

Disease

Molybdenum cofactor deficiency A^{1, 2, 3}

Gene

MOCS1

Inheritance	AR
Current Treatments	cyclic pyranopterin monophosphate (fosdenopterin)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	urinary xanthine, uric acid, S-sulfocysteine, sulfite, thiosulfate and plasma uric acid ⁴
Disease Frequency (per 100,000)	0.495 ^{5, 6}
Genes Included in Disease Frequency	MOCS1, MOCS2, MOCS3 and GPHN cause molybdenum cofactor deficiency (2/3 of molybdenum cofactor deficiency cases are due to MOSC1. There is no treatment for molybdenum cofactor deficiency due to the other 3 genes)
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Molybdenum&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/20385644>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/26343839>
- ³ <https://omim.org/entry/252150>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/20385644/>
- ⁵ <https://ghr.nlm.nih.gov/condition/molybdenum-cofactor-deficiency#genes>
- ⁶ <https://www.ncbi.nlm.nih.gov/pubmed/31201073>