

Disease

Niemann-Pick disease, type C, NPC1^{1, 2}

Gene

NPC1

Inheritance	AR
Current Treatments	Miglustat, Miplyffa (arimoclomol) and Aqneursa (levacetylleucine)
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>