

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

**Wednesday, July 23, 2025**

Topic/Session	Session Chair/Confirmed Speakers
<b>Session 1: Diagnosis &amp; Genetics of RYR-1-RD</b>	<b>Livija Medne, MS, LCGC</b>
<i>RYR1</i> Database	Joshua Todd, PhD & Tokunbor Lawal, PhD
ClinVar VCEP Efforts for <i>RYR1</i> Variant Curation	TBD
Functional Readouts	Robert T. Dirksen, PhD
<b>The prevalence of RYR1-related disease - an international, collaborative multicentre study and needs to be inserted between</b>	<b>Heinz Jungbluth, MD, PhD</b>
<b>Session 2: Clinical Trial Readiness</b>	<b>Payam Mohassel, MD</b>
Natural History and Registries	Nicol Voermans, MD, PhD
Muscle Imaging	TBD
Wearables	Ashkan Vaziri, PhD
RYR-1-RD Severity Classification System	TBD
<b>Session 3: Novel Insight into RyR1 Interplay and Cell Pathophysiology</b>	<b>Johanna Lanner, PhD</b>
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
<b>Session 4: RYR1 and Knowledge Gained from Animal Models</b>	<b>Johanna Lanner, PhD</b>
From Zebrafish to Rodents	TBD
Dog Model	TBD
Neuromuscular Regeneration in Organoids	Francesco Saverio Tedesco, MD, PhD
<b>Session 5: Therapeutic Pipeline</b>	<b>Michael Goldberg, MD, MPH</b>
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
Using HSV as a viral vector for RYR1	Selene Ingusci
Two strategies for restoring RyR1 protein expression	Afroz Rashnonejad
Muscle Regeneration	Nicolas Dumont, PT, PhD
<b>Session 6: Physical Therapy &amp; Exercise Approaches in RYR-1-RD</b>	<b>Mairéad Ryan, BS, V27</b>
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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