

## Disease

# Mucopolysaccharidosis I<sup>1</sup>

## Gene

### IDUA

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

## References

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/books/NBK1162/>
- <sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/30143438/>
- <sup>3</sup> <https://www.mdpi.com/2409-515X/4/3/24/htm>