

Disease

Metachromatic leukodystrophy^{1, 2}

Gene

ARSA

Inheritance	AR
Current Treatments	Hematopoietic stem cell transplantation (HSCT) - bone marrow transplant, Skysona (elivaldogene autotemcel (Lenti-D), atidarsagene autotemcel (Libmeldy)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	arylsulfatase A enzyme activity in leukocytes, urinary sulfatides ¹
Disease Frequency (per 100,000)	1.565 ³
Genes Included in Disease Frequency	ARSA
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Metachromatic+Leukodystrophy&term=&cntry=&state=&city=&

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1130/>
- ² <https://pubmed.ncbi.nlm.nih.gov/28976817/>
- ³ <https://www.ncbi.nlm.nih.gov/books/NBK1130/#mld.Summary>