

Disease

Mucopolysaccharidosis IVA¹

Gene

GALNS

Inheritance	AR
Current Treatments	elosulfase alfa enzyme replacement
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	leukocyte N-acetylgalactosamine 6-sulfatase enzyme activity ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC1010
Disease Frequency (per 100,000)	0.335 ^{3, 4}
Genes Included in Disease Frequency	GALNS
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Mucopolysaccharidosis+IV&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK148668/>
- ² <https://pubmed.ncbi.nlm.nih.gov/30143438/>
- ³ <https://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-iv#genes>
- ⁴ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4251694/>