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Major NIH Grant Awarded to Members of the RYR-1 Foundation's Scientific Advisory Board

August 10, 2020- A highly competitive grant from the National Institutes of Health (NIH), worth **over \$2 million**, was just awarded to Drs. Robert Dirksen and James Dowling, members of the Scientific Advisory Board (SAB) of the RYR-1 Foundation. This grant will fund their continued research of recently developed mice with RYR-1-Related Diseases (RYR-1-RD).

The title of their grant is: "Pathophysiology and Treatment of Recessive RYR1 Related Myopathy." More details on this grant can be found at: www.ryr1.org/nihgrant.

The data presented in their NIH grant application were based, in part, on research that was funded by the RYR-1 Foundation. Furthermore, both of the mouse lines to be studied in this research grant were *created at the direction of the RYR-1 Foundation's SAB and with funding from the RYR-1 Foundation*. Each of these mouse lines has severe weakness due to their RYR1 mutations, making them ideal for research, including the assessment of potential therapies. The creation of RYR-1 mice with severe weakness was a major advancement in RYR-1 research. One of these mouse lines has been carefully studied and described in a recently published article in a peer-reviewed journal.¹

"The creation of these mouse lines has already led to important advances in the field of RYR-1-RD," says Michael F. Goldberg, MD, MPH, President of the RYR-1 Foundation. "In addition, ***it is particularly gratifying when funding from the RYR-1 Foundation can help researchers obtain substantially larger grants from premier organizations like the NIH. It***

¹ Brennan S, Garcia-Castañeda M, Michelucci A, et al. Mouse model of severe recessive RYR1-related myopathy. *Human Molecular Genetics*, 2019;28:3024–3036. The article can be accessed by clicking [HERE](#).

is a major 'return on investment' for the RYR-1 Foundation, its benefactors, and the entire RYR-1 community."

"Using the animal models of severe, early-onset recessive RYR1-RD developed by the RYR-1 Foundation, this award provides the resources needed to uncover the underlying disease mechanisms and to develop new and effective therapeutic interventions for this devastating disease," Dr. Robert Dirksen said. ***"These advances would simply not be possible without the tremendous support of the foundation, patients, and family members living with RYR1-RD. Their collective commitment to an effective treatment or cure for RYR1-RD inspires us every day."***

Drs. Dirksen and Dowling are long-standing members of the SAB of the RYR-1 Foundation. Dr. Dirksen is the Lewis Pratt Ross Professor and Chair of Pharmacology and Physiology at the University of Rochester Medical Center.

Dr. Dowling is a clinician-scientist who is focused on gene discovery and therapy for childhood muscle diseases. He is an Associate Professor of Neurology at the University of Toronto, a neurologist at the Hospital for Sick Children in Toronto, and the inaugural Mogford Campbell Chair in Pediatric Clinical Neuroscience at Toronto's Hospital for Sick Children. For more information on Drs. Dirksen and Dowling, please visit: www.ryr1.org/sab.

Since 2015, the RYR-1 Foundation has awarded 14 research grants to outstanding scientists in the United States, Australia, Canada, Hungary, and Italy, representing a commitment of over \$1.3 million. Updates on their currently funded research were presented by grant recipients at a virtual Scientific Advisory Board meeting on July 31, 2020. For more information on current and prior research grants, please go to: www.ryr1.org/grants.