

Lars Schlotawa

List of Publications by Year in descending order

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