

Disease

Myotonia congenita^{1, 2}

Gene

CLCN1

Inheritance	AR and AD
Current Treatments	Mexiletine. Lamotrigine. Phenytoin and carbamazepine have been reported to have beneficial effects. Quinine, dantrolene, or acetazolamide may be beneficial in some cases. Avoid certain pharmacologic agents.
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	electromyography test ²
Disease Frequency (per 100,000)	1.225 ²
Genes Included in Disease Frequency	CLCN1 and SCN4A
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Myotonia+Congenita&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1355/#myotonia-c.Summary>

² <https://pubmed.ncbi.nlm.nih.gov/32270509/>