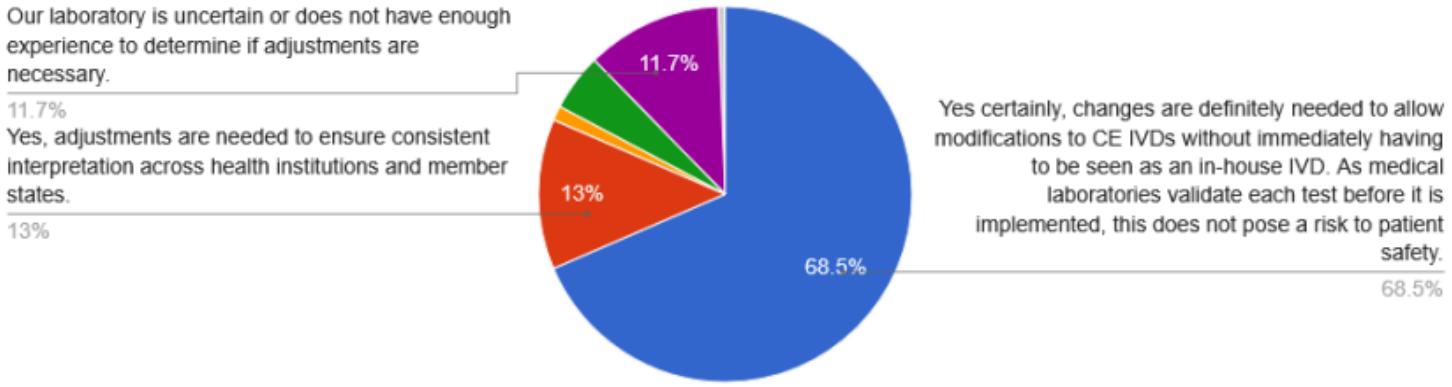
The European Society of Human Genetics (ESHG) is a professional organization with approximately 4,000 members. We have participated as a stakeholder in meetings organized by the MDCG through BioMed Alliance.
In January 2024, the Commission announced actions aimed at ensuring the availability of In Vitro Diagnostics (IVD), including addressing the challenges related to orphan devices and fostering the development of innovative devices. The focus on reducing costs and administrative burdens for implementing legislation provides some hope. As ESHG society, we recognize the significance of the In Vitro Diagnostic Regulation (IVDR) 2017/746 in establishing a robust framework that emphasizes patient safety while encouraging innovation.
To better understand the challenges genetic testing laboratories face in implementing the IVDR, we conducted a survey among our members to gather valuable insights. A total of 562 participants began the survey, with 143 completing it in full. For most questions, we received feedback from 100 to 270 respondents, as detailed in the annex. The participants represented 36 countries across Europe, with 22 members from the European Union. While a comprehensive report of the survey findings will be published in the coming months, we are providing some key figures and data in the annex of this letter to highlight the challenges genetic services encounter under the current IVDR legislation.
Challenges which would like to address are:

1. Harmonization in monitoring safety of IVD devices in health institutions
2. Clarification of the scope of an CE-IVD or an in-house IVD device
3. Lack of CE-IVD devices in genetic testing - need to keep innovation enabled
4. The requirement to the use of CE-IVD equipment
5. IVDR declaration of conformity – and the added value
6. In house IVD for orphan diagnostics
7. Legal entity of health institution including the genetic laboratory

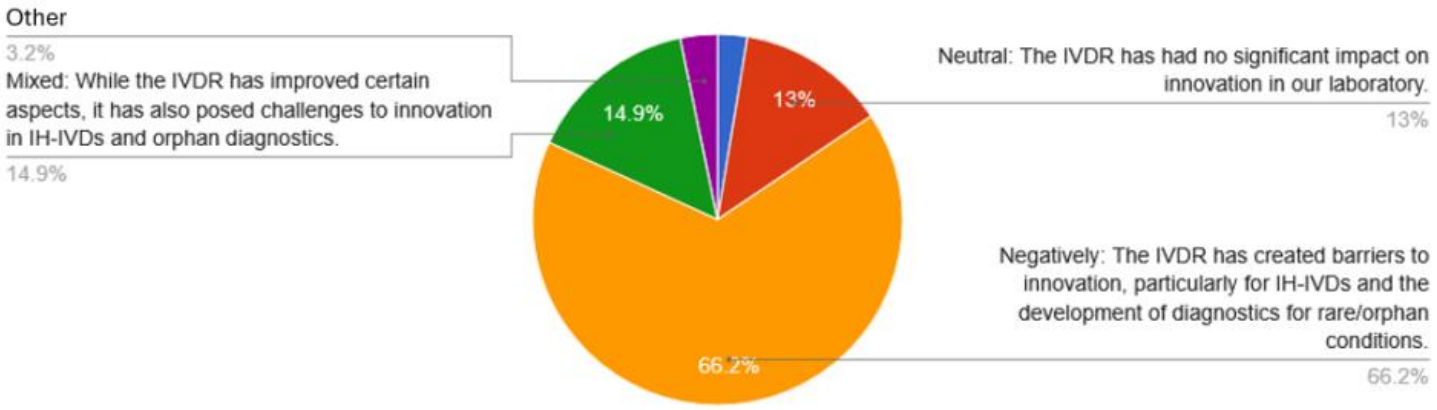
This annex also presents proposed solutions centred on the needs of patients across Europe within our field. This letter highlights the challenges from genetic services face under the current IVDR legislation and presents proposed solutions centred on the needs of patients across Europe in our field of work.

European Society of Human Genetics
Call of Evidence – IVDR 2017/746 Page 1 of 13

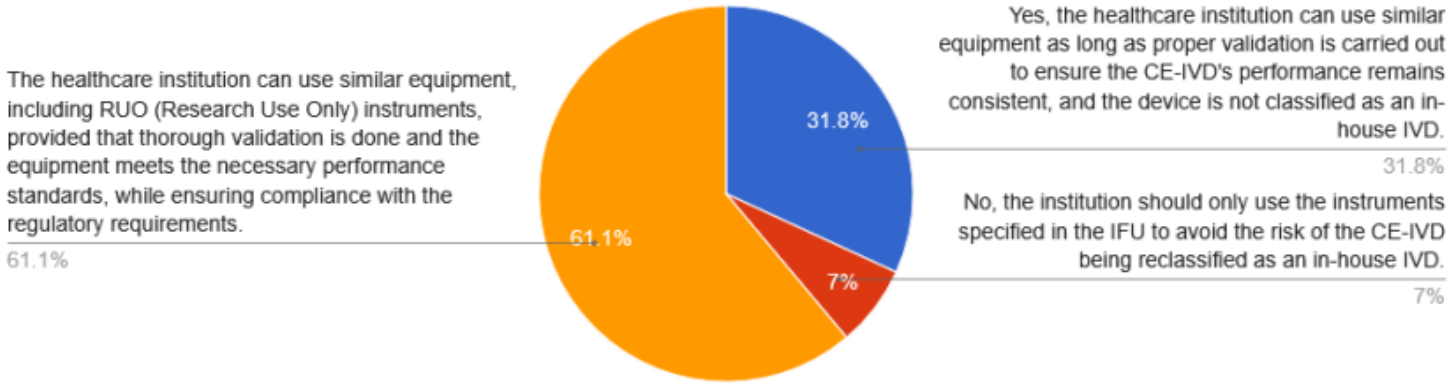
Do you think the scope of CE-IVDs and their transintion to in-house IVDs needs to be clarified?



How does IVDR impact innovation in your lab, esp. in context of IH-IVDs and orphan diagnostics?



The instructions for use of CE-IVD kits often restrict the use to specific instruments.
Can the lab its own equipment (similar but not exactly the same) without the CE-IVD being classified as in-house IVD?



IVDR Survey

1. Ensuring harmonization in monitoring IVD device safety within healthcare institutions.
2. Clarifying the distinction between CE-IVD devices and in-house IVD devices.
3. Addressing the shortage of CE-IVD devices while maintaining space for innovation.
4. Assessing the requirement to use CE-IVD equipment.
5. Evaluating the added value of the IVDR declaration of conformity.
6. Securing the use of in-house IVDs for orphan diagnostics.
7. Defining the legal entity status of healthcare institutions, including genetic laboratories.

https://ec.europa.eu/info/law/better-regulation/have-your-say/initiatives/14155-EU-rules-on-medical-devices-and-in-vitro-diagnostics-targeted-evaluation/F3530219_en

Persistent poor performance in external quality assessment

Working group lead by Weronika Gutowska-Ding (UK), Nicola Wolstenholme (UK), Christine Vianey-Saban (France), Els Dequeker (Belgium), Isabel Carreira (Portugal), with representation of all EQA providers

- Harmonisation of the definition of poor performance
- Define how to act on persistently poorly performing laboratories in the absence of performance monitoring national bodies.

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WE WOULD LIKE TO PRESENT THE IMPORTANCE OF EXTERNAL QUALITY ASSESSMENT AND THE NEED TO REVIEW POOR PERFORMANCE AT YOUR ANNUAL NATIONAL MEETING

-> 10-15 MIN ONLINE PRESENTATION: importance of EQA, examples of remediation

Please contact us at eurogentest@eshg.org

EuroGentest Committee

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