European Reference Networks

S&D Group of the Progressive Alliance of Socialists & Democrats in the European Parliament

BRAINS FOR BRAIN

HLM 4RARE

HLM4RARE 2026

ERN COORDINATORS CALL

10 FEBRUARY, 2026

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HLM RARE 2025

The European Reference Networks, MEP Vytenis Andriukaitis (S&D, Lithuania) and the Brains for Brain Foundation, hosted in Brussels from 9-11 December 2025 the first three-day High-Level Meeting on a European Innovation and Care Ecosystem for Rare and Complex Diseases (HLM Rare 2025). A key milestone of this meeting was the launch of a community-led Declaration.

HLM RARE 2025: THE HIGH-LEVEL AGENDA

	DAY 1 RESEARCH AND INNOVATION	**DAY 2** EU INFRASTRUCTURE AND SKILLS	**DAY 3** POLICY AND FUNDING
Venue	**Residence Palace**		**European Parliament**
Structure of the day	• High-Level keynote speeches • Panel discussion 1: scene setting • Panel discussions 2-4: deep dive on a Declaration recommendation		• High-Level keynote speeches • Policy discussion 1-2: deep dive on a Declaration recommendation
Topics under discussion	▪ EU Leadership in Clinical Trials ▪ Integrated infrastructure for research, innovation & care ▪ Public Private Partnerships for research	▪ Newborn screening ▪ Real world evidence ▪ Artificial Intelligence ▪ Workforce development and capacity building	▪ WHO Resolution on Rare Diseases ▪ Early access models ▪ ERN's funding
EU Legislations under discussion	EU Pharmaceutical Package EU Regulation on ATMPs EU Clinical Trials Regulation EU Biotech Act	EU Regulation on Health Technology Assessment EU AI Regulation Act European Health Data Space European Union of Skills EU Life Sciences Strategy	Multiannual Financial Framework (2028-2034) EU Directive on the application of patients' rights in cross-border healthcare EU Biotech Act EU Critical Medicines Act

WE LAUNCHED THE DECLARATION ON A EUROPEAN INNOVATION AND CARE ECOSYSTEM FOR RARE AND COMPLEX DISEASES

What it is	The objective	The short term goal	The medium-to-long term goal
A political and strategic commitment to fundamentally transform the European ecosystem for rare and complex diseases. **A bold shared vision** for better alignment between research, innovation, health systems, patients and industry.	To **define clear strategic priorities** to set the direction for future action and mobilize strong political support from EU institutions, relevant agencies, national authorities, and the wider stakeholder community.	**The establishment of Working Groups,** tasked with developing concrete and measurable targeted Plans of Action to achieve these ambitions.	**Accelerate progress for rare and complex diseases**, notably by increasing the number of available treatments and by universalizing newborn screening and advanced diagnostics.

WE LAUNCHED THE DECLARATION ON A EUROPEAN INNOVATION AND CARE ECOSYSTEM FOR RARE AND COMPLEX DISEASES

8 Recommendations

- Prioritise the EU Action Plan on Rare Diseases with a Clear Governance unifying the Rare Disease Community
- Strengthen Workforce Development and Capacity Building for Rare Diseases and formalise the ERN Academy
- Accelerate Equitable Access to Diagnostics and enable Early Treatment Onset with Innovative Orphan Drugs for Unmet Medical Needs
- Foster EU Leadership in Clinical Trials for Rare Diseases through Inclusive Collaboration between Academy, Patient Groups and Industry to accelerate Innovation for people living with rare diseases
- Create, in each Member State, at least one comprehensive rare disease infrastructure cluster (CoRDIC) for research, innovation and care
- Boost real-world evidence generation by making Rare-Disease Data High Quality and Clinical Utility, Interoperable and Integrated under the European Health Data Space
- Explore new business models and mechanisms to prioritise equitable access to innovative orphan therapies and diagnostics, also supported by a European Guarantee Fund
- Ringfence ERNs funding under the 2028-2034 Multiannual Financial Framework

2025 EVIDENCES OF SUCCESS

500 people

250 registered for in-person attendance across the 3 days and + 250 watched the livestreaming each day

+ 80 speakers

were part of the programme, including 2 EU Commissioners, 1 MoH and 11 MEPs, as well as 6 EC and 2 EMA representatives

+60 organizations

signed the Declaration during or after the conference

POLITICO

HC Newsletter

Mentions from Politico during the days of our event

36 earned media

mentions on LinkedIn from different key stakeholders, including Emer Cooke and Hans Kluge

10,214

HLM Rare unique page views in December 2025 alone

HLM RARE 2025: SOME OF THE STAKEHOLDERS PRESENT

EU and national policy- and decision-makers

European Commission

HaDEA

EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

MINISTRY OF HEALTH
OF THE REPUBLIC OF LITHUANIA

ESPAÑA
REPRESENTACIÓN
PERMANENTE EN LA
UE

Medical and research community

European Reference Networks

EDERA
European Rare Diseases Research Alliance

ISNS
International Society for Neonatal Screening

ILAE-EUROPE

THE CYPRUS INSTITUTE OF NEUROLOGY & GENETICS

Realise D

Inserm

orphanet

EUROPEAN PAEDIATRIC NEUROLOGY SOCIETY

screen4rare

Bambino Gesù
OSPEDALE PEDIATRICO · IRCCS

JARDIN

SJD Sant Joan de Déu
Barcelona · Hospital

Patient community

EURORDIS
RARE DISEASES EUROPE

Childhood Cancer International
EUROPE

IPOPI
INTERNATIONAL
PATIENT ORGANISATION
FOR PRIMARY IMMUNODEFICIENCIES

CHILDREN'S TUMOR FOUNDATION
ENDING NF THROUGH RESEARCH
EUROPE

IMPSN
International MPS Network
Mucopolysaccharide and Related Diseases

fundacja SMA

Industry

EUCOPE
European Confederation of Pharmaceutical Entrepreneurs AISBL

efpia

ultragenyx

Chiesi

sanofi

DENALI
THERAPEUTICS

ACADIA

BIOMARIN

EVERSANA

HLM RARE 2025: THE HOSTS AND ERN COORDINATORS LEADING THE MEETING

In order of appearance: 1. HLM Rare 2025 – room at the Residence Palace; 2. HLM Rare 2025 – room at the European Parliament; 3. & 4. Maurizio Scarpa and MEP Vytenis Andriukaitis, hosts of the HLM Rare 2025, opening the meeting; 5. Ruth Ladenstein, PaedCan Coordinator, 6. Holm Graessner, ERN-RND Coordinator; 7. Mar Manu Pereira, ERN-EuroBloodNet Coordinator; 8. Luca Sangiorgi, ERN-BOND Coordinator; 8. Alexis Arzimanoglou, EpiCare Coordinator

HLM RARE 2025: SOME OF THE HIGH LEVEL POLICY-MAKERS INVOLVED

In order of appearance: 1. Ursula von der Leyen, European Commission President; 2. Roberta Metsola, President of European Parliament; 3. António Costa, President of the European Council; 4. Marija Jakubauskienė, Lithuanian Health Minister; 5. Olivér Várhelyi, European Commissioner for Health and Animal Welfare; 6. Ekaterina Zaharieva, Commissioner for Startups, Research and Innovation; 7. Teresa Ribera (Executive Vice-President for a Clean, Just and Competitive Transition; 8. Emer Cooke, European Medicines Agency, Executive Director

WANT TO KNOW MORE?

Click in one of the following boxes (in presentation mode)

CHECK OUT THE FINAL PROGRAMME AND SPEAKER LINE UP

WHATCH THE RECORDINGS

READ THE DECLARATION

CONSULT THE LIST SIGNATORIES

The Declaration remains open for signature at this link.

2026 PLAN

Building on the momentum and commitments generated in 2025, the Brains for Brains Foundation, together with the European Reference Networks and MEP Vytenis Andriukaitis are already planning for the next steps. In 2026, HLM Rare is evolving into the Health Leadership Mission for Rare Diseases (HLM4RARE), a strategic, mission-oriented forum designed to move from political alignment to coordinated delivery.

OBJECTIVES OF THE HEALTH LEADERSHIP MISSION FOR RARE DISEASES (HLM4RARE)

In 2026, the HLM4Rare will aim to:	
Main objectives	**Targets**
• **Garner community support for the Declaration**, including through a growing number of official endorsing organisations as well as individuals	• Growing from+60 to 500 endorsing organizations • Registration of European Citizens' Initiative and official launch at HLM4Rare 2026 meeting
• **Convene multi-stakeholder Working Groups** that will come together to develop targeted Plans of Action to achieve the ambitions set out by each of the recommendations outlined in the Declaration.	• Convening 8 WG, ensuring the involvement of 3-5 EU Institutions or MS representatives across the different WG as well key advocacy stakeholders that focus on more common diseases • Developing targeted Plans of Action (roadmaps)
• **Launch targeted Plans of Action** at launch at HLM4Rare 2026 meeting	• 500 in-person (depending on room availability) and 500 remote participants (across all days of the conference) • Participation of European Patient Advocacy Groups (ePAGs) from at least 14 EU Countries

GOVERNANCE

Core Team (CT)

- Maurizio Scarpa
- Ruth Ladenstein
- Mar Manu Pereira
- Holm Graessner
- Alexis Arzimanoglou
- Luca Sangiorgi
- Birute Tumiene
- MEP Vytenis Andriukaitis

- Chairing the working groups that will lead on development of the sub-action plans
- Shaping the HLM4Rare 2026 conference programme and preparatory activities based on the strategic direction provided by the SC.
- Overseeing day-to-day implementation of the work plan by the HLM Rare secretariat.

Steering Committee (SC)

Members include:

- ERN Coordinators
- EURORDIS
- ERNs Patient Advocates Delegation
- FESCA
- Children's Tumour Foundation
- Rare Disease International
- International MPS and Related Diseases Network
- European Business Summit
- EUCOPE
- International Society for Neonatal Screening
- Innovation for Global Health Institute
- IPOPI
- EFPIA
- Other partners & sponsors

- Provides strategic direction to CT, helping define priorities, themes and agendas
- Participates in Working Groups and HLM4Rare Activities (e.g., policy briefings)

Policy Ambassador Network (PAN)

Members (some still TBC):

- MEP Vytenis Andriukaitis
- Manuel Heitor
- Enrico Letta
- Michael Marmot
- Axel Pries

- Provides high-level political endorsement
- Promotes political action at a national and EU level
- Supports cross-fertilisation

OUR 4 KEY EXTERNAL MILESTONES

External

Rare Disease Day
28 February

Conference on Rare Diseases by the Cyprus Presidency of the Council of EU
5-6 March, Nicosia

ERN Meeting (led by the EU Commission)
26 March, Brussels

European Conference on Rare Diseases (ECRD) 2026
3-4 June, Prague

European Health Forum Gastein
29 September - 2 October, Bad Hofgastein (Austria)

3rd Policy session: World Health Summit session
11-13 October, Berlin

World Orphan Drug Congress Europe
26-28 October, Barcelona

HLM4Rare 2026 meeting
1 – 2, December, Brussels

Invitation only

1st Policy Briefing session
March, Brussels

2nd Policy Briefing session
June, Brussels

Objective: exchange on policy topics between selected HLM stakeholders, Members of the European Parliament and Commission officials on relevant topics for the realisation of the European ecosystem for rare and complex diseases and the completion of the European Health Union

During HLM4Rare. we aim to host an exhibition. This exhibition will showcase, for 8 rare and complex diseases, the transformative impact of scientific and policy progress, including innovative treatments. We will do this by showcasing, through real patient stories, elements such as quality-of-life improvements, extended life expectancy and impacts on HC systems.

DEEP DIVE ON WORKING GROUPS

About the Working Groups (WG)

Why	**To develop 8 targeted Plans of Action** to achieve the ambitions defined by the recommendations set out by the Declaration launched at HLM Rare. Each Plan of Action will be dedicated to one recommendation set out by the Declaration.
Co-Chairs	**2-3 ERN Coordinators per WG** whose responsibility will be to A) co-lead the development of the targeted Plan of Action, B) co-chair WG meetings, and C) co-lead on the preparatory work needed to deliver on A and B
Members	**Circa 10 stakeholders in each WG** including patient organisation representatives, medical societies, industry and EU Institutions or MS representatives
Targets	Involving 3-5 EU Institutions or MS representatives across the different WG
Members Workload	**Up to 12 hours throughout the year** Including 3-4 x 2h meetings throughout the year as well as 1h of offline before or after each meeting

Indicative timeline

The 8 Working Groups

WG 1: Prioritise the EU Action Plan on rare diseases with a clear governance unifying the rare disease community

ERN Coordinator Lead:
Alexis Arzimanoglou

WG 2: Strengthen workforce development and capacity building for rare diseases and formalise the ERN Academy

ERN Coordinator Lead:
Maurizio Scarpa

WG 3: Accelerate equitable access to diagnostics and enable early treatment onset with innovative orphan drugs and digital innovation for unmet medical needs

ERN Coordinator Lead:
Luca Sangiorgi

WG 4: Foster EU leadership in clinical trials for rare diseases through inclusive collaboration between Academy, ERNs, patient groups and industry to accelerate innovation for PLWRD

ERN Coordinator Lead:
Holm Graessner

WG 5: Create, in each Member State, at least one comprehensive rare disease infrastructure cluster for research, innovation and care (CoRDIC)

ERN Coordinator Lead:
Ruth Ladenstein

WG 6: Boost the generation of real-world evidence by ensuring that rare-disease data are high-quality, interoperable, and clinically useful, to accelerate biomarker discovery and support drug development in rare diseases within the EHDS and EU AI Act Regulation

ERN Coordinator Lead:
Mar Manu Pereira

WG 7: Explore new early access models and mechanisms to prioritise equitable access to innovative orphan therapies and diagnostics, also supported by a European Guarantee Fund

ERN Coordinator Lead:
Maurizio Scarpa

WG 8: Ringfence European Reference Networks funding under the 2028-2034 Multiannual Financial Framework

Coordinator:
MEP Vytenis Andriukaitis (S&D, LT)

Questions for discussion

As we deep dive in each WG, some questions for discussion:

1. **Who wants to co-lead some of these working groups together with the already assigned ERN coordinators?**
2. **Anyone else who should be invited to contribute to each?**
3. **If there is time: What should be the focus of the Plans of Action and of its 3-4 preparatory meetings? What key topics should be discussed?**

PRIORITY ACTION 1: EU ACTION PLAN FOR RARE DISEASES

THE CHALLENGE

- Rare disease policy remains fragmented across initiatives, legislations and funding streams, limiting coherence and impact.
- The absence of a single governance mechanism hinders alignment between EU institutions, Member States, ERNs and stakeholders.
- There is no shared framework to monitor implementation and results across rare disease policies at EU level.

WHAT THE DECLARATION CALLS FOR

- Adoption of a comprehensive EU Action Plan on Rare Diseases, anchored in a cross-cutting Rare Disease Mission.
- Establishment of a high-level Consultative Group to assess coherence, reduce regulatory overlaps and optimise cross-border healthcare frameworks.
- Development of a shared dashboard with clear indicators, targets and metrics to track progress and outcomes.

PRIORITY ACTION 1: HLM RARE 2025 PANEL

Panel discussion	Prioritise an EU Action Plan and Mission on Rare Diseases that unifies the rare disease stakeholder community
Conference day	Day 3: Policy & funding

Questions discussed

- How do you see the role of the ERNs in relation to innovation and competitiveness?
- What are the current building blocks on rare disease in the EU's portfolio of activities?
- The imperative for an overarching EU action plan on rare diseases has long been acknowledged. However, this has not been put on the agenda. What specific, immediate political steps are necessary to ensure its successful implementation?
- How can we collectively work to ensure the next Multiannual Financial Framework (MFF) effectively ring-fences dedicated funds for the European Reference Networks?
- How will this funding guarantee a cohesive and coordinated European approach that aligns rare disease research with patient care priorities?

Stakeholders involved

Moderator
Alexis Arzimanoglou
European Reference Network for all rare and complex epilepsies (EpiCare) Coordinator

Joanna Drake
Deputy Director-General for Planet, People and Science for Policy in the European Commission's Directorate-General for Research and Innovation (DG RTD)

Nikos Papandreou
Member of the European Parliament
S&D, Greece

Vlad Voiculescu
Member of the European Parliament
Renew, Romania

Leonidas A. Phylactou
CEO and Medical Director of The Cyprus Institute of Neurology and Genetics

Birute Tiumene
Co-Chair of ERN Board of Member States

Till Voigtländer
Coordinator of Joint Action on Integration of ERNs into National Healthcare Systems (JARDIN)

Avril Daly
President, EURORDIS

Alexander Natz
Secretary General of EUCOPE

Working Group 1: EU Action Plan on rare diseases

Co-chairs	• Alexis Arzimanoglou

Potential members

- **EURORDIS** representative
- **Birute Tiumene**, Co-Chair of ERN Board of Member States
- **European Economic and Social Committee** Representative
- World Health Summit Representative
- **DG SANTE** representative
- **Cypriot Council of the EU Presidency** representative
- **Ireland Council of the EU Presidency** representative
- **Lithuanian Council of the EU Presidency** representative
- **Joanna Drake**, Deputy Director-General for Planet, People and Science for Policy in the European Commission's Directorate-General for Research and Innovation (DG RTD)
- **Nikos Papandreou**, Member of the European Parliament (S&D, Greece)
- **Vlad Voiculescu**, Member of the European Parliament (Renew, Romania)
- Up to 3 e-PAGs
- Up to 3 other ERN coordinators on a voluntary basis
- Pharmaceutical industry representative(s)

Key take-aways from HLM Rare 2025

- A strong consensus emerged for a comprehensive EU Action Plan/Mission on Rare Diseases to overcome current fragmentation
- Emphasizing crucial elements like newborn screening, streamlined diagnostic processes, and making Europe a more attractive hub for clinical trials is essential for patient access to innovative therapies.
- ERNs are recognized as a European success story that needs continued, ring-fenced funding to fully realize their potential in care, research, and innovation.
- Future efforts must strategically align research (e.g., via ERDERA, JARDIN, , Biotech Act) with patient needs, foster public-private partnerships.

Working Group 1: EU Action Plan on rare diseases

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

Working Group 1: EU Action Plan on rare diseases

Topics for targeted Plan of Action

The Rare Disease Mission

Role of the ERNs in the Action Plan

Methodology, governance and Accountability Mechanisms

PRIORITY ACTION 2: WORKFORCE DEVELOPMENT

THE CHALLENGE

- Rare disease expertise remains insufficiently embedded in health education and training, contributing to delayed diagnosis and uneven care.
- Workforce development and knowledge exchange are fragmented and under-resourced, with limited structured EU-level coordination.
- Gaps in digital and AI-related skills limit the effective use of innovation and cross-border collaboration within ERNs.

WHAT THE DECLARATION CALLS FOR

- Formalise the ERN Academy as the EU framework for training, skills development and knowledge exchange in rare diseases.
- Embed rare disease education in medical and nursing curricula, aligned with the “Choose Europe” initiative and the Fifth Freedom.
- Strengthen EU and national investment in workforce capacity, including digital and AI-enabled skills, and support training for patients and families.

PRIORITY ACTION 2: HLM RARE 2025 PANEL

Panel discussion	Strengthening Workforce Development and Capacity Building for rare diseases and formalizing the ERN academy
Conference day	Day 2: EU Infrastructure and skills

Questions discussed

- How can the EU attract and retain the next generation of rare disease specialists?
- What role can the ERN Academy play in reducing disparities in rare diseases expertise?
- How can we better support the upskilling of general practitioners to improve early recognition and referral of people with rare diseases?
- What is currently needed to improve rare-disease specialists' skills and to better support their needs?

Stakeholders involved

Donata Meroni
Head of Unit B3 'Health monitoring and cooperation, Health networks', DG SANTE, European Commission

Cristophe Clergeau
Member of the European Parliament (S&D, France)

Vlad Voiculescu
Member of the European Parliament (Renew, Romania)

Alberta Spreafico
Senior Vice President Health Innovation, EVERSANA; President, Innovation for Global Health Institute

Ilaria Galetti
Vice-President Federation of European Scleroderma Associations & ePAG ReCONNET

Marta Mosca
European Reference Network of Connective Tissue and Musculoskeletal diseases (ERN ReCONNET) Coordinator

Maurizio Scarpa
MetabERN Coordinator, President and Founder of B4B Foundation

Jens K. Habermann
Director General Biobanking and Biomolecular Resources Research Infrastructure – European Research Infrastructure Consortium

Working Group 2: Workforce development and capacity

Co-chairs	• Maurizio Scarpa

Potential members

- BBMRI-ERIC
- EVERSANA
- DG EMPL
- DG SANTE
- Knowledge Generation cross-ERNs WG
- Up to 3 ePAGs
- Up to 3 other ERN coordinators on a voluntary basis
- Pharmaceutical industry representative(s)

Key take-aways from HLM Rare 2025

- Skills gaps drive delayed diagnosis, uneven care, weaker registries, fewer trials and slower research translation.
- ERN education already exists but is fragmented. This is why the ERN Academy is needed to unify and certify it.
- Talent retention needs clear career paths and support.
- GPs need rare-disease awareness and “red flags” training, as well as clear referral pathways.
- Digital and data competencies are now essential, but clinicians must be appropriately trained guide, validate and implement these tools.

Working Group 2: Workforce development and capacity

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

Working Group 2: Workforce development and capacity

Topics for targeted Plan of Action

- Formalisation and governance of the ERN Academy
- Standardised, certified training programmes for rare diseases
- Upskilling General Practitioners and frontline healthcare professionals
- Reducing geographical and expertise disparities across Member States
- Integration of digital and data skills into clinical training
- Linking training to research, registries and clinical trials

PRIORITY ACTION 3: EQUITABLE ACCESS TO DIAGNOSTICS AND EARLY TREATMENT

THE CHALLENGE

- Access to early and accurate diagnosis remains uneven across Member States, with significant delays for many people living with rare and complex diseases.
- Newborn screening programmes, digital tools and advanced diagnostics are implemented inconsistently, limiting their impact and scalability.
- Fragmented data collection and evaluation frameworks slow the responsible uptake of digital and AI-enabled solutions across care pathways.

WHAT THE DECLARATION CALLS FOR

- Accelerate equitable access to early diagnosis, including through the advancement of evidence-based newborn screening programmes across the EU.
- Support coordinated EU-level action to align rare disease data collection and enable the harmonised assessment and responsible use of digital and AI-enabled applications.
- Strengthen integrated care pathways, ensuring that early diagnosis translates into timely treatment onset and improved outcomes across Member States.

PRIORITY ACTION 3: HLM RARE 2025 PANEL

Panel discussion	Accelerating equitable access to screening programmes, advanced diagnostics and innovative orphan drugs
Conference day	Day 2: EU Infrastructure and skills

Questions discussed

- How can Europe accelerate equitable access to newborn screening technologies? And how to ensure the wide adoption of a minimum number of newborn screening programmes?
- What are the most promising population-based screening procedures that can improve early detection?
- What do you feel the role and voice of the patient advocacy community is in highlighting the existing needs and gaps in screening and early diagnosis?
- How can referral pathways be strengthened to ensure that patients move quickly from suspicion of a rare disease to specialised expertise, regardless of the differences in national care pathways?
- What is the role of the EU to enable regulators, industry and research community to work together to make newborn screening more equally accessible and a vehicle for earlier access to the latest approved treatments?

Stakeholders involved

Moderator
Luca Sangiorgi
European Reference Network on Rare Bone Diseases (ERN-BOND) Coordinator

James R. Bohnam
President of the International Society of Neonatal Screening (ISNS)

Jane Cooper
Senior Vice-President, Head of EMEA, Ultragenyx

Alessandra Ferlini
University of Ferrara, Italy & Scientific Coordinator of the EU-IHI Screen4Care project

Peter Mulders
European Reference Network for Rare Uro-Recto-Genital Diseases and Complex Conditions (EUROGEN) Coordinator

Martine Pergent
President, International Patient Organisation for Primary Immunodeficiencies (IPOPI)

Anton Ussi
Operations and Finance Director, EATRIS

Working Group 3: Equitable access to diagnostics and early treatment

Co-chairs	• Luca Sangiorgi

Potential members

- International Society of Neonatal Screening
- EU-IHI Screen4Care
- DG Connect
- DG SANTE
- European Medicines Agency
- Trio Presidency
- Up to 3 ePAGs (incl. International Patient Organisation for Primary Immunodeficiencies)
- Up to 3 other ERN coordinators on a voluntary basis
- Pharmaceutical industry representative(s)

Key take-aways from HLM Rare 2025

- Scaling newborn screening requires strong evidence, political support and ERN-led consensus.
- Screening must link to ERNs through clear referral pathways, CPMS expert review and sustainable registries for follow-up care. Data sharing and sustainable registries are essential to move from expert opinion to high-quality evidence and improved outcomes.
- Digital tools (AI/apps) can support early suspicion, but must be validated, well governed and never replace expert clinical judgment.

Working Group 3: Equitable access to diagnostics and early treatment

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

Working Group 3: Equitable access to diagnostics and early treatment

Topics for targeted Plan of Action

- Defining a European minimum set of newborn screening programmes
- Assessing most promising population-based screening approaches
- Designing standardised referral pathways from screening and early-suspicion to ERNs
- Role of digital tools in early detection
- Linking early diagnosis to early access pathways for orphan drugs
- Patient pathway navigation after early diagnosis

PRIORITY ACTION 4: FOSTER EU LEADERSHIP IN CLINICAL TRIALS FOR RARE DISEASES

THE CHALLENGE

- Europe's share of global rare-disease clinical trials is declining, with development increasingly shifting to the US and Asia.
- Lengthy and fragmented approval processes, particularly for multi-country trials, delay patient access and deter investment.
- Limited coordination between academia, patient groups, industry, ERNs and regulators slows the translation of trial results into clinical practice.

WHAT THE DECLARATION CALLS FOR

- Strengthen inclusive public–private collaboration between academia, patient organisations and industry to accelerate rare-disease clinical research.
- Reduce timelines and administrative barriers through a risk-based, fast-track regulatory approach for rare-disease clinical trials.
- Support EU-level funding and coordination mechanisms that reward partnerships, harmonise ethical review processes and facilitate rapid uptake of validated therapies across Member States.
- Fully harnessing the 28th regime to enable Europe to act as one, helping European regain competitiveness, leadership and independence including on clinical trials.

PRIORITY ACTION 4: HLM RARE 2025 PANEL

Panel discussion	Fostering EU leadership in clinical trials for rare diseases
Conference day	Day 1: research & innovation

Questions discussed

- Why is Europe losing ground in rare-disease clinical trials? What are the most critical roadblocks today? Which two or three changes would have the greatest impact on restoring Europe's attractiveness for rare-disease trials?
- Have recent EU initiatives (e.g. ERDERA, RealiseD, ERNs), been able to meaningfully unify Europe's fragmented research and clinical trial efforts?
- How can ERNs and other EU infrastructures move from fragmented, project-based roles to becoming the stable backbone for rare-disease trials?
- How should public–private partnerships be designed so they truly focus on unmet needs, meaningfully involve patients and speed up access to effective therapies across all Member States?
- Will the proposed recommendations successfully help the EU lead the way on rare diseases research and clinical trials? Do these address the most urgent gaps?
- Who should take the lead in implementing these recommendations?

Stakeholders involved

Moderator
Josep Figueras
Director Emeritus
European Observatory on Health Systems and Policies

Ralf Dieter Hilgers
RealiseD Coordinator

Michael Berntgen
Head of Scientific Evidence Generation Department, EMA

Virginie Hivert
Acting Chief Executive Officer and Head of Therapies & Access, EURORDIS-Rare Diseases Europe

Niklas Blomberg
Executive Director, Innovative Health Initiative (IHI)

Daria Julkowska
European Rare Diseases Research Alliance (ERDERA) Coordinator

Holm Graessner
European Reference Network for Neurological Disorders (ERN-RND) Coordinator

Enrico Piccinini
Senior Vice President, EU & International, Rare Diseases, Chiesi Group

WORKING GROUP 4: EU leadership in clinical trials

Co-chairs	• Holm Graessner

Potential members

- Ralf Dieter Hilgers, RealiseD Coordinator
- Daria Julkowska, European Rare Diseases Research Alliance (ERDERA) Coordinator
- Mark Turner Chief Executive Officer, Conect4Children Stichting
- Jacques Demotes-Mainard, ECRIN Director General
- Niklas Blomberg Executive Director, Innovative Health Initiative (IHI)
- Michael Berntgen Head of Scientific Evidence Generation Department, EMA
- Up to 3 ePAGs
- Up to 3 other ERN coordinators on a voluntary basis
- Pharmaceutical industry representative(s)

Key take-aways from the discussion

- ERNs are not formally recognised or financed as trial networks, despite being “ideally placed” to run innovative designs, use registries as external controls, and connect patients to studies.
- Existing instruments— e.g. ERNs, registries, RealiseD methods, Connect4Children templates— can be combined to deliver faster, high-quality trials.
- For patients priority issues include: access to cross-border trials when no study exists in a patient’s home country; meaningful involvement in trial design, especially in paediatrics; and smart use of AI to improve recruitment and protocol feasibility.

WORKING GROUP 4: EU leadership in clinical trials

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

WORKING GROUP 4: EU leadership in clinical trials

Topics for targeted Plan of Action

- Formal recognition and financing of ERNs as clinical trial networks
- Integration of existing resources (e.g. ERNs, ERDERA, RealiseD, Connect4Children, etc)
- Challenges for multi-country clinical trials
- Incentives for Pubic Private Partnerships dedicated to innovative diagnostics and therapies
- Access to cross-border clinical trials
- Standardisation of collaboration between ERNs and regulatory agencies
- Risk-based regulatory approach and fast track approval pathway for rare disease clinical trials

PRIORITY ACTION 5: COMPREHENSIVE RARE DISEASE INFRASTRUCTURE CLUSTERS (CORDIC)

THE CHALLENGE

- Research, innovation and care for rare diseases remain dispersed across institutions, limiting efficiency, visibility and patient navigation.
- Not all Member States have sufficient infrastructure to support advanced diagnostics, translational research and clinical trials.
- Interoperability between biobanks, registries, genomic initiatives and centres of expertise remains incomplete, constraining pan-European collaboration.

WHAT THE DECLARATION CALLS FOR

- Establish at least one Comprehensive Rare-Disease Infrastructure Cluster (CoRDIC) in each Member State, integrating research, innovation and care capacities.
- Ensure CoRDICs function as identifiable access points for patients, supporting early diagnosis, clinical trials and state-of-the-art care.
- Strengthen interconnections and interoperability between national infrastructures and EU-level initiatives, with sustained and predictable funding to support a pan-European ecosystem.

PRIORITY ACTION 5: HLM RARE 2025 PANEL

Panel discussion	Creating comprehensive rare disease infrastructure clusters (CRDIC) for research, innovation and care
Conference day	Day 1: research & innovation

Questions discussed

- With the current infrastructure in place, namely the ERNs and existing ERDERA clinical research network, does the EU have what it takes to lead rare disease research, innovation and care? Or do we need more?
- What could be the benefits of having centralised clusters of research, innovation and care for the rare disease community?
- Do you believe the proposed recommendation will successfully help accelerate research, innovation and care?
- Who should take the lead in implementing it?
- Is it feasible to implement such clusters in all EU countries?
- Where should investment be coming from?

Stakeholders involved

Moderator
Ruth Ladenstein
European Reference Network for Paediatric Oncology (PaedCan) Coordinator

Georgios Margetidis
Head of Sector, Health and Food, EU4Health, HaDEA

Jacques Demotes-Mainard
ECRIN Director General

Alexander Natz
Secretary General, EUCOPE

Arjon van Hengel
Senior Policy Officer, Health Innovations & Ecosystems, DG RTD

Mark Turner
Chief Executive Officer, Conect4Children Stichting

Holm Graessner
European Reference Network for Neurological Disorders (ERN-RND) Coordinator

Sheela Upadhyaya
Together4Rare Steering Group Chair

Virginie Hivert
Acting Chief Executive Officer and Head of Therapies & Access, EURORDIS-Rare Diseases Europe

Working Group 5: Comprehensive rare disease infrastructure clusters

Co-chairs	• Ruth Ladenstein

Potential members

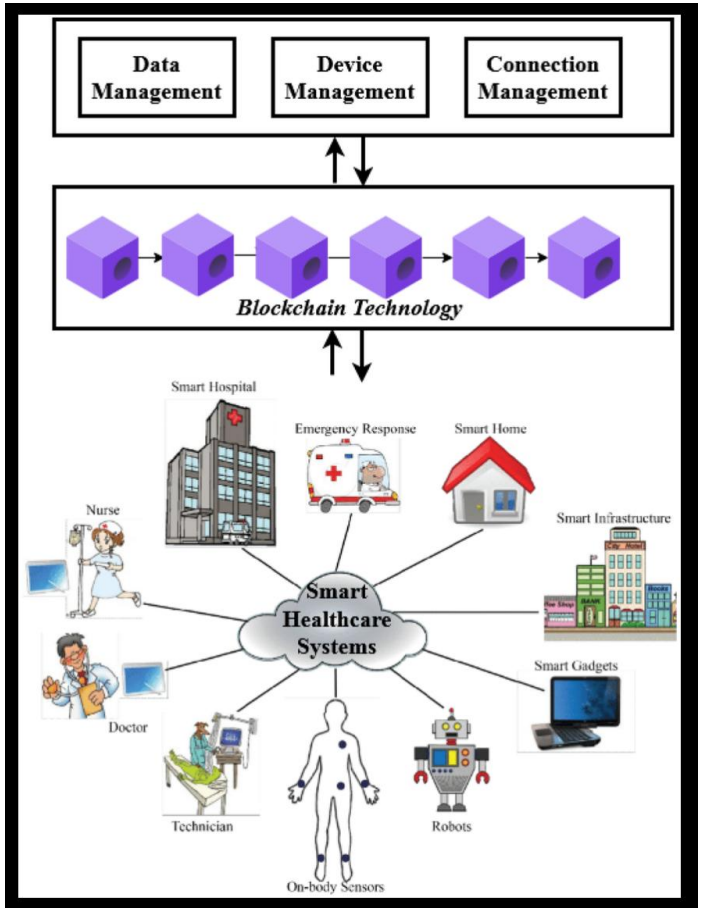
- Mark Turner Chief Executive Officer, Conect4Children Stichting
- Jacques Demotes-Mainard, ECRIN Director General
- Georgios Margetidis Head of Sector, EU4Health grants on cancer, NCDs, mental health, rare diseases and health workforce, HaDEA
- Up to 3 ePAGs
- Up to 3 other ERN coordinators on a voluntary basis
- Pharmaceutical industry representative(s)

Key take-aways from HLM Rare 2025

- Rare disease trials demand pan-European scale, building on ECRIN (multinational sponsor support via 130 units for regulatory/ethical/data services), C4C (pediatric site quality/training), and ERNs (disease-specific science/patient access).
- CoRDICs would evolve from ERNs and initiatives like Together for Rare, operationalizing Biotech Act and clinical trial streamlining via centralized hubs for harmonized screening and pre-qualified trial centers,
- CoRDICs would need integration into healthcare systems.

At least One CORDIC /Member State : An Integrated Virtual House for Rare & Complex Disease Care

One VIRTUAL roof – one navigation core – vertically integrated infrastructures



CoRDIC
Central Navigator Function
(Patient & Knowledge Navigation Function
(Visible, bidirectional, continuous)

CARE
Hospital Care Units & ERNs
Specialized care • Multidisciplinary teams • Cross-border expertise
expertise

Rare Disease Patient Welcome Desk
Entry point • Case coordination • Navigation through complex journeys

Diagnostic facilities
Imaging • Pathology • Molecular diagnostics • Functional testing

Overarching Clinical Trial Unit
Trial design • Coordination • Regulatory • Access across diseases

Shared „Omics" Infrastructures
Genomics • Proteomics • Metabolomics • Bioinformatics

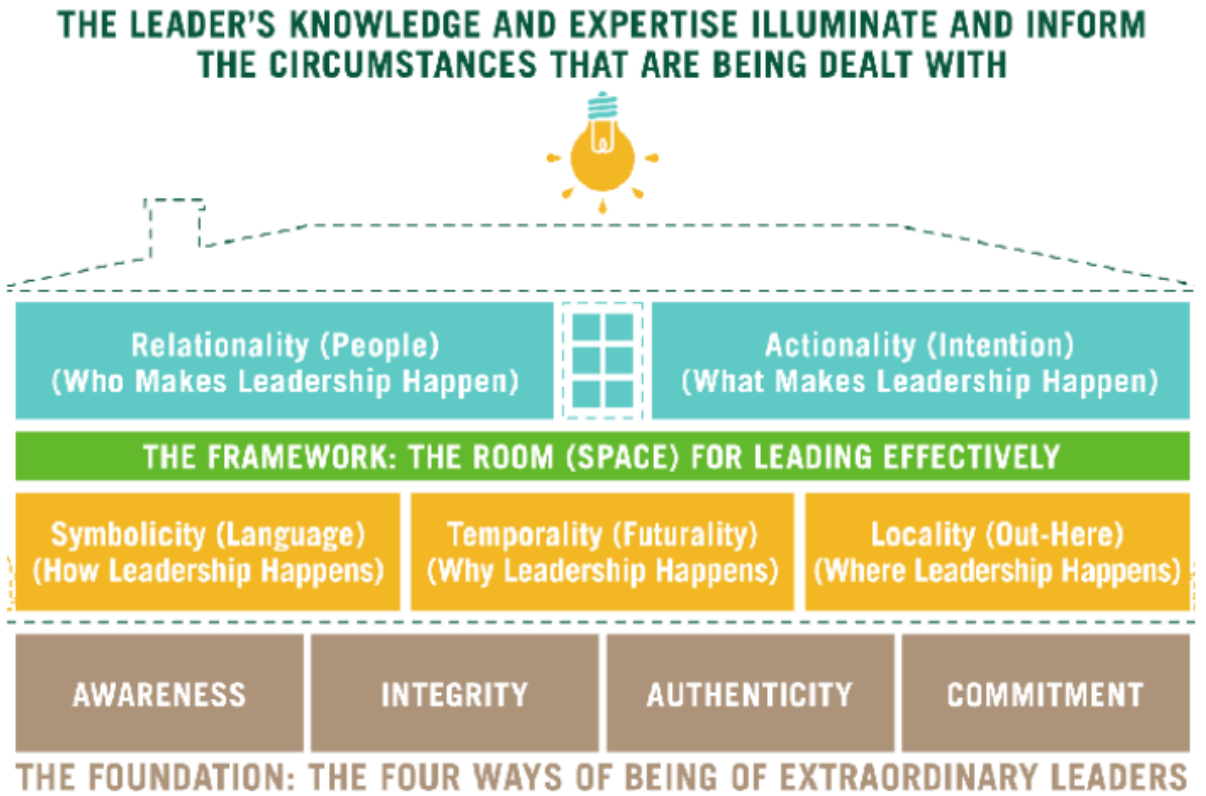
Supportive Care & Long-Term Follow-Up
Psychosocial care • Rehabilitation • Survivorship • Lifelong monitoring

Basic & Translational Research
Disease mechanisms • Models • Biomarkers • Bench-to-bedside translation

Other ?

AIM: Shorten time to Dx to < 1 Year! BENEFIT: Central Visibility and Access
TARGET: Equality across EU MS – EU Funding Mechansimn for Infratsructurs

WE HAVE IT IN OUR HANDS



The House of Leadership. The house metaphor provides the leader with a solid foundation for being a leader and a practical framework for exercising leadership effectively. The leader's know-how and knowwhat serve to inform the leader in making choices and decisions.

Working Group 5: Comprehensive rare disease infrastructure clusters

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

Working Group 5: Comprehensive rare disease infrastructure clusters

Topics for targeted Plan of Action

- Integration of research, innovation and care capacities at national level (and country-specific considerations)
- Integration of these hubs into national healthcare systems
- Interconnections and interoperability between national infrastructures and EU-level initiatives
- Sustained and predictable funding
- Leveraging existing infrastructure (ERNs, ECRIN, C4C), initiatives (Together4Rare) and regulations (e.g. EU Biotech Act)

PRIORITY ACTION 6: BOOST REAL-WORLD EVIDENCE GENERATION

THE CHALLENGE

- Rare-disease data are fragmented, heterogeneous and uneven in quality, limiting their clinical and regulatory usefulness.
- Limited interoperability between registries, biobanks and EHRs constrains cross-border research, care coordination and evidence generation.
- The absence of shared standards and analytics frameworks slows the responsible uptake of AI-enabled innovation.

WHAT THE DECLARATION CALLS FOR

- Coordinate end-to-end rare-disease data collection and governance across ERN registries, biobanks and EHRs, aligned with FAIR principles and the EHDS.
- Improve data quality and clinical utility through harmonised standards, shared validation metrics and integration of real-world data and PROMs.
- Enable federated analytics and common data models to support privacy-preserving cross-border collaboration, AI/ML development and regulatory-grade evidence.

PRIORITY ACTION 6: HLM RARE 2025 PANEL

Panel discussion	Making rare-disease data high quality and clinical utility, interoperable and integrated under the EHDS
Conference day	Day 2: EU skills

Questions discussed

- Given the central role of OrphaCodes in rare-disease visibility, what concrete steps should Member States and EU institutions take to ensure full end-to-end integration of OrphaCodes into national health systems?
- What concrete steps should Europe take to accelerate the generation of robust, interoperable datasets and improve access to existing clinical-trial data?
- What improvements in data accessibility, quality and interoperability would most accelerate the development of diagnostics and therapies, and how can rare-disease clinical-trial data be better integrated into the EHDS to support faster regulatory assessment, HTA decisions and equitable patient access?
- How can the ERNs and patients advocacy groups strengthen their role as facilitators of high-quality, interoperable evidence generation?
- How can the European Commission, and specifically DG Connect, and the Member States work together to make the EHDS a truly functional, pan-European ecosystem for rare-disease data?

Stakeholders involved

Moderator
Maria del Mar Mañú Pereira
European Reference Network on Rare Hematological Diseases (ERN-EuroBloodNet) Coordinator

Christel Schaldemose
Vice-President (S&D, DK), European Parliament

Dariusz Adamczewski
Managing Director, Children's Tumor Foundation Europe

Michael Berntgen
Head of Scientific Evidence Generation Department, EMA

Szymon Bielecki
Head of Sector, AI health tech and infrastructure, DG CONNECT

Teresinha Evangelista
European Reference Network for neuromuscular diseases (ERN EURO-NMD) Coordinator

Michael Ostland
Head of Development Europe, Denali Therapeutics

Ana Rath
Orphanet Director, INSERM

Working Group 6: Real-world evidence

Co-chairs	• Mar Manu Pereira

Potential members

- Joint Research Centre (European Commission)
- Orphanet / Inserm
- European Medicines Agency
- DG SANTE
- Up to 3 ePAGs (incl. Children's Tumor Foundation Europe)
- Up to 3 other ERN coordinators on a voluntary basis
- Pharmaceutical industry representative(s)

Key take-aways from HLM Rare 2025

- EHDS needs high-quality, interoperable rare-disease data to reduce fragmentation and enable real-world evidence.
- Orpha codes and standardised phenotyping are the backbone for consistent identification and cross-border interoperability.
- RWE must be decision-grade: shared data quality standards, strong registries and federated infrastructure support regulators/HTA.
- AI needs trust and representativeness: manage bias/opt-out risk and keep experts, patients and data stewards in the loop.

Working Group 6: Real-world evidence

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

Working Group 6: Real-world evidence

Topics for targeted Plan of Action

- Data quality standards for decision-grade real-world evidence
- Strengthening rare disease registries under the EHDS
- Integration of clinical trials data under the EHDS
- Role of ERNs as data stewards and evidence facilitators
- AI and advanced analytics in rare disease data use
- End-to-end integration of OrphaCodes in national health systems

PRIORITY ACTION 7: NEW BUSINESS MODELS AND MECHANISMS FOR EQUITABLE ACCESS

THE CHALLENGE

- Access to innovative orphan therapies and diagnostics remains uneven across Member States, particularly affecting smaller countries with limited fiscal capacity.
- Existing early access, procurement and reimbursement pathways are fragmented and poorly coordinated, delaying patient access.
- Structural inequities limit the ability of national systems to absorb and sustain innovation, despite EU-level approvals.

WHAT THE DECLARATION CALLS FOR

- Develop financially sustainable early access models for orphan therapies and diagnostics, based on transparent, value-based and patient-centred criteria.
- Strengthen pan-European pathways and cross-country collaboration with industry on early access, procurement and reimbursement mechanisms.
- Pilot a European Guarantee Fund, on a small and targeted scale, to address structural inequities and assess its feasibility in improving timely access across the EU.

PRIORITY ACTION 7: HLM RARE 2025 PANEL

Panel discussion	Exploring new business models to prioritise equitable access to innovative orphan therapies and diagnostics
Conference day	Day 3: policy & funding

Questions discussed

- What are today the main challenges to timely and equitable access to effective orphan medicines and therapies in Member States and where should EU and national decision-makers prioritise action?
- What practical risk-sharing mechanisms with industry could work best in the EU context to reduce uncertainty, increase market availability and ultimately improve access to medicines and diagnostics for people living with rare diseases?
- Thinking about moving from pilots to scale, and linked to the above, there are already a number of pilot schemes and collaborative initiatives under way; which examples show the greatest potential, and what would be needed in practice to scale them up across more Member States?
- Regarding EU-level tools and joint public procurement, what could be the role of EU-level or joint public procurement in addressing access problems for orphan medicines and diagnostics, particularly for smaller Member States and very small patient cohorts?
- Looking specifically at a possible European Guarantee Fund, what key design features would be essential to make it both politically feasible and attractive in practice?

Stakeholders involved

Moderator
Josep Figueras
Director Emeritus
European Observatory on Health Systems and Policies

Peter Liese
Member of the European Parliament
EPP, Germany

Stine Bosse
Member of the European Parliament
Renew, Denmark

Silvio Brusaferro
Former President of the Italian Institute of Health (2019-2023)

Alexis Arzimanoglou
European Reference Network for all rare and complex epilepsies (EpiCare) Coordinator

Alexandra Heumbler Perry
CEO, Rare Diseases International

Johan Prevot
Executive Director, International Patient Organisation for Primary Immunodeficiencies (IPOPI)

Giacomo Chiesi
Executive Vice President, Global Rare Diseases, Chiesi Group

Working Group 7: Early access models and mechanisms

Co-chairs	• Maurizio Scarpa

Potential members

- **DG SANTE** representative
- **Health Emergency Preparedness and Response** Authority representative
- **Enrico Letta**, President of the Jacqued Delors Institute
- **Manuel Heitor**, Former Portuguese Minister of Science, Technology and Higher Education
- **Stine Bosse**, Member of the European Parliament (Renew, Denmark)
- **Silvio Brusaferro**, Former President, Italian Institute of Health (2019-2023)
- **Victor Maertens**, Government Affairs Director, EUCOPE
- **Copenhagen Economics** representative
- Up to 3 ePAGs
- Up to 3 other ERN coordinators on a voluntary basis
- Pharmaceutical industry representative(s)

Key take-aways from HLM Rare 2025

- Current administrative hurdles and market failures cause delays in rare disease diagnosis and access, making early intervention costly to society.
- New legislative efforts (Pharma Review, Biotech Act) aim to significantly speed up approvals, offer tailored pathways, and boost incentives for rare disease innovation within Europe.
- ERNs are recognized as an existing infrastructure for cross-border collaboration, expert advice, and decentralized trials.
- The goal is to transform Europe into a leader in rare disease innovation and therapy, preventing talent and treatments from moving to other regions like the US or China.

Working Group 7: Early access models and mechanisms

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

Working Group 7: Early access models and mechanisms

Topics for targeted Plan of Action

The 28th Regime and Regulatory Innovation

European Guarantee Fund

Funding Mechanisms for Accelerated Multi-Country Clinical Trials

Strategic alignment of EU Funding programs to support early access and diagnostic pathways

Strategic alignment for public/private funding programs to incentivise the development of new drugs

PRIORITY ACTION 8: RINGFENCE ERNS FUNDING UNDER MFF

THE CHALLENGE

- Funding for the ERNs and rare disease actions remains fragmented, short-term and project-based, limiting sustainability and strategic planning.
- Insufficient coordination between EU and national funding streams creates gaps and discontinuities in ERN operations and impact.
- Complex administrative procedures and overlapping governance reduce efficiency and long-term investment capacity.

WHAT THE DECLARATION CALLS FOR

- Ringfence ERN and rare disease funding within the 2028–2034 Multiannual Financial Framework.
- Ensure continuity across EU and national programmes, linking successors of Horizon Europe and EU4Health.
- Move from project-based funding to durable mechanisms, including multi-country consortia and pooled investments.
- Simplify governance and administrative procedures to support long-term planning and reduce duplication.

PRIORITY ACTION 8: HLM RARE 2025 PANEL

Panel discussion	Ringfencing EU funding for ERNs and translational research
Conference day	Day 1: research & innovation

Questions discussed

- How can ringfencing funding for the ERNs, scale up cross-border healthcare, clinical trials and translational research?
- What are the benefits of having an EU Action Plan and Mission on rare diseases and what would be the impact?
- What concrete initiatives should be prioritised for scaling cross-border healthcare, clinical trials and translational research?
- How can we make sure that within the target of 1400 clinical trials jointly established by European Commission (EC), the Heads of Medicines Agencies (HMA) and EMA, a significant percentage is covering rare diseases?
- How can the EU maximise the impact of EU Research & Innovation programmes on rare diseases, especially through the next Multiannual Financial Framework (2028–2034)?

Stakeholders involved

Moderator
Holm Graessner
European Reference Network for Neurological Disorders (ERN-RND) Coordinator

Ruth Ladenstein
European Reference Network for Paediatric Oncology (PaedCan) Coordinator

Andras Kulja
Member of the European Parliament (EPP, Hungary)

Vinciane Pirard
Global Medical Affairs – Rare Diseases, Sanofi

Alberto Pereira
European Reference Network for Rare Endocrine Conditions (Endo – ERN) Coordinator, Adult Chair

Enrique Terol
Health Attaché to the Permanent Representation of Spain to the EU

Working Group 8: Multiannual Financial Framework

Co-chairs	• MEP Vytenis Andriukaitis

Potential members

- Cypriot Council of the EU Presidency representative
- Ireland Council of the EU Presidency representative
- Lithuanian Council of the EU Presidency representative
- Enrico Letta, President of the Jacques Delors Institute
- Manuel Heitor, Former Portuguese Minister of Science, Technology, and Higher Education
- Till Voigtländer, Coordinator of Joint Action on Integration of ERNs into National Healthcare Systems (JARDIN)
- MEP Nicolás González Casares (S&D, Spain)
- DG SANTE representative
- Up to 3 Patient Advocates representing their respective ERNs
- 6 ERN coordinators (Core Team)
- Pharmaceutical industry representative(s)

Key take-aways from HLM Rare 2025

- The need for direct, non-competitive grants treating ERNs as Europe's singular healthcare system, not MFF competitive calls.
- The need for ringfenced ERN funding in the MFF to avoid that world-leading networks vanish due to insuficiente funding

Working Group 8: Multiannual Financial Framework

Topics for targeted Plan of Action: your perspective

Focus

What should be the focus of this Plan of Action?

Key topics

What key topics should be discussed or considered by the WG from your perspective?

Working Group 8: Multiannual Financial Framework

Topics for targeted Plan of Action

Dedicated health and research budget line in the 2028 MFF for ERN funding (non-competitive)

Continuity of funding at both EU and national levels, to maintain steady financing for coordination, clinical operations, knowledge sharing, data infrastructure, and capacity building.

Simplification of administrative procedures and governance

Collaboration with future presidencies of the Council of the EU (Cyprus, Ireland and Lithuania)

Biotech Act (with funding potentially integrated into the 2028-2034 MFF).