Rx-Genes
Treatment Guidance at the Point of Care

**Disease**

# Congenital myasthenic syndrome-14[1, 2]

**Gene**

## ALG2

| | |
|---|---|
| **Inheritance** | AR |
| **Current Treatments** | 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, SLC25A12 |
| **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