

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

Wednesday, July 23, 2025

Topic/Session	Session Chair/Confirmed Speakers
Session 1: Diagnosis & Genetics of RYR-1-RD	Livija Medne, MS, LCGC
<i>RYR1</i> Database	Joshua Todd, PhD & Tokunbor Lawal, PhD (tentative)
ClinVar VCEP Efforts for <i>RYR1</i> Variant Curation	Pamela Ajuyah Robertson, PhD
Functional Readouts	Robert T. Dirksen, PhD
The prevalence of RYR1-related disease - an international, collaborative multicentre study and needs to be inserted between	Arti Mistry, PhD
Session 2: Clinical Trial Readiness	Payam Mohassel, MD
Natural History and Registries	Nicol Voermans, MD, PhD
Muscle Imaging	Giorgio Tasca, MD, PhD
Wearables	Ashkan Vaziri, PhD
RYR-1-RD Severity Classification System / Clinical Trial Outcomes	Panel: TBD
Session 3: Novel Insight into RyR1 Interplay and Cell Pathophysiology	Johanna Lanner, PhD
Ion and Small Molecule Interplay with RyR1 Function	Montserrat Samso, PhD
Reduced RyR1 Content and Cellular Stress	Nadège Zanou, MD, PhD
MH and Altered Glucose Handling in Muscle	Eshwar Tammineni, PhD

Patient testimonials will be featured throughout the Research Workshop.

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Thursday, July 24, 2025

Topic/Session	Session Chair/Confirmed Speakers
Session 4: <i>RYR1</i> and Knowledge Gained from Animal Models	Johanna Lanner, PhD
From Zebrafish to Rodents	TBD
Large Animal Model	TBD
Neuromuscular Regeneration in Organoids	Francesco Saverio Tedesco, MD, PhD
Session 5: Therapeutic Pipeline	Michael Goldberg, MD, MPH
Gene Editing and Gene Replacement Therapy for RYR-1-RD: Opportunities and Challenges	Isabelle Marty, PhD
Novel Gene Transfer Approaches: Split Inteins	Hichem Tasfaout, PhD, PharmD, MS
Using HSV as a viral vector for <i>RYR1</i>	Selene Ingusci, PhD
Two strategies for restoring RyR1 protein expression	Afrooz Rashnonejad, MSc, PhD
Muscle Regeneration	Nicolas Dumont, PT, PhD
Session 6: Physical Therapy & Exercise Approaches in RYR-1-RD	Mairéad Ryan, BS, V27
Physical Therapy General Guidelines for RYR-1-RD	Minal Jain, PT, MS, DSc
Exercise: Strength vs Cardiovascular & Muscle Function	Johanna Lanner, PhD

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