RYR-1 FOUNDATION

STRENGTH IN NUMBERS

RYR-1-Related Diseases Patient-Led International Research Workshop:

Novel Perspectives, Treatments & Interventions

Pittsburgh, PA | USA

July 23 - 24, 2025

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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 *RYR1* Database Project	Joshua Todd, PhD, MBA
9:30 am - 10:00 am	ClinGen VCEP Efforts for *RYR1* 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 *Patient Representative Co-Directors identified this as a key priority*	**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*

Agenda

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 *in Vitro* 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 *RYR1*	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** *Patient Representative Co-Directors identified this as a key priority*	**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*

Sponsors & Donors

The RYR-1 Foundation gratefully acknowledges the generous support of our sponsors for the 2025 RYR-1-Related Diseases Patient-Led International Research Workshop: Novel Perspectives, Treatments & Interventions.

Their partnership and commitment to advancing research in RYR-1-Related Diseases (RYR-1-RD) play a critical role in making this Research Workshop possible. We are deeply appreciative of their investment in scientific collaboration and progress.

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In addition, The RYR-1 Foundation extends heartfelt thanks to the following individual donors for their generous contributions in support of this Research Workshop. Their support fosters innovation, strengthens the community, and drives meaningful progress in the field of RYR-1-RD research.

Drew Huseth
Dr. Laurie Jayne Toomajanian & Mr. Randolph Pepper
Dr. Halah Stern

Grant Support

The RYR-1 Foundation sincerely thanks our grant supporters for their generous funding and commitment to advancing research, education, and patient support for those affected by RYR-1-Related Diseases.

Their invaluable support enables us to host this critical Research Workshop, foster collaboration among experts, and empower affected individuals and families around the world. We are deeply grateful for their partnership in our mission to accelerate the discovery of treatments and, ultimately, a cure.

Co-Directors

**Remote Participation*

The RYR-1 Foundation extends its deepest gratitude to the Co-Directors of the 2025 RYR-1-Related Diseases Patient-Led International Research Workshop: Novel Perspectives, Treatments & Interventions for their vision, expertise, and generous commitment of time. This important Research Workshop would not have been possible without their leadership and dedication.

Michael F. Goldberg, MD, MPH

Dr. Michael F. Goldberg is a board-certified neuroradiologist and serves as the Director of Neuroradiology at the Allegheny Health Network. As someone personally affected by an RYR-1-RD, Mike understands firsthand the importance of building a supportive community of individuals and families facing similar challenges. As a physician, he has leveraged his medical and scientific background to lead The RYR-1 Foundation's efforts to fund critically important RYR-1-RD research. In a relatively short period, the organization's research program has made a significant international impact, contributing to major scientific and medical breakthroughs. Dr. Goldberg currently serves as the board president and co-chair of research at The RYR-1 Foundation. He is also a co-founder of the organization, along with his wife and parents.

Dr. Michael Goldberg serves as President of The RYR-1 Foundation's Board of Directors and Co-Chair of Research.

Johanna Lanner, PhD

Dr. Johanna Lanner is an Associate Professor of Physiology at Karolinska Institutet (KI), Stockholm Sweden, where she leads the Molecular Muscle Physiology and Pharmacology lab. She earned her PhD in Cellular Physiology from KI, with a research exchange at Ulm University in Germany, and completed her postdoctoral training in Professor Susan Hamilton's laboratory at Baylor College of Medicine in Houston, USA. Her research focuses on the molecular underpinnings of muscle function and dysfunction, particularly in the context of calcium signaling, redox biology, as well as mitochondrial function. With over 70 peer-reviewed publications and an h-index of 27, she is recognized in the field, frequently invited as a speaker at international conferences. She holds several editorial roles, including Associate Editor for *Acta Physiologica*, and has received numerous competitive grants and honors. Dr. Lanner also plays an active role in academic leadership and education, including serving as President-elect of the Scandinavian Physiological Society and chairing educational committees at KI.

Dr. Lanner is a member of The RYR-1 Foundation's Scientific Advisory Board.

Livija Medne, MS, LCGC

Ms. Livija Medne is a senior genetic counselor in the Divisions of Human Genetics and Neurology and a co-director of the Roberts Individualized Medical Genetics Center (RIMGC) at Children's Hospital of Philadelphia (CHOP). For the last 15 years, she has specialized in neuromuscular and neurogenetic disorders and served as a CHOP co-investigator for several clinical research trials and patient registries. As a genetic counselor, her goal is to facilitate accurate and genetically confirmed diagnoses for all children with suspected neuromuscular disorders and other diagnoses. More recently, she has developed expertise in large-scale genomic testing and played a critical role in developing the RIMGC program at CHOP to allow for the incorporation of genomic testing into clinical practice. With the increasing number of testing options and modalities, it is critically important that each and every individual gets access to the right test at the right time. Ms. Medne has developed significant expertise in interpreting genetic testing results for neuromuscular disorders, including RYR-1-RD. While many genetic neuromuscular diagnoses still lack targeted therapies, they can be notably better managed and treated when the exact molecular diagnosis is known.

Ms. Medne is a member of The RYR-1 Foundation's Scientific Advisory Board.

Payam Mohassel, MD

Dr. Payam Mohassel is a physician-scientist and associate professor of Neurology at Johns Hopkins University School of Medicine. He is also the co-director of Johns Hopkins Myositis Center. Dr. Mohassel obtained his medical degree from Johns Hopkins University School of Medicine, where he remained for a medical internship, residency training in neurology, and clinical fellowship training in neuromuscular medicine. He then joined the National Institute of Neurological Disorders and Stroke Neurogenetics branch as a clinical research fellow before returning to Johns Hopkins as a faculty member. Dr. Mohassel's research focuses on translational studies in neuromuscular disorders, and it spans gene discovery efforts, mechanistic studies to identify therapeutic targets, and early phase interventional clinical trials. He was also the principal investigator for the S 48168 (ARM 210) for the Treatment of RYR1-related Myopathies (RYR1-RM) for the first-in-patient trial phase 1 clinical trial.

Dr. Mohassel is a member of The RYR-1 Foundation's Scientific Advisory Board.

Jinson Paul, MBA*

Jinson Paul is a mechanical engineer and MBA in Finance. Based out of Bangalore, India, he currently works with a major European bank. Living with an RYR-1-RD, Jinson understands the unique challenges faced by those affected by this condition and is passionate about making a positive impact in their lives. Having experienced the invaluable support of The RYR-1 Foundation during his diagnosis process, Jinson is eager to contribute to the organization's efforts in finding a cure for RYR-1-RD and helping others affected by the condition.

Jinson serves as an Advisor on The RYR-1 Foundation's Board of Advisors.

Emily Pedersen, MS, CTRS

Emily Pedersen lives in southern New Jersey, where she works as a Recreation Therapist at Magee Rehabilitation Hospital in Philadelphia. She is married and has two daughters, Lillian and Klara, her youngest, who was born with a recessive form of Central Core Disease. Since Klara's birth, Emily has learned the importance of advocacy and raising awareness for both individuals and their families impacted by disabilities. She advocates tirelessly for accessibility and inclusion in her local community and school district where she has served on the school board for six years, ensuring there is a voice for students and families with disabilities. For the past 14 years, the Pedersen Family has been actively raising awareness of RYR-1-RD and supporting The RYR-1 Foundation by hosting a variety of fundraising events. Emily prides herself on being someone people can come to for advice and support. She is always researching new ways to improve accessibility and inclusion for her daughter in every aspect of her life.

Emily serves as a Director on The RYR-1 Foundation's Board of Directors.

Mairéad Ryan, BS, V27

Mairéad Ryan has been involved with The RYR-1 Foundation since 2016 and volunteered at the past Family Conferences. She is one of the Patient Co-Directors for this year's Patient-Led Research Workshop. She was diagnosed with a mild form of Central Core Disease in 2010. Mairéad graduated from the University of California, Davis in 2022 with a degree in Animal Science and a specialization in captive exotic animals. She is currently in her third year of veterinary school at Tufts University where she is also part of the International Veterinary Medicine Program. She hopes to work in the intersection of Conservation Medicine and Theriogenology. She has worked as a veterinary assistant in various countries including Kenya, Costa Rica, and the United States.

In 2017, Mairéad interned for the Beggs' Laboratory at Boston Children's Hospital where she assisted with and learned about research involving animal models with RYR-1-RD. She was the Tufts Elephant Conservation Alliance Fellow for the 2024-2025 Academic Year. In her free time, Mairéad enjoys riding her horse, Trigger, and playing with her cat, Kingston.

Mairéad serves as an Advisor on The RYR-1 Foundation's Board of Advisors.

Expert Speakers

**Remote Participation*

Tom Crawford, MD

Dr. Tom Crawford has been a member of the Department of Neurology at Johns Hopkins University School of Medicine since 1987. He is co-director of the Muscular Dystrophy Association (MDA) Clinic for Neuromuscular Disorders and Neurologist for the Ataxia Telangiectasia Clinical Center at Johns Hopkins. His practice involves general child neurology with a principal interest in caring for children with neuromuscular, neuromotor, and ataxic disorders. His primary research interests involve the basic science and clinical characterization of two important neurologic disorders that affect children: Spinal Muscular Atrophy and Ataxia Telangiectasia. He is also actively involved in the Biology of neurofilaments by characterization of transgenic animal models. He is on the Medical and Scientific Advisory Boards of Families of Spinal Muscular Atrophy, and the Medical Advisory Committee for the MDA. He is the Neurologist for the Ataxia Telangiectasia Clinical Center at Johns Hopkins, which has evaluated almost half of the known patients with this disorder in the United States. Additional specific clinical interests include evaluation and treatment of children with brachial plexus palsies.

Dr. Crawford received his medical degree from the University of Southern California. He completed a pediatric internship and residency at the Pediatric Pavilion of the Los Angeles County / University of Southern California Medical Center, followed by a pediatric chief residency. He completed his training in Neurology with Special Qualification in Child Neurology at Los Angeles Children's Hospital. He then traveled east to the laboratory of Dr. John Griffin at Johns Hopkins for a fellowship in Neuromuscular Disorders. Prior to medical training and Neurology residency, he majored in Psychology and Religion at Yale College.

Dr. Crawford has published extensively and presented nationally and internationally. He has an active role in teaching medical students and residents in neurology. In addition, Dr. Crawford has special interest and experience in EMG studies of children and adults.

Nicolas Dumont, PT, PhD

Dr. Nicolas Dumont earned his PhD at Université Laval (Québec City) and completed his postdoctoral fellowship at the University of Ottawa. In 2016, he joined the Université de Montréal as an assistant professor and became a principal investigator at the CHU Sainte-Justine Research Center. His lab focuses on understanding the regulation of muscle stem cells in regeneration and disease. His research is structured around three main axes: elucidating mechanisms that govern muscle stem cell fate decisions, characterizing how these cells are altered in muscular disorders, and developing precision therapies to restore their function. Dr. Dumont holds the Canada Research Chair in Stem Cells and Neuromuscular Diseases, and his lab is funded by agencies such as the Canadian Institutes of Health Research. He has published greater than 50 articles in leading journals, including *Nature Medicine, Nature Communications, Cell Stem Cell, and Science Advances.*

Selene Ingusci, PhD

Dr. Selene Ingusci was appointed Research Instructor at the University of Pittsburgh, Department of Microbiology and Molecular Genetics, in January 2023. Her expertise lies in molecular biology, with a focus on HSV vector engineering and manufacturing. She earned her BS, MS, and PhD from the University of Ferrara (Italy), where she developed a strong foundation in HSV-mediated gene therapy. Her doctoral research emphasized engineering replication-defective (rd) HSV vectors, while her postdoctoral studies focused on developing tumor-specific oncolytic HSV (oHSV) for glioblastoma therapy.

Dr. Ingusci's current research centers on advancing rdHSV vectors for applications in neurodegenerative and muscular disorders. Her work has led to the identification of HSV genomic loci and engineering designs that support robust, long-term transgene expression in brain and muscle tissues out to one year, without cytotoxicity or inflammation. Leveraging the large genetic capacity of HSV, she has also demonstrated the vector's ability to deliver multiple transgenes under independent transcriptional control across different cell types. These studies provided valuable insights into how noncoding genomic elements regulate cell-type-specific transcription and how to modulate the rdHSV epigenetic landscape to optimize gene expression. Her research continues to enhance the potential of HSV-based gene therapy by optimizing vector stability, tissue specificity, and long-term efficacy.

Looking ahead, Dr. Ingusci is leveraging her expertise to develop an HSV-based gene complementation strategy for RYR1–related myopathies. She will employ engineered rdHSV vectors to deliver the full-length *RYR1* cDNA to skeletal muscle, establishing the foundation for a novel therapeutic approach for congenital muscle diseases.

Dr. Ingusci has been awarded a grant through The RYR-1 Foundation's 2025–2027 Grant Cycle.

John Lueck, PhD

Dr. John Lueck is an associate professor in the Department of Pharmacology and Physiology at the University of Rochester School of Medicine and Dentistry. His lab investigates the mechanistic underpinnings of skeletal muscle weakness in myotonic dystrophy and develops suppressor tRNA-based technologies with hopes for use as therapies for diseases caused by nonsense mutations. His scientific foundation was built during his graduate work in the Dirksen and Thornton labs where he investigated ion channel dysfunction in myotonic dystrophy type 1. This work resulted in high-impact scientific output and sparked his long-standing interest in translational research.

He expanded his expertise in the Campbell lab where he focused on membrane protein biochemistry, skeletal muscle physiology, and gene therapy approaches in the context of muscular dystrophy. A growing interest in genetic code expansion led him to the Ahern lab where he published five studies in three years on suppressor tRNA strategies for cystic fibrosis. Now, as an independent investigator, he continues to explore the therapeutic potential of stop codon suppression *in vivo*, leveraging his broad background in molecular biology, membrane biophysics, and mouse genetics. His long-term goal is to develop new treatments for cystic fibrosis, Duchenne muscular dystrophy and other diseases caused by nonsense mutations, guided by understanding the mechanistic underpinning of each disease.

Arti Mistry, PhD

Dr. Arti Mistry is a Postdoctoral Research Associate at the Randall Centre of Cell & Molecular Biophysics at King's College London. She is currently investigating the prevalence of RYR-1-RDs in a multicentre study in two countries, the United Kingdom and the Netherlands. Arti successfully completed her PhD in the Department of Women and Children's Health (KCL), on "The role of skeletal muscle ryanodine receptor type 1 (RyR1) in uterine vascular and myometrial smooth muscle function during pregnancy," with several related publications currently under review or being prepared for submission. Arti graduated from St George's University of London in 2018 with an MSci in Biomedical Science.

Dr. Mistry is a contributing member of "The Prevalence of RYR1-Related Disease—An International, Collaborative Multicentre Study" supported by a research grant from The RYR-1 Foundation.

Jessica Rose Nance, MD

Dr. Jessica Rose Nance is a pediatric neurologist with specialized training in pediatric neuromuscular disorders. She is an Assistant Professor of Neurology and the Director of the Johns Hopkins Pediatric Neuromuscular Clinical Trials Group. Dr. Nance cares for children and families affected by genetic nerve and muscle diseases in her clinics at Johns Hopkins Hospital and the Kennedy Krieger Institute in Baltimore, Maryland. She is an investigator in several multi-center, industry-sponsored clinical trials evaluating neuromuscular therapies. Dr. Nance's research focuses on the development and validation of measurements of motor function in patients with neuromuscular disorders.

Dr. Nance serves as an Advisor on The RYR-1 Foundation's Board of Advisors.

Afrooz Rashnonejad, MSc, PhD

Dr. Afrooz Rashnonejad is an Assistant Professor of Pediatrics at The Ohio State University College of Medicine and a Principal Investigator at the Jerry R. Mendell Center for Gene Therapy at Nationwide Children's Hospital (NCH). Her research focuses on developing adeno-associated virus (AAV)-based gene therapies for neuromuscular diseases. As part of her doctoral work, she developed a prenatal gene therapy approach for Spinal Muscular Atrophy (SMA) under the mentorship of Dr. Guangping Gao (UMass Medical School) and Dr. Ferda Ozkinay (Ege University). She later completed her postdoctoral training in Dr. Scott Harper's lab at NCH, where she developed RNA-targeting therapies, including AAV.CRISPR-Cas13, miRNA, and U7-snRNA–mediated exon skipping for FSHD. Her lab investigates therapies for neuromuscular diseases such as ACTA1-related Nemaline Myopathy, RYR1-related myopathy, and CMT1B neuropathy. She is also advancing *in-utero* therapies for severe congenital neuromuscular diseases. Dr. Rashnonejad has chaired the American Society of Gene & Cell Therapy (ASGCT)'s New Investigator Committee and serves on the Musculoskeletal Cell and Gene Therapy Committee, and also the Prenatal Cell and Gene Therapy Committee. She was honored to receive the ASGCT Excellence in Research Award in two consecutive years, 2019 and 2020. Dr. Rashnonejad also serves on the scientific advisory boards of several patient foundations.

Dr. Rashnonejad has been awarded a grant through The RYR-1 Foundation's 2025–2027 Grant Cycle.

Pamela Ajuyah Robertson, PhD

Dr. Pamela Ajuyah Robertson is a Genomics Specialist and Senior Biocurator for the Clinical Genome Resource (ClinGen) program at the Broad Institute of MIT and Harvard. She is a variant and gene curation framework expert across several rare disease ClinGen expert panels and affiliated projects. Pamela is co-chair of the ClinGen Curation of ClinVar Working Group, focused on improving the accuracy of ClinVar's aggregate variant classifications. Previously in Australia, she was a Clinical Curation Scientist at the ZERO Childhood Cancer program. There she was involved in several aspects of the program including variant analysis, creating molecular educational materials and workshops, and conducting research. Prior to that she worked in industry for a few years at a biotech start up and an IVF company where she did a mix of genomics analysis and quality control. Pamela completed a PhD investigating the molecular biology of microRNAs in Head and Neck Cancer.

Montserrat Samsó, PhD

Dr. Montserrat Samsó is an Associate Professor in the Department of Cellular, Molecular and Genetic Medicine and Director of the cryo-EM Facility at Virginia Commonwealth University (VCU). She received her PhD degree in Biochemistry and Molecular Biology from the Autonomous University of Barcelona and did postdoctoral training at the Biological Microscopy and Image Reconstruction Unit of the Wadsworth Center (Albany, NY) and then became Instructor and later Assistant Professor at the Anesthesia Department at the Brigham and Women's Hospital/ Harvard Medical School (Boston, MA). Her research focuses on structural biology, using advanced techniques like cryo-electron microscopy (cryo-EM) to study the architecture and function of membrane proteins and ion channels. She concentrates on the ryanodine receptor (RyR) and its disease-associated mutants to better understand and help cure RyR-related disorders. Recently, she identified the binding site and mechanism of dantrolene, a drug used to treat malignant hyperthermia. Dr. Samsó also teaches at the VCU Medical School and mentors students and postdoctoral fellows.

Anna Sarkozy, MD, PhD*

Dr. Anna Sarkozy is a Consultant in Neuromuscular Disorders at the Dubowitz Neuromuscular Centre at Great Ormond Street Hospital for Children and Honorary Senior Lecturer at University College London (UCL) with a major role in the national highly specialised diagnostic and advisory service for congenital muscular dystrophies and congenital myopathies.

After earning her medical degree, completing medical training, and obtaining a PhD at La Sapienza University in Rome, Italy, Anna worked at the Newcastle Muscle Centre from 2008 to 2014 as a specialty doctor in neuromuscular disorders, and then in London from 2014. As part of her clinical activity, Anna runs specialist clinics for patients with rare early-onset neuromuscular conditions.

Her research focuses on improving care for neuromuscular patients and identifying new disease genes, particularly for congenital muscular dystrophies and congenital myopathies. In the last years, she has focused on trial readiness, and is the principal investigator of a UK prospective natural history study for recessive TTN gene-related congenital myopathy.

Dr. Sarkozy is a contributing member of "The Prevalence of RYR1-Related Disease—An International, Collaborative Multicentre Study" supported by a research grant from The RYR-1 Foundation.

Eshwar Tammineni, PhD

Dr. Eshwar Tammineni is a Research Instructor in the Department of Physiology and Biophysics at Rush University Medical Center in Chicago. His research focuses on skeletal muscle physiology, with particular interest in rare skeletal muscle disorders and the role of calcium dysregulation in driving muscle insulin resistance and metabolic disorders such as type 2 diabetes. Dr. Tammineni earned his PhD in Pharmacology, where he studied calcium channel subunits in cardiac muscle. His postdoctoral work expanded into investigating the pathophysiological mechanisms underlying the rare muscle condition malignant hyperthermia.

Giorgio Tasca, MD, PhD

Dr. Giorgio Tasca is a Clinical Professor of Neuromuscular Science at the John Walton Muscular Dystrophy Research Centre, Newcastle University, United Kingdom, since January 2023. He received his residency in Neurology in 2011 and completed a PhD in Neuroscience in 2014 at the Catholic University School of Medicine in Rome. His primary areas of research in neuromuscular disorders include muscle imaging in different adult-onset myopathies and muscular dystrophies, and the study of the molecular mechanisms and biomarkers in Facioscapulohumeral muscular dystrophy (FSHD). He is co-chair of the Neuromuscular Imaging group of the Euro-NMD project (European Reference Network).

Hichem Tasfaout, PhD, PharmD, MS

Dr. Hichem Tasfaout is an Assistant Professor in the Neurology Department of the University of Washington (Seattle, USA). His research interests focus on developing novel gene therapy methods that combine split inteins and myotropic vectors to deliver and express large proteins. Using this approach, multiple protein fragments are delivered specifically to striated muscles using potent myotropic adeno-associated viral (AAV) vectors. Upon their expression, these fragments are then joined into highly functional proteins.

Dr. Tasfaout has been awarded a grant through The RYR-1 Foundation's 2025–2027 Grant Cycle.

Francesco Saverio Tedesco, MD, PhD, FHEA, FRCPCH

Dr. Francesco Saverio Tedesco is a clinician-scientist with expertise in neuromuscular diseases, muscle regeneration and disease modelling. He is Professor of Neuromuscular Biology and Regenerative Medicine at University College London, Senior Group Leader at the Francis Crick Institute and Honorary Consultant Paediatric Neurologist in the Dubowitz Neuromuscular Centre at Great Ormond Street Hospital for Children in London, UK. Prof. Tedesco graduated in Medicine and Surgery with honours at the Sapienza University of Rome (Italy) studying muscle stem cell biology at the Institut Pasteur (Paris, France); he was then awarded his PhD at the San Raffaele Scientific Institute of Milan (Italy) and completed his clinical specialty training in paediatrics and paediatric neurology in London.

Prof. Tedesco was awarded several major fellowships and research grants, including an NIHR Academic Clinical Fellowship followed by an NIHR Clinical Lectureship, a European Research Council (ERC) Starting Grant and the leadership of a large Horizon Europe consortium (10M EUR; 17 institutions; 22 PIs; **MAGIC)**. He received the 2015 Young Investigator Award by the European Society of Gene and Cell Therapy, the 2020 Simon Newell Investigator of the Year award by the Royal College of Paediatrics and Child Health and the 2021 MacKeith Prize by the British Paediatric Neurology Association. In 2024, he was made Fellow of the Royal College of Paediatrics and Child Health.

The Tedesco laboratory harnesses the regenerative potential of muscle stem cells to create innovative models to study and develop therapies for incurable neuromuscular disorders. Their work pioneered engineering and pre-clinical translation of artificial chromosomes as gene therapy vectors (Tedesco et al., *Sci Transl Med* 2011; Benedetti et al., *EMBO Mol Med* 2018), as well as generation, differentiation, genetic correction and pre-clinical testing of human induced pluripotent stem cells (iPSCs) from patients with muscular dystrophy (Tedesco et al., *Sci Transl Med* 2012; Maffioletti et al., *Nat Protoc* 2015). More recently, they developed the first 3D human muscle model containing up to four distinct isogenic cell lineages capable to model disease-associated phenotypes with high resolution and fidelity, and to support testing of advanced therapies such as gene therapy vectors and myogenic cell transplantation (e.g., Maffioletti et al., *Cell Reports* 2018; Choi et al., *EMBO Mol Med* 2022; Pinton et al., *Nat Protoc* 2023). The overall goal of the Tedesco laboratory is the translation of regenerative strategies into novel therapies to improve future outcomes for children with neuromuscular disorders.

Dr. Tedesco has been awarded a grant through The RYR-1 Foundation's 2025–2027 Grant Cycle.

Joshua Todd, PhD, MBA

Dr. Joshua Todd is a Staff Scientist (Clinical) with the National Institute of Neurological Disorders and Stroke (NINDS/NIH), where he holds a joint position with the Clinical Trials Unit and the Neuromuscular and Neurogenetic Disorders of Childhood Section (Bönnemann Team). Here, he leads translational and clinical development of gene and transcript-directed therapies for a wide range of rare neurological disorders. Dr. Todd was a co-investigator on the first two clinical trials for RYR1-RM (NAC and Rycal) and is Principal Investigator of the *RYR1* database project.

Dr. Todd received a grant for the project "RYR1 Genetic Database Development" through the Muscular Dystrophy Association (MDA), in collaboration with The RYR-1 Foundation.

Ashkan Vaziri, PhD

Dr. Ashkan Vaziri is a Founder and CEO of BioSensics, an FDA-registered medical device company focused on the development of wearable sensor and digital health technologies for clinical trials and research. Dr. Vaziri's work presents a unique combination of academic research, product development, and small business experience. He co-founded BioSensics in 2007, when he was a Research Associate and Lecturer at the Harvard School of Engineering. Until 2019, Dr. Vaziri was a tenured full professor of Engineering at Northeastern University, where his research was mainly focused on materials science, biomechanics, computational modeling, and numerical simulations. Dr. Vaziri has authored 175+ journal papers and over 280+ conference proceedings with 13k+ citations (h-index=65), 6 issued/pending patents, and served as Principal Investigator on 25+ NIH/NSF-funded projects.

Nadège Zanou, MD, PhD

Dr. Nadège Zanou is Head of Research and Senior Lecturer at the Institute of Sport Sciences, University of Lausanne.

She holds a medical degree from Benin and a Master's and PhD in Biomedical Sciences from the Catholic University of Louvain, where she studied the role of transient receptor potential (TRP) channels in muscle development and in the pathophysiology of Duchenne muscular dystrophy.

Between 2013 and 2020, she completed two postdoctoral fellowships—first at the Catholic University of Louvain and later at the University of Lausanne in Switzerland—where she developed strong expertise in the translational study of muscle responses to exercise and disease-related plasticity.

Between 2017 and 2018, Dr. Zanou spent six months as a visiting researcher in the laboratory of Professor Andrew Marks at Columbia University, where she investigated post-translational modifications of the ryanodine receptor (RyR1) in response to exercise.

Her current research focuses on the pathophysiology of myopathies and the molecular and cellular mechanisms underpinning skeletal muscle adaptations to exercise.

RYR-1-RD Community Representatives

**Remote Participation*

Justin Michael Bonny*

Justin Michael Bonny is a British-American musician, instructor, and speaker based in New Hampshire. Diagnosed with Centronuclear Myopathy as an infant, he received an official diagnosis of an RYR1-related myopathy in 2013 from MassGeneral. As Youth Development Coordinator for the Dover Housing Authority, he plays a pivotal role in mentoring and empowering young people. After his day job, he continues his dedication to education and the arts as a Front Ensemble Instructor for the Dover High School Marching Band, proudly serving the program as an alumnus.

A recipient of numerous accolades—including multiple Maine Band Directors Association Gold Medals and the CYCC (Children and Youth with Chronic Conditions) Champion Award—Justin has been a featured speaker for CYCC and the New Hampshire Department of Health and Human Services. His passion for music and education continues to shape his work.

In his spare time, he enjoys playing music, travelling, and ice fishing.

Daniel Eppel

Daniel Eppel is a composer and music producer based in Cape Town, South Africa. Born in Johannesburg and raised in Memphis, Tennessee, Daniel's multicultural upbringing shapes his dynamic and versatile musical voice.

He received a scholarship to attend the prestigious Berklee College of Music in Boston, where he studied performance, film scoring, and music engineering, graduating *cum laude* in 2003.

In 2014, Daniel founded Edible Audio Studios—a world-class, sound facility designed to meet the highest international acoustic standards. His work spans music composition and audio post-production for feature films, commercials, documentaries, and animation.

Ben Huseth

Ben Huseth is a member of Plumbers & Pipefitters Local Union 6 located in Rochester, Minnesota. He has three siblings, two of whom are affected by RYR-1-RD, and is attending the Research Workshop to share his personal experiences growing up in a family impacted by RYR-1-RD.

Currently, Ben resides with his spouse and two children, keeping busy with plenty of opportunities for tinkering, repairing, or reconstructing around the house. Ben looks forward to his next travel destination, walking the dogs, a day on the water or golfing, and becoming an empty nester.

Curt D. Keller, DMin

Curt Keller is a retired United Methodist Church minister and retired licensed Marriage and Family Therapist.

Curt has an RYR-1-RD, which is associated with Malignant Hyperthermia Susceptibility (MHS). His son died of an MH episode, which occurred outside of a medical setting. His father had an MH episode during heart surgery and passed away a week later. He has been a board member of the Malignant Hyperthermia Association of the United States (MHAUS) for approximately 10 years and currently chairs the Patient Liaison Committee. He began competitive swimming at the age of seven and continues to swim competitively through US Masters Swimming. He is married and has a surviving son who is a musician in the Nashville, Tennessee area.

Sarah Keller*

Sarah Keller is a dedicated School Counselor with Virginia Beach City Public Schools and a passionate advocate for medically complex families. A proud mother of two boys and four bonus children, Sarah's journey as a medical mom began in 2015 when her son Bennett was born with an RYR-1-RD and other related conditions.

Her personal experience has inspired a deep commitment to supporting families facing similar challenges. She has served on both the NICU Family Advisory Council and the Hospital Family Advisory Council at Children's Hospital of the King's Daughters (CHKD), where she works closely with families as they transition from hospital to home life. Through her professional role and volunteer work, Sarah brings empathy, insight, and strength to every family she supports.

Melanie Moore

Melanie Moore is the mother to Trelyn Moore (age 14), her son, who was diagnosed with an RYR-1-RD when he was almost eight. Since learning about his diagnosis, she has set forth to learn as much as possible and connect with other parents, caregivers, doctors, researchers, and specialists to ensure that Trelyn receives the best care. Trelyn faced medical challenges prior to his RYR-1-RD diagnosis, having been born prematurely at just 25 weeks. She didn't know that all of the therapies due to prematurity, also helped him with his RYR-1-RD symptoms. She is excited to be a part of the Research Workshop as a Parent Speaker.

Melanie, in her professional career, is an expert Human Resources professional with nearly 30 years of experience.

Avi Orseck

Avi Orseck is a Sophomore studying Philosophy at Wofford College in Spartanburg, South Carolina. He was diagnosed with an RYR-1-RD as a toddler and has had multiple surgeries to correct complications from this life long condition. He enjoys writing, Dungeons & Dragons, and being a social advocate.

Patricia Powers, MDiv, PhD*

Trish Powers has 35 years of proven command experience and leadership in military and civilian sectors, including operations management, command and control, and human research protections. Her expertise also extends to religious institutions, higher education, national volunteer organizations (e.g., peer mentor for Tragedy Assistance Programs for Survivors (TAPS); Special Olympics Coach), and in publishing, where she has contributed to theological review, editing, writing, and the production of magazines and catechetical and homiletic materials. Trish's specialties include: Catholic Moral Theology (Christian Ethics), bioethics (clinical and research), applied ethics, undergraduate and graduate level teaching and research in philosophy and theology, and pastoral counseling. Her educational background is in political science and philosophical ethics, including familiarity with American and European political/strategic issues, bioethics and law.

Jenny C. Roller

Jenny C. Roller is a rare patient advocate. She is actively involved with state and federal legislators to improve the current healthcare system as well as proposing changes to ensure the rare patient community is represented. She is currently working with other patient advocates to increase funding for the NIH. Major cuts made by the new administration have deeply impacted patients with RYR-1-RD.

Jenny was born with Malignant Hyperthermia, Central Core Disease, and King Denborough Syndrome. She has many anomalies associated with the *RYR1* gene variants. Over the years, she has developed serious health issues and endures constant pain. She fights hard to maintain her quality of life and is determined to help others who struggle with the hurdles of having a rare disease. Jenny is always the optimist and can find something to laugh about even when she is struggling. She lives with her husband, Ed, in Arkansas.

The RYR-1 Foundation's Scientific Advisory Board Members

**Remote Participation*

Robert T. Dirksen, PhD (Chair)

Dr. Robert T. Dirksen obtained his PhD in 1991 from the University of Rochester, where he studied the mechanisms that underlie neuromodulation of cardiac excitability and contractile function. He then completed a postdoctoral fellowship with Dr. Kurt Beam at Colorado State University focused on the mechanisms of skeletal muscle excitation-contraction coupling. In 1997, he moved back to the University of Rochester as an independent faculty member in the Department of Pharmacology and Physiology. Dr. Dirksen's research program focuses on elucidating the cellular and molecular mechanisms by which *RYR1* mutations lead to altered skeletal muscle function and the development of mouse models of RYR1-related myopathy both to better understand disease pathophysiology and to test new therapeutic interventions.

Carsten G. Bönnemann, MD, habil

Dr. Carsten G. Bönnemann is a pediatric neurologist specializing in neuromuscular disorders and neurogenetics. He is a Senior Investigator in the Neurogenetics Branch and Chief of the Neuromuscular and Neurogenetic Disorders of Childhood Section at the National Institute of Neurological Disorders and Stroke at the National Institutes of Health in Bethesda.

Dr. Bönnemann is also an Adjunct Professor of Neurology at the University of Pennsylvania and the Children's Hospital of Philadelphia (CHOP), and he is a member of the Executive Board of the World Muscle Society as well as Co-Editor-in-Chief of the *Journal of Neuromuscular Diseases*. Dr. Bönnemann is an internationally recognized expert in the diagnosis of neuromuscular diseases. His research is dedicated to understanding the molecular genetics and cell biology of muscular dystrophies and early onset myopathies in order to develop molecular-based treatments. Dr. Bönnemann was a Pew Scholar in the Biomedical Sciences and received the Derek-Denny Brown Neurological Scholar Award of the American Neurological Association.

James Dowling, MD, PhD*

Dr. James Dowling recently became the Director at Penn Neurogenetics Therapy Center. He is a clinician-scientist who is focused on gene discovery and therapy for childhood muscle diseases. Previously, he was a Professor of Paediatrics and Molecular Genetics at the University of Toronto and a staff clinician at the Hospital for Sick Children in Toronto. He was also the inaugural Mogford Campbell Chair in Pediatric Clinical Neuroscience at Toronto's Hospital for Sick Children. In addition, he is a member of the Executive Board of the World Muscle Society and a member of the Board of Directors of TREAT-NMD.

Dr. Dowling is considered one of the world's leading experts in RYR-1-RD. His research spans the continuum from new gene discovery to disease pathogenesis to therapy development. He is engaged in both targeted and non-biased drug discovery for RYR-1-RD. His laboratory employs both the zebrafish and mouse model systems, along with complementary *in vitro* analyses. Dr. Dowling is the recipient of numerous NIH and private foundation research grants and has recently published detailed results of his RYR1-related research.

Minal S. Jain, PT, DSc, FAPTA

Dr. Minal S. Jain is a research physical therapist and the physical therapy section research coordinator within the Rehabilitation Medicine Department at the NIH Clinical Center. She has been an associate investigator on many protocols related to neuromuscular diseases, including RYR-1-related diseases. This includes determining the most appropriate motor assessments to use, assisting with developing a standardized protocol, data collection, data analysis and interpretation of the data for dissemination of the findings. Her research interests are in the areas of measuring motor changes, using standard assessments and innovative technologies such as wearable sensors. She has written manuscripts and presented lectures at local, national, and international conferences.

Heinz Jungbluth, MD, PhD*

Dr. Heinz Jungbluth is Professor of Paediatric Neurology at King's College London and Consultant Paediatric Neurologist at the Evelina Children's Hospital, Guy's & St Thomas' NHS Foundation Trust, London, UK, with more than 25 years clinical experience in Paediatric Neurology.

His main research interest is in early-onset neuromuscular and neurodevelopmental disorders. He has been particularly interested in the clinical and genetic characterization of the congenital myopathies, in particular those affecting excitation-contraction coupling due to mutations in the skeletal muscle ryanodine receptor (*RYR1*) gene, and related episodic disorders such as malignant hyperthermia (MH) and (exertional) rhabdomyolysis (ERM). He and his team have also introduced the concept of congenital disorders of autophagy, a novel class of inborn neuromuscular and neurometabolic conditions linking aberrant neurodevelopment with common adult-onset neurodegenerative disorders such as dementia, Parkinson's disease and ALS.

He has published extensively and is a member of national and international expert consortia concerned with improving the care and developing therapies for early-onset neuromuscular and neurological disease.

Dr. Jungbluth is the Principal Investigator of "The Prevalence of RYR1-Related Disease—An International, Collaborative Multicentre Study" supported by a research grant from The RYR-1 Foundation.

Andrew R. Marks, MD*

Dr. Andrew R. Marks received his undergraduate degree from Amherst College (with honors in both Biology and English), and his MD from Harvard Medical School. Following an internship and residency in internal medicine at the Massachusetts General Hospital (MGH), he was a postdoctoral fellow in molecular genetics at Harvard Medical School, and then a clinical cardiology fellow at the MGH. He is board certified in internal medicine and in cardiology.

Dr. Marks is Chair and Professor of the Physiology and Cellular Biophysics Department at Columbia University. He is a member of the National Academy of Sciences (2005), National Academy of Medicine (2004), and American Academy of Arts and Sciences (2005). From 2002-2007 Dr. Marks was **Editor-in-Chief** of the *Journal of Clinical Investigation*. He received the Doctor of Science, *Honoris causa*, Amherst College (2009), and from de l'Université de Montpellier (2016), the **ASCI Stanley J. Korsmeyer Award** (2010), the Pasarow Foundation Award for Cardiovascular Research (2011), the Ellison Medical Foundation Senior Scholar in Aging Award (2011), Glorney-Raisbeck Award from NY Academy of Medicine (2016), Columbia University Dean's Award for Excellence in promoting Diversity (2017), the Naranjan Dhalla Award for Innovative Investigators in Cardiovascular Sciences, International Academy of Cardiovascular Sciences (2018).

In 2002, he founded the Summer Program for Undergraduate Rising Stars (SPURS) at Columbia. SPURS provides mentored research training at Columbia University for under-represented and economically disadvantaged students primarily from the New York City public colleges and universities.

Dr. Marks has published over 200 articles in which he has contributed new understandings of fundamental mechanisms that control muscle contraction, heart function, lymphocyte activation, and cognitive function. He has discovered novel causes of human diseases including heart failure, cardiac arrhythmias, muscular dystrophy, diabetes, and neurodegenerative disorders. He developed novel treatments for coronary artery disease (in 2003 the FDA approved the first drug eluting stent), and a new class of drugs called Rycal®, one of which is being tested in patients with heart and muscle disorders.

Dr. Marks is the founding scientist of RyCarma Therapeutics (formerly ARMGO Pharma, Inc.) a biotech company focused on developing drugs that target leaky ryanodine receptors.

Isabelle Marty, PhD

Dr. Isabelle Marty has more than 30 years of experience studying *RYR1* focusing on understanding its precise functions and dysfunctions. She is now an expert in developing therapies for RYR-1-RD, and specifically in gene therapy approaches. She is also deeply involved in the French patient organization, Association Française contre les Myopathies (AFM-Téléthon), and engages with patients and families to help interpret research progress.

Ichizo Nishino, MD, PhD*

Dr. Ichizo Nishino is the Director of the Department of Neuromuscular Research at the National Institute of Neuroscience (NIN), National Center of Neurology and Psychiatry (NCNP) in Tokyo, Japan. He earned his MD in 1989 and PhD in 1998 from Kyoto University and completed postdoctoral training, including two years at Columbia University. Since 2001, he has led a national referral center for muscle disease, analyzing around 80% of muscle biopsies in Japan. His lab has amassed over 24,000 muscle biopsy samples.

Dr. Nishino is a Visiting/Guest Professor at seven universities across Asia and serves as President of the Asian-Oceanian Myology Center. He is also an Executive Board member of the World Muscle Society (WMS) and a corresponding Fellow of the American Academy of Neurology (FAAN) and the American Neurological Association (FANA). His research focuses on understanding the mechanism of muscle diseases, particularly congenital muscle diseases including those caused by *RYR1* gene mutations.

Sheila Riazi, MSc, MD, FRCPC

Dr. Sheila Riazi is a Professor at the Department of Anesthesia, Site Head of Anesthesia at Toronto Western and Women's College Hospitals, a staff anesthesiologist at University Health Network, and clinician scientist at Toronto General Research Institute. Dr. Riazi directs the only Canadian diagnostic center for malignant hyperthermia (MH), where she assesses at-risk individuals and provides genetic testing and caffeine-halothane contracture test (CHCT). Her research interests include pathophysiology of MH, links with exertional heat illnesses, and phenotypic variability in MH susceptible patients with funding from various sources such as NIH, and CIHR. She is the chair of the MH International Professional Advisory Council, a member of the board of directors of MH Association of the United States (MHAUS), and a member of the NIH-led panel on *RYR1* interpretation.

Filip Van Petegem, PhD

Dr. Filip Van Petegem obtained his PhD in 2002 from Ghent University in Belgium, where he used X-ray crystallography to study the 3D structures of proteins. He then moved to San Francisco for postdoctoral studies at UCSF, where he applied this methodology to understand the function of ion channels, specialized proteins that generate electrical signals.

In 2007, he moved to Vancouver (Canada) to start as a faculty member in the Department of Biochemistry and Molecular Biology at The University of British Columbia. He established a research program to investigate the structure and function of ion channels involved in cardiac and skeletal muscle function. This includes multiple studies on the RyR1 protein, elucidating its 3D structure, how it is affected by MH mutations, and how they bind various molecules that are either beneficial or detrimental to their normal function.

Nicol C. Voermans, MD, PhD

Dr. Nicol C. Voermans is a distinguished neurologist specializing in neuromuscular disorders at Radboud University Medical Center (Radboudumc) in Nijmegen, Netherlands.

Since joining Radboudumc as a medical specialist in 2008, she has focused on congenital myopathies and facioscapulohumeral muscular dystrophy (FSHD). Her research emphasizes genotype-phenotype correlations, trial readiness, natural history of rare inherited myopathies, patients' experiences in trials, and the development of optimal symptomatic treatments. She has a special interest in (exertional) rhabdomyolysis.

She has collaborated with esteemed researchers, including Prof. Jungbluth in London and Prof. Treves in Basel. In recognition of her significant contributions, she was appointed Professor of Muscular Diseases at Radboudumc, effective October 1, 2023. Prof. Voermans's dedication to bridging the gap between neuromuscular research and clinical practice continues to enhance the understanding and treatment of rare genetic muscle disorders.

Special Guests

**Remote Participation*

Morton F. Goldberg, MD, FACS

Along with his son, Dr. Michael Goldberg, Dr. Morton Goldberg is the Co-Chair of Research of The RYR-1 Foundation. He is also an ophthalmologist specializing in retinal disease and genetics. He is the Joseph Green Professor of Ophthalmology at the Johns Hopkins School of Medicine and the Director Emeritus of the Wilmer Eye Institute. He is a member of the Institute of Medicine of the National Academies of Science of the United States.

Dr. Morton Goldberg is Director Emeritus of The RYR-1 Foundation's Board of Directors, a co-founder, and currently serves as Co-Chair of Research for the Organization.

Susan Hamilton, PhD

Dr. Susan Hamilton is the Chair and Professor in the Department of Integrative Physiology at Baylor College of Medicine. Her research over the past 30 years has contributed to the understanding of mechanisms of excitation-contraction coupling in skeletal muscle and RYR-1-RD, including myopathies, Malignant Hyperthermia Susceptibility, and Enhanced Sensitivity to Heat Stroke.

Her research is designed to identify new targets and interventions for RYR-1-RD. Her lab has created many new mouse models of RYR-1-RD and has identified several potential therapeutic interventions. Dr. Hamilton is the senior author on numerous peer-reviewed publications, and her research program has been funded continuously by NIH grants.

Dr. Hamilton is the former Senior Vice President and Dean of Research at Baylor College of Medicine, a former Chair on the Board of Trustees of the Gordon Research Conferences, and a current member of The RYR-1 Foundation Board of Directors.

Dr. Hamilton served as a member of The RYR-1 Foundation's Scientific Advisory Board from 2015 to 2020. She currently serves as a Director on The RYR-1 Foundation's Board of Directors.

Susan Treves, PhD*

Dr. Susan Treves' laboratory at the University of Basel (Switzerland) focuses on studying intracellular calcium homeostasis in mammalian cells under normal and pathological conditions, with particular emphasis on skeletal muscle excitation-contraction coupling. One of the models she uses to study calcium dysregulation in skeletal muscle is unique to her laboratory since they use biopsy-derived human skeletal muscle cells differentiated into myotubes *in vitro*.

As a result of her PhD work in the laboratory of Prof. Tullio Pozzan, one of the pioneers in the development of fluorescent Ca2+ indicators, she has a broad understanding on the use, pitfalls, and advantages of the available methods to study calcium homeostasis with fluorescent calcium indicators. This expertise coupled with access to patient material has allowed her laboratory to directly assess the effect of mutations in the human ryanodine receptor calcium channel in muscles obtained from patients affected by neuromuscular disorders.

Her laboratory applies a broad range of techniques, including cell and tissue culture, molecular biology, cell biology, biochemistry, microscopy and imaging, and fluorescence measurements of intracellular [Ca2+].

Dr. Treves served as a member of The RYR-1 Foundation's Scientific Advisory Board from 2018 to 2025.

Students, Trainees, & Young Investigators

**Remote Participation*

Ting Chang, PhD

Dr. Ting Chang obtained her PhD degree from Baylor College of Medicine this March. She works as a postdoctoral associate in Dr. Hamilton's lab. The lab created some of the first mouse models of RYR1-related myopathies to delineate the disease mechanisms and develop the interventions for the diseases.

Lourdes Carolina Figueroa, PhD

Dr. Lourdes Carolina Figueroa is an Assistant Professor in the Department of Physiology and Biophysics at Rush University Medical Center in Chicago, USA. Her research group investigates the complexities of RyR1 dysfunction in various contexts, including muscular diseases, chemotherapy-induced myopathy, diabetes, and aging. Their goal is to develop innovative therapeutic strategies using patient-derived cells and tissues, as well as animal models.

Ainsley Johnson

Ainsley Johnson is a third-year undergraduate with Southern Methodist University and a research assistant in cancer biology and rare disease genomics. Driven by her unique experiences living with an *RYR1* variant and an inherited collagenopathy, she has leveraged her perspective as a patient-scientist towards furthering research efforts in rare and undiagnosed genetic disorders. She aspires to pursue a PhD in genetics and eventually, a career in rare disease research.

Nick Kruijt, MD*

Dr. Nick Kruijt is a Dutch physician, currently working as a family medicine doctor and PhD candidate in neurology. He earned his MD from Radboud University Medical Center in 2019 and has a clinical and academic focus on rhabdomyolysis and exertional heat stroke.

By combining his role as a general practitioner with ongoing research, he bridges the gap between frontline care and scientific research. His research has led to several peer-reviewed publications examining the genetic and physiological mechanisms of neuromuscular disorders, with particular emphasis on RYR1-related and exercise-related conditions.

His work has appeared in leading journals such as *Sports Medicine*, *European Journal of Neurology*, and *Neuromuscular Disorders*, offering not only clinical insights but also patient-centered perspectives grounded in family medicine.

Changseok Lee, PhD

Dr. Changseok Lee is an assistant professor at Baylor College of Medicine in Houston, Texas. After joining the laboratory of Dr. Susan L. Hamilton (chair of the department) as a postdoctoral associate, he has developed his career and has focused on rare skeletal muscle diseases related to calcium handling proteins, including type 1 ryanodine receptor, for about 15 years.

Marco C. Miotto, PhD

Dr. Marco C. Miotto is an Assistant Professor at Columbia University with a background in structural biology. His research combines biophysical techniques, particularly cryogenic electron microscopy, to uncover the molecular and atomic details of biological processes.

In 2019, he joined the Marks Lab at Columbia to investigate diseases linked to Ryanodine Receptors, including cardiac conditions like Heart Failure and Catecholaminergic Polymorphic Ventricular Tachycardia, as well as skeletal muscle disorders such as RYR1-related myopathies. Dr. Miotto is currently focused on developing tools and insights to aid in the diagnosis and treatment of these diseases, such as creating a structure-based machine learning model to predict the pathogenicity of RyR2 variants and studying the binding properties of activating compounds to advance therapies for RYR1-related myopathies.

Courtney Perrin

Courtney Perrin is a PhD student who will be starting this Fall at Upstate Medical University. She holds a BS in molecular genetics and a BA in Spanish at the University of Fredonia. Since 2016, Courtney has been actively fundraising for The RYR-1 Foundation, starting by collecting bottles and cans alongside her mother. Together with her community, they have raised over $50,000 to support The RYR-1 Foundation. Courtney has always had a passion for fundraising and aspires to research RYR-1-RD as well after earning her PhD.

Parisa Shamsesfandabadi, MD

Dr. Parisa Shams is a graduating radiation oncology resident at Allegheny Health Network with a passion for reimagining how we teach, learn, and innovate in the oncology field. She brings a dynamic blend of research, advocacy, and creative educational leadership to the field. Her work spans MR Linac-guided radiation therapy to initiatives improving global access to radiation care.

Kara Soler-Sala*

Kara Soler-Sala is currently a Registered Nurse practicing at the Mayo Clinic with a strong foundation in evidence-based patient care. She has had over eight years in Oncology/Bone Marrow Transplant nursing care in Boston, Massachusetts and presently in Jacksonville, Florida.

Kara is currently advancing her clinical knowledge and leadership skills as a Family Nurse Practitioner (FNP) student at Georgetown University, where she is training to provide holistic, primary care across the lifespan. In addition to her professional and academic pursuits, she is deeply passionate about advancing research related to RYR-1-RD—a cause that has become profoundly personal since the birth of her nephew in October 2024. His diagnosis has fueled her dedication to raising awareness and supporting efforts that improve understanding, treatment, and outcomes for individuals affected by this rare condition.

Lizan Stinissen, MSc

Lizan Stinissen is a PhD candidate under the supervision of Dr. Nicol Voermans at the department of Neurology at the Radboud University Medical Center (the Netherlands). She obtained her Master's degree in Biomedical Sciences at the Radboud University in 2023. Her PhD focuses on the patient perspective and patient participation of patients with inherited neuromuscular disorders. This includes the impact of the treatment perspectives on the daily lives of patients, and the lessons learned during the first clinical studies.

Throughout her PhD, she has collaborated with patients and patient organizations in various countries. This has resulted in several papers, including a paper on the online patient survey and testimonials presented during the 2022 RYR-1-Related Diseases International Research Workshop in Pittsburgh. Recently, she started a study that extends this research, aiming to explore the clinical trial preferences of individuals with RYR-1-RD in more detail. This study was introduced in a webinar in collaboration with The RYR-1 Foundation last March.

Pomi Yun, MD

Dr. Pomi Yun is a third-year pediatric neurology resident at Johns Hopkins University where she has joined the lab of Dr. Payam Mohassel. Past research experiences include four years at the NIH while under the mentorship of Dr. Carsten Bönnemann.

Staff

Lindsay Goldberg, BSN, RN

Lindsay Goldberg is the Executive Director & Patient Liaison and also a co-founder of The RYR-1 Foundation. She serves as an advocate for the RYR-1-RD community and liaison between affected individuals/their families and physicians, genetic testing companies, and scientists. She has a BSN from the University of Michigan and spent the first 10 years of her career as a bedside registered nurse specializing in pediatric cardiac intensive care (CICU).

Lena Leghart, BSW

Lena Leghart became a part of The RYR-1 Foundation team in 2022 as the Administrative Assistant and was promoted to Program Manager in February 2024. In her current role, Lena oversees and facilitates key programs, including the Family Conferences, Research Workshops, webinars, CCG, and more. She is also responsible for coordinating daily operations, managing our website, and overseeing social media.

Patricia Raffaele, MA, CAE

Patricia Raffaele joined The RYR-1 Foundation in February 2024 as the Development, Public Relations, & Research Director. Pat focuses on development to support the mission of the organization, communications to “tell the story” of The RYR-1 Foundation, and advocacy to build awareness of the organization and those it serves. She holds a Master’s degree in Corporate Communications and a Bachelor’s degree in Journalism, both from Duquesne University. She is also a Certified Association Executive through the American Society of Association Executives.

Industry & External Organization Guests

Mike Greenberg, MD

Mike Greenberg, MD, an emergency physician by training, is the Vice President of Medical Affairs at Eagle Pharmaceuticals where he is responsible for data generation, scientific and clinical information exchange and drug safety across Eagle's acute care/hospital and hematology/oncology portfolios.

In addition to his experience as an emergency physician and medical affairs professional, Dr. Greenberg has also worked with the Federal Agency for Healthcare Research and Quality (AHRQ) to raise awareness of comparative effectiveness research, and as acted as a consultant to teams focusing on Risk Evaluation and Mitigation Strategies (REMS) and Drug Safety Communications at the FDA Center for Drug Evaluation and Research (CDER).

Mike earned his medical degree at the State University of New York Upstate Medical University and completed a residency in Emergency Medicine at the University of Massachusetts.

Angela Lek, PhD

Angela Lek, PhD, is the interim Chief Research Officer at the Muscular Dystrophy Association (MDA). Dr. Lek joined MDA over three years ago as Vice President of Research, where she managed the organization's research grant programs, collaborative research programs, venture philanthropy investments, and served as scientific lead of MDA's Kickstart program. In her new role as interim Chief Research Officer, she will expand her leadership to oversee MDA's entire discovery, translational, and clinical research portfolio, bringing both strategic and operational expertise to the organization.

Prior to working at MDA, Dr. Lek completed her PhD in the Department of Medicine at the University of Sydney and conducted postdoctoral training at Boston Children's Hospital under the mentorship of Professor Louis Kunkel, PhD. She was honored with several internationally recognized fellowships, including those from the Australian National Health & Medical Research Council and the American Australian Association.

Dr. Lek's research focused on understanding the genetic basis of muscular dystrophies and developing innovative therapies. Dr. Lek has led her own research program at Yale University, supported by fellowships from MDA and the Hood Foundation. She currently serves as a program consultant for the NIH's Somatic Cell Genome Editing program and collaborates with numerous advocacy groups in the neuromuscular disease space to drive drug development, advocacy, and patient outreach efforts. The diagnosis of her husband, Monkol Lek, PhD with limb-girdle muscular dystrophy deepens her passion and commitment to understanding and advancing treatments for neuromuscular diseases.

Elizabeth Tarka, MD

Elizabeth Tarka, MD, joined RyCarma Therapeutics in 2025 as Chief Medical Officer (CMO), bringing over 20 years of experience in the pharmaceutical and biotechnology industry including the design, implementation, and medical oversight of late-stage clinical trials. She previously served as CMO for XyloCor Therapeutics, a clinical stage biopharmaceutical company developing novel gene therapies for cardiovascular diseases and CMO at Idera Pharmaceuticals, advancing its rare disease and immuno-oncology pipeline.

Earlier in her career, Elizabeth was the Clinical Program Lead for Xarelto® (rivaroxaban) at Janssen Pharmaceuticals and spent over ten years in the metabolic pathways and cardiovascular therapeutic areas at GlaxoSmithKline where she helped to lead multiple cardiovascular programs including a Phase 3 global cardiovascular outcomes study with a novel drug candidate.

Elizabeth earned a bachelor's degree in biochemistry from the University of Pennsylvania and a medical degree from the University of Pennsylvania School of Medicine where she also completed her residency and fellowship training. Prior to her industry roles, Elizabeth was on the faculty at the University of Pennsylvania and currently serves as Adjunct Faculty Associate Professor. She has been board certified in cardiology and internal medicine and has been published in several peer-reviewed journals.

RYR-1 FOUNDATION

STRENGTH IN NUMBERS

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