RWE4Decisions

Public Webinar

Improving Rare Disease Real-World Data Availability and Quality for HTA/Payers

8 April 2026

15:00-16:30 CEST

Via Zoom

RWE4Decisions REAL WORLD EVIDENCE

Mission

HTA/Payer-led, multi-stakeholder Learning Network

RWE4Decisions brings together experts from all stakeholder groups to engage in dialogues that consider how fit-for-purpose RWE can be generated over the life cycle of **highly innovative medicines** through:

- horizon scanning systems that identify medicines which are most likely to need RWE
- identifying what RWE is needed to inform HTA/Payer decisions
- clarifying how RWE should be generated by stakeholders and evaluated by HTA/Payers
- aligning planning and execution of effective Post Launch Evidence Generation (PLEG) studies.

RWE4Decisions REAL WORLD EVIDENCE 2026 Steering Group

HTA BODIES / PAYERS

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INAMI-RIZIV

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The Secretariat is provided by FIPRA funded by EUCOPE and member companies

RWE4Decisions
Public Webinar

Improving Rare Disease Real-World Data Availability and Quality for HTA/Payers

Co-Moderators

Dr. Karen Facey
RWE4Decisions Facilitator

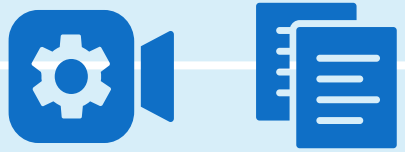
Dr. Victoria Hodgkinson
Executive Director, Canadian Neuromuscular Disease Registry

RWE4Decisions
Public Webinar

Improving Rare Disease Real-World Data Availability and Quality for HTA/Payers

Agenda

CEST 15:00 – 15:10	**Welcoming remarks** • **Dr. Karen Facey** • **Dr. Victoria Hodgkinson**
15:10 – 15:25	**Keynote presentation** **Innovating data collection across stakeholders to deliver value in rare diseases** • **Prof. Maurizio Scarpa, MD, PhD** (MetabERN)
15:25 – 15:50	**Panel Reflections** • **Alessio Amadasi,** Vice President, Medical Affairs for Rare Diseases, Chiesi • **François Houÿez,** Information/Access to Therapies Director & Health Policy Advisor, EURORDIS • **Prof. Wim Goettsch,** Special Advisor in HTA, ZIN (Healthcare Institute Netherlands • **Dr. Nicole Mittmann, PhD,** Chief Scientist and Vice-President of Evidence, Canada's Drug Agency
15:50 - 16:28	**Multi-stakeholder discussion: Q&A from audience**
16:28 – 16:30	**Closing remarks and next steps**



Improving Rare Disease Real-World Data Availability and Quality for HTA/Payers

Setting the Scene

Dr. Victoria Hodgkinson
Executive Director,
Canadian Neuromuscular Disease Registry

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Improving Rare Disease Real-World Data Availability and Quality for HTA/Payers

Prof. Maurizio Scarpa (MD, PhD)
Coordinator, European Reference Network for Rare Hereditary Metabolic Diseases (MetabERN)

Keynote presentation:

Innovating data collection across stakeholders to deliver value in rare diseases

Innovating Data Collection Across Stakeholders to Deliver Value in Rare Diseases

Maurizio Scarpa, MD, PhD

Coordinator, European Reference Network for Hereditary Metabolic Diseases (MetabERN)

Director, Regional Coordinator Centre for Rare Diseases, University Hospital of Udine, Italy

RWE4Decisions
REAL WORLD EVIDENCE

300 Million People Worldwide Cannot Afford to Wait for Better Evidence

- Over **6,000 clinically defined rare diseases** affect an estimated **30 million million people in Europe** and 300 million worldwide — 72% are genetic, genetic, 70% begin in childhood.
- **95%** of rare diseases have no approved treatment; the average diagnostic diagnostic odyssey spans **5–7 years**.
- Small, geographically dispersed patient populations make conventional conventional randomised controlled trials structurally inadequate.
- The evidence gap is not a scientific failure — it is a **systemic data collection failure**.
- Real-World Data (RWD) from registries, electronic health records, and patient-reported outcomes represents the most viable path to generating actionable evidence at scale.
- The question is no longer *whether* to collect RWD — it is *how to collect it better, together, and for multiple purposes simultaneously.*

> **The rare disease community has a unique opportunity — and responsibility — to lead the transformation of health data infrastructure in Europe.**

Data from The Everylife Foundation.Org

24 ERNs Connect Over 900 Highly Specialised Centres Across 26 EU Member States

- The **24 European Reference Networks (ERNs)**, established under EU Directive 2011/24/EU, are virtual networks connecting highly specialised healthcare providers across Europe.
- Between 2018 and 2024, new patient referrals to ERN centres increased by **160%**, demonstrating growing clinical impact.
- Each ERN is mandated to build a **central patient registry** — a structured, longitudinal data repository capturing clinical, genetic, and treatment data.
- ERN registries represent the most comprehensive cross-national clinical datasets on rare diseases worldwide.
- **MetabERN** (Hereditary Metabolic Diseases) exemplifies this: the **U-IMD (Unified Registry for Inherited Metabolic Disorders)** is the first European registry encompassing all 1,400+ inherited metabolic disorders.
- ERN registries are not merely administrative tools — they are **living scientific instruments** generating natural history data, safety signals, and treatment outcomes in real time.

ERNs have built the infrastructure. The challenge now is to unlock its full potential for all stakeholders — regulators, HTA bodies, payers, researchers, and patients.

Clinicians Have Invested Enormously — Now the System Must Reciprocate

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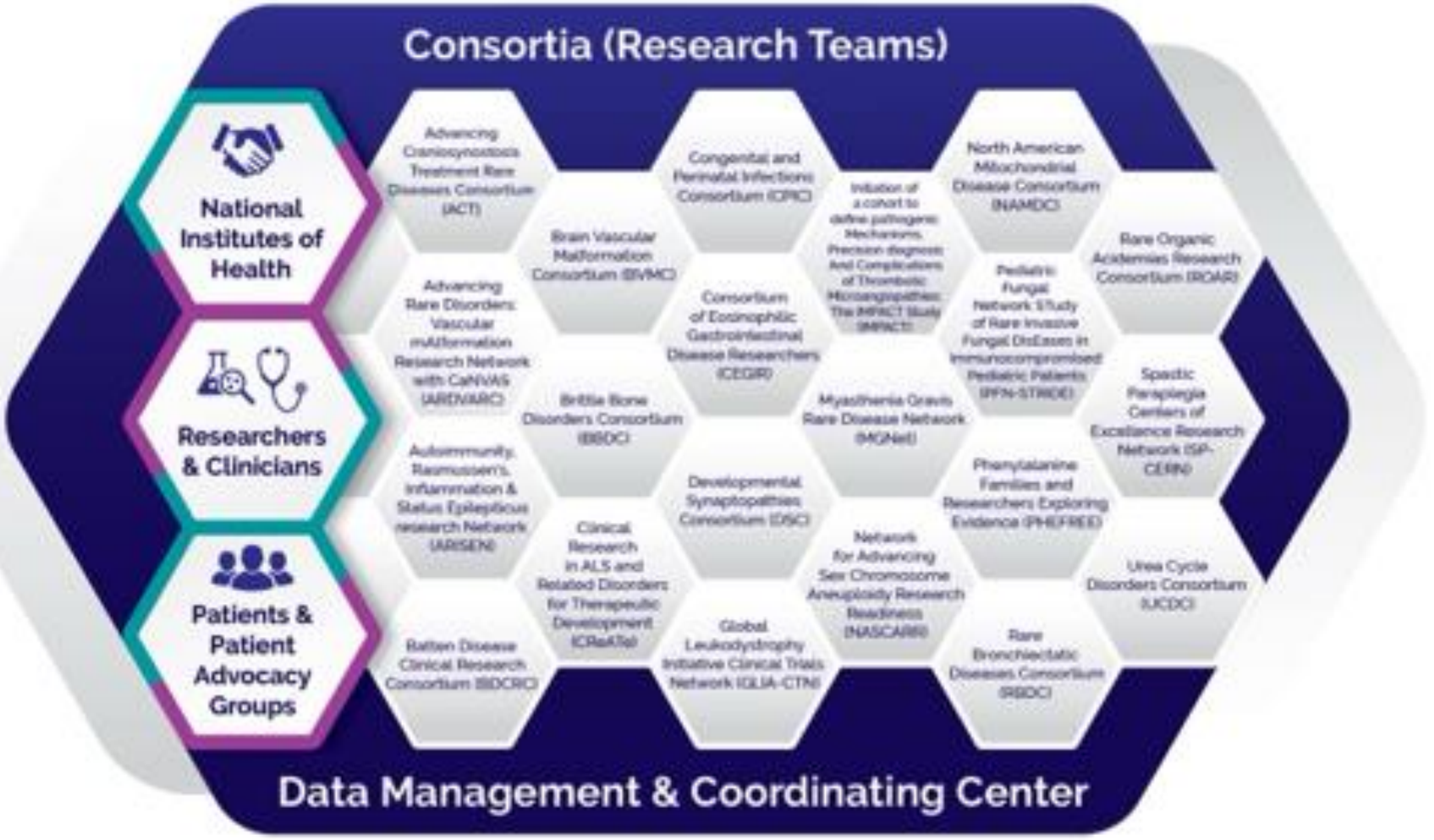

Physicians are the primary data generators. Sustainable RWD collection requires recognising their contribution and designing systems that serve clinical care, not just downstream users.

https://www.rarediseasesnetwork.org/consortia-directory

One Dataset, Many Decisions — The Multi-Purpose Value of ERN Registry Data

ERN registry data is increasingly being leveraged across the full evidence ecosystem:

PURPOSE	APPLICATION	STATUS
Regulatory	EMA post-authorisation studies, PASS, label extensions	Active pilots
HTA / Payer	Joint Clinical Assessments (EU HTA Regulation), national reimbursement	Emerging — JCA for ATMPs from 2025
Clinical Research	Natural history studies, comparative effectiveness, biomarker discovery	Established
Patient Empowerment	Patient-reported outcomes, quality of life, diagnostic journey	Growing
Public Health	Epidemiological surveillance, burden of disease estimates	Foundational
Industry R&D	Trial design, endpoint selection, comparator arms	Pre-competitive collaboration

- The **EU HTA Regulation** (effective January 2025 for ATMPs, expanding to orphan medicines in 2028) creates a formal demand for RWE from ERN registries in **Joint Clinical Assessments**.
- ZIN (Netherlands), HAS (France), and NICE (UK) have all initiated pilots using rare disease registry data in HTA processes.
- The challenge: registry data was designed for clinical care — it must now be **prospectively designed** to serve HTA/payer needs without compromising clinical utility.

> **The multi-purpose use of RWD is not a future aspiration — it is a present reality requiring deliberate design and governance.**

The European Health Data Space Transforms RWD from Aspiration to Legal Framework

The European Health Data Space (EHDS) Regulation entered into force on 25 March 2025 — a landmark moment for rare disease data governance.

What EHDS changes:

- Establishes a **legal right of secondary use** of electronic health data for research, policy, and public interest purposes across all EU Member States.
- Creates **Health Data Access Bodies (HDABs)** in each Member State to manage data access requests.
- Mandates **interoperability standards** for electronic health records, including rare disease data.
- Enables **cross-border data linkage** — critical for rare diseases where no single country has sufficient patient numbers.
- Provides a **FAIR data framework** that aligns with ERN registry standards.

Implications for ERNs:

- ERN registries must align with EHDS interoperability requirements — a significant but achievable technical challenge.
- The EHDS creates a **legal pathway** for HTA bodies and payers to access ERN registry data under defined conditions.
- Patient consent and privacy frameworks must be harmonised across 26 Member States.
- ERNs are uniquely positioned to serve as **model use cases** for EHDS secondary data use in rare diseases.

> **EHDS is not a compliance burden — it is the legal and technical foundation that makes pan-European RWD use possible at scale.**

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 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

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

HLM RARE

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

https://www.hlm4rare.eu

HLM Rare 2025 Produced a Political Commitment to Transform the European European Rare Disease Ecosystem

THE MEETING CONTEXT

The **High-Level Meeting on European Research and Innovation for Rare Diseases (HLM Rare 2025)** took place in Brussels, **9–11 December 2025**, bringing together:

- EU policymakers and European Commission representatives
- Coordinators of all 24 European Reference Networks (ERNs)
- Patient organisations (led by EURORDIS)
- Industry leaders and academic researchers
- HTA bodies and health technology developers

KEY OUTCOME: THE DECLARATION

The Declaration on the European Innovation and Care Ecosystem for Rare and Complex Diseases establishes:

- A **political and strategic commitment** to fundamentally transform the rare disease R&I ecosystem.
- A **bold shared vision** with clear strategic priorities and measurable objectives.
- A call for a **coherent EU-level framework** integrating research, clinical care, data infrastructure, and market access.
- Explicit links connecting rare disease data infrastructure to the **EU Biotech Act** and the **EU Life Sciences Strategy** (July 2025).

HLM Rare 2025 elevated rare diseases from a niche clinical concern to a strategic European policy priority — with data infrastructure at its core.

The Strategic Priorities That Will Shape European Rare Disease Policy Through 2030

The Declaration and action plan from HLM Rare 2025 identified six interconnected priorities:

1. **Sustainable ERN Funding**

   Transition ERNs from project-based to structural EU funding, recognising them as permanent health infrastructure.

2. **Integrated Data Ecosystems**

   Connect ERN registries, national registries, and EHR systems under the EHDS framework; adopt FAIR principles universally.

3. **Accelerated Diagnostics**

   Leverage AI, genomics, and cross-border data sharing to reduce the diagnostic odyssey from years to months.

4. **Equitable Access to Therapies**

   Reform orphan medicine incentives and HTA processes to ensure approved therapies reach patients across all Member States.

5. **Patient-Centred Research**

   Embed patient-reported outcomes and patient co-design into all registry and research frameworks.

6. **Biotech Act Alignment**

   Ensure the forthcoming EU Biotech Act includes specific provisions for rare disease data, clinical trial flexibility, and RWE integration.

> **These are not aspirations — they are action items with named stakeholders, timelines, and accountability mechanisms.**

The Biotech Act Proposed on 16 December 2025 Must Embed RWE at Its Core

The **European Commission's Biotech Act** (proposed 16 December 2025) is a comprehensive regulatory framework aimed at strengthening EU competitiveness in biotechnology and biomanufacturing:

WHAT THE BIOTECH ACT OFFERS RARE DISEASES

- **Streamlined clinical trial authorisations** across Member States — reducing time-to-trial for rare disease therapies.
- **Adaptive trial design provisions** — enabling platform trials, basket trials, and rolling reviews suited to small populations.
- **Enhanced orphan medicine incentives** — reforming the regulatory pathway to maintain innovation attractiveness.
- **RWE integration** — formal recognition of real-world evidence from registries as valid evidence for regulatory and HTA decisions.
- **Pre-competitive data sharing** — legal frameworks for industry collaboration on RWD infrastructure without competition concerns.

THE HLM RARE 2025 INFLUENCE

- The Declaration explicitly calls for Biotech Act provisions that recognise ERN registries as **validated RWE sources**.
- ERN coordinators, EURORDIS, and patient organisations are actively engaged in the legislative process.
- The window for influence is **now** — the Act is in its parliamentary negotiation phase.

The Biotech Act is the legislative vehicle through which the commitments of HLM Rare 2025 can be codified into binding EU law.

ERNs + EHDS + Biotech Act = A Transformative Ecosystem for Rare Disease Evidence

ERN REGISTRIES	EHDS FRAMEWORK	EU BIOTECH ACT
Clinical data	Legal access	Regulatory use
Natural history	Interoperability	HTA integration
Multi-disease	Cross-border	Adaptive trials
Patient-centred	Privacy-compliant	Pre-competitive

The Multi-Stakeholder Imperative:

- **Physicians/ERNs:** Design registries for multiple purposes from inception; adopt EHDS-compliant data standards.
- **HTA bodies/Payers:** Engage early in registry design; define what data they need and when.
- **Industry:** Invest in pre-competitive data infrastructure; use ERN registries as external control arms.
- **Patient organisations:** Co-design data collection; ensure patient-reported outcomes are captured.
- **Regulators (EMA/EC):** Provide clear guidance on RWE acceptability; validate ERN registries as trusted data sources.
- **Policymakers:** Fund ERN data infrastructure sustainably; embed RWD requirements in the Biotech Act.

No single stakeholder can solve this alone. The architecture exists — what is needed is coordinated action and shared governance.

From Evidence to Action — What Each Stakeholder Group Must Do Next

This session brings together the stakeholders who can make the difference. The questions before us today:

FOR HTA BODIES AND PAYERS (ZIN, CDA-AMC)

- How do we formally embed registry data requirements into HTA processes for rare diseases?
- What governance structures ensure data quality and independence?

FOR INDUSTRY (CHIESI AND BEYOND)

- When no robust RWD system exists, what is the industry's responsibility to build one?
- How can pre-competitive collaboration be structured under EHDS and the Biotech Act?

FOR PATIENT ORGANISATIONS (EURORDIS)

- How do patients move from data contributors to data co-owners?
- What mechanisms ensure patient groups receive meaningful feedback on the data they provide?

FOR ERN CLINICIANS

- How do we design the next generation of registries to serve HTA/payer needs without compromising clinical utility?
- What resources and recognition are needed to sustain physician engagement?

The rare disease community has never been better positioned to act. The policy window, the legal framework, and the clinical infrastructure are all in place. What remains is the will to act together.

Innovating Data Collection Means Building a System Where Every Stakeholder Both Contributes and Benefits

> ***"Real-world data for rare diseases is not a technical problem. It is a governance, incentive, and collaboration problem — and those are problems that policy can solve."***

SUMMARY OF KEY MESSAGES

1. **ERN registries** have built an unparalleled clinical data infrastructure — now it must be designed for multi-purpose use.
2. The **EHDS** provides the legal and technical framework to make pan-European RWD use a reality.
3. **HLM Rare 2025** has produced a political commitment and action plan that must now be translated into concrete Measures in the **HLM4RARE** action.
4. The **EU Biotech Act** is the legislative vehicle to codify these commitments — the window for influence is open.
5. **Multi-stakeholder collaboration** is not optional — it is the only model that works for rare diseases.

> **The discussion that follows in this webinar is not academic. It is the beginning of the action-oriented work that will shape how rare disease patients across Europe access life-changing therapies — and how the evidence to support those decisions is generated, shared, and used.**

Thank You

& Discussion

SPEAKER CONTACT

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ACKNOWLEDGEMENTS

RWE4Decisions Secretariat
www.rwe4decisions.com

HLM Rare 2025 Declaration
www.brains4brain.eu/eu-activities-polices/hlm-rare/declaration/

UP NEXT: PANEL DISCUSSION

Moderated by Victoria Hodgkinson
University of Calgary & Canadian Neuromuscular Disease Registry

KEY REFERENCES

- **EHDS Regulation (EU) 2025/327** — entered into force 25 March 2025
- **EU Biotech Act Proposal COM(2025)1022** — 16 December 2025
- **HLM Rare 2025 Declaration** on the European Innovation and Care Ecosystem for Rare and Complex Diseases — December 2025
- **ERN First Monitoring Report 2025** — European Commission
- **EU HTA Regulation (EU) 2021/2282**

RWE4Decisions REAL WORLD EVIDENCE

Improving Rare Disease Real-World Data Availability and Quality for HTA/Payers

Panel discussion - Introductory Remarks

Prof. Dr. Ir. Wim Goettsch
Special Advisor HTA, ZIN

Dr. Nicole Mittmann
Chief Scientist and VP of Evidence, Canada's Drug Agency (CDA-AMC)

François Houÿez
Director of Information & Access to Therapies, Health Policy Advisor, EURORDIS

Alessio Amadasi
VP, Medical Affairs EU/International, Rare Diseases, Chiesi Group

Thank you!

A **report** will be published on our website: www.rwe4decisions.com

In the meantime, follow us on LinkedIn: @RWE4Decisions

Save the date for our next RWE4Decisions event:

- **Invitation-only Roundtable:** "*How can we use RWE to explain patient benefit?*", 3 June, 14:30-17:00 CEST
- **Get in touch at** secretariat@rwe4decisions.com if you are interested to receive an invitation.