Proposed Agenda: Rare Diseases Conference – Bridging Research and Care for Patients, Day 2 (April 11th, 2025)

Hosted by: Medical University of Warsaw

Under the patronage of: Polish Ministry of Health and European Economic and Social Committee

With support of: ERDERA, MRA 5ABM), EURORDIS, Krajowe Forum Orphan
Format: Hybrid

08:00 – 08:45 | Registration & Welcome Coffee

Participants check-in and networking opportunity

08:30 – 09:15 | Opening Remarks

- Prof. Krzysztof Szczałuba, Director of the Center of Excellence for Rare and Undiagnosed Diseases, Medical University of Warsaw, Poland
- Prof. Urszula Demkow, Secretary of State, Ministry of Health, Poland
- Ms. Malogrzata Bogusz, member of the EESC, president of the Kulski Foundation for Polish-American Relationsand president of the Institute for Social Development
- Prof. Wojciech Fendler, President of the Medical Research Agency (MRA), Poland
- Mr. Stanislaw Maćkowiak, President of National Forum ORPHAN, Poland
- Dr. Virginie Bros-Facer, CEO of EURORDIS

- Dr. Daria Julkowska, coordinator of ERDERA, INSERM, France

Conference Chair: Ms. Marzena Nelken, National Forum Orphan

09:15 – 10:45 | Session 1: Advancing Diagnostics for Rare Diseases

Chair: Prof. Holm Graessner

- **Newborn screening initiatives & future directions – the Polish example of diagnosis and treatment of Spinal Muscular Atrophy (SMA) ,** Prof. Anna Kostera-Pruszczyk, Head of the Department and Clinic of Neurology at UCK WUM, University of Warsaw, Poland (20 min)

- **Advancements in NBS - Screen4Care** – Prof. Allessandra Ferlini, director of the Medical Genetics section at the University of Ferrara & Screen4Care coordinator, Italy (15 min)

- **Innovation in diagnostics - beyond genetic testing and towards clinical decision support tools** – Prof. Holm Graessner, Director of the Rare Disease Centre Tübingen, University of Tübingen, Germany (15 min)

- **From diagnosis to innovative craniofacial surgeries** – Prof. Dawid Larysz, Head of the Clinic of Head and Neck Surgery for Children and Adolescents at the Provincial Specialist Children's Hospital in Olsztyn, Poland (15 min)

- **Challenges and opportunities in diagnostics from the perspective of patients** – Wojciech Nadolski , Association of Families with Fabry Disease Poland (10 min)

- **Q&A (15 min)**

10:45 – 11:00 | Coffee Break & Networking

11:00 – 11:50 | Session 2: Innovative Therapies and Clinical Trials – Key projects supported by Medical Research Agency (MRA)

Chair: Prof. Wojeciech Fendler

- **MRA's actions for Rare Diseases- success stories and future prospects**, Prof. Wojciech Fendler, President of the Medical Research Agency (MRA), Poland

- **Clinical Trials as a key component of effective treatment in children with rare diseases based on MRA's projects experienced**, Prof. dr hab. n. med. Anna Raciborska, Head of the Department of Oncology and Oncological Surgery for Children and Adolescents, Institute of Mother and Child, Poland

- **MRA grants in orphan diseases in children: lessons from TSC**, Prof. dr hab. n. med. Katarzyna Kotulska-Jóźwiak; Head of the Department of Neurology and Epileptology, The Children's Memorial Health Institute, Poland
- **Q&A** (10 min)

11:50 – 13:15 | Networking & Lunch Break

13:15 – 15:00 | Session 3: Strengthening National & European Collaboration in translational research

Chair: Prof. Luis Pereira de Almeida

- **Opening by Hélène Le Borgne**, Policy Officer, DG RTD, European Commission (15 min)
- **Translational medicine platforms: a way to strengthen national & European ecosystem**, Prof. Luis Pereira de Almeida, President of the Centre for Innovative Biomedicine and Biotechnology, University of Coimbra, Portugal (15 min)
- **RareBridges – Translational Research Platform for Rare Diseases**, Prof. Maciej Giefing, Director of IGC PAN (10 min)
- **EATRIS – European Infrastructure for Translational Medicine**, Anton Ussi, Operations & Finance Director of EATRIS (10 min)
- **PORT- Polish Center for Technology Development, - Advancing translational research via public-private collaborations**, Agnieszka Krzyzosiak, Head of Laboratory of Mechanisms of Neurodegeneration Life Sciences & Biotechnology Center, PORT (10 min)
- **Patient organisation as driver for translational research**, Małgorzata & Piotr Kośla, PACS2 Research Foundation (10 min)
- **Q&A (30 min)**

15:00 – 16:00 | Discussion Panel: RD research: From national registries to European Health Data Space and back

Chair: Mr. Tomasz Grybek

Panelists & topics:

- **How to build FAIR registries**, Dr. Bruna Dos Santos Vieira, Data Steward, Center for Molecular and Biomolecuar Informatics, Radboud UMC, The Netherlands (5 min statement)

- **How to build quality data sets in the clinical environment**, Prof. Mar Manu Pereira, Head of research Lab in rare anemia disorders, Scientific Director of ERN-EuroBloodNet, Val d'Hebron Insitute of Research, Spain (5 min statement)

- **Unlocking the Power of Medical Registries for Rare Disease Research and Patient Care**, Robert Lugowski, Chief Executive Officer, CliniNote (5 min statement)
- **Importance of quality data for regulatory decisions**, Tomasz Grybek, Member of the EURORDIS Board of Directors (5 min statement)
- **Open forum discussion with audience participation (20 min)**

16:00 – 16:15 | Coffee Break
16:15 – 17:45 | Holistic approach to care

Chair: Dr. Daria Julkowska
This session will be dedicated to discussing the links between the national RD plans/strategies and the holistic approach to care. The role of the RD National Mirror Groups and how they can support national communities will also be presented with the objective to initiate the creation of the Polish National Mirror Group.

- **State-of-the-art of the organisation of Polish RD community**, Prof. Alicja Chybicka, Poland (10 min)

- **RD National Mirror Groups as key element to support national community and strategic integration**, Daria Julkowska, ERDERA coordinator, France (10 min)

- **Example of French National Plan for Rare Diseases – holistic approach to care in National Plan - learning from best practices**, Ms. Anne-Sophie Lapointe, Ministry of Health, France (10 min) – TBC

- **Social and psychological support for patients with RD** – Ms. Vinciane Quoidbach, European Brain Council, Belgium (10min)

- **Reports on patients needs – two perspectives one goal**, Mr. Stanislaw Maćkowiak National Forum Orphan, Poland (10min)

- **Open forum discussion with audience participation** (30 min)

17:45 – 18:00 | Summary End of Conference

08:00 – 08:45	Registration & Welcome Coffee
08:30 – 09:15	Opening Remarks: Welcome by Krzysztof Szczałuba (Medical University of Warsaw) and other high level invited speakers
09:15 – 10:45	Session 1: Advancing Diagnostics for Rare Diseases
10:45 – 11:00	Coffee Break & Networking
11:00 – 11:50	Session 2: Innovative Therapies and Clinical Trials – Key projects supported by Medical Research Agency (MRA)
11:50 – 13:15	Networking & Lunch Break
13:15 – 15:00	Session 3: Strengthening National & European Collaboration in translational research
15:00 – 16:00	Discussion Panel: RD research: From national registries to European Health Data Space and back
16:00 – 16:15	Coffee Break
16:15 – 17:45	Session 4: Holistic approach to care
17:45 – 18:00	Summary End of Conference