Rx-Genes
Treatment Guidance at the Point of Care

Disease

Mucopolysaccharidosis I[1]

Gene

IDUA

Inheritance	AR
Current Treatments	laronidase enzyme replacement, Bone marrow transplantation Hematopoietic Stem Cell Transplantation (HSCT)
Evidence for Treatment	guideline[2]
Non-Molecular Testing Availability	yes
Non-Molecular Test	Alpha-L-iduronidase enzyme activity in leukocytes[1]
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC006
Disease Frequency (per 100,000)	0.55[3]
Genes Included in Disease Frequency	IDUA
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Mucopolysaccharidosis+I&term=&cntry=&state=&city=&dist=

References

[1] https://www.ncbi.nlm.nih.gov/books/NBK1162/

[2] https://pubmed.ncbi.nlm.nih.gov/30143438/

[3] https://www.mdpi.com/2409-515X/4/3/24/htm