Conclusions and recommendations

The conference, organised by the EESC in cooperation with the Spanish presidency of the Council of the EU and the authorities of the Basque Country, aimed to take stock of what has been achieved and address some of the main challenges facing the EU policy framework on rare diseases and European Reference Networks (ERNs).

The conference responded to both the call for action issued by the Czech presidency of the Council of the EU and the political priority of the previous trio of presidencies. Plans are already underway to keep up this project under the Belgian and Hungarian presidencies.

Four main challenges were addressed:

- Strengthening and integrating ERNs and national networks of highly specialised providers: what has been learned from the five-yearly evaluation of the ERNs and what is planned for the Joint Action on integration of ERNs;
- Integrating social and health care for patients with rare diseases: the burden on families and unmet social and health care needs;
- Research priorities and resources: what is not covered? The opportunity offered by the European Health Data Space Regulation;
- Affordability and development of new treatments and therapies: how to improve access to safer, more effective medicines while ensuring that they are affordable and healthcare systems are sustainable.

The conference brought together stakeholders from the rare diseases community (civil society organisations, patients' associations, medical practitioners, researchers, healthcare managers and policy makers), health ministers and representatives of the EU institutions (European Economic and Social Committee, European Parliament, European Commission, European Council).

The conference sought to ensure continuity in terms of the work and political commitments of EU Member States and EU institutions since 2004, given that in recent years the EU has implemented a range of measures spanning various policy areas and providing a comprehensive approach to rare diseases. These measures include:

- establishing the 24 ERNs for Rare Diseases connecting rare diseases experts and specialised healthcare providers from across Europe in 2017;
- the future joint action by Member States, which aims to integrate the ERNs into national healthcare systems;
- establishing the European Platform on Rare Disease Registries and 24 patient registries under the ERNs;
- the EU's pharmaceutical reform, which aims to give companies more incentives to develop more effective and affordable medicines, including for patients living with rare diseases;
- the European Health Data Space legislation which would also benefit patients with rare diseases by making real world health data available for research, innovation and policy making;
- the planned Partnership on Rare Diseases under the Horizon Europe Programme.

The conference gave more visibility to the need to fund research into rare diseases, boosted awareness among civil society at EU level and in the EU institutions and Member States, conveyed the views of patients' associations, and cited Amyotrophic Lateral Sclerosis as an example of the importance of activism by patients' organisations. In all, the conference brought together 50 speakers, 36 EESC members, the president and several members of the Economic and Social Council of the Basque Country, two Members of the European Parliament and around 900 participants from all over Europe, both online and physically. The event, which was sponsored by the Spanish presidency of the Council of the EU, was opened by the EESC president, Mr Röpke, the Spanish Minister for Health, Mr Miñones, the Basque Minister for Health, Ms Sagardui, and the director-general of the European Commission's DG Sante, Ms Gallina.

The main conclusions and recommendations of the Conference are as follows:

1. On the organisational models and good practices regarding social and health care for rare diseases

- Rare diseases are a major burden for patients and their families. Centralised care coordination, ideally in institutions specialising in rare diseases, is crucial and increases the likelihood of a longer, better life for patients. Providing patients and their families with honest, reliable information is key as it gives them hope.
- The treatments are expensive and complex and should be addressed at European level, to help cover costs not met by national healthcare funding mechanisms.
- Recent developments, including pilot projects, have shown that it is possible to adapt professionals and healthcare systems to a complex public health strategy which will seek to reduce mortality of young children and address inequality in the field of health and well-being.

- Examples show that national rare disease plans can improve treatment, including through better data management and increased focus on innovation.
- The early diagnosis of rare diseases is crucial for future joint actions.
- A European Action Plan on Rare Diseases needs to be set up by the time the new European Commission takes office in 2024, tasked with:
  - making the implementation of national plans more efficient;
  - allowing for effective data collection;
  - enhancing cooperation between healthcare systems and agencies across Europe.
- A European Solidarity Fund should help offset unequal access to health services by covering costs not met by social security systems and other funding mechanisms.
- Decades of pilot projects on early screening of rare diseases must be followed by structured, system-wide public health programmes and strategies, which give priority to early diagnosis by scaling up pre-conception and pre-natal screening and screening of newborns.
- The next step is to step up the focus on innovation. An EU strategy built around the ERNs is needed to make national plans more efficient, allow for effective data collection and enhance cooperation between health bodies in Europe.
- Rare diseases are forerunners for precision medicine applied to common diseases, and rare diseases experts are the drivers of modern medicine.
- Rare diseases are high on the agenda in Europe, and the European Commission has already done a good job by encouraging Member States to develop their own national plans and developing the ERNs. The time has now come to devise a European action plan setting out common goals to strive for perfection in this field.

2. On the consolidation, future and integration of European Reference Networks into EU health systems

- ERNs should be further strengthened and integrated into general healthcare systems, thus making it easier for people to find the best treatment for their disease in good time, as envisaged by the Joint Action on integration of ERNs.
- A cultural shift in healthcare is needed to break down silos and promote integration.
- It is important to have fair, equitable access to healthcare for all, regardless of the rarity of the disease in question, concentrating knowledge and expertise in specialised centres.
- Healthcare planning must be based on patients' needs and not solely on the availability of therapies.
- Addressing rare diseases is not just a healthcare matter: it is also a reflection of European values and solidarity within the European Union.
- ERNs are essential for knowledge creation and sharing, structured collaboration and specialised healthcare in rare diseases and should therefore be seen as an investment rather than a cost, as they improve patients' health and promote prevention.
- A patient-centric approach is key: the goal is to enable expertise and knowledge to travel to patients, rather than having patients travel for specialised care.
- Financial sustainability is a major challenge, especially as ERNs aim to shift budgetary responsibility toward Member States while maintaining European support and developing efficient reimbursement models in the event of cross-border healthcare.

- Addressing the diversity of healthcare systems across the 27 Member States is a significant challenge, as each country has its own unique system.
- Developing comprehensive and interoperable European health data infrastructure is essential.

3. On ways to generate, exchange and apply knowledge

- Information on rare diseases is essential for understanding the origin, prognosis and evolution of the diseases and for solving urgent problems concerning time to diagnosis and access to treatment.
- The role of patients has been instrumental in drawing public attention to rare diseases, generating valuable data and increasing scientific interest. This has also motivated European policies and mechanisms, such as the ERNs which have a significant impact at national level and should be maintained.
- ERN registries provide a unique European platform linking real world data on excellence in complex care to research. Priority must be given to standardising all registers and ensuring the interoperability of data on rare diseases in order to guarantee that knowledge is shared and improve the management of rare diseases.
- Rare diseases are not part of the medical curriculum; it is therefore paramount that teaching programmes be amended to increase awareness of rare diseases.
- ERNs have developed all the necessary tools to increase knowledge of rare diseases and contribute to earlier diagnosis and appropriate treatment; they are able to carry out all core activities and clearly demonstrate structural pathways to science.
- The European Rare Disease Research Coordination and Support Action consortium, of which all 24 ERNs are part, is a very successful start-up that now needs to mature into professional organisations for 30 million rare disease patients in the EU.
- Clinical practice guidelines are important for reducing inappropriate care, minimising geographic variations in practice patterns and making effective use of healthcare resources. However, the development of guidelines for rare diseases presents many challenges owing to limited data availability and scarce professional knowledge and experience due to low disease prevalence.
- A more straightforward version of clinical practice guidelines, adapted to the various healthcare systems in Europe, must be developed and more widely disseminated in order to improve the level of care provided to patients.
- Patient-friendly versions of clinical practice guidelines and patients' lifestyle guidelines could play a major role in empowering patients in the management of their disease and supporting shared decision making with healthcare professionals.
- ERNs are an opportunity to develop clinical practice guidelines for rare diseases, as ERNs play a role in knowledge generation and registries and include both expert healthcare professionals and patients. In addition, ERNs are an ideal structure for disseminating, implementing and adapting clinical practice guidelines for both healthcare professionals and patients (development of lay version).
- The ERNs have developed a joint strategy for training and education to reconcile the increasing need for awareness about rare and complex diseases and the lack of specific training in the medical curriculum. Rare diseases desperately need a young generation of

medical professionals who are fully informed about rare diseases in terms of diagnosis, management and treatment. The ERNs have created a model that could help establish a European education programme in the area of rare diseases and complex conditions.

- However, new challenges need to be addressed: 1) interest in rare diseases drops in the wake of pandemics, 2) the shortage of healthcare professionals across the EU and 3) the need to master new technologies at molecular level and in particular at IT level (artificial intelligence); this requires collaboration with other professional profiles that are sometimes less closely related to the medical world.
- An evidence-based newborn screening strategy is also key. The ERN NBS Expert Platform is free of national bias and provides reliable, high-quality information to support decision making at national level. ERNs have the necessary expertise and play a key role in promoting equal access to newborn screening for rare diseases in the EU. The presidencies of the Council of the EU and the Commission must continue to support newborn screening as a key component of future EU policy on rare diseases.

4. On the remaining challenges linked to research into rare diseases

- Progress has been made in improving the lives of people with rare diseases, but serious challenges remain: factors such as the comparatively low number of patients for each disease, the limited funding for research, the lack of concerted European efforts and restricted access to data make rare diseases a difficult area to research. As a result, it takes an average of five years to receive a rare disease diagnosis.
- Emphasis must be placed on the importance of collaboration and data sharing, enhanced interoperability in data exchange, improved study designs and advanced genomic analysis in rare disease research.
- Coordination is needed between basic, clinical, transitional and social research due to the systemic nature of many rare diseases.
- Bridging the gap between patient needs and regulatory requirements will require:
  - harmonisation of regulatory pathways to facilitate rapid and equal access to diagnosis and therapies for rare diseases;
  - enhanced communication between healthcare professionals, patients, carers and experts to enhance the care of rare disease patients through the ERNs.
- A number of factors can speed up progress in rare disease research: efficient use of research, ensuring high standards and accessibility of data registries, changing regulations to enable non-sponsored clinical trials on rare diseases, accurately codifying rare diseases, guaranteeing data fairness and interoperability at data source level, adopting advanced genomic analysis techniques and prioritising data security and high standards in healthcare systems.
- In terms of collaboration at European level, the focus should be on:
  - improving study designs for the approval of medicines for ultrarare diseases in close collaboration with ERNs;
  - developing a common European framework for promoting data interoperability;
  - facilitating data sharing between accredited medical teams and ERN members;
  - ensuring that the Member States, the European Commission and research funders provide the resources needed to maintain knowledge bases on rare diseases.

These conclusions and recommendations will be disseminated to all participants, the European Parliament and the European Commission, as well as to the health ministries of EU Member States. They will be specifically forwarded to the future Belgian presidency of the Council of the EU, which has already undertaken to enhance the well-being of individuals affected by rare diseases, as well as to the Hungarian presidency as part of the current trio.