Lars Schlotawa

List of Publications by Year
in descending order

Version: 2024-02-01

15 papers	476 citations	932766 10 h-index	996533 15 g-index
15 all docs	15 docs citations	15 times ranked	597 citing authors

#	Article	IF	Citations
1	Molecular basis of multiple sulfatase deficiency, mucolipidosis II/III and Niemannâ€“Pick C1 disease â€“ Lysosomal storage disorders caused by defects of non-lysosomal proteins. Biochimica Et Biophysica Acta - Molecular Cell Research, 2009, 1793, 710-725.	1.9	86
2	A152T tau allele causes neurodegeneration that can be ameliorated in a zebrafish model by autophagy induction. Brain, 2017, 140, 1128-1146.	3.7	84
3	Molecular analysis ofSUMF1 mutations: stability and residual activity of mutant formylglycine-generating enzyme determine disease severity in multiple sulfatase deficiency. Human Mutation, 2008, 29, 205-205.	1.1	74
4	SUMF1 mutations affecting stability and activity of formylglycine generating enzyme predict clinical outcome in multiple sulfatase deficiency. European Journal of Human Genetics, 2011, 19, 253-261.	1.4	63
5	Multiple Sulfatase Deficiency: A Disease Comprising Mucopolysaccharidosis, Sphingolipidosis, and More Caused by a Defect in Posttranslational Modification. International Journal of Molecular Sciences, 2020, 21, 3448.	1.8	32
6	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	0.5	31
7	Natural history of multiple sulfatase deficiency: Retrospective phenotyping and functional variant analysis to characterize an ultraâ€rare disease. Journal of Inherited Metabolic Disease, 2020, 43, 1298-1309.	1.7	23
8	Rapid degradation of an active formylglycine generating enzyme variant leads to a late infantile severe form of multiple sulfatase deficiency. European Journal of Human Genetics, 2013, 21, 1020-1023.	1.4	19
9	Cerebellar ataxia, mental retardation and dysequilibrium syndrome 1 (CAMRQ1) caused by an unusual constellation of VLDLR mutation. Journal of Neurology, 2013, 260, 1678-1680.	1.8	15
10	A systematic review and metaâ€analysis of published cases reveals the natural disease history in multiple sulfatase deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 1288-1297.	1.7	14
11	Recognition and ER Quality Control of Misfolded Formylglycine-Generating Enzyme by Protein Disulfide Isomerase. Cell Reports, 2018, 24, 27-37.e4.	2.9	12
12	Expanding the genetic cause of multiple sulfatase deficiency: A novel SUMF1 variant in a patient displaying a severe late infantile form of the disease. Molecular Genetics and Metabolism, 2017, 121, 252-258.	0.5	11
13	Severe neonatal multiple sulfatase deficiency presenting with hydrops fetalis in a preterm birth patient. JIMD Reports, 2019, 49, 48-52.	0.7	7
14	A homozygous missense variant of SUMF1 in the Bedouin population extends the clinical spectrum in ultrarare neonatal multiple sulfatase deficiency. Molecular Genetics & Genomic Medicine, 2020, 8, e1167.	0.6	4
15	Unexpected Phenotype Reversion and Survival in a Zebrafish Model of Multiple Sulfatase Deficiency. Frontiers in Cell and Developmental Biology, 2022, 10, .	1.8	1