



www.eshg.org
**European Society
of Human Genetics**

Administrative Office:
ESHG c/o Vienna Medical Academy
Alser Strasse 4
1090 Vienna Austria

Phone: +43 1 405 13 83 20
Fax: +43 1 405 13 83 23
Email: office@eshg.org
membership@eshg.org

2020/09/22

Dear colleague,

The **European Society of Human Genetics** has **drafted and approved a statement** in which we clarify our position regarding the Regulation EU 2017/746 (the [IVD Regulation](#)) and the potential threat to both precision medicine, and **in particular diagnostics of rare diseases**. We would ask you for all the **support and/or formal endorsement** you can provide in this important matter, namely by:

- a. Endorsing the enclosed ESHG position statement and publicise this on your website. As an example could serve the Czech Society of Medical Genetics and Genomics ([in Czech](#));
- b. Contacting either your Ministry of Health or any other competent authority since we learned that transposition laws of the IVD Regulation are currently being prepared in various EU Member states;
- c. Enlisting the respective [national Alliance for Rare Diseases](#) and ask them for their support;
- d. Lobbying for change with your [EU Parliament MEPs from the ENVI committee](#) and claim that for the Medical Devices Directive the ENVI Committee of the EU Parliament already [voted vis-a-vis the Covid-19 crisis for its amendment](#), hence there is a recent precedent for our “cause”;
- e. Referring to the [European MedTech initiative](#); and the illustrative [Editorial by Thomas Liehr](#) addressing the effects of the Regulation on molecular cytogenetics.
- f. Enlisting other stakeholders, namely other professional societies in your country or at the European level, such as pathologists, microbiologists who have been supportive of amending the aforementioned EU Regulation and have a shared interest/concerns on its implementation in the current unprecedented situation in Europe, and beyond.

We thank you very much in advance for all and any support you can provide for the successful outcome of this concerted effort!

We remain at your disposal for any further questions.

With best regards,

Maurizio Genuardi

President
European Society of Human Genetics

Links included in this document:

- The IVD Regulation: <https://eur-lex.europa.eu/legal-content/EN/TXT/PDF/?uri=CELEX:32017R0746&from=cs>
- Endorsement Czech Society of Medical Genetics and Genomics: <http://slg.cz/aktuality/stanovisko-eshg-k-narizeni-eu-ivd/>
- National Alliances for Rare Diseases: <https://www.eurordis.org/content/national-alliances-rare-diseases>
- EU Parliament MEPs from ENVI committee: <https://www.europarl.europa.eu/committees/en/envi/home/members>
- Amendment Medical Devices Directive: https://www.politico.eu/wp-content/uploads/2020/04/Parliament-MDR-delay-clean.pdf?utm_source=POLITICO.EU&utm_campaign=54c9a9bc7e-EMAIL_CAMPAIGN_2020_04_15_05_56&utm_medium=email&utm_term=0_10959edeb5-54c9a9bc7e-189624705
- European MedTech initiative: <https://www.med-technews.com/news/medtech-europe-calls-for-pause-on-implementation-of-new-eu-r/>
- Editorial J GENET GENOM Vol 4:2,2020 “Is Molecular Cytogenetic Diagnostics of Rare Diseases in Europe Close to Extinction” by T. Liehr: <https://www.hilarispublisher.com/open-access/molecular-cytogenetic-diagnostics-of-rare-diseases-in-europe-close-to-extinction.pdf>