

# ERICA ERN Research Conference

11-13 December 2024 (Udine, Italy)

 programme: **Wednesday, December 11<sup>th</sup> 2024**  
 hosted by: MetabERN / ASU FC


programme

DAY 1	
13:30 - 14:00	Registration & Coffee
Conference Opening	
Setting the Scene for ERN, RD Research across	
Welcome	
14:00 – 14:10	By host <i>Maurizio Scarpa</i> , MetabERN and <i>Alberto Pereira</i> , ERICA/Endo-ERN Opening words by <i>David Turello</i> , Chief Medical Director of ASUFC
14:10 – 14:30	ERN Research: The Expected ERICA Legacy By <i>Hélène Le Borgne</i> , European Commission
14:30 – 14:40	Key achievements from ERICA Chaired by <i>Alberto Pereira</i> (ERICA Coordinator)
Work Packages sessions (WP2 - WP6) WP-leads à 20min (15min+5min Questions)	
14:40 – 15:00	<b>WP2</b> 'Data Collection, Integration and Sharing' By <i>Franz Schaefer</i> and <i>Eduardo Lopez Granados</i>
15:00 – 15:20	<b>WP3</b> 'Patient Centered Research' By <i>Ana Rath</i> and <i>Céline Desvignes-Gleizes</i>
15:20 – 15:40	<b>WP4</b> 'Clinical Trial Support' By <i>Luca Sangiorgi</i> and <i>Ralf-Dieter Hilgers</i>
15:40 – 16:00	<b>WP5</b> 'Translation and Innovation' By <i>Ruth Ladenstein</i> and <i>Anton Ussi</i>
16:00 – 16:25	Coffee break
16:25 – 16:45	<b>WP6</b> 'Integration, Outreach & Dissemination' By <i>Ana Rath</i> , <i>Alberto Pereira</i> and <i>Maurizio Scarpa</i>
16:45 – 17:00	Conclusions & Future Prospectives
17:00– 17:30	Poster pitches à 3 min
17:30 – 18:30	Poster Presentations & Networking reception
18:30	End DAY 1
20:00 – 22:00	Faculty Dinner

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DAY 2	
ERN Research	
8:30 – 9:00	Gathering & Coffee
<b>Session 1: Diagnostic Research</b> (Data Readiness, Genome re-analysis research pipeline, Genomic innovation to shorten time to diagnosis)	
9:00 – 9:15	Plenary: The impact of SOLVERD to research & future of RD Diagnostics Moderated by <i>Holm Graessner</i> , PI SolveRD/ERN-RND
9:15 – 9:30	Solving the unsolved rare disease cases by working together in a shared analysis infrastructure with flexible variant calling and interpretation to accommodate for specific research questions By <i>Lennart Johansson</i> , MOLGENIS, UMCG (online)
9:30 – 9:45	The genetic landscape of dilated cardiomyopathy in Hungary By <i>Beáta Csányi</i> ERN GUARD-Heart
9:45 – 10:00	ERN ReCONNET Red Flags for primary care: An approach to promote early diagnosis in rare and complex conditions By <i>Rosaria Talarico</i> ReCONNET
<b>Session 2: Outcome Research</b> (Real World Data, Clinical Outcome Assessment )	
10:00 – 10:15	Using primary healthcare data for clinical research: Directions and Barriers By <i>Franz Schäfer</i> , ERKNet
10:15 – 10:30	Achieving continuous quality improvement in a european rare disease network (Insert ERN): A conceptual framework By <i>Olivia Spivack</i> ERNICA
10:30 – 10:45	Accelerating and Reducing the Cost of Literature Reviews and Regulatory Dossiers for Rare Diseases with AI By <i>Meelis Lootus</i> , Tehistark, UK
10:45 – 11:00	European registries for rare endocrine and bone conditions: Electronic tools for mapping and studying rare diseases By <i>Tess de Rooij</i> , Endo-ERN
11:00 – 11:25	Coffee break
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	<b>DAY 2 (continued)</b>
	<b>Session 3: Innovative Therapies (Advanced Therapeutic Medicinal Products , N-of-few approach)</b>
<b>11:25 – 11:45</b>	A roadmap to tailored genetic interventions in patients with neurodegenerative disorders <i>By Willeke van Roon-Mom, LUMC</i>
<b>11:45 – 12:00</b>	Obinutuzumab in rituximab-intolerant Anca-associated vasculitis patients <i>By Jolijn van Leeuwen ERKNet and ERN-RITA</i>
	<b>Session 4: Clinical Trials (Drug Trials, inc. nr of few clinical trials; Non-Pharmacological interventions)</b>
<b>12:00 – 12:15</b>	Plenary: Clinical Trials Methodological considerations <i>Moderated by Ralf-Dieter Hilgers, RealiseD</i>
<b>12:15 – 12:30</b>	Overcoming challenges in epilepsy clinical trials: The role of european consortium for epilepsy trials (ECET) <i>By Sébile Tchaicha, ERN-EpiCARE</i>
<b>12:30 – 12:45</b>	The Impact of Allocation Bias in Rare Disease Clinical Trials with Multiple Endpoints <i>By Stefanie Schoenen, RWTH Aachen</i>
<b>12:45 – 13:00</b>	Thyroid hormone analogue (TRIAC) therapy for resistance to thyroid hormone in children: Proposal for an international multi-center clinical trial <i>By Edward Visser, Endo-ERN</i>
<b>13:00 – 13:20</b>	Summary Panel: Lessons Learned and ERICA output / ERNs Research & Clinical Research Network(CRN) <i>Moderated by Alberto Pereira, ERICA with ERICA WP-leads and Session leads</i>
<b>13:20 – 14:10</b>	<b>Lunch</b>
	<b>Clinical Trials &amp; Supporting Networks</b>
<b>14:10 – 14:30</b>	PLUTO Program- research in forgotten disorders <i>By Diego Ardigo, Chiesi, IRDiRC</i>
<b>14:30 – 14:50</b>	EMA ACT-EU (Accelerate Clinical Trials in EU) <i>By Kristina Larsson, EMA</i>
<b>14:50 – 15:05</b>	Connect4Children (c4c), Success stories from PPPs <i>By Ricardo Fernandes, c4c</i>
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	<b>DAY 2 (continued)</b>
	<b>Clinical Trials &amp; Supporting Networks</b>
<b>15:05 – 15:25</b>	<b>RealiseD, other IHI Projects</b> By <i>Ralf-Dieter Hilgers &amp; Solange Rohou</i> , EFPIA
<b>15:25 – 15:40</b>	<b>Panel Discussion: Needs &amp; Hurdles</b> With <i>Kristina Larsson</i> , EMA, <i>Heidrun Hildebrand</i> EFPIA/c4c and <i>Diego Ardigò</i> , Chiesi Beside conducting CTs also methodological support, contacting expertise etc. Chaired by <i>Ralf-Dieter Hilgers</i>
<b>15:40 – 16:00</b>	<b>Coffee break</b>
<b>16:00 – 16:45</b>	<b>Oxford Debate – Does AI work for RD?</b> Moderated by <i>Anton Ussi</i> , EATRIS <b>Favor:</b> <i>Maurizio Scarpa</i> , MetabERN <b>Against:</b> <i>Ruth Ladenstein</i> , ERN-PaedCan
<b>16:45 – 18:15</b>	<b>Innovation Workshop</b> Chaired by <i>Ruth Ladenstein</i> , ERN-PaedCan with <i>Gilles Vassal</i> & invited Panelists
<b>18:15</b>	<b>End DAY 2</b>
<b>20:00 – 22:00</b>	<b>ERN Research Party Social Programme/Dinner including Abstract Award Ceremony</b>

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Future of the ERN Research	
8:30 – 9:00	Gathering & Coffee
9:00 – 9:20	<b>Building together the ERN Research Roadmap &amp; Continuation of ERN Research cooperation</b> Current status of ERN Research and GAP-Analysis (ERN involvement in JTCs). How to respond to all the research needs of the ERNs. Need of ERN Research Conference By <i>Franz Schäfer &amp; Alberto Pereira</i>
Alignment of ERDERA & Jardin & Together4RD	
9:20 – 9:45	<b>Research funding opportunities &amp; ERNs within ERDERA</b> By <i>Daria Julkowska</i> (20+5min)
9:45 – 10:05	<b>The role of the National Mirror Groups - NMGs</b> By <i>Sonja van Weely</i> (15+5min)
10:05 – 10:25	<b>Jardin &amp; ERN Research</b> By <i>Luca Sangiorgi</i> , ERN-Bond/Jardin (online, 15+5min)
10:25 – 10:45	<b>Together4RD Industry &amp; ERN Cooperation</b> By <i>Sheela Upadhyaya</i> (15+5min)
10:45 – 11:10	Coffee break
11:10 – 11:45	<b>ERN Research strategy alignment</b> <b>ERN Alignment ERDERA WP25.4</b> Panel discussion on corporate identity of ERN Research With ERDERA Moderated by <i>Maurizio Scarpa</i> and <i>Alexis Arzimanoglou</i> with all ERICA WP-leads
11:45 – 12:00	Take home messages and final words
12:00 – 13:00	Goodbye lunch