

Disease

Mucopolysaccharidosis II¹

Gene

IDS

Inheritance	XL
Current Treatments	idursulfase Elaprase enzyme replacement, Bone marrow transplantation Hematopoietic Stem Cell Transplantation (HSCT)
Evidence for Treatment	guideline ^{2, 3}
Non-Molecular Testing Availability	yes
Non-Molecular Test	iduronate 2-sulfatase (I2S) enzyme activity in white cells, fibroblasts, or plasma ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1001
Disease Frequency (per 100,000)	0.795 ⁴
Genes Included in Disease Frequency	IDS
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Mucopolysaccharidosis+II&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1274/#hunter.Summary>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/30143438>
- ³ <https://www.nature.com/articles/s41436-020-0909-z>
- ⁴ https://www.ncbi.nlm.nih.gov/books/NBK1274/#hunter.Genetically_Related_Allelic_Disor