

2025 RYR-1-RELATED DISEASES PATIENT-LED INTERNATIONAL RESEARCH WORKSHOP: NOVEL PERSPECTIVES, TREATMENTS & INTERVENTIONS

Wednesday, July 23, 2025

Time	Topic/Session	Speaker
7:30 am - 8:30 am	Breakfast (Allegheny ABC)	
8:30 am - 8:50 am	Patient Testimonials	Jinson Paul, MBA* Melanie Moore
8:50 am - 9:00 am	Session 1: Diagnosis & Genetics of RYR-1-Related Diseases (RYR-1-RD)	Livija Medne, MS, LCGC
9:00 am - 9:30 am	An Update on the <i>RYR1</i> Database Project	Joshua Todd, PhD, MBA
9:30 am - 10:00 am	ClinGen VCEP Efforts for <i>RYR1</i> Variant Curation	Pamela Ajuyah Robertson, PhD
10:00 am - 10:30 am	Functional Readouts	Robert T. Dirksen, PhD
10:30 am - 11:00 am	Break	
11:00 am - 11:20 am	Patient Testimonials	Mairéad Ryan, BS, V27 Ben Huseth
11:20 am - 11:50 am	The Prevalence of RYR1-Related Disease - an International, Collaborative Multicentre Study	Arti Mistry, PhD & Anna Sarkozy, MD, PhD*
11:50 am - 12:00 pm	Session 2: Establishing Clinical Trial Readiness for RYR-1-Related Diseases (RYR-1-RD)	Payam Mohassel, MD
12:00 pm - 12:30 pm	Natural History Studies and Registries	Nicol C. Voermans, MD, PhD
12:30 pm - 1:00 pm	Muscle Imaging as a Diagnostic and Prognostic Biomarker of Neuromuscular Disease	Giorgio Tasca, MD, PhD
1:00 pm - 2:00 pm	Lunch (Allegheny ABC)	
2:00 pm - 2:30 pm	Advancing Clinical Trials in Neuromuscular Disorders with Wearable and Digital Health Technologies	Ashkan Vaziri, PhD
2:30 pm - 2:50 pm	Stratification Tools and Disease Severity Scales for Neuromuscular Clinical Trials	Jessica Rose Nance, MD
2:50 pm - 3:20 pm	Panel on Establishing Clinical Trial Readiness for RYR-1-RD <i>Patient Representative Co-Directors identified this as a key priority</i>	Moderator: Payam Mohassel, MD Panelists: Tom Crawford, MD Jessica Rose Nance, MD Giorgio Tasca, MD, PhD Ashkan Vaziri, PhD Nicol C. Voermans, MD, PhD
3:20 pm - 3:40 pm	Patient Testimonials	Avi Orseck Patricia Powers, MDiv, PhD*
3:40 pm - 3:50 pm	Break	
3:50 pm - 4:00 pm	Session 3: Novel Insight into RyR1 Interplay and Cell Pathophysiology	Johanna Lanner, PhD
4:00 pm - 4:30 pm	Not all RyR1 Mutations are Created Equal: Insights From CryoEM and Effect of Dantrolene	Montserrat Samsó, PhD
4:30 pm - 5:00 pm	Reduced Skeletal Muscle Ryanodine Receptor Type 1 Content Induces Endo/Sarcoplasmic Reticulum and Metabolic Stress	Nadège Zanou, MD, PhD
5:00 pm - 5:30 pm	Elevated Basal Calcium in Muscle is a Type 2 Diabetes Risk Factor	Eshwar Tammineni, PhD
6:30 pm - 7:30 pm	Dinner (Allegheny ABC)	

*Remote Participation

2025 RYR-1-RELATED DISEASES PATIENT-LED INTERNATIONAL
RESEARCH WORKSHOP: NOVEL PERSPECTIVES,
TREATMENTS & INTERVENTIONS

Thursday, July 24, 2025		
Time	Topic/Session	Speaker
7:30 am - 8:30 am	Breakfast (Allegheny ABC)	
8:30 am - 8:50 am	Patient Testimonials	Daniel Eppel Sarah Keller*
8:50 am - 9:00 am	Session 4: Translational Studies & Therapeutics Development Part I	Johanna Lanner, PhD
9:00 am - 9:30 am	An Inducible and Muscle Specific RyR1 KO Mouse Model and its Therapeutic Interest	Isabelle Marty, PhD
9:30 am - 10:00 am	Targeting Defective Muscle Stem Cells in Myopathies	Nicolas Dumont, PT, PhD
10:00 am - 10:30 am	Advanced <i>in Vitro</i> Modeling of Neuromuscular Diseases and Therapeutics	Francesco Saverio Tedesco, MD, PhD, FHEA, FRCPCH
10:30 am - 11:00 am	Break	
11:00 am - 11:20 am	Patient Testimonials	Emily Pedersen, MS, CTRS Justin Michael Bonny
11:20 am - 11:30 am	Session 5: Therapeutic Pipeline	Michael Goldberg, MD, MPH
11:30 am - 12:00 pm	Gene Editing and Gene Replacement Therapy for RYR-1-RD: Opportunities and Challenges	Isabelle Marty, PhD
12:00 pm - 12:30 pm	Novel Gene Transfer Approaches: Split Inteins	Hichem Tasfaout, PhD, PharmD, MS
12:30 pm - 1:30 pm	Lunch (Allegheny ABC)	
1:30 pm - 2:00 pm	Using HSV as a Viral Vector for <i>RYR1</i>	Selene Ingusci, PhD
2:00 pm - 2:30 pm	Restoring RyR1 Expression via a Novel RNA End-Joining Strategy	Afrooz Rashnonejad, MSc, PhD
2:30 pm - 3:00 pm	Development of ACE-tRNA Technologies as Platform Nonsense Therapeutics	John Lueck, PhD
3:00 pm - 3:10 pm	Break & Group Photo	
3:10 pm - 3:30 pm	Patient Testimonials	Jenny C. Roller Curt D. Keller, DMin
3:30 pm - 3:40 pm	Session 6: Physical Therapy & Exercise Approaches in RYR-1-RD <i>Patient Representative Co-Directors identified this as a key priority</i>	Mairéad Ryan, BS, V27
3:40 pm - 4:10 pm	A Pathway to Progress: Physical Therapy in RYR1-RD	Minal S. Jain, PT, DSc, FAPTA
4:10 pm - 4:40 pm	Introduction to Exercise and Muscle Adaptation in Health and Disease	Johanna Lanner, PhD
5:00 pm - 9:00 pm	Kickoff of The RYR-1 Foundation International Family Conference 2025	

*Remote Participation