Advancement of Treatments FOR RARE DISEASES

European Economic and Social Committee

The European Life Science Strategy along with the Strategy on Research and Technology infrastructures with focus on the treatment of rare diseases

(*exploratory opinion requested by the Cyprus Presidency of the Council of the EU*)

Ass. Prof. Milena ANGELOVA, PhD

Organisers: THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS; DEPUTY MINISTRY OF RESEARCH, INNOVATION AND DIGITAL POLICY REPUBLIC OF CYPRUS; CY2026.EU CYPRUS PRESIDENCY OF THE COUNCIL OF THE EU

Funded by the European Union

The conference is funded by the European Union (project no 101297283)

Advancement of Treatments FOR RARE DISEASES

European Economic and Social Committee

The European Economic and Social Committee:

- Backs the call of the European Parliament and asks for a **comprehensive, goal-driven European Action Plan for Rare Diseases to be swiftly adopted**, including by reviving and **further developing the Tartu call for a healthy lifestyle and embracing the 'One Health' approach** as to enhance the rare disease pathway and foster European collaboration in this critical area;
- Suggests the Action Plan to include clear and measurable objectives that directly address unmet needs, ensure equal opportunities across all Member States, and **improve health outcomes by reducing diagnostic times, minimizing inequalities, and fostering innovation;**
- Asks for **swift alignment and clarification of how 'rare diseases' are define -** to support the coordinated implementation of EU initiatives in this area (including the European Strategy on Research and Technology Infrastructures and Choose Europe);
- Underlines that **Research and technology infrastructure** (including that focused on rare diseases) **is a key enabler** for the development of the life sciences and the longevity economy;
- Suggests **cross-cutting support actions** to include the promotion of talented and young entrepreneurs, data integration, IP protection and innovation financing in the 2028-2034 multiannual financial framework (MFF);
- Sees the development of the **European Health Data Space (EHDS), 1+MG genomic and BBMRI-ERIC biobanking infrastructure and the ELIXIR bioinformatics ecosystem** as critical for delivering precision medicine, genomic diagnostics, molecular epidemiology and translational research, as the EU life sciences are evolving into data-driven sciences.

Organisers: THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
DEPUTY MINISTRY OF RESEARCH, INNOVATION AND DIGITAL POLICY REPUBLIC OF CYPRUS
CY2026.EU CYPRUS PRESIDENCY OF THE COUNCIL OF THE EU
Funded by the European Union
The conference is funded by the European Union (project no 101297283)

European Economic and Social Committee

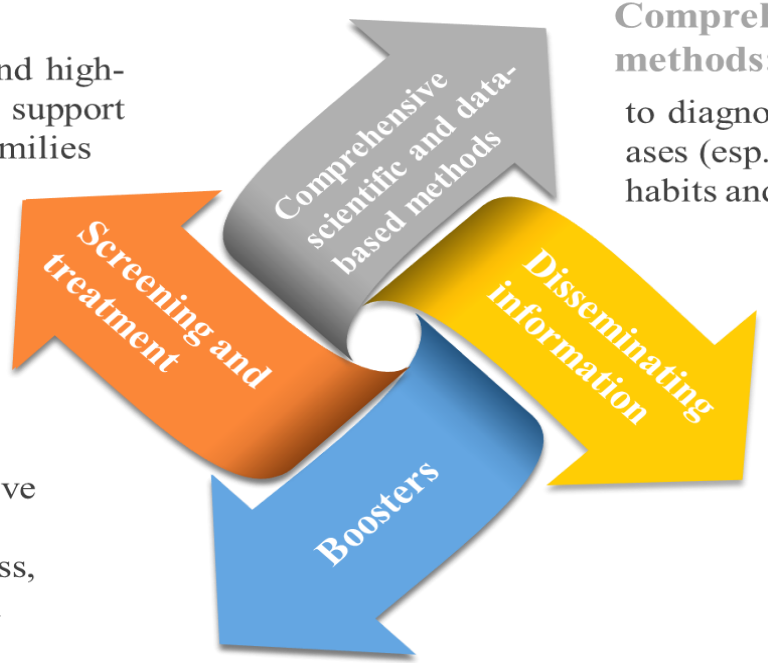
❖ **Building blocks of the holistic approach include**:

Screening and treatment:

effective and accessible treatment; and high-quality care and accessible support infrastructure for patients and their families

Comprehensive scientific and data-based methods:

to diagnose and understand the causes of diseases (esp. rare diseases, e.g. genetics, unhealthy habits and lifestyles, pollution, stress, etc.)



Boosters

- targeted development of R&I;
- sufficient funding;
- simpler, future-proof legislative framework anchored in reality;
- promotion of competitiveness, excellence, talent and digitalisation

Disseminating information:

on prevention and early diagnosis (e.g. pre-conception)

❖ **Translational centres**, specialised in rare diseases, to be built in each Member State so as to gather the medical community, patients, social workers and bio-banks, and ensure that they are coordinated and supported at EU level. They can be used to build a more structured approach in tackling rare diseases and to set clearer goals and priorities, applying the holistic and integrated approach;

❖ **Strengthening the European Reference Networks (ERNs)** and increasing their outreach.

Advancement of Treatments FOR RARE DISEASES

European Economic and Social Committee

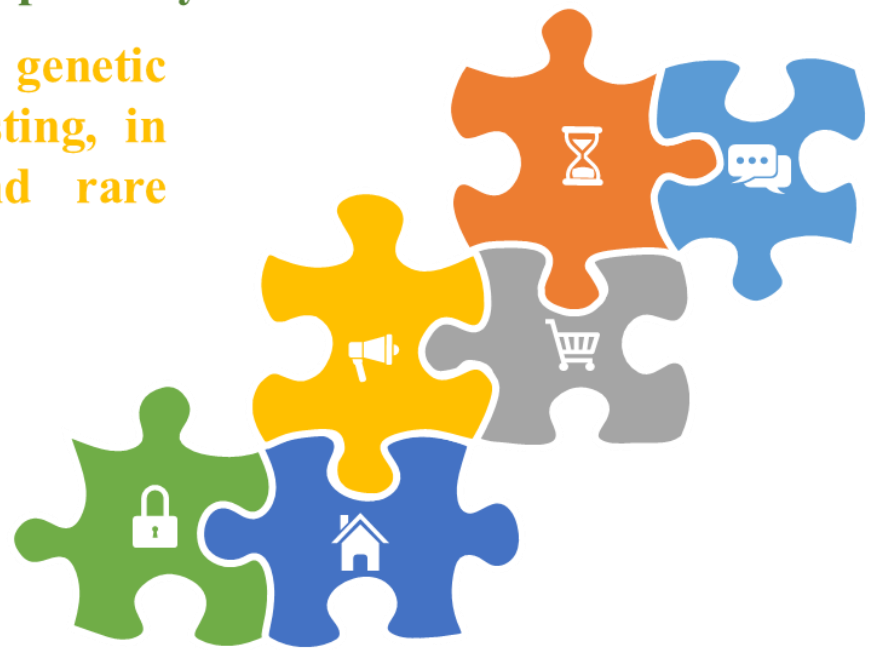
To address the challenges related to the diagnosis and treatment of rare diseases, the **EESC suggests focus on**

1. Establishing individual patients pathways

2. Early effective diagnosis, incl. genetic screening and preconception testing, in cooperation with biobanks and rare disease organisations.

3.Translation of scientific advances into therapeutic solutions by accelerating approval pathways, including for orphan drugs

4. Aligning pediatric and adult treatment pathways to ensure a lifecycle approach



5. Ensuring sustainable, long-term funding and the coordinated development of genomic research and infrastructure, including sequencing centres, bioinformatics platforms, secure cloud environments and reference databases. Training of specialists (clinical genetics specialists and counsellors).

6.Improving public understanding of rare diseases, early detection and prevention and the overall culture of pursuing a healthier lifestyle.

Organisers: THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
DEPUTY MINISTRY OF RESEARCH, INNOVATION AND DIGITAL POLICY
REPUBLIC OF CYPRUS
CY2026.EU CYPRUS PRESIDENCY OF THE COUNCIL OF THE EU
Funded by the European Union
The conference is funded by the European Union (project no 101297283)

- EU health systems differ considerably: need for a **model to prioritise coordination** rather than uniformity, using regional specificities to test, validate and scale health innovations:
  - ✓ Support for alignment of interfaces, coordination of objectives and structured exchange of evidence and good practices;
  - ✓ Ensure interoperable data flows and comparable clinical and regulatory evidence, including through EDHS-compliant data environments and shared methodological frameworks for JCA;
  - ✓ Strategic priority on ATMPs development, incl. gene, cell and tissue therapies: they are among the most significant therapeutic groups for rare diseases, particularly those of monogenic origin;
  - ✓ EMA, health technology assessment bodies and 'notified bodies' to coordinate evidence requirements, early dialogue and consultations more quickly and improve the flexibility of standardised procedures and regulatory sandboxes, so that innovations can be evaluated under local conditions; that evidence travels, even if practices differ; and that regional and clinical JSAs/JCAs feed into EU-level understanding;
- **Genomics and molecular biology are the scientific foundation** of contemporary life sciences: to be integrated as a cross-cutting component of EU health and research cooperation. **National coordination points to monitor** sequencing infrastructure, research capacity, workforce skills and data integration readiness, ensuring alignment with national health strategies, digitalisation efforts and the 1+MG initiative;
- **ERNs to be reinforced and integrated** with the EHDS and genomic initiatives;
- **Orphanet to be strengthened** so that it can publish its work in all EU official languages and ensure that patients and professionals have the information they need.

The conference is funded by the European Union (project no 101297283)

Advancement of Treatments FOR RARE DISEASES

European Economic and Social Committee

Rare diseases:

- Access to rare disease therapies varies across EU - **harmonised HTA frameworks, joint clinical assessments and strengthened cross-border mechanisms are essential for ensuring equitable access**;
- Commitment to **interoperable data formats**, **harmonised consent models and metadata standards** for rare disease data, in line with the EHDS;
- **Common standards** for genomic data generation, biobank sample processing, metadata models and mechanisms for linking records between biobanks and clinical registries are vital in order to harmonise biobank procedures and integrate genomic data into biobanking and registry infrastructure relevant to rare diseases;
- **Innovation potential** - constrained by structural regulatory barriers that delay market access, increase compliance costs and reduce the EU's attractiveness as a location for research and investment. Core obstacles to be removed:
  - ✓**Streamlining and simplifying** overly lengthy, complex and duplicated authorisation processes, addressing the lack of proportionate and risk-based pathways;
  - ✓**Removing inefficiencies** in the pre-submission, risk-assessment and risk-management phases;
  - ✓**Scaling the disproportionate requirements** under the Transparency Regulation;
  - ✓Providing **robust impact assessments and proportionate rules** and adequately considering the effects of new legislation and administrative procedures on competitiveness and SMEs; and
  - ✓Incentivising Member States to apply **mutual recognition** in order to ensure a shared minimum level of compatibility.

Organisers: THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
DEPUTY MINISTRY OF RESEARCH, INNOVATION AND DIGITAL POLICY REPUBLIC OF CYPRUS
CY2026.EU CYPRUS PRESIDENCY OF THE COUNCIL OF THE EU
Funded by the European Union
The conference is funded by the European Union (project no 101297283)

Modernise the legislative framework and facilitate market and global access for biotech products, taking into account the broad spectrum of health-related biotechnology, including products, technologies and industrial applications:

- **Continued support to advanced and high technology** such as vaccines, CAR-T cell therapies and personalised medicine;
- **Strong biotechnological foundation** (especially regarding immunology and oncology), vital role of biosimilars as established therapeutic options for chronic and life-threatening diseases, which contribute significantly to patient access and healthcare system sustainability;
- **Clearer, predictable and efficient rules** tailored to patient needs and the realities of biotech innovation can boost the advancement, development and application of health-related biotechnology (including ATMPs, molecular diagnostics and novel therapeutic modalities) with a view to excelling in prevention, diagnostics and treatment;
- Member States to address **systemic inefficiencies in clinical trial authorisation and management** and to develop mutual trust in the assessment of reporting;
- **Expansion of the EMA's DARWIN-EU** federated data network would enable broader use of real-world data in decision-making, on the condition that EMA and national authorities are resourced with top expertise and cloud-based infrastructure to optimise workflows;
- **Strengthening intellectual property protection and regulatory data protection**, and ensuring efficient authorisations and audit processes (in research and manufacturing, e.g. import licences for biotech platforms, SHO and environmental permits) can support scale-up. Access to venture capital and funding instruments.

The conference is funded by the European Union (project no 101297283)

Thank you!

Organisers: THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS
DEPUTY MINISTRY OF RESEARCH, INNOVATION AND DIGITAL POLICY
REPUBLIC OF CYPRUS
CY2026.EU
CYPRUS PRESIDENCY OF THE COUNCIL OF THE EU
Funded by the European Union
The conference is funded by the European Union (project no 101297283)