

Disease

Mucopolysaccharidosis type VII^{1, 2}

Gene

GUSB

Inheritance	AR
Current Treatments	Vestronidase alfa enzyme replacement, Bone marrow transplantation Hematopoietic Stem Cell Transplantation (HSCT)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	leukocyte beta-glucuronidase enzyme activity ³
Disease Frequency (per 100,000)	0.13 ¹
Genes Included in Disease Frequency	GUSB
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Mucopolysaccharidosis+VII&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/31661765>
- ² <https://www.sciencedirect.com/science/article/pii/S1096719219307851>
- ³ <https://pubmed.ncbi.nlm.nih.gov/28962937/>