

Disease

Niemann-Pick disease, type A and type B^{1, 2, 3}

Gene

SMPD1

Inheritance	AR
Current Treatments	recombinant human acid sphingomyelinase enzyme replacement therapy - Xenpozyme - olipudase alfa, miglustat
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	leukocyte acid sphingomyelinase enzyme activity ¹
Disease Frequency (per 100,000)	0.91 ⁴
Genes Included in Disease Frequency	SMPD1
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/books/NBK1370/>
- ² <https://pubmed.ncbi.nlm.nih.gov/33875845/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/35471153/>
- ⁴ <https://www.mdpi.com/2409-515X/4/3/24/htm>