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RYR-1 FOUNDATION
STRENGTH IN NUMBERS

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:

- Support research leading to an effective treatment or a cure for RYR-1-RD (RYR-1-RD)
- Raise awareness about RYR-1-RD within the medical community
- Provide education, outreach, and advocacy for individuals and families affected by RYR-1-RD

RYR-1-RD are a heterogeneous group of inherited neuromuscular conditions caused by pathogenic variants in the *RYR1* gene, the most common genetic cause of congenital myopathies. The *RYR1* gene encodes the skeletal muscle ryanodine receptor (RyR1), a calcium-release channel essential for excitation-contraction coupling. Disruption of RyR1 expression or function can impair intracellular calcium regulation and skeletal muscle contraction, resulting in a broad spectrum of clinical manifestations, including myopathy, malignant hyperthermia susceptibility, and exertional rhabdomyolysis. Disease severity and progression vary considerably, with many individuals experiencing a stable or slowly progressive clinical course.

Available Resources

The RYR-1 Foundation has published Clinical Care Guidelines: What Patients & Families Need to Know About RYR-1-Related Diseases (CCG), available free of charge to the public in nine languages. To learn more or download a copy, visit: **www.ryr1.org/ccg**.

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

If you would like assistance connecting with expert medical professionals, please contact info@ryr1.org.