

Disease

Congenital myasthenic syndrome-18^{1, 2}

Gene

SNAP25

Inheritance	AD
Current Treatments	3,4-diaminopyridine. Acetylcholineesterase (AChE) 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, CHRNB1, SCN4A, SNAP25, SLC25A23
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>