RYR-1 FOUNDATION

www.ryr1.org

If you have a patient with an RYR-1-related disease, The RYR-1 Foundation is here for you.

The mission of The RYR-1 Foundation is to: 1) Support research leading to an effective treatment or a cure for RYR-1-related diseases, 2) Raise awareness about RYR-1-related diseases within the medical community, and 3) Provide education, outreach, and advocacy for individuals and families affected by RYR-1-related diseases. RYR-1-related diseases are inherited forms of myopathy resulting in a wide range of symptoms, including muscle weakness, a potentially fatal reaction to general anesthesia (malignant hyperthermia), and rhabdomyolysis. Mutations or changes in the *RYR1* gene are the most common cause of congenital muscle myopathy. The RyR1 receptor is a channel in muscle cells that regulate the flow of calcium, a critical component of muscle contraction. Therefore, a reduced number and/or abnormal RyR1 channels lead to dysfunctional muscle contraction. There is a wide range of symptoms of RYR-1-related diseases, but they are typically either non-progressive or very slowly progressive.

In September 2020, The RYR-1 Foundation published the ***Clinical Care Guidelines: What Patients & Families Need to Know About RYR-1-Related Diseases***. This document, freely available to the public at no cost. To learn more and to download a copy of the CCG, please visit: **www.ryr1.org/ccg**. On this website, there is also a brief video that introduces the CCG and explains some of its unique features.

For a list of free online RYR-1-related medical literature, please visit: **www.ryr1.org/literature**.

If you would like assistance with being put in contact with expert medical professionals, please email Lindsay Goldberg at **lindsay@ryr1.org**.

PO Box 13312, Pittsburgh, PA 15243
E: lindsay@ryr1.org W: www.ryr1.org