

Disease

GNE myopathy^{1, 2, 3}

Gene

GNE

Inheritance	AR
Current Treatments	N-acetylmannosamine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma n-acetylneuraminic acid (Neu5Ac) concentration by tandem mass spectrometry ³
Disease Frequency (per 100,000)	0.1 ²
Genes Included in Disease Frequency	GNE
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=gne&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/605820>
- ² <https://pubmed.ncbi.nlm.nih.gov/29720219/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34257421/>