Rx-Genes
Treatment Guidance at the Point of Care

Disease

Niemann-Pick disease, type C, NPC2[1, 2]

Gene

NPC2

Inheritance	AR
Current Treatments	Miglustat and Miplyffa (arimoclomol)
Evidence for Treatment	guideline[3]
Non-Molecular Testing Availability	yes
Non-Molecular Test	oxysterol analysis[3]
Disease Frequency (per 100,000)	1[4]
Genes Included in Disease Frequency	NPC1, NPC2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Niemann-Pick+Diseases&term=&cntry=&state=&city=&dist=

References

[1] https://www.ncbi.nlm.nih.gov/pubmed/30732631

[2] https://www.ncbi.nlm.nih.gov/books/NBK1296/

[3] https://www.ncbi.nlm.nih.gov/pubmed/29625568

[4] https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0785-7#Sec25