

Disease

Fucosidosis^{1, 2}

Gene

FUCA1

Inheritance	AR
Current Treatments	Bone marrow transplantation Hematopoietic Stem Cell Transplantation (HSCT)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	fucosidase activity in serum or plasma ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Fucosidosis&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/230000>

² <https://pubmed.ncbi.nlm.nih.gov/33266441/>