

Disease

Congenital myasthenic syndrome-3^{1, 2}

Gene

CHRND

Inheritance	AR and AD
Current Treatments	3,4-diaminopyridine, Acetylcholine-esterase 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, SLC25A9
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>