

Disease

Congenital myasthenic syndrome-2^{1, 2}

Gene

CHRNA1

Inheritance	AR and AD
Current Treatments	Fluoxetine. Acetylcholine-esterase inhibitors ineffective
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, CHRNA1, SCN4A, SNAP25, SLC25A22
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>