

“Integrating Research and Healthcare for Rare Diseases: a structured cooperation with high community added value”

Parallel session to the Informal Meeting of EU Health Ministers

March 20th, 2017

From 9h00 to 16h30

Popes' Room

Grand Master's Palace, Valletta, Malta



A) Rationale and objectives of the meeting

“Rare diseases” is an area in the health sector that brings about **significant innovation and advances by enhancing access to novel and highly specialised services, medicines and technologies**, but also faces a **number of challenges**.

Stakeholders in the field have recognised that this is an area with high European added value, in which the most effective strategies are cross-border and EU-wide, with an international perspective. Since the adoption of the Regulation on Orphan Medicinal Products in 1999, significant advances in research have been made, strengthened ten years later by the Commission Communication on rare diseases: Europe's challenges COM(2008)679, and the Council Recommendation on an action in the field of rare diseases (2009/C 151/02), that promoted comprehensive policies at EU and national level to improve care and research for rare diseases. The Cross-Border Health Care Directive (Dir.2011/24/EU) included specific provisions on rare diseases and provided the legal ground for the development of European Reference Networks. At the end of 2016, the announcement of 23 European Reference Networks (with approval of a 24th in early 2017), is a stand at a point in history - by connecting patients, experts and hospitals, ERNs will have a radical impact on the provision of care for patients with rare diseases and on the development of research over the next decade.

There is a major interest for collaboration between rare diseases researchers in Europe and beyond. All efforts employed until now demonstrated that managing of rare diseases research in a coordinated way is clearly possible, and very much needed. The drivers of innovative health technologies appear in new science, new technologies and new processes e.g. medicines, medical devices, imaging, data collection and management, patient engagement. They provide huge opportunities to improve patient health outcomes, but they come with challenges in affordability and sustainability. In the specific context of the introduction of -omics into care practice, on one hand, and of the ongoing structuring of rare disease care centres in European Reference Networks, on the other hand, it is crucial to move one step further to maximise the potential of already existing tools, programmes and projects, by adapting them, scaling them up, and integrating them under the same umbrella to create a sustainable ecosystem of knowledge (expertise and data) generation, allowing a virtuous circle between rare disease care, research and medical innovation.

In this fast-moving and promising context, the **objectives of this meeting** are:

- 1) to outline high-level strategic organisation and performance of research activities in a structured and transversal manner in order to achieve a higher level of integration, optimisation of resources and synergy between EU research infrastructure and highly specialised healthcare through European Reference Networks;
- 2) to design operational proposals to address the specific challenges of prompt translation of innovative health technologies from research to care, structured cooperation for training of healthcare providers in health services for highly specialised rare diseases, structured cooperation in patient registrations and disease registries to generate the data needed both for clinical excellence and performance of national health systems;
- 3) to identify instruments and solution to ensure longer-lasting support to rare disease research in an integrated manner.

The ultimate goal is to establish a long-awaited single pipeline covering research, tools and clinics leading to optimisation and exploitation of results (and means), faster drug discovery and improved patients' care.

Synergies within and across the research and healthcare rare diseases communities will be presented, as well as gaps and existing challenges. Participants will work to design shared solutions that address existing needs. Specific focus will be on the infrastructural potential of European Reference Networks where research for rare diseases can be incubated and developed, and on other solutions for integrating research and healthcare activities in a sustainable manner.

B) Provisional agenda

Practical information: *There will be a registration desk for this event at the Grand Hotel Excelsior on the 19th March from 5pm to 7pm. Delegates can also register on the 20th March from 8am-9am at the Presidency Registration Office in Archbishop's Street, Valletta (opposite front door of the Grandmaster's Palace). The meeting shall start at 9am so please leave plenty of time to register. A bus transfer has been arranged from the Grand Hotel Excelsior Hotel to the Grandmaster's Palace at 8am.*

Programme 20th March 2017:

- **9h00 –9h15: INTRODUCTION**

- Welcome addresses:
 - **Hon Dr. Helena Dalli**, Minister for Social Dialogue, Consumer Affairs, and Civil Liberties within the Maltese Government
 - **Dr. Clarence Pace**, Director General, Ministry of Health, Malta
 - **Professor Alfred J. Vella**, Rector, University of Malta
- Introducing the meeting- **Yann Le Cam**, Chief Executive Officer, EURORDIS

- **SESSION 1: SYNERGIES, COMPLEMENTARITY & GAPS IN CURRENT INITIATIVES**

9h15– 11h15

*The panellists will paint the background by presenting the different research collaboration initiatives forming the current landscape and they will discuss existing gaps. In a short presentation (10 minutes each) panellists are invited to illustrate, in particular, how each initiative or infrastructure contributes to translational research for rare diseases (**Chair: Ana Rath**)*

- RD-Connect (**Victoria Hedley**, RD-ACTION Thematic Coordinator, Newcastle University John Walton Muscular Dystrophy Research Centre)
- BBMRI-ERIC (**Jan-Eric Litton**, Director General, Biobanking and Biomolecular Resources Research Infrastructure (BBMRI-ERIC))
- Orphanet (**Ana Rath**, Director, INSERM US14-Orphanet)
- IRDiRC (**Diego Ardigo**, Chair of the Therapies Scientific Committee, IRDiRC)
- E-Rare (**Daria Julkowska**, Programme Coordinator, E-Rare)
- JRC - EU Platform for Rare Diseases Registration (**Simona Martin**, Project Leader Directorate General Joint Research Centre, European Commission)
- Rare Diseases Registries –State of the Art (**Domenica Taruscio**, Director, National Center for Rare Diseases Istituto Superiore di Sanità (ISS), Italy)

- **11h15 – 11h30: COFFEE BREAK**

- **SESSION 2: HOW TO OPTIMISE A RESEARCH-READY NETWORK?**

11h30 – 12h30

*In this session, existing models for rare disease clinical research will be presented and the opportunities for networks like the European Reference Networks (ERNs) to adopt or adapt these models for their own research needs will be explored. In addition, the specific question of diagnosis for undiagnosed patients and how it can be achieved within the ERN system will be discussed. During the debate, questions like: ‘how can ERNs establish their research side?’ and ‘what are their priorities and what needs to be put in place to support this?’ will be further explored (**Chair: Matthew Bolz-Johnson**)*

- The EORTC model (**Denis Lacombe**, Director General, European Organisation for Research and Treatment of Cancer (EORTC))
- Clinical Research Network for RD in the US (**Petra Kaufmann**, Director, Office of Rare Diseases Research and the Division of Clinical Innovation, National Center for Advancing Translational Sciences (NCATS), USA, *video message*)
- RD-ACTION –ERNs’ research needs and opportunities (**Victoria Hedley**, RD-ACTION Thematic Coordinator, Newcastle University John Walton Muscular Dystrophy Research Centre)
- Developing a central undiagnosed hub to rare disease ERNs? (**Olaf Rieß**, Head of the Institute of Medical Genetics and Applied Genomics, Universitätsklinikum Tübingen, Germany)

Debate and Q&A

- **12h30 – 14h00: LUNCH at Casino Maltese, Valletta**

- **SESSION 3: RARE DISEASES RESEARCH IN THE FUTURE: PERSPECTIVES, NEEDS AND OPPORTUNITIES**

14h00– 16h00

*Each panellist will present a few slides with their vision for rare diseases research, taking into account their own needs and concerns, and highlighting the opportunities they see in the future. The chairs will highlight compatible and diverging elements and will encourage an active discussion to identify opportunities and a common-ground for sustainable European rare disease research (**Chairs: Alex Felice and Daria Julkowska**)*

- Patient representatives (**Avril Daly**, Vice-President of the EURORDIS, Chief Executive, Retina International, and **Simona Bellagambi**, Member of EURORDIS Board of Directors)

- European Commission:
 - DG Research (**Irene Norstedt**, Head of Unit, Innovative and Personalised Medicine, DG Research and Innovation (DG RDT), European Commission)
 - DG SANTE (**Jaroslav Waligora**, Policy Officer, Unit Health programme and chronic diseases, DG Health and Food Safety (DG SANTE), European Commission)
- National Perspectives:
 - Malta (**Alex Felice**, Professor and Consultant Geneticist, University of Malta / BBMRI.mt)
 - France (**Anne Paoletti**, Directorate-General for Research and Innovation, Ministry of Education, Higher Education and Research, France)
 - Italy (**Maria Luisa Lavitrano**, Professor at the School of medicine and surgery, University of Milano-Bicocca, Italy / BBMRI.it)
 - Germany (**Ralph Schuster**, Project leader, Clinical research and structural funding, DLR Project Management Agency, Germany)
 - The Netherlands (**Sonja van Weely**, Scientific officer at The Netherlands Organization for Health Research and Development ZonMw)
- Researchers (**Ana Rath**, Director, INSERM US14-Orphanet, and **Kurt Zatloukal**, Vice-chair of the institute of Pathology, Medical University of Graz, Austria / BBMRI.at)
- ERN Case Studies: perspectives from ERN coordinators (**Luca Sangiorgi**, Head of Medical Genetic Department, Rizzoli Orthopaedic Institute, Italy and Coordinator of the Rare Bone Disease ERN / BBMRI.it)

Debate and Q&A

- **WRAP UP: Yann Le Cam. 16h00 – 16h15**
- **16h15 – 16h30 ‘FAMILY’ PHOTO**
- **16h30: END OF MEETING**
- **18h45: BUS Transfer from Grand Excelsior Hotel to Mdina**
- **19h30: SOCIAL EVENT AND RECEPTION/DINNER AT MDINA**
- **22h30: BUS Transfer from Mdina to Grand Excelsior Hotel, Valletta**