36th Workshop of the
EURORDIS Round Table of Companies (ERTC)
10th October 2023
09.30-17.00 CET
Barcelona

Overcoming Barriers to Rare Disease Diagnosis

Overarching objective and scope

This workshop aims to reflect on the challenges associated with diagnosing rare diseases and to explore practical solutions to improve the path to diagnosis. By identifying bottlenecks in the diagnostic pipeline and bringing together patients, policy makers, developers, diagnostic solution providers and researchers, the workshop seeks to foster collaboration and facilitate knowledge exchange for the benefit of people living with a rare disease in Europe and beyond.

Evidence collected from thousands of people living with a rare disease in the context of the EURORDIS Rare Barometer latest surveys on Diagnosis and Newborn Screening will be used as a backdrop to the discussions. This will set the scene to help workshop participants learn from patients and their carers about their journey to diagnosis and their perspectives on the factors influencing the process of obtaining an accurate and timely diagnosis.

Latest insights into European infrastructure and technological advances contributing to streamlining the diagnostic pathway will then be presented. Participants will have the opportunity to discuss the potential of these solutions to facilitate patient access to diagnosis. Another key focus of the workshop will be to reflect on the potential of clinical diagnostic networks in addressing the devastating impact of undiagnosed conditions on the lives of patients and their families, and to discuss collaboration opportunities between researchers and developers to advance diagnostic research.

The workshop will also investigate the potential of personalised treatments based on genomic data and examine the European political landscape surrounding the expansion of newborn screening, highlighting emerging technologies and best practices through pilot studies. Patient data being paramount in our collective work, the need for clear guidelines for patients on how and where to share data will also be emphasised.

Objectives

- Understand the goals set by EURORDIS and IRDiRC to improve and shorten the path to rare disease diagnosis
- Hear from people living with a rare disease about diagnosis delays and the challenges in the healthcare system they are still facing
- Understand what clinical diagnostic networks are and explore the potential of a close collaboration between industry and research in contributing to advancing diagnostic research and progress on undiagnosed cases.
- Recognise the need for clear guidelines for patients on how, where, and why to share data, emphasising the importance of data shareability
- Investigate the potential of personalised treatments based on genomic data
- Examine the political landscape surrounding the expansion of newborn screening programmes
- Foster discussions and strategies to promote a balanced approach between treatment-oriented pipelines and diagnostic pipelines
- Network, build connections and share perspectives and experiences

PROGRAMME

Co-Chairs:
Shirlene Badger, Global Patient Advocacy Lead, Illumina
Anna Arellanesová, Chairperson, Rare Diseases Czech Republic
Deputy General Secretary, EURORDIS-Rare Diseases Europe

9.00 – 9.30	**REGISTRATION**
9.30 – 9.45	**Welcome & Introduction by the Co-Chairs**
9.45 – 10.00	**Setting the scene** **Yann Le Cam,** Chief Executive Officer, EURORDIS-Rare Diseases Europe **Sergi Beltran**, Bioinformatics Unit Head, Centre Nacional d'Anàlisi Genòmica CNAG-CRG
10.00 – 11.00	• **EURORDIS Rare Barometer key survey findings** Perspectives from people living with a rare disease on diagnosis and newborn screening (NBS) **Jessie Dubief**, Rare Barometer Programme Senior Manager, EURORDIS-Rare Diseases Europe • **Case study from Spain** Analysis on diagnostic delays in rare diseases

	Mario Gómez-Martínez, PhD student, Institute of Rare Diseases Research (IIER), Instituto de Salud Carlos III
11.00–11.30	**MORNING COMFORT BREAK**
11.30–13.00	**Potential Solutions and Lessons Learnt** • Better organisation of healthcare in a national system. Best practices in NBS in Greece (from the perspective of a lab). **Eleana Petropoulou,** Chief Newborn Screening Dietitian for Greece Department of Newborn Screening Institute of Child Health • Developing infrastructure and best practices in early diagnosis, including in the field of NBS, at the European and international levels. **Laurence Faivre,** Professor in clinical genetics, University hospital of Dijon • Analysis and reanalysis of results and overall advances in bioinformatics: Solve-RD **Sergi Beltran,** Bioinformatics Unit Head, Centre Nacional d'Anàlisi Genòmica CNAG-CRG • Delivering better health outcomes through technology (linked to AI / IT solutions that use medical symptoms to accelerate diagnosis). **Julián Isla-Gómez,** Founder, Foundation 29
13.00-14.30	**LUNCH**
14.30-15.45	**Breakout sessions: what are the missing steps to help overcome persisting barriers to diagnosis?** **New perspectives on rare disease research** ***Patient case stories:*** **Claudia Beard,** SWAN UK ***Discussion topics:*** why should diagnosis and treatment always be linked? How can prediction models enhance the natural history data and real-world data informing the prognosis, in the absence of a molecular marker?

	Moderator: **Alexandre Méjat,** Deputy Director, International Scientific Networks, AFM-Téléthon and EURORDIS-Rare Diseases Europe Board of Directors *Rapporteur:* **Walter Atzori,** Senior Director, Head of International Patient Advocacy, Alexion AstraZeneca Rare Disease **Improving interactions between industry and healthcare professionals in primary care settings** *Patient case study:* **Jennifer Reyes,** President, Association La Maison 8p ***Discussion topics:*** how can industry work efficiently with healthcare providers (e.g. improved interoperability and data protection, better incentives)? Which advances and practical solutions in technology can support these interactions? *Moderator:* **Antoni Montserrat,** Vice President, ALAN Maladies Rares Luxembourg *Rapporteur:* **Anne-Sophie Chalandon,** Head of Public Affairs, Rare Diseases Policy, Sanofi **Improving knowledge and awareness** *Patient case study:* **Daniel de Vicente,** President, Asociación de pacientes ASMD España ***Discussion topics:*** best practices around sharing knowledge on rare diseases with patients, families and healthcare settings, including understanding of genomic data and delivery of genomic results. How can industry work with the rare disease community to help advance advocacy messages on this topic? *Moderator:* **David Rintell,** SVP, Head of Patient Advocacy, Bridge Bio *Rapporteur:* **Nick Meade,** Director of Policy, Genetic Alliance UK
15.45 – 16.05	**AFTERNOON COMFORT BREAK**
16.05 – 16.35	**Reconvene and Share Highlights from breakout sessions** Rapporteurs and Co-Chairs
16.35 – 16.40	**Key takeaways from a healthcare corporate** **Grainne Crowley,** Head of EU Patient Advocacy and Public Affairs, Ultragenyx
16.40-16.55	**Summary and wrap up,** including perspectives from the Rare Disease Moonshot initiative

	Klaus Viel, Head Global Medical Affairs Operations, Boehringer Ingelheim
16:55-17:00	**Closing and Next Steps by co-Chairs**