

Disease

Congenital myasthenic syndrome-4^{1, 2}

Gene

CHRNE

Inheritance	AR and AD
Current Treatments	Acetylcholine-esterase inhibitors (note: for some patients pyridostigmine and 3,4-diaminopyridine may be ineffective or may worsen the phenotype), albuterol, salbutamol, fluoxetine and drug combinations
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, SLC25A1
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>