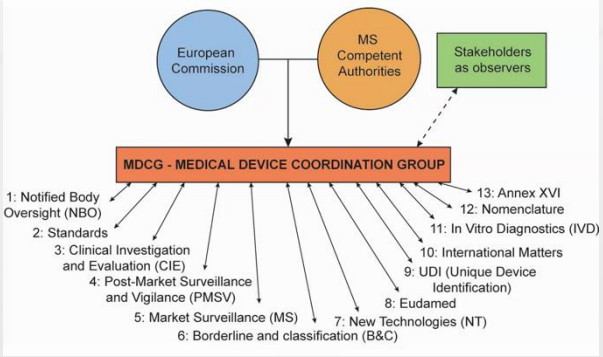
IVDR & Rare Diseases – update and important deadlines to know!

Prof dr E Dequeker, Belgium
Liaison for the ESHG in the Task Force of IVDR Biomedical Alliance
Head of the Quality Department of Medical Diagnostics – University of Leuven

IVDR 2017/746 (May, 26th 2022) – *Harmonization*

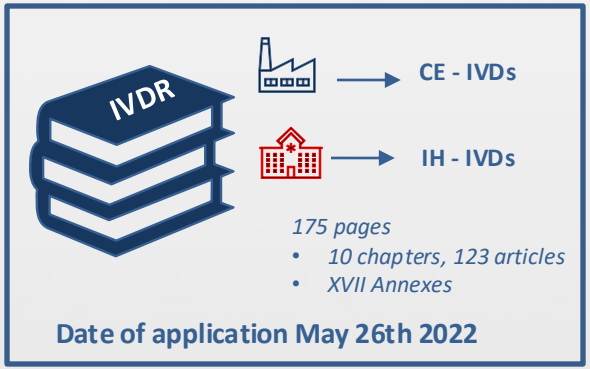
- **European regulation**
  - CE-IVD & In-house (IH)-IVD
  - Industry & Health Institution

- **EU guidelines / interpretation documents – MDCG**



MDCG 2022-15,
MDCG 2022-22 rev1,
MDCG 2022-9, …

MDCG Guidelines



Impact of IVDR

for CE-IVD's and in-house-IVD's

HemaPolicy
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Critical Implications of IVDR for Innovation in Diagnostics: Input From the BioMed Alliance Diagnostics Task Force

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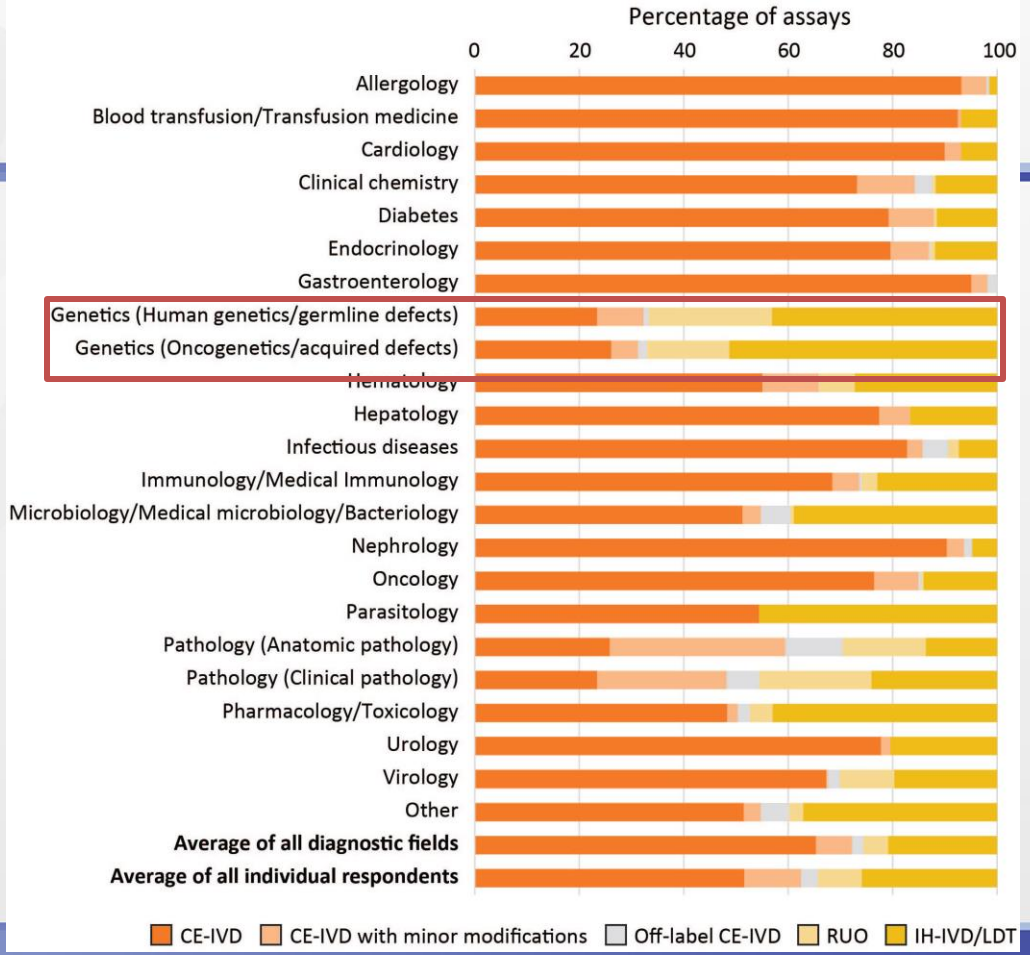
Hemasphere. 2022 May 20;6(6):e724



Initiatives ESHG took to help genetic diagnostic laboratories

Initiatives taken by ESHG

1. **Policy work**

IVDR Task Force Activities

EU presidency Czech Republic – Prague (Milan Macek – Els Dequeker)

Quite a **challenge** for **new tests for "rare diseases"** to preserve the final purpose of the regulation - Call for **embedding an incubation period**

EU presidency Spain - Bilboa (Milan Macek - EURORDIS)

EU presidency Belgium – Gent (Gert Matthijs / Els Dequeker)

Meeting with all the national CAMD (competent authorities medical devices) and DG Sante

"Challenges for in house – Orphan Diagnostics"

Letter to DG Sante

2024-02-08

Rare diseases diagnostics should be exempt from the Vitro Diagnostic Medical Devices Regulation

We express our concerns on the impact of certain provisions on our community and urge the European Commission to take action.

More information

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

EURORDIS
RARE DISEASES EUROPE

Sandra Gallina
Director General – DG SANTE
European Commission
Rue de la Loi 200 - 1049 Brussel
Belgium

22 December 2023

Ref.: Rare diseases diagnostics (i.e. "orphan diagnostics") should be exempt from the Vitro Diagnostic Medical Devices Regulation (EU) 2017/746 in its Article 5.5

Dear Ms. Gallina,

We are jointly writing you on behalf of the European Society of Human Genetics (ESHG), EURORDIS-Rare Diseases Europe and the **European Reference Networks (ERNs) Coordinators Group** to express our concerns on the impact of certain provisions of the In Vitro Diagnostic Medical Devices Regulation (EU) 2017/746 (henceforward IVDR) on our community and urge the European Commission to take action.

As we will illustrate below, in its current form the IVDR negatively impacts the diagnosis and treatment of patients with rare diseases in Europe and hampers the development of rare disease diagnostics by the European private and public sector.

We also share a more general concern raised by Member States at the last EPSCO meeting that the transition to the new regulatory IVDR is not as advanced as necessary and there is a consequent, serious risk of shortages of devices, especially class D devices, which may seriously hinder the availability of in vitro devices in the Union.

We therefore call on the European Commission to:

- **Grant an exemption of rare diseases/rare cancers diagnostics (or, as we propose, "orphan diagnostics") from IVDR regulation in Article 5.5;**
- **Take all necessary measures, including those of legislative nature where appropriate, to reduce or avoid shortages of classes of devices that are essential for patients. This may include a further postponement of IVDR implementation.**

Accessibility to medical devices necessary for diagnostics of rare diseases is of crucial importance to reduce the protracted journeys towards diagnosis that people living with rare disease experience, as recently confirmed by the Rare Barometer global survey on diagnosis (*pending publication of results*).

More than 70% of clinical decisions and therapy guidance are based on laboratory examinations. This is particularly impactful in the field of rare diseases, with more than 80% of them being of a genetic nature. Rare disease diagnostics are also highly relevant for the entire field of rare cancers where delayed diagnosis could have disastrous consequences. The number of EU citizens needing these tests is not small: with rare diseases affecting less than 1 in 2,000 inhabitants, cumulatively these individuals constitute around 5% of the general population, an estimated over 30 million citizens in Europe with a risk of developing one of the 6,000 rare diseases. In paediatrics, rare disease patients constitute a major part of all hospital admissions and are responsible for a substantial part of morbidity and mortality in minors.

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Commission proposes measures to improve the availability of IVD

Brussel, 23 January 2024

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

Quote(s)

"A priority of a strong European Health Union is to ensure that medical devic and diagnostics are available to patients, whenever they need them. We mu take immediate action to improve their availability. Today's proposal will prov relief for the sector, without compromising patient safety and care. Going forward, we are determined to analyse the root causes that slow the transitio and committed to take appropriate action."

7. **What other actions is the Commission taking to ensure availability of medical devices and *in vitro* diagnostics?**

Specifically for devices intended for small patient populations like children or persons with a rare disease ("**orphan devices**") a **new guidance**, is being developed. The guidance is expected to **significantly help the certification of existing orphan devices** in accordance with the MDR/IVDR, by addressing the specific challenges of clinical evidence requirements for these types of devices.

Finally, the Commission will start already in 2024 preparatory work for a **targeted evaluation of the legislation**. The evaluation should assess in particular the impact of the legislation on the availability of devices, especially devices responding to special needs such as "orphan devices", and the development of innovative devices in Europe. Special attention in the assessment may also be given to costs and administrative burdens stemming from the implementation of legislation, especially for SMEs.

Initiatives ESHG took to help genetic diagnostic laboratories

Initiatives taken by ESHG

1. Policy work
2. **Informing & sharing knowledge**

"IVDR – Beneficial or just an expensive straight-jacket?"

June 22, 2022, 10:00 – 16:30 hrs

The **new IVD EU regulation (IDVR 2017/746)** has major implications on the use, availability and associated costs of in vitro diagnostic tests. But on the other hand, it also builds in many ways new assurance points to improve and monitor the quality of our diagnostic services.

To help laboratories prepare for the IVDR, including compliance with IVDR requirements for in-house devices, the ESHG is hosting a free of charge full-day webinar on IVDR, providing the basics and latest updates on implementation, as well as tips and tricks for working towards compliance.

During these webinar presentations we invite speakers from the EU Commission, Notified Bodies, Compent authorities, national IVDR working groups, lab people working on the implementation of the IVDR will share their knowledge and experience.

2022

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Program – 8 speakers & discussion sessions

- Introduction on IVDR
- Role of different organisation: EC, CA, NB, Reference labs, observers
- IH-IVD's and hospital exemption
- MDCG guidelines and the guideline of in-house devices

Participation is free of charge, registration is necessary !

High attendance > 300 persons

2023

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ESHG, Glasgow

Get2gether : IVDR is a challenge for all of us, the greatest challenge is keeping "orphan" diagnostics available

Speakers

- **Insight into the main elements of IVDR and initiatives ESHG took to help genetic diagnostic laboratories**
  Els Dequeker, University of Leuven, Liaison for the ESHG in the Task Force of IVDR Biomed Alliance
- **Process for CE-marking of a genetic test**
  Alex Laan, Head of the IVD Notified Body BSI The Netherlands
- **Where can genetic diagnostic laboratories help industry and vice versa**
  Maurizio Suppo, Co-owner/VP QARAD

Panel discussion

With persons a laboratory, clinician, a diagnostic company, a Notified Body and a consultant

2024-2025

(One-day) online webinar on EuroGentest projects and IVDR

Needs to confirm – February – March 2025

the program for seeing topics for:

- medical geneticists
- clinical geneticists
- technicians

and specific topics for IVDR

Initiatives ESHG took to help genetic diagnostic laboratories

Initiatives by ESHG – EuroGentest

1. Policy work
2. Informing & sharing knowledge
3. **Actions of the ESHG-EUGT Task Force**

IVDR Task force – actions

Action 1 Workgroup with national expert in IVDR

Profile:
- Persons with a scientific background and working in a genetic lab
- knowledge of relevant guidelines, norms and product-specific standards
- knowledge of a quality management system and ISO 15189: 2022
- person have (voluntary) time to invest for active participation

Working group: regular meetings (1 month) with projects to help preparing information for the EU

Action 2

IVDR Task force – actions

Action 2

Launch survey to get picture

- on the national implementation legislation of IVDR
- national working groups on IVDR
- cooperation per country with the competent authority
- try to get a picture of the situation in each country

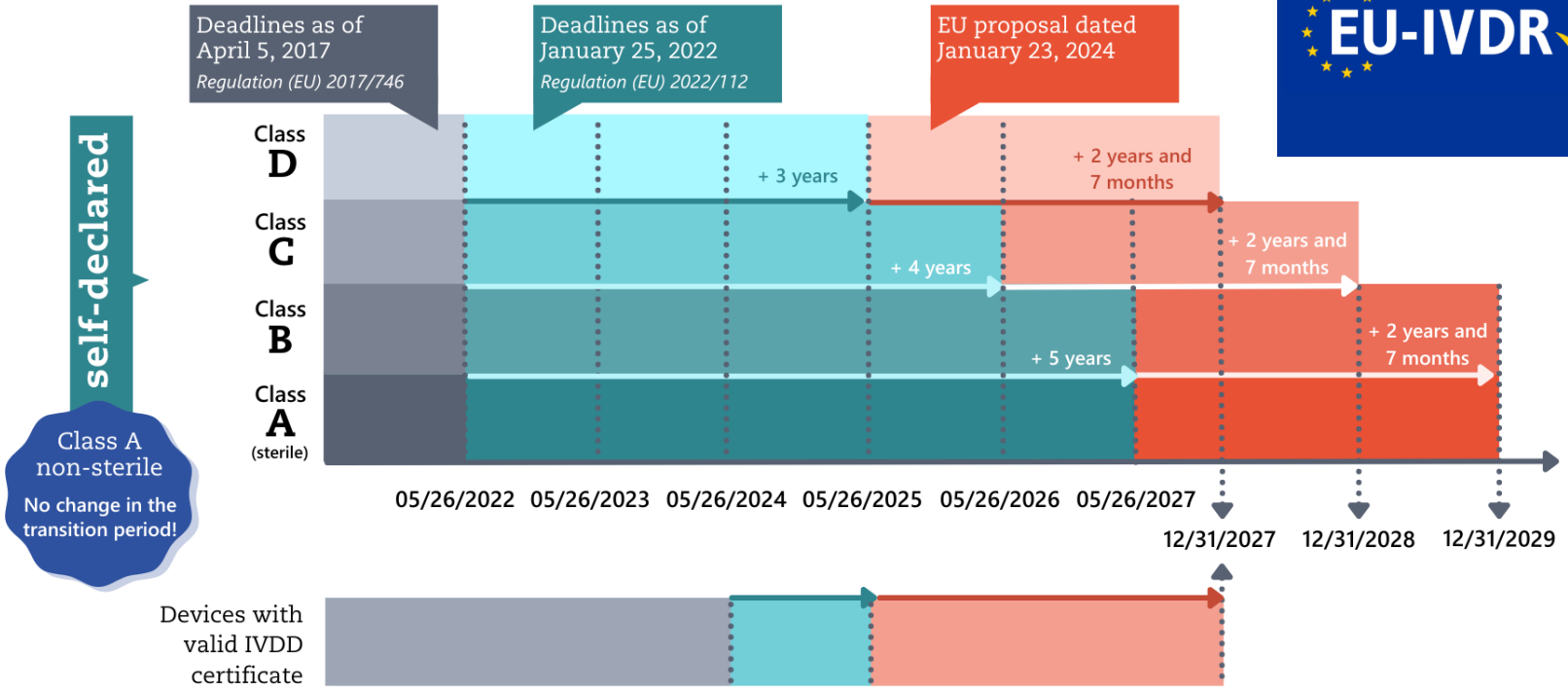
-> Information useful for apply further political pressure if necessary and maintain harmonisation within Eu

Initiatives ESHG took to help genetic diagnostic laboratories

Initiatives by ESHG – EuroGentest

1. Policy work
2. Informing & sharing knowledge
3. Actions of the ESHG-EUGT Task Force
4. **Important deadlines**

CE-IVD


Deadlines as of
April 5, 2017
Regulation (EU) 2017/746
Deadlines as of
January 25, 2022
Regulation (EU) 2022/112
EU proposal dated
January 23, 2024
EU-IVDR
self-declared
Class A
non-sterile
No change in the
transition period!
Class
D
Class
C
Class
B
Class
A
(sterile)
+ 3 years
+ 2 years and
7 months
+ 4 years
+ 2 years and
7 months
+ 5 years
+ 2 years and
7 months
05/26/2022
05/26/2023
05/26/2024
05/26/2025
05/26/2026
05/26/2027
12/31/2027
12/31/2028
12/31/2029
Devices with
valid IVDD
certificate

In house IVD

May 2022 - Description of in house IVD and completion of Annex 1 with relevant documentation

May 2024
- QMS is also adapted for in-house developed devices (see ISO 13485)
- register of IVDs in-house is available and you should follow the instructions of your local competent authority regarding reporting of these devices

May ~~2028~~ 2030
- justification no equivalent IVD device on the market
- state of the art of your in-house device

ESHG board and ESHG-EUGT IVDR Task Force are doing their utmost to take necessary actions

but this takes time and a European law cannot and should not be ignored.

There are sanctions attached to IVDR legislation so laboratories must take initiatives.