Rx-Genes
Treatment Guidance at the Point of Care

Disease

Congenital myasthenic syndrome-16[1, 2]

Gene

SCN4A

Inheritance	AR
Current Treatments	acetylcholineesterase (AChE) inhibitors
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	repetitive nerve stimulation test[3]
Disease Frequency (per 100,000)	0.92[4]
Genes Included in Disease Frequency	AGRN, ALG14, SYT2, PREPL, GFPT1, SLC5A7, CHRNA1, CHRND, COLQ, DOK7, ALG2, MUSK, CHAT, SLC18A3, COL13A1, LRP4, RAPSN, DPAGT1, VAMP1, MYO9A, CHRNE, CHRNB1, SCN4A, SNAP25, SLC25A25
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Congenital+Myasthenia&term=&cntry=&state=&city=&dist=

References

[1] https://www.ncbi.nlm.nih.gov/books/NBK1168/#cms.Summary

[2] https://www.ncbi.nlm.nih.gov/pubmed/30808424

[3] https://pubmed.ncbi.nlm.nih.gov/30808424/

[4] https://www.ncbi.nlm.nih.gov/pubmed/24500997