RYR-I FOUNDATION

STRENGTH IN NUMBERS

INTERNATIONAL FAMILY CONFERENCE

Pittsburgh, PA | USA

July 24 - 27, 2025

RYR-I FOUNDATION

STRENGTH IN NUMBERS

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Agenda

THE RYR-1 FOUNDATION INTERNATIONAL FAMILY CONFERENCE 2025 AGENDA

Thursday, July 24, 2025		
Time	**Open Hours: Registration, Lounges, and Kids' Club**	**Room**
12:00 pm - 9:00 pm	Registration Desk	Glenn Foyer
12:00 pm - 9:00 pm	The Social Hub: Adult Lounge Experience	Earhart A
12:00 pm - 4:00 pm 5:00 pm - 9:00 pm	The Funhouse Clubhouse (Kids' Club for Ages 5 - 12)	Lindberg AB
12:00 pm - 4:00 pm 5:00 pm - 9:00 pm	The Teen Spot (Teen Lounge for Ages 13 - 17)	Wright B
Time	**Event**	**Room**
5:00 pm - 9:00 pm	Happy Hour Starts Now: Bar Open (Cash Bar)	Regency Foyer & Allegheny Foyer
5:30 pm - 7:00 pm	Mix & Mingle: Welcome Cocktail Soirée	Regency Foyer Allegheny Foyer Allegheny ABC
7:00 pm - 9:00 pm	Strength In Numbers: Welcome & Family Introductions	Regency Ballroom AB

Agenda

THE RYR-1 FOUNDATION INTERNATIONAL FAMILY CONFERENCE 2025 AGENDA

Friday, July 25, 2025			
Time	**Open Hours: Registration, Lounges, and Kids' Club**		**Room**
7:30 am - 5:00 pm	Registration Desk		Glenn Foyer
8:00 am - 5:00 pm	The Social Hub: Adult Lounge Experience		Earhart A
9:00 am - 12:00 pm 1:00 pm - 5:00 pm	The Funhouse Clubhouse (Kids' Club for Ages 5 - 12)		Lindberg AB
9:00 am - 12:00 pm 1:00 pm - 5:00 pm	The Teen Spot (Teen Lounge for Ages 13 - 17)		Wright B
Time	**Event**	**Speaker**	**Room**
8:00 am - 9:00 am	**Breakfast** Natural History Study Opportunity - NIH		**Regency Ballroom**
9:00 am - 9:30 am	Overview of RYR-1-Related Diseases (RYR-1-RD)	Carsten G. Bönnemann, MD, habil	Regency Ballroom
9:30 am - 10:00 am	Genetics 101	Livija Medne, MS, LCGC	Regency Ballroom
10:00 am - 10:30 am	Interpreting Genetic Test Results	Andrew McCarty, MS, LCGC	Regency Ballroom
10:30 am - 11:00 am	Mechanism and Functional Readouts of *RYR1* Variants	Robert T. Dirksen, PhD Filip Van Petegem, PhD	Regency Ballroom
11:00 am - 11:30 am	**Expert Panel:** Sequela of RYR-1-RD	**Moderator:** Lindsay Goldberg, BSN, RN **Panelists:** Carsten G. Bönnemann, MD, habil Payam Mohassel, MD Nicol C. Voermans, MD, PhD	Regency Ballroom
11:30 am - 12:00 pm	**Q&A:** RYR-1-RD Session	All of the Above	Regency Ballroom
12:00 pm - 1:00 pm	**Lunch & Group Photo** Effective Fundraising: Tactics That Work - Emily Pedersen, Amy & Courtney Perrin, & Courtney Woomer		**Regency Ballroom**
1:00 pm - 1:45 pm	**Psychosocial Insights:** An Introductory Guide Breakout Groups	Julie Bowden, PsyD	Regency Ballroom
1:45 pm - 2:15 pm	**Patient-Led Panels:** Accommodate and Thrive: A Patient-Led Panel on School Support Challenges and Triumphs: Living with Malignant Hyperthermia Phenotype Dating with Confidence: A Patient-Led Conversation **Concurrent Session:** Psychosocial Breakout Groups	Lauren Heft, Emily Pedersen, & Amy Perrin Bob Hanich, Cody Hodgson, & Curt D. Keller Jeni Ryan Julie Bowden, PsyD	Allegheny AB Yeager AB Earhart CD Regency Ballroom
2:15 pm - 2:30 pm	**Break**		
2:30 pm - 3:00 pm	Gene Therapy: Current Progress in Treating Neuromuscular Diseases and Future Hopes for RYR1-Related Myopathies	Isabelle Marty, PhD	Regency Ballroom
3:00 pm - 3:30 pm	**Expert Panel:** Fueling Innovation: The RYR-1 Foundation's Research Grants	**Moderator:** Michael Goldberg, MD, MPH **Panelists:** Selene Ingusci, PhD Afrooz Rashnonejad, MSc, PhD Hichem Tasfaout, PhD, PharmD, MS	Regency Ballroom
3:30 pm - 4:00 pm	**Expert Panel:** Exploring Current Treatment Options	**Moderator:** Michael Goldberg, MD, MPH **Panelists:** Payam Mohassel, MD Sheila Riazi, MSc, MD, FRCPC Nicol C. Voermans, MD, PhD	Regency Ballroom
4:00 pm - 4:30 pm	**Expert Panel:** The Road Ahead: Practical Perspectives on Future Treatments and Cure	**Moderator:** Michael Goldberg, MD, MPH **Panelists:** Robert T. Dirksen, PhD Susan Hamilton, PhD	Regency Ballroom
4:30 pm - 5:00 pm	**Q&A:** Therapies and Treatment Options Session	All of the Above	Regency Ballroom

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THE RYR-1 FOUNDATION INTERNATIONAL FAMILY CONFERENCE 2025 AGENDA

Friday, July 25, 2025			
Time	**Event**	**Speaker**	**Room**
One-on-One Consults			
9:00 am - 12:00 pm	Navigating Your Child's Individualized Education Program: Personalized Help (15 minute consults)	Jeremy G. Armann, PhD, NCSP	Foerster
1:00 pm - 2:15 pm 3:45 pm - 5:30 pm	Personalized Genetic Interpretation (15 minute consults)	Carsten G. Bönnemann, MD, habil (1st Timeframe) Livija Medne, MS, LCGC (1st & 2nd Timeframe)	Foerster
1:00 pm - 2:15 pm	Personalized Variant Interpretation: Structural and Phenotypic (Symptoms) Analysis (15 minute consults)	Robert T. Dirksen, PhD Filip Van Petegem, PhD	Allegheny C
Sessions			
9:00 am - 12:00 pm 2:00 pm - 5:00 pm	Strength in Movement: Ability-Based Physical Therapy Groups (30 minute sessions)	Michael Brucker, PT, DPT, CSCS, CNC	Earhart B
1:00 pm - 1:45 pm	Accessible Yoga: Chair-Based Movement for Everyone (45 minutes)	Jennifer Orseck	Earhart B
Special Programming for Kids & Teens			
2:30 pm - 3:00 pm	Curiosity Corner: Q&A with Our Carnival Medical Experts	Carsten G. Bönnemann, MD, habil Livija Medne, MS, LCGC	Lindbergh AB
3:00 pm - 3:30 pm	Inside Scoop: Teen Q&A with the Medical Experts	Carsten G. Bönnemann, MD, habil Livija Medne, MS, LCGC	Wright B

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THE RYR-1 FOUNDATION INTERNATIONAL FAMILY CONFERENCE 2025 AGENDA

Saturday, July 26, 2025

Time	Open Hours: Registration, Lounges, and Kids' Club	Room
7:30 am - 9:00 pm	Registration Desk	Glenn Foyer
8:00 am - 9:00 pm	The Social Hub: Adult Lounge Experience	Earhart A
9:00 am - 12:00 pm 1:00 pm - 5:00 pm 6:00 pm - 9:00 pm	The Funhouse Clubhouse (Kids' Club for Ages 5 - 12)	Lindberg AB
9:00 am - 12:00 pm 1:00 pm - 5:00 pm 6:00 pm - 9:00 pm	The Teen Spot (Teen Lounge for Ages 13 - 17)	Wright B

Time	Event	Speaker	Room
8:00 am - 9:00 am	**Breakfast**		**Regency Ballroom**
9:00 am - 10:00 am	Malignant Hyperthermia What You Need to Know Is It Safe to Have Dental Care in the Office Setting in the *RYR1* and MH Patient?	Sheila Riazi, MSc, MD, FRCPC Andrew Herlich, DMD, MD, FAAP, FASA, FAAOMS(H)	Regency Ballroom
10:00 am - 11:00 am	**Concurrent with Q&A Sessions:** Escalation and Then De-escalation of Respiratory Care During an Acute Illness (Pediatric) Bleeding Issues and Abnormalities Related to Menstruation and Pregnancy Understanding Your Options: Family Planning and Genetics Understanding Rhabdomyolysis and Malignant Hyperthermia in RYR-1-Related Diseases	Oscar Henry Mayer, MD Arti Mistry, PhD Livija Medne, MS, LCGC Sheila Riazi, MSc, MD, FRCPC & Nicol C. Voermans, MD, PhD	Allegheny AB Yeager AB Earhart CD Regency Ballroom
11:00 am - 12:00 pm	**Concurrent with Q&A Sessions:** Managing Heat Sensitivity: A Guide for Those with Malignant Hyperthermia Susceptibility Breathing Matters: Acute Adult Pulmonary Care RYR-1-RD: The Patient Perspective and Patient Participation (Clinical Trial Readiness Survey) Strength Within Limits: Exercise Options for RYR-1-RD	Susan Hamilton, PhD Jennifer Newitt, MD Lizan Stinnisen, MSc Minal S. Jain, PT, DSc, FAPTA; Sheila Riazi, MSc, MD, FRCPC; Nicol C. Voermans, MD, PhD	Allegheny AB Yeager AB Earhart CD Regency Ballroom
12:00 pm - 1:00 pm	**Lunch** Congressional Advocacy: Making Your Voice Heard - Amy Perrin & Courtney Woomer		**Regency Ballroom**
1:00 pm - 2:00 pm	Legal Planning Tools for Holding and Transferring Wealth (Q&A included)	Megan Ingram, Esq	Regency Ballroom
2:00 pm - 3:00 pm	**Concurrent with Q&A Sessions:** Hypoventilation Assessment and Management in *RYR1* Myopathy (Pediatric & Adult) Understanding Your Rights Under the Americans with Disabilities Act Schooling with RYR-1-RD: Survey Results & Key Considerations	Oscar Henry Mayer, MD & Jennifer Newitt, MD Jenee' Oliver, Esq Jeremy G. Armann, PhD, NCSP; Tammy L. Hughes, PhD, ABPP; Christine Porter, MA, JD	Allegheny AB Yeager AB Earhart CD
3:00 pm - 3:15 pm	**Break**		
3:15 pm - 4:15 pm	**Concurrent with Q&A Sessions:** Comprehensive Pediatric to Adult Transition of Care - Process More Than Event Movement Matters: PT, Exercise and Equipment Needs	Oscar Henry Mayer, MD & Jennifer Newitt, MD Minal S. Jain, PT, DSc, FAPTA	Allegheny AB Yeager AB
4:15 pm - 4:45 pm	**Patient-Led Panels:** Stronger Together: A Patient-Led Discussion on Staying Active Navigating the World: Travel Insights from a Patient's Perspective From Patients, For Patients: Redefining Patient-Centered Care Fueling Our Lives: Patient Perspectives on Adult Nutrition	Mairéad Ryan Marcie Epstein & Emily Pedersen Ilaria Salvatori Bob Hanich	Allegheny AB Yeager AB Earhart CD Regency Ballroom
6:00 pm - 9:00 pm	**Game On & Dine: A Night of Food, Fun, and Family**		**Regency Ballroom**

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Saturday, July 26, 2025			
Time	**Event**	**Speaker**	**Room**
One on One Consults			
8:30 am - 9:45 am 11:15 am - 12:00 pm	Personalized Genetic Interpretation (15 minute consults)	Livija Medne, MS, LCGC Payam Mohassel, MD	Foerster
9:00 am - 12:00 pm 1:00 pm - 3:00 pm	Personalized Variant Interpretation: Structural and Phenotypic (Symptoms) Analysis (15 minute consults)	Robert T. Dirksen, PhD Filip Van Petegem, PhD	Allegheny C
10:00 am - 11:00 am 1:00 pm - 2:00 pm	Navigating Your Child's Individualized Education Program: Personalized Help (15 minute consults)	Jeremy G. Armann, PhD, NCSP Tammy L. Hughes, PhD, ABPP	Foerster
2:00 pm - 5:00 pm	Financial Guidance for Individuals with Disabilities (15 minute consults)	Megan Ingram, Esq	Foerster
Sessions			
9:00 am - 9:45 am	Accessible Yoga: Chair-Based Movement for Everyone (45 minutes)	Jennifer Orseck	Earhart B
10:00 am - 12:00 pm 1:00 pm - 5:00 pm	Strength in Movement: Ability-Based Physical Therapy Groups (30 minute sessions)	Michael Brucker, PT, DPT, CSCS, CNC	Earhart B
Special Programs for Kids & Teens			
11:00 am - 12:00 pm	Beyond the Surface: Teen Voices on Life and Feelings with Dr. Kristin	Kristin Lindquist, PsyD	Wright B
1:00 pm - 2:00 pm	The Caring Carousel: Kids' Chat with Dr. Kristin	Kristin Lindquist, PsyD	Lindbergh AB
2:00 pm - 3:00 pm	**Patient-Led Panel:** Level Up: Career Talks for Teens and Young Adults	Mairéad Ryan	Wright B

Agenda

THE RYR-1 FOUNDATION INTERNATIONAL FAMILY CONFERENCE 2025 AGENDA

Sunday, July 25, 2025		
Time	**Open Hours: Registration, Lounges, and Kids' Club**	**Room**
7:30 am - 2:00 pm	Registration Desk	Glenn Foyer
9:00 am - 2:00 pm	The Funhouse Clubhouse (Kids' Club for Ages 5 - 12)	Lindberg AB
9:00 am - 2:00 pm	The Teen Spot (Teen Lounge for Ages 13 - 17)	Wright B
Time	**Event**	**Room**
10:00 am - 12:00 pm	**Brunch Before Goodbye: A Warm Send-Off**	**Regency Ballroom**
12:00 pm - 2:00 pm	Closing Connections: Final Social Time	Regency Ballroom

Sponsors

The RYR-1 Foundation extends its deepest gratitude to the generous sponsors and donors of The RYR-1 Foundation International Family Conference 2025.

Your support made this meaningful gathering possible and helped bring together individuals and families affected by RYR-1-Related Diseases (RYR-1-RD) from around the world. We are deeply thankful for your partnership and commitment to our mission.

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- Chris & Kate Reinmiller
- The Van Nice Family
- Mary Beth Walsh
- Courtney & Adam Woomer
- Carla Zingariello




Family Conference Planning Committee & Volunteers

Family Conference Planning Committee Members

We extend our heartfelt thanks to the incredible members of the Family Conference Planning Committee. Your time, creativity, and unwavering dedication have been essential in bringing this event to life.

The Family Conference Planning Committee is made up of dedicated Directors and Advisors of The RYR-1 Foundation, including **Ms. Jeni Ryan (Chair), Mr. Winston Chung, Dr. Robert Hanich, Mrs. Lauren Heft, Mrs. Sofia Leon, Mrs. Emily Pedersen, Ms. Mairéad Ryan, and Ms. Brentney Simon.** From shaping the agenda to creating an inclusive and welcoming experience for all, your efforts have made a meaningful difference. We are truly grateful for your passion, collaboration, and commitment to the RYR-1-RD community.

Volunteers

We are extremely grateful to all the amazing volunteers who gave their time and energy to help make the 2025 Family Conference run smoothly.

Your support made a huge difference and helped create a warm, inclusive experience for our entire community.

We truly couldn't have done it without you—thank you!

Important Information

- **Lena Leghart - Program Manager:** Cell Phone: **(412) 223-7236** | Email: **lena@ryr1.org**
  Please direct any questions, problems, or issues to Lena during the weekend and she will be happy to assist you. She can be reached in person, by phone (call and/or text), or by email.

- **Melissa Flaherty - Family Conference Coordinator:** Cell Phone | **(412) 223-7236**
  Please direct any questions, problems, or issues to Melissa during the weekend and she will be happy to assist you. She can be reached in person or by phone (call and/or text).

- **Front Desk at the Hyatt Regency Pittsburgh International Airport:** 1111 Airport Blvd, Pittsburgh, PA 15231 | **(724) 899-1234**

Additional Information

Toilet seat risers have been installed in the accessible stalls of the public restrooms in the *Regency Foyer* convention space to help individuals safely and comfortably lift oneself from the toilet. The accessible stalls in the public restrooms *near the lobby in the Glenn Foyer* remain unmodified for those who prefer not to use risers.

For details about the hotel, Pittsburgh dining and shopping, airport services, transportation, nearby pharmacies and hospitals, and local activities, visit our **FAQs Page**!

It's packed with helpful information about the Family Conference, travel, accommodations, accessibility, and more.

The Funhouse Clubhouse & The Teen Spot

The Funhouse Clubhouse, located in Lindbergh AB, is led by Michelle Archibald and Lee Noviello. Open to children ages 5 and older, The Funhouse Clubhouse will be running from Thursday, July 24 through Sunday, July 27.

This space offers a wonderful opportunity for kids to make new friends, engage in creative activities, and—most importantly—have fun! Each day features thoughtfully planned programming aligned with our "Carnival" theme, designed to include children of all interests and abilities. While the kids enjoy themselves, parents can fully participate in the Family Conference.

Be sure to check out The Funhouse Clubhouse Agenda for the full schedule of events!

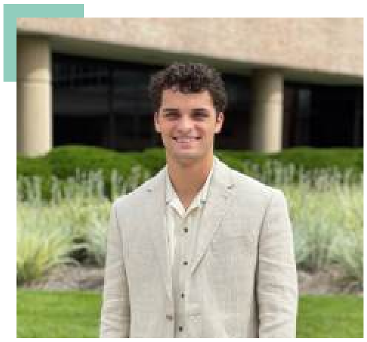
The Teen Spot, located in Wright B, is designed as a hangout for teens ages 13–17 who want a space to connect, relax, or take a break from sessions. In addition to The Teen Spot, there will be special programming for teens and young adults, which can be found in the **Family Conference Agenda.**

Teens ages 15 and up are also welcome to volunteer and assist in The Funhouse Clubhouse—an awesome way to get involved and support younger participants!

REMINDER:
Any special assistance your child may need during the **Kids' Club** will be the responsibility of a family member or designated adult. Please ensure the **Kids' Club Questionnaire** is completed before your child attends.

If your child has any allergies, be sure to inform the Kids' Club Coordinators and provide the appropriate allergy-identifying sticker for their ID badge.

Expert Speakers



Jeremy G. Armann, PhD, NCSP

Dr. Jeremy G. Armann is a clinical fellow in school-based mental health and special education at Harvard Medical School, working within the Manville School at the Baker Center for Children and Families. In this role, he provides therapeutic services, case management, and evidence-based interventions to students with complex needs. Dr. Armann recently earned his PhD in School Psychology from Duquesne University, where his dissertation, *"The Educational Experiences of Children and Adolescents with an RYR1-Related Disease,"* explored school-based challenges and supports for children and adolescents within the RYR-1-RD community. He has presented at The RYR-1 Foundation Family Conference 2022 and delivered a webinar to raise awareness about educational needs for individuals with an RYR-1-RD. His work blends clinical expertise with a deep commitment to advocacy, consultation, and research-driven practice for families impacted by RYR-1-RD.

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.

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

Julie Bowden, PsyD

Dr. Julie Bowden is a licensed clinical psychologist in the state of Pennsylvania as well as the co-owner and director of Resilient Minds Behavioral Health. Employing a primarily cognitive-behavioral approach, Dr. Bowden strongly believes in the connection between one's thoughts and feelings, and in the ability to make changes in this area to support personal growth.

After earning her undergraduate degree in Human Development and Family Studies from Penn State University (University Park campus), Dr. Bowden obtained her master's and doctoral degrees in Clinical Psychology from Indiana University of Pennsylvania. She completed her predoctoral internship at Torrance State Hospital and was licensed as a psychologist in Pennsylvania in 2009. Since this time, she has worked with hundreds of individuals in various settings to assist them in improving their overall functioning and mental health.

Dr. Bowden has extensive experience in assessing, diagnosing, and treating individuals ages 12 years and up with a variety of issues including anxiety, depression, bipolar disorder, life stress, and self esteem concerns. In addition, she has a special interest and passion for working with mothers in all stages, from preconception to postpartum and beyond, having a particular compassion and awareness for treating those who are suffering from various challenges related to motherhood. Dr. Bowden continues to expand her practice to focus on women who are struggling with postpartum depression and anxiety.

In addition to her clinical work, Dr. Bowden finds speaking to groups at various retreats, conventions, conferences, and trainings incredibly fulfilling. She particularly enjoys training others to improve their overall functioning and mental health via the use of self care, boundary setting and stress management. She is honored to bring her skills and expertise to The RYR-1 Foundation Family Conference 2025.

Robert T. Dirksen, PhD

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.

Dr. Dirksen is the Chair of The RYR-1 Foundation Scientific Advisory Board.

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.

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.

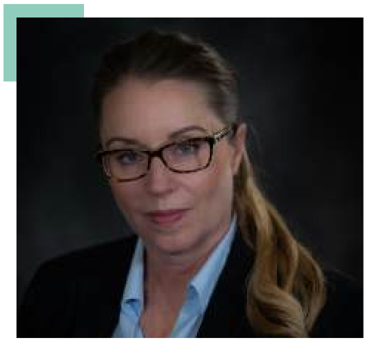
Her research is designed to identify new targets and interventions for RYR-1-RD. Her lab has created many new mouse models of RYR-1-RD and has identified several potential therapeutic interventions. Dr. Hamilton is the senior author on numerous peer-reviewed publications, and her research program has 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 serves as a Director on The RYR-1 Foundation's Board of Directors.

Andrew Herlich, DMD, MD, FAAP, FASA, FAAOMS(H)

Dr. Andrew Herlich is a recently retired pediatric anesthesiologist. He has provided over 20 years of service as a Hotline Consultant for MHAUS and remains active in this role. He has both dental and medical degrees which helped him navigate the challenges of anesthesia safety in both the hospital and office-based environments. He is the American Society of Anesthesiologists' Liaison to the American Dental Association, the American Association of Oral and Maxillofacial Surgeons, and the American Dental Society of Anesthesiologists.



Tammy L. Hughes, PhD, ABPP

Dr. Tammy L. Hughes is a Duquesne Professor of School Psychology at Duquesne University in Pittsburgh. She is a Past President of both the Division of School Psychology (Division 16) of the American Psychological Association (APA) and the Trainers of School Psychologists (TSP). She currently co-chairs the APA Presidential Task Force on Safety of Schools (SOS). She is also the past chair of the Coalition for Psychology and Education in Schools, where she is the project leader of CDC grant-funded initiative to address Education Disparities and the use of psychological science to support classroom management for teachers and parents. Dr. Hughes is committed to identifying strategies that result in promoting student health, mental health and educational attainment that support positive lifetime outcomes. She developed her interests as a result of working as a psychologist treating youth, as a consultant and collaborator to school teams and community agencies as well as promoter of best practices via training to justice personnel. This work often engages decision makers in effecting change in their policies, and bringing together school and community personnel, parents and child advocates and other experts to work together on socially complex challenges. She is experienced in solving wicked problems– the types of problems where there is a number and diversity of stakeholders whose influence can undermine progress if their priorities are not considered (Conklin, 2001).

Dr. Hughes is a school psychologist, a licensed psychologist and is Board Certified in School Psychology. She is a Fellow of the American Psychological Association. Dr. Hughes' research focuses on marginalized youth particularly those who are at high risk for disciplinary actions that result in justice contact.

Megan Ingram, Esq

Megan Ingram is an attorney with the law firm of Steinbacher, Goodall & Yurchak, an elder care and special needs law firm with seven offices across Pennsylvania. Megan works out of their State College office and leads the firm’s Special Needs Planning department. She also works in the firm’s Wealth Protection Planning department to craft estate plans for clients who have a net worth of two million dollars or more, with a special emphasis on business succession planning and federal estate tax planning. She was honored to be designated as a Rising Star Attorney by Super Lawyers for 2022-2025, and she serves on the Board of Directors of the Centre Region Estate Planning Council.

Megan resides in Lewistown, Pennsylvania with her son, Jonathan, and their goldendoodle.

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.

Dr. Jain is a member of The RYR-1 Foundation’s Scientific Advisory Board.

Kristin Lindquist, PsyD

Dr. Kristin Lindquist is a licensed clinical psychologist and registered art therapist with over 15 years of experience specializing in children and adolescents' emotional and developmental needs. With a warm and integrative approach, Dr. Lindquist brings together evidence-based psychological interventions and the expressive power of art therapy to support young people navigating a wide range of challenges.

Her clinical background includes work in schools, hospitals, and therapeutic day programs, where she has provided assessment, therapy, and consultation for individuals with autism spectrum disorders, ADHD, anxiety, depression, trauma, and learning differences. She is particularly skilled in supporting children with complex emotional and behavioral profiles, helping them build self-regulation, resilience, and connection.

Dr. Lindquist is passionate about creating a safe, developmentally attuned space where young clients can explore their feelings, strengthen their sense of self, and develop new ways of relating to others. She also works closely with families to foster understanding and growth across home and school environments. Currently, she works with AHN Women's Behavioral Health at the Alexis Joy D'Achille Center for Perinatal Mental Health to treat mothers struggling with mental health challenges during the perinatal period.

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.

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

Oscar Henry Mayer, MD

Dr. Oscar Henry Mayer is a Professor of Clinical Pediatrics at the Perelman School of Medicine at the University of Pennsylvania, and an attending pulmonologist within the Division of Pulmonology at Children's Hospital of Philadelphia (CHOP). He is the medical director of the CHOP Pulmonary Function Testing (PFT). He has been actively involved in the specialized care of patients with neuromuscular disease and complex chest wall and spinal deformity. He has been the pulmonologist in the Neuromuscular Center for the last 23 years and in the Center for Thoracic Insufficiency Syndrome since its inception 20 years ago. He is on a number of national committees and study groups involved in pediatric pulmonary function testing, the assessment and management of children with complex thoracic spinal disorders and neuromuscular disorders, on which he has lectured nationally and internationally and has published over 90 journal articles and book chapters on his research and clinical interest.

Andrew McCarty, MS, LCGC

Andrew McCarty is the founder of Clover Genetics and an experienced board-certified genetic counselor whose work is driven by a passion for expanding access to genetic care through telehealth.

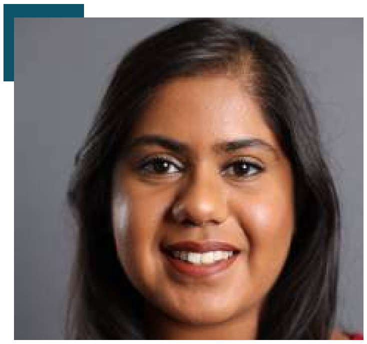
Clinically, Andrew has supported patients and families across pediatrics, oncology and, most pertinently for the RYR-1-RD community, neuromuscular conditions. He's committed to demystifying complex genetic information and empowering individuals to play an active role in their health and wellness.

In addition to his private-practice work, Andrew holds a supervisory faculty appointment at the University of Pittsburgh School of Public Health and Wake Forest Genetic Counseling Programs, where he mentors emerging genetic counselors and delivers lectures on genetics topics.

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.



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.

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.

Jennifer Newitt, MD

Dr. Jennifer Newitt obtained her MD from Albany Medical College in 2012 and then completed a pediatrics residency at Indiana University in 2016. She then came to the University of Pittsburgh, where she completed a fellowship in Pulmonary and Critical Care fellowship, followed by a fellowship in Sleep Medicine. Dr. Newitt is currently a Clinical Instructor at UPMC and a Postdoctoral Scholar at the University of Pittsburgh.

Her research interests focus on the use of non-invasive ventilation in patients with neuromuscular disease.

Jenee' Oliver, Esq

Ms. Jenee' Oliver has worked as a staff attorney assigned to DRP's Pittsburgh Office since 2023. She received her BA from Point Park University in Legal Studies and her JD from Duquesne University Law School. While in Law School, Jenee' clerked for the Honorable Maureen Lally-Green in the Superior Court of Pennsylvania and interned with the Allegheny County District Attorney's Office. Ms. Oliver is an experienced litigator having litigated a range of cases including Criminal Defense, Civil Rights and Family Law. At DRP, Ms. Oliver has worked on a range of matters, including employment discrimination, housing discrimination, vocational rehabilitation services, and transportation matters. She is currently focusing on issues impacting access to health care for people with disabilities, including physical and communication barriers, and how to address them through education, training, and policy advocacy.

Sheila Riazi, MSc, MD, FRCPC

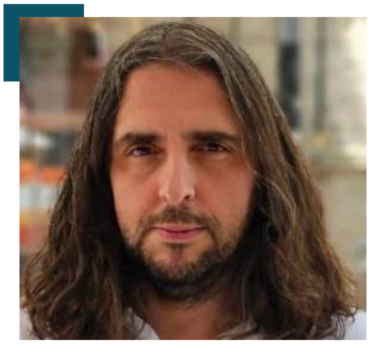
Dr. Sheila Riazi is a Professor at the Department of Anesthesia, Site Head of Anesthesia at Toronto Western and Women's College Hospitals, a staff anesthesiologist at University Health Network, and clinician scientist at Toronto General Research Institute. Dr. Riazi directs the only Canadian diagnostic 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.

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

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.



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.

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

Christine Porter, MA, JD

Christine Porter (she/her) is a staff attorney in the Pittsburgh office of the Education Law Center - PA. Prior to joining the Education Law Center, she was the Deputy Director of the Allegheny County Public Defender's Juvenile Division, where she designed the division's first education advocacy program. Christine has represented families as an education attorney with a private firm in Seattle and practiced disability and elder law with Neighborhood Legal Services in Pittsburgh. She has served on the boards of nonprofits advocating for juvenile justice reform and for the rights of individuals with disabilities.

Christine received her MA and JD degrees from the University of Washington in Seattle. Before law school and graduate school, Christine worked in public school classrooms through an education nonprofit in Florida.

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.

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

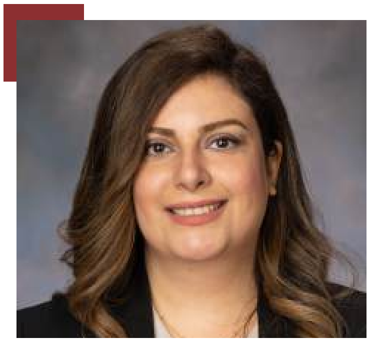
The RYR-1 Foundation Research Grant Awardees

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.



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.

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.

Group Session Instructors

Michael Brucker, PT, DPT, CSCS, CNC

Dr. Michael Brucker holds a Doctor of Physical Therapy degree and is a Certified Strength and Conditioning Specialist as well as a Certified Nutrition Coach. Michael attended Saint Francis University (Pennsylvania), where he received his Bachelor of Science in Health Science with a minor in Movement Science. He then went on to receive his Doctor of Physical Therapy degree from Saint Francis University. He has been a practicing Physical Therapist (PT) for six years and has demonstrated experience working in multiple therapy settings. Specialties include orthopedic/neuromuscular rehabilitation, corrective exercises and posture, injury prevention, sport specific training, flexibility and mobility, and weight loss/nutrition. Michael has always had a passion for health and wellness. He believes that the most effective treatment approach takes every aspect of wellness into consideration including nutrition, psychological well-being, and physical wellness. Actively listening to a patient's concerns and goals is crucial in building a strong and meaningful patient-therapist rapport. Keeping up with the latest research-based treatment approaches, Michael is constantly striving to better himself as a clinician to be of greater value to his patients. What Michael enjoys most as a PT is being able to influence his patients to take an active interest in their own health, and to improve their overall quality of life and longevity.

Jennifer Orseck

Jennifer Orseck is leading chair yoga, which is a gentle and accessible form of yoga that can be practiced while seated in a wheelchair or using a chair for support and balance.

Jen received her 200 hour yoga teacher training in 2018 at Asheville Yoga Center. One of her three children has RYR1-related myopathy and is severely impacted by the disease. Jen was found to also have RYR1-related myopathy after her daughter’s diagnosis. She is actively involved in fundraising and advocacy for the disability community at home in Spartanburg, South Carolina.

Patient-Led Panelists

Marcie Epstein

Marcie Epstein is a dedicated volunteer, advocate, and proud mother. She lives near Philadelphia with her husband, Steven, and their son, Jackson. Both Steven and Jackson have Central Core Disease, a form of RYR-1-RD. Jackson is more severely affected and uses a manual wheelchair for mobility. He has been an inspiration for Marcie's active involvement in the disability community.

Marcie has been involved with The RYR-1 Foundation since 2014 and previously served on the Board of Advisors. Her family was also featured in the organization's documentary, helping to raise awareness about RYR-1-RD. She also volunteers with Children's Hospital of Philadelphia's Family Partners Program, where she provides input on programs and services from a patient family's perspective and presents during new employee orientation sessions.

In addition to her advocacy work, Marcie serves as secretary of Jackson's high school Home and School Association and plays an integral role in supporting the school's marching band and indoor percussion ensemble, both of which Jackson participates in. As a dedicated band mom, she contributes to fundraising, manages uniforms, and chaperones events, helping to ensure students have the resources and support they need to thrive.

Outside of her volunteer commitments, Marcie enjoys crafting and traveling with her family. Together, they have explored Europe, Canada, the Caribbean, and many parts of the United States, including Alaska. She takes pride in planning accessible travel experiences for Jackson, ensuring every trip is inclusive and memorable for the whole family.

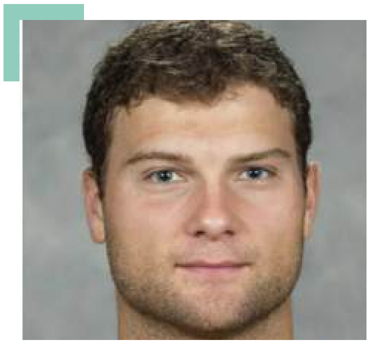
Robert Hanich, MD, FACC

Bob retired from Asheville Cardiology Associates in western North Carolina after thirty years as an electrophysiologist and interventional cardiologist. He holds an AB from Harvard College and an MD from the University of Pennsylvania. He completed his Internal Medicine residency at University of Pennsylvania and his Cardiovascular Disease fellowship at Johns Hopkins. His dominant RYR1 mutation presents as the Malignant Hyperthermia/rhabdomyolysis phenotype.

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

Lauren Heft

Lauren Heft is the parent of a teenager with an RYR-1-RD. Her son, Charlie, is 14 years old and was diagnosed at age five with RYR1 myopathy caused by a de novo dominant mutation. She lives with her family in Dublin, Ohio, and for the past 12 years has been a stay-at-home parent and volunteer for her children's schools and other local nonprofits. She and her husband, Bryan (who serves as a director on The RYR-1 Foundation's Board of Directors), are proud supporters of The RYR-1 Foundation and its mission.



Cody Hodgson

Cody Hodgson is a Canadian former professional ice hockey player, playing six seasons in the National Hockey League with the Vancouver Canucks, Buffalo Sabres, and Nashville Predators. In 2016, Cody retired from playing professional hockey after being diagnosed with Malignant Hyperthermia (a form of RYR-1-RD). He has also become a spokesman for The RYR-1 Foundation and is committed to raising awareness for those suffering from RYR-1-RD.

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

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.

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.

Amy Perrin, MS, FNP-C

Amy Perrin is a family nurse practitioner in primary care. She is currently a PhD student at Binghamton University. Her passion for advocating for The RYR-1 Foundation is due to the support and community The RYR-1 Foundation has provided to her family. She gives back through her fundraising efforts. Her daughter was diagnosed with RYR1 myopathy through whole exome sequencing, and her muscle biopsy was inconclusive. She firmly believes that, as a family unit, the Perrins do well thanks to the support of her RYR-1 "family."

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

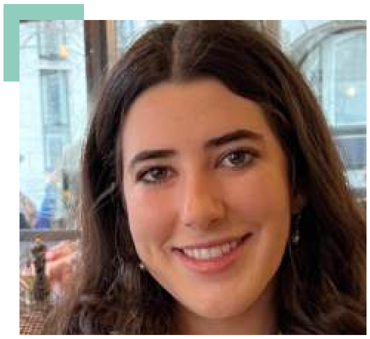
Courtney Perrin

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.

Jennifer Ryan, MBA

Jeni Ryan is a trustee of the Schooner Foundation and holds an MBA in Operations Management from Simmons College. She is the former COO of Integral Resources, a telemarketing firm specializing in small donor acquisition for nonprofits. She has Central Core Disease (CCD), an RYR-1-RD caused by a mutation in the RYR1 gene. Jeni is the chair of The RYR-1 Foundation International Family Conference 2025 Planning Committee. Her passion for bringing together the RYR-1-RD community has been a cornerstone of her involvement with The RYR-1 Foundation.

Jeni serves as Vice President of 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.

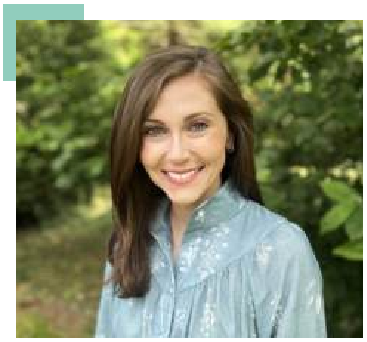
Ilaria Salvatori

Ilaria Salvatori is a patient living with a RYR1-related myopathy, a radiographer with over a decade of clinical experience, and currently a PhD candidate in Pedagogy at the University of Verona (Italy), specializing in healthcare professional education. Her academic work explores the intersection of technological competencies, medical humanities, and the development of professional identity among healthcare technologists.

Ilaria's commitment to education is deeply shaped by her lived experience. Diagnosed at the age of 29, after a lifelong journey marked by misdiagnoses since early childhood, she brings a unique, triple-perspective lens to her work: that of a person living with a rare disease, a healthcare professional, and a researcher. She strongly advocates for the active engagement of patients and caregivers, not merely as a right, but as a pedagogical and ethical imperative grounded in empathy, narrative competence, and relational ethics.

She has served as an adjunct professor and clinical tutor at several Italian universities and is actively involved in public education and advocacy on rare diseases. Ilaria is a member of the Italian Society for Medical Pedagogy (SIPeM) and serves on the scientific committee of FASTeR, the Federation of Scientific Associations of Radiographers in Italy, which supports professional development, research, and continuing education for radiologic technologists nationwide.

Her unwavering dedication to promoting equitable access to diagnosis, care, and knowledge stems from both her personal and professional journey, a commitment she continues to carry forward through her research, teaching, and public engagement.



Courtney Woomer

Courtney, a Lexington, Kentucky native, graduated from the University of Kentucky in 2011. She serves as the HR Director for Kentucky Primary Care Association, a nonprofit with the mission of providing healthcare to the underserved in rural areas of Kentucky. She volunteers as a peer mentor in the NICU at University of Kentucky Hospital, where she provides support, and also receives strength from other parents who share similar family experiences. Most importantly though, Courtney is devoted to her family, including her husband and three daughters; her youngest daughter, Maisie, has a recessive form of RYR-1-RD. Both professionally and personally, Courtney champions for those who struggle, and admires their resilience and ability to overcome.

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

Staff & Coordinators

Lindsay Goldberg, BSN, RN / Executive Director & Patient Liaison

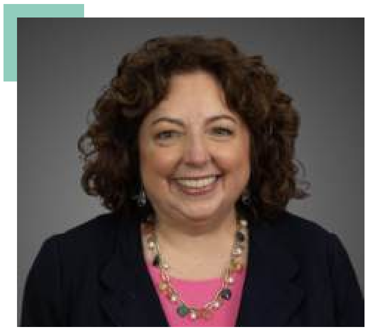
Lindsay is also a co-founder of The RYR-1 Foundation. She serves as an advocate for the RYR-1-RD community and liaison between affected individuals/their families and physicians, genetic testing companies, and scientists. She has a BSN from the University of Michigan and spent the first 10 years of her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

“The RYR-1 Foundation has a very special place in my heart as a co-founder and family member of an affected individual. As a nurse, I have seen firsthand how important it is to have an organization that supports individuals and families affected by a specific disease.”

Lena Leghart, BSW / Program Manager

Lena 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.

“The RYR-1 Foundation has shown me how one organization can make such a difference in a small community. Being a part of this team allows me to challenge myself to make a difference, no matter how small, to better the lives of others.”



Patricia Raffaele, MA, CAE / Development, Public Relations, & Research Director

Pat joined The RYR-1 Foundation in February 2024 as the Development, Public Relations, & Research Director. 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.

With her background in healthcare associations and nonprofit organizations, she brings experience in development, communications, advocacy, and governance. In her role at The RYR-1 Foundation, she focuses on development to support the mission of the organization; communications to “tell the story” of The RYR-1 Foundation and those it serves; and manages the research grant program.

Pat is passionate about the mission of The RYR-1 Foundation and is excited to bring her experience to support its work with researchers throughout the world; families and individuals affected by RYR-1-RD; and the medical community.

Melissa Flaherty, JD

Melissa Flaherty, JD is the Drug Free Communities Grant Coordinator for the Pawtucket Prevention Coalition in Pawtucket, Rhode Island. She has been working in the public health field for over 20 years.

She earned her bachelor's degree in communications from the University of Rhode Island and her Juris Doctor from Southern New England School of Law, and she is a Rhode Island Certified Mediator. Prior to working in Pawtucket, she served as the Substance Abuse Task Force Director in a rural community, addressing underage drinking and tobacco use.

She has also served as the Resident Services Director at the Pawtucket Housing Authority, where she developed and implemented programs on health and wellness topics, including healthy food initiatives, parenting forums, domestic violence prevention workshops, and childhood safety. She has implemented youth programs in community centers, police substations, and public housing sites, and has collaborated with local community partners to assemble and distribute thousands of Narcan kits to address the opioid crisis.

Melissa has also been active in mobilizing her community around issues of housing and homelessness, mental health, and cancer prevention. She currently serves as the Board Chair of The Blackstone Academy Charter School in Pawtucket, Rhode Island.

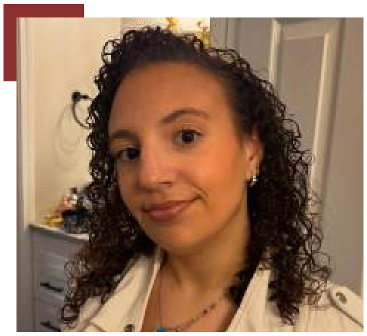
Melissa is supporting Lena on-site in coordinating the Family Conference.

Michelle Archibald

Michelle is a 44-year-old nature enthusiast, born in the Upper Peninsula of Michigan. After getting married, she moved to Sault Ste. Marie, Ontario in Canada for 11 years before moving to the Pittsburgh area four years ago. She is a mother to two grown boys, Dylan, 24 and Breydan, 16.

She has always loved children and worked with them in one capacity or another. For the last 10 years, she has lovingly worked in special education as a teaching assistant, and for the last two years she has been working as Lee Noviello's assistant—they couldn't be a better team! She looks forward to meeting all the wonderful kids and individuals associated with such a meaningful organization!

Michelle is one of the Kids' Club Coordinators.



Lee Noviello

Lee is originally from the Philadelphia area, but moved to Pittsburgh for college, where she met her amazing husband, Nick. They just got married in June!

Lee attended the University of Pittsburgh, where she received both her undergraduate and Master's in Education. She is qualified to teach both General Education and Special Education and has been teaching in a special education role for the last three years. Currently, she is a special education teacher at a school in Beaver County, where she works alongside her assistant, Michelle Archibald. Together they will be running The Funhouse Clubhouse! She is excited for this opportunity!

Lee is one of the Kids' Club Coordinators.

Our Team

Board of Directors

We are deeply grateful for the leadership and unwavering support of our Board of Directors. Their dedication and commitment are essential to the success and smooth operation of The RYR-1 Foundation. Without their guidance and tireless efforts, we would not be able to fulfill our mission or serve our community as effectively.

Our Board of Directors includes remarkable individuals who bring diverse expertise and passion to our organization: **Dr. Michael F. Goldberg (Board President), Ms. Jennifer Ryan (Board Vice President), Mr. Drew Huseth (Secretary), Mr. Randolph Pepper (Treasurer), Mr. Michael Legum (Assistant Treasurer), Dr. Susan Hamilton, Dr. Robert Hanich, Mr. Bryan Heft, Dr. Justin McArthur, Mrs. Emily Pedersen, Mrs. Courtney Woomer, and Dr. Donald Zack.**

We are especially honored to have our co-founders, **Dr. Morton F. Goldberg and Mrs. Myrna Goldberg**, continue their involvement as Director Emeriti, providing invaluable historical insight and ongoing support.

To learn more about the backgrounds and contributions of our Directors, please visit the Our Team: Board of Directors page.

Scientific Advisory Board (SAB)

We are extremely grateful to the members of The RYR-1 Foundation's SAB who have been instrumental in the growth of The RYR-1 Foundation and in the understanding of RYR-1-RD. This group is composed of the world's leading authorities in RYR-1-RD. The SAB helps to determine the research priorities of The RYR-1 Foundation. We are extremely grateful to **Dr. Robert T. Dirksen (Chair), Dr. Carsten G. Bönnemann, Dr. James Dowling, Dr. Minal S. Jain, Dr. Heinz Jungbluth, Dr. Johanna Lanner, Dr. Andrew R. Marks, Dr. Isabelle Marty, Ms. Livija Medne, Dr. Payam Mohassel, Dr. Ichizo Nishino, Dr. Sheila Riazi, Dr. Filip Van Petegem, and Dr. Nicol C. Voermans** for their continued guidance and participation in our many important programs.

To learn more about our Scientific Advisory Board members and read their bios, please visit the Our Team: SAB page.

Board of Advisors

The RYR-1 Foundation's Board of Advisors is composed of dedicated individuals who offer valuable experience, insight, and strategic guidance to support our mission. Their expertise helps strengthen our programs, outreach, and long-term impact across the RYR-1-RD community.

To learn more about the backgrounds and contributions of our Advisors, please visit the Our Team: Board of Advisors page.

Join The RYR-1 Foundation Patient Registry

Enrolling in the RYR-1-Related Diseases (RYR-1-RD) Patient Registry is one of the most effective ways for you to support RYR-1-RD research. Enrollment in the patient registry will encourage scientists, physicians, and pharmaceutical companies to pursue research to find a treatment and/or cure for RYR-1-RD. Your enrollment in the registry could also make you potentially eligible for clinical trials for treatments of RYR-1-RD.

We highly encourage you to enroll—just fill out the Patient Registry Form online to get started!

- **There are several reasons why this project is so critical. This database will serve to:**

1) Increase our understanding of RYR-1-RD: RYR-1-RD is a rare group of diseases, though likely more common than often assumed. *RYR1* gene-related abnormalities have been associated not only with congenital myopathy, but also with malignant hyperthermia, intolerance of certain cholesterol-lowering drugs (“statin myopathy”), and exercise intolerance. Building a patient database will allow physicians, researchers, and genetic counselors to better understand the different forms of RYR-1-RD, their natural history, and their prevalence.

2) Conduct clinical trials: As a rare disease, an RYR-1-RD can present a challenge to researchers, pharmaceutical companies, and biotech firms who want to test potential therapies. Without easy access to patients who can serve as study subjects, clinical trials cannot be performed. And without clinical trials, hope for a cure is lost. Thus, by developing a database of RYR-1-RD patients, we can collaborate with those who have found potentially promising therapies.

3) Find a cure: This is the ultimate mission of The RYR-1 Foundation. Clinical trials are essential to developing and approving effective treatments—and ultimately, a cure. By joining the patient registry, you are directly contributing to this life-changing goal.

Advance Research: Participate in the NIH Clinical Study on RYR-1-Related Disorders

The Natural History Study on RYR-1-Related Disorders (RYR-1-RD) being conducted at the National Institutes of Health (NIH) is now accepting participants! The study aims to better understand the clinical features and progression of RYR-1-Related Disorders (RYR-1-RD) in both children and adults.

Leading the study is Tokunbor A. Lawal, PhD, FNP-BC, an independent research scholar in the Division of Intramural Research at the NIH.

The details are below:

Who Can Participate?

- Individuals 7 years or older
- Confirmed genetic diagnosis of RYR-1-RD
- Resides in the United States

Who Is Not Eligible?

- Individuals with other neuromuscular diseases causing muscle weakness

Interested in Participating?

To learn more or to enroll, contact the NIH Office of Patient Recruitment: call **(800) 411-1222** or email **ccopr@nih.gov**. Please refer to Research Study #001737-CC.

Participation in the study could help advance research and future treatments for RYR-1-RD!
Find additional information in the **Natural History Flyer.**

About The RYR-1 Foundation

The RYR-1 Foundation is a nonprofit public charity, designated 501(c)(3) by the IRS. Members of the Goldberg Family started The RYR-1 Foundation in 2014. Their family has been affected by an RYR-1-Related Disease (RYR-1-RD). Currently, there is no other organization that exists solely to advocate for and serve the needs of patients with RYR-1-RD. The goal of The RYR-1 Foundation is to fill this much-needed void.

Mission Statement: The mission of The RYR-1 Foundation is to support research leading to an effective treatment or a cure for RYR-1-RD.

To achieve this mission, The RYR-1 Foundation pursues the following goals:

1) Support Research: The RYR-1 Foundation makes grants to researchers interested in RYR-1-RD. As the only organization solely dedicated to RYR-1-RD, we fund, promote, and highlight research in this area. Our ongoing development of a **patient registry** is also key to promoting clinical trials of potential therapies.

2) Medical Professional Education: The vast majority of medical professionals have never heard of RYR-1-RD. We raise awareness through resources on our website, including the latest **medical literature**, as well as direct meetings with medical professionals around the world.

3) Patient/Family Support and Advocacy: Due to the rarity of RYR-1-RD, receiving this diagnosis can be an anxiety-provoking and isolating experience for an affected individual and their families. We serve as a resource for patients and their families through our **website**, other forms of social media, and **Family Conferences.**

Engage with The RYR-1 Foundation Online:

We'd love to see your photos!

Be sure to share your favorite moments from the Family Conference on social media and tag us so we can see and share your posts!

Social Media Handles:

Facebook: **@ryr1foundation**

Instagram: **@theryr1foundation**

LinkedIn: **@the-ryr-1-foundation**

X (formerly Twitter): **@RYR1foundation**

Don't Forget to Use Our Official Hashtags:

#RYR1FamilyConference / #StrengthInNumbersRYR1

Let's spread awareness and celebrate this incredible community together!

National & International Affiliations of The RYR-1 Foundation

In our ongoing efforts to promote our mission and activities—both in funding research and providing family support and outreach—we have established strong partnerships with several key organizations. These include the **National Organization of Rare Disorders (NORD), AFM-Téléthon (Association Française contre les Myopathies), Cure Congenital Muscle Disease (Cure CMD), Congenital Muscle Disease International Registry (CMDIR), Malignant Hyperthermia Association of the United States (MHAUS), MTM-CNM Family Connection, ZNM – Zusammen Stark!,** and the **Muscular Dystrophy Association (MDA)**.

Additionally, we regularly participate in briefing sessions with pharmaceutical and companies as well as consultants across the United States and world.

NORD®
National Organization for Rare Disorders

CMDIR
congenital muscle disease international registry
leading the way to a treatment and cure

MTM CNM Family Connection, Inc
Resources•Research•Relationships

ZNM – Zusammen Stark! e.V.
Verein für Zentronukleäre Myopathien

MDA® Muscular Dystrophy Association

MHAUS
MALIGNANT HYPERTHERMIA ASSOCIATION OF THE UNITED STATES

cure CMD
a cure is among us

AFMTÉLÉTHON
CURE THROUGH INNOVATION

Looking Ahead

Family Conference Feedback & Survey :

Please fill out the Family Conference 2025 Survey.

The RYR-1 Foundation kindly asks each attendee of the 2025 Family Conference to complete the survey linked above. Please submit only one survey per person. The survey should be completed at the conclusion of the Family Conference and will remain online until September 1, 2025.

Please email Lena Leghart, Program Manager, **lena@ryr1.org**, with any questions you have.

Future Family Conferences:

If you are interested in attending future Family Conferences, we welcome volunteers, **donations**, and ideas.
For more information, please contact Lena Leghart, Program Manager, at **lena@ryr1.org.**

Resources & Helpful Literature

The RYR-1 Foundation's **Clinical Care Guidelines: What Patients & Families Need to Know About RYR-1-Related Diseases**

American with Disabilities Association Literature and Materials

The Muscular Dystrophy Association Quest Magazine

Guide for Families with a Congenital Myopathy

RYR-1-RD Medical Journal Articles on PubMed

Muscular Dystrophy Association

MyID First Responders Bracelet

American Medical ID

Congenital Muscle Disease International Registry

Malignant Hyperthermia Association of the United States

Your generosity makes our work possible.

To make a donation to **The RYR-1 Foundation,**

please visit our donation page.

Thank you for your continued support!

STRENGTH IN NUMBERS

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