# 30th Workshop of the EURORDIS Round Table of Companies (ERTC)

## Newborn Screening: Shaping the future

15-16 October 2020 ONLINE

### FINAL PROGRAMME

**Thursday 15th OCTOBER**  
13:00-18:00  
Session chaired by:  
**Dr. Gulcin Gumus**, Research and Policy Project Manager, EURORDIS-Rare Diseases Europe

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| 13:00 – 13:10 | Welcome introduction, setting the scene & goals for the workshop  
Yann Le Cam, Chief Executive Officer, EURORDIS-Rare Diseases Europe |
| 13:10-13:15  | Programme of the day  
Dr. Gulcin Gumus, Research and Policy Project Manager, EURORDIS-Rare Diseases Europe |
| 13:15 – 13:30 | Newborn Screening Landscape  
EU perspective- **Dr. Martina Cornel**, Professor of Community Genetics & Public Health Genomics, Amsterdam University Medical Center |
| 13:30 – 15:15 | Session 1: Learning from experiences and pilots (Comfort break after 45 min)  
Panel discussion with representatives from different stakeholder groups in newborn screening.  
- UK Newborn Screening programme – **Sara Hunt**, Chief Executive Officer, Alex, The Leukodystrophy Charity  
- Italian Extended Newborn Screening programme – **Prof. Giancarlo La Marca**, Head, Newborn Screening, Clinical Chemistry and Pharmacology Lab, Meyer Children's Hospital  
- French Newborn Screening programme - **Marie-Christine Ouillade**, President, SMA NBS Alliance  
- SMA newborn screening: perspectives and learnings from pilots - **Wildon Farwell**, Global Medical Head of Neuromuscular Diseases, Biogen  
- Q&A |
| 15:15 – 15:30 | Break |

**Ethical, social and economic ramifications**
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| 15:30 – 16:30 | • Ethical & Social implications of Newborn Screening (15 min)  Prof. Pascal Borry, Interfaculty Centre for Biomedical Ethics and Law, University of Leuven  
• Economic implications of Newborn Screening (15 min)  Prof. Jim Chilcott, Professor of Healthcare Decision Modelling, School of Health and Related Research, University of Sheffield  
• Q&A | (Online facilitated networking session) |
| 16:30 – 18:00 | Meet the participants | |

**Friday 16th OCTOBER**

**13:00-17:00**

Session chaired by: Dr. Virginie Bros Facer, Scientific Director, EURORDIS-Rare Diseases Europe

| Workshop session 1: Newborn Screening as a whole system, not just a test (Comfort Break 14:30-14:45) |
| Introduction on Newborn Screening: Working Group & EURORDIS Draft position statements (10 minute video) – Dr. Gulcin Gumus, Research and Policy Project Manager, EURORDIS-Rare Diseases Europe |
| **Moderator:** Nick Meade, Director of Policy, Genetic Alliance UK  
**Rapporteur:** Sarah Creviston, Global Head, Patient Advocacy and Government Affairs, Novartis Gene Therapies |

**Workshop session 2: Empowerment & follow up support (Comfort Break 14:30-14:45)**

Introduction on Newborn Screening: Working Group & EURORDIS Draft position statements (10 minute video) – Dr. Gulcin Gumus, Research and Policy Project Manager, EURORDIS-Rare Diseases Europe

**Moderator:** Simona Bellagambi, UNIAMO, EURORDIS Board of Directors  
**Rapporteur:** Charlotte Chanson, Director, Diagnostics EMEA, Orchard Therapeutics

**Workshop session 3: From shaping the future to concrete implementation (Comfort Break 14:30-14:45)**

Introduction on Newborn Screening: Working Group & EURORDIS Draft position statements (10 minute video) – Dr. Gulcin Gumus, Research and Policy Project Manager, EURORDIS-Rare Diseases Europe

**Moderator:** Valentina Bottarelli, Public Affairs Director, Head of European and International Advocacy, EURORDIS  
**Rapporteur:** Gabriella Pasciullo, European Regional Medical Lead, Bluebird Bio
## Workshop session 4: Maximising the potential  
*(Comfort Break 14:30-14:45)*

Introduction on Newborn Screening- Working Group & EURORDIS Draft position statements (10 minute video) – **Dr. Gulcin Gumus**, Research and Policy Project Manager, EURORDIS-Rare Diseases Europe  
**Moderator:** Dr. Cor Oosterwijk, Director, VSOP – Dutch National Patient Alliance for Rare and Genetic Diseases  
**Rapporteur:** Shirlene Badger, Patient Advocacy Manager, Illumina Cambridge Ltd.

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<td>15:40-16:00</td>
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| 16:00 – 16:45 | **Feedback from breakout sessions**  
**Moderator:** Dr. Virginie Bros Facer, Scientific Director, EURORDIS-Rare Diseases Europe  
**Rapporteurs:** Refer to breakout sessions above |
| 16:45 – 16:50 | **Key take-home messages:** The importance for industry to support the advocacy of Newborn Screening  
**Sian O’Neill**, Senior Director, Patient Engagement EMEA, PTC Therapeutics |
| 16:50 – 17:00 | **Concluding remarks**  
**Yann Le Cam**, EURORDIS-Rare Diseases Europe, Chief Executive Officer |
| 17:00 | **Meeting ends** |