CONCLUSIONS AND RECOMMENDATIONS

This conference aimed to help develop a European policy framework for rare diseases. This framework would integrate strategies across various fields, including research, digital technologies, healthcare and social protection, and by doing so, complement existing legislation and stimulate new actions on rare diseases at both European and national levels.

The conference highlighted the European Economic and Social Committee's (EESC) exploratory opinion *Leaving No One Behind: European Commitment to Tackling Rare Diseases*, prepared at the request of the Hungarian Presidency of the Council of the EU, in which it urges the European Commission to publish a Communication on a European action plan for rare diseases for 2025-2029. The aim is to ensure that patients with rare diseases receive a diagnosis within one year of the onset of symptoms.

At the conference the Hungarian Presidency and the EESC called on the new European Commission and on the EU Member States to propose and implement a comprehensive European action plan for rare diseases, setting common and measurable objectives to improve the treatment of rare diseases and strengthen European collaboration in this field. The main recommendations of the conference, together with the main conclusions of the EESC's exploratory opinion, were brought to the attention of the EPSCO Council on 3 December 2024.

The conference also brought together a wide range of stakeholders, including civil society organisations, patient associations, doctors, researchers, health ministries and European institutions. This diverse participation ensured the continuity of the work and political commitment that EU Member States, civil society organisations and relevant stakeholders have been pursuing since 2004.

In all, the conference brought together 32 speakers, 30 EESC members and some 235 participants from all over Europe, both online and in person. The programme is included in the annex to this document.

Main conclusions and recommendations of the Conference

1. On achieving a comprehensive European action plan for rare diseases by 2030

- Political support is needed, with a commitment from the European Commission to establish a coherent framework that guides national policies and encourages collaborative action, beyond the European Reference Networks (ERNs).
- A steering group should be set up for the European action plan for rare diseases and composed of experts from the Member States, the EESC and EURORDIS to ensure horizontal coordination and cooperation, as well as monitoring and supervision of the action plan. It is crucial that this steering group, which would represent all stakeholders in the rare disease ecosystem, include patient representatives as full members. This inclusion would ensure that the lived experiences and priorities of rare disease patients directly influence policy decisions.
- It is essential to close the gaps in patient care pathways, addressing inequalities across the EU and significant barriers to access to screening, diagnosis, treatment and timely and coordinated care, including social care and comprehensive and integrated psychosocial support, and ensuring that those who live with a rare disease are able to participate fully in community life.
- It is vital to integrate healthcare and social services for patients with rare diseases, and to recognise those living with rare diseases as a disadvantaged group, ensuring that they receive appropriate allowances and benefits, much in the same way as people with disabilities.
- Europe needs to address the huge shortage of healthcare staff in the EU, including those working on mapping rare diseases. Existing human resources for patients are under disproportionate and extreme pressure in many countries, so further analysis and discussion is needed in this area.
- To significantly improve diagnostics and therapies for people with rare diseases in Europe, we need to strengthen early diagnosis through advanced molecular technologies and digital health, reinforce referral networks, promote collaborative research and data sharing, increase awareness and training among healthcare professionals, and ensure equitable access to care for all patients.
- It is important to take a holistic approach to healthcare, rather than the current fragmented approach that overly focuses on individual specialised areas that are not interconnected.
- Research on orphan drugs should be encouraged and adapted to daily practice.

- To strengthen diagnostic capacities, specific training is needed not only for healthcare workers, but also for social workers and social welfare staff, in order to enable them to care more effectively for people with rare diseases, help them in their daily lives and meet their psychosocial needs.
- Teachers and educators in childcare facilities and schools should also receive training to promote prevention and diagnostics.
- It is crucial that Member States and the European Commission join efforts to secure and extend funding for rare diseases in order to ensure the sustainability of all the targeted tools and models developed under JARDIN. This collaborative approach would ensure that the necessary resources are available to support ongoing research, improve diagnostics and therapies, and maintain equitable access to care for all patients across Europe.
- It is of paramount importance that ERNs be better integrated into all relevant sectors of national healthcare systems.
- Supporting and enhancing the implementation of national plans on Rare Diseases, with a coordination role for the EU, is crucial.
- Collaboration and exchanges with international organisations are vital for advancing rare disease initiatives. The World Health Organization's development of a Global Network for Rare Diseases (GNRD) in collaboration with Rare Diseases International (RDI) is a promising step forward. This initiative aims to replicate and adapt the successes of the ERNs on a global scale, leveraging digital innovations to pool expertise and improve accessibility.

A European action plan for rare diseases should:

- contain common and measurable objectives that directly address unmet needs and ensure equal opportunities in all Member States through improved health outcomes (reduced diagnostic time) and reduced inequality, and by boosting innovation;
- include measures to support national, regional and local health authorities and civil society organisations (patient organisations, among others) in their efforts to provide access to high-quality and affordable rare disease care;
- include SMART targets that can be achieved by 2030 to enable the diagnosis of rare diseases within one year of the onset of symptoms;
- introduce an effective horizontal coordination model for Commission services dealing with rare diseases and cross-border healthcare;
- encourage agreements with Member States on the content, updating, application and monitoring of national plans for rare diseases;
- include the collection and dissemination of best practices across the European Union; guarantee financing and feasibility, ensure the planning of EU health programmes, Horizon Europe and other financial programmes and ensure significant budgetary resources for health in the 2028-2035 multiannual financial framework;

- give guidance and earmark funding (EU4Health programme and the research programme) for initiatives aimed at providing better access to diagnosis, care and support for patients and families;
- include targeted incentive mechanisms to promote research and innovation, particularly in the areas of advanced diagnostics, precision medicine and gene therapy.

2. On uniting civil society and patient associations to improve the health and social situations of people with rare diseases

- It is crucial to prioritise the realistic needs of people living with rare diseases. All information and support should be tailored to these needs to ensure that patients receive the most relevant and effective care.
- It is also crucial in the fight against rare diseases to provide the right information. To ensure that patients and their families are well-informed, the information they receive must be adequate, accurate, real, medically approved, understandable and accessible.
- Cooperation among non-governmental organisations, researchers, healthcare professionals and decision-makers at local, national and European level is key. Building a common platform that is easy to use and accessible to all parties can facilitate effective collaboration.
- It is essential to integrate psychosocial support into healthcare, especially considering the long journey many rare disease patients face before receiving a diagnosis.
- An independent, transversal and global approach, supported by appropriate funding, is essential for addressing rare diseases effectively. This can be best achieved through national plans that actively involve patients and associations.
- The involvement of empowered patient organizations is crucial for the holistic, cross-sectoral and integrated management of rare diseases. These organisations can provide valuable insights and support in various areas, such as healthcare, disability and social affairs, education, work, family life and housing. By involving patient organisations in these areas, we can create a more supportive and inclusive environment for rare disease patients.
- A collaboration is needed between the public and private sector in order to tackle rare diseases.
- Civil society organisations are drivers for change, as they strive to create awareness and provide support and solutions for the challenges that people living with rare diseases face. The rare disease national alliances are key stakeholders in national plans, bringing expert knowledge and concrete solutions to the table.
- The rare disease national alliances need European initiatives, such as a comprehensive action plan for rare diseases, as a springboard for national efforts. It should be reinforced at European level in order to integrate health, social care, research and data management policies; promote access to diagnosis and treatment; empower patients; ensure cross-

border healthcare; create clear, measurable goals; and to set the course for anyone who can help address all unmet needs throughout the journey of patients with rare diseases.

- It is of utmost importance that civil society organisations – particularly patient organisations – be involved in monitoring and scrutinising national policies and the implementation of these.

3. On the progress made and challenges ahead for the European Health Data Space (EHDS)

- The EHDS offers great potential to address the unique challenges in the field of rare diseases. By harmonising data standards and connecting platforms at European level, the EHDS can enhance diagnosis, research and treatments for rare diseases. While national implementation is primarily the responsibility of Member States, this cannot succeed without relying on existing best practices and professional cooperation at the European level.
- It is of utmost importance to support and implement a pilot of a common European methodology and model for data interoperability for secondary use under the framework of the EHDS Regulation, and to facilitate data sharing for primary use between accredited medical teams and ERNs members.
- In order to strengthen the ERNs and ensure they are integrated into Member States' health systems, a European Health Data Space that is interoperable within the EU needs to be put in place.
- Patients must have control over their personal health data. They should be provided with the possibility to update sharing preferences and to receive clear explanations on how their data is used in care. Ethical management strengthens trust and enables the effective use of primary data to deliver better care.
- The EHDS offers a great potential for patients with rare diseases by enabling faster diagnoses, improving cross-border care and driving research. However, to truly succeed, it must prioritise ethical governance, actively involve patients in decision-making and give them control over their data. Only by balancing these elements can the EHDS transform healthcare while safeguarding patient rights and trust.
- Driving engagement is essential. Ensuring patients and healthcare providers understand and interact effectively with the EHDS requires education and support to bridge gaps in understanding.
- It is only if citizens and patients are able to trust the EHDS system that there is a prospect of success. All stakeholders, including healthcare providers, policymakers and technology developers, must therefore collaborate to ensure that the EHDS achieves its full potential, respecting patient rights and advancing public health.
- ERNs represent an excellent operational use case for the EHDS.
- ERNs can use health data for both primary purposes (direct patient care) and secondary purposes (research, policy-making and improving healthcare systems). ERNs are at the

forefront of leveraging health data, making them pioneers in the European health data ecosystem. They can set standards and best practices for other entities to follow.

- The superposition of ERNs and the EHDS missions with primary (Clinical Patient Management System) and secondary (ERNs-Registries) uses of health data can significantly enhance the management of rare diseases.
- International Classification of Diseases (ICD) codes for rare diseases are used in routine health statistics. However, these useful codes do not cover all rare diseases, and distinct ICD codes do not exist for each rare disease. A remarkable proportion of rare diseases do, however, have useful ICD codes. Epidemiological evaluations for those rare diseases are possible thanks to cause-of-death statistics and discharge records used by the National Health Insurance Fund. Both systems cover the whole population in Hungary. These indicators are therefore informative and valid in describing the epidemiological trends for a significant proportion of rare diseases.
- A common language is needed to achieve high-quality data on rare diseases at national, trans-national and European levels (including on undiagnosed cases). This would ensure consistency and comparability of data across different regions and systems.
- The significance of a unified language for rare diseases was acknowledged in the 2009 Council recommendation, and the European Commission has continuously endorsed the use of ORPHAcodes since then. The next crucial step is to make a definitive decision and demonstrate strong political commitment to consistently implement and use ORPHAcodes across all relevant systems.
- The Orphanet portal, which is responsible for developing and updating the ORPHAcodes system for coding rare diseases, is a vital resource. However, its current availability in only seven EU languages limits its accessibility. Establishing a new EU agency that provides support and consultation in all official EU languages would greatly enhance its utility. This would ensure that all stakeholders, regardless of their language, can access and benefit from the information and services provided.

These conclusions and recommendations will be disseminated to all participants, to the European Parliament and to the European Commission, as well as to the health ministries of the EU Member States. They will be specifically forwarded to the Hungarian Presidency and to the next EU trio of Presidencies.

Annex

PROGRAMME

08:30 | WELCOME COFFEE AND REGISTRATION

09:00 – 09:40 | OPENING SESSION

Moderator: Claude Rolin, EESC member

- **Baiba Miltoviča**, President of the Section for Transport, Energy, Infrastructure and the Information Society, EESC
- **Péter Takács**, State Secretary for Health, Hungary
- **Antonio Parenti**, Director, DG SANTE, European Commission (video message)
- **Tomislav Sokol**, Member of the European Parliament (video message)

09:40 – 11:20 | SESSION 1

How to achieve a comprehensive European action plan on rare diseases by 2030?

Moderator: Donata Meroni, Head of Unit, Health Monitoring and Cooperation, DG SANTE, European Commission

Setting the scene

- **Ágnes Cser**, EESC rapporteur: presentation of the EESC opinion at the request of the Hungarian Presidency *Leaving No One Behind: European Commitment to Tackling Rare Diseases*
- **Virginie Bros-Facer**, Chief Executive Officer, EURORDIS
- **Haris Hajrulahovic**, World Health Organisation Representative to Hungary

Discussion with the participation:

- **María Fernández García**, Deputy Director-General for Public Health and Quality, Health Ministry, Spain
- **György Pfliegler**, Head of Division, University of Debrecen, Hungary, Member of the ERN Board of Member States (BoMS)
- **Till Voigtländer**, Coordinator, Joint Action JARDIN
- **Luca Sangiorgi**, ERN BOND Coordinator, Coordinator of the ERNs Coordinators Group
- **Mária Judit Molnár** Director, Institute of Genomic Medicine and Rare Disorders, Semmelweis University, Hungary

Debate

11:20 – 13:00 | SESSION 2

How can civil society and patient organisations join forces to improve the health and social situation of people living with rare diseases?

Moderator: Alain Coheur, EESC Rapporteur on *Ensuring strong European solidarity for rare disease patients*

- **Borszékiné dr Cserháti Erika**, Érintettek Parents Association, Hungary
- **Daniel Theisen**, Director, ALAN, Luxembourg
- **Gábor Pogány**, President, RIROSZ, National Association of People with Rare and Congenital Disorders, Hungary
- **Jean-Philippe Plançon**, Vice-Chairman, *Alliance Maladies Rares*, France
- **Lene Jensen**, Chief Executive Officer, Rare Diseases Denmark
- **Annalisa Scopinaro**, President, UNIAMO, Italy

Debate

13:00 – 14:00 | LUNCH

14:00 – 15:30 | SESSION 3

For a European Health Data Space – progress made and challenges ahead

Moderator: Lech Pilawski, EESC member

- **Bas Nijhuis**, Health Counsellor, Permanent Representation of Netherlands to the EU
- **Tamás A. Kovács**, Health Informatics Service and Development Centre, Hungary
- **Stefan Živković**, National Organisation of Rare Diseases of Serbia
- **Hélène Dollfus**, Coordinator of ERN-EYE
- **János Sándor**, Head of the Institute of Public Health and Epidemiology, University of Debrecen, Hungary
- **Ana Rath**, Director, Orphanet

Debate

15:30 – 16:00 | CLOSING SESSION

Next steps and commitment to change

Moderator: Enrique Terol García, Health Counsellor, Permanent Representation of Spain to the EU

- **Mónica García Gómez**, Minister for Health, Spain (video message)
- **Judit Bidló**, Deputy State Secretary for the Professional Management of Health, Hungary
- **Urszula Demkow**, Undersecretary of State, Ministry of Health, Poland
- **Ágnes Cser**, EESC rapporteur