

"Rare Diseases Research across the Life Continuum"

RARE DISEASE DAY.ORG
#RARE DISEASE DAY



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Rare Disease Day is organised by EURORDIS-Rare Diseases Europe and 65 National alliances of patient organisations for rare diseases.

Date 25th February 2022

Time: 13.00-17.00 (GMT)

Register for our free [Zoom Webinar](#) here



Chairs: Prof Amy Jayne McKnight & Anne Lawlor		
13.00 – 13.10	Welcome Address: Prof Thilo Kroll , Professor of Health Systems Management UCD Centre for Interdisciplinary Research, Education and Innovation in Health Systems (UCD IRIS)	
13.10 – 13.30	Keynote Address: Associate Prof Emma Tonkin (Genomic Healthcare) Genomics Policy Unit, University of South Wales Genomic Medicine Service Alliances and role of nurses and midwives within the new structure	
13.30-13.45	Prof. Amy Jayne McKnight and Dr Katie Kerr, Queen’s University Belfast	Biomolecular approaches are improving rare disease diagnosis
13.45-14.00	Dr Edmund Gilbert Postdoctoral Researcher RCSI University of Medicine and Health Sciences	Genomic insights into the population structure and history of the Irish Travellers
14.00-14.15	Dr David Issom , Director of Global Health Information and Education European Sickle Cell Federation.	Sickle Cell Patient Association and Patient led Research
14.15-14.30	Dr. Fionnuala Mone , Belfast NHS Trust/QUB	Evolution of Prenatal Exome Sequencing in the NHS?
14.30-14.45	Dr. James O’Byrne , UCD Medicine /Mater Misericordiae University Hospital	Precision Medicine: A focus on Clinical Utility
14.45-15.00	Comfort Break	
Chairs: Dr. Suja Somanadhan & Rhoda Walker		
15.00-15.30	Les Martin, Anne Lawlor Julie Power -RAINDROP- PPI Expert Advisory Group, UCD	TRUE PPI – Rare Diseases Research
15.30-15.45	Dr Carol Somers , University College Cork (UCC)	Understanding the experiences of parenting a child with Ehlers-Danlos Syndrome.
15.45-16.00	Mary Ellen O’Shea , Munster Technological University	The self-expressed quality of life of Irish adults with early diagnosed Phenylketonuria on long term dietary therapy
16.00-16.15	Dr Maria Caples , University College Cork (UCC)	22q11.2 Deletion Syndrome: Factors that Influence Family Adaptation
16.15-16.30	Dr. Karen LeGrow , Associate Professor in the Daphne Cockwell School of Nursing, Ryerson University. Canada	Impact of COVID-19 on pediatric home care services for children with rare diseases and/or complex healthcare needs: Parents' and healthcare practitioners' experiences
16.30-17.00	Closing Keynote Address “Innovation at the Centre of Rare Diseases Therapy”	