RYR-1-related diseases are classified as an orphan disease. The prevalence is approximately 1/90,000 in the United States. The disorder most likely goes misdiagnosed or undiagnosed making it difficult to determine the true frequency in the general population. There are also reports of slightly increased prevalence in certain ethnic and geographic populations. The most definitive diagnostic test for RYR-1-related diseases is genetic testing. A genetic test is often ordered due to clinical suspicion related to clinical signs and symptoms, family history, muscle biopsy, and muscle MRI. Muscle biopsy evaluates for changes in the muscle cell that may be associated with mutations in the RYR-1 gene (e.g. CCD, MMD, CNM, CFTD). Muscle MRI allows the physician to evaluate for muscle damage throughout the body, with varying patterns being associated with various forms of muscular dystrophies and myopathies, including subtypes of RYR-1 myopathy.