Maria Fuller

List of Publications by Year
in descending order

82 papers	2,575 citations	172457 29 h-index	206112 48 g-index
83 all docs	83 docs citations	83 times ranked	2767 citing authors

#	Article	IF	Citations
1	Identification and Characterization of Ambroxol as an Enzyme Enhancement Agent for Gaucher Disease. Journal of Biological Chemistry, 2009, 284, 23502-23516.	3.4	260
2	Newborn screening for lysosomal storage disorders. Molecular Genetics and Metabolism, 2006, 88, 307-314.	1.1	145
3	Early Neurodegeneration Progresses Independently of Microglial Activation by Heparan Sulfate in the Brain of Mucopolysaccharidosis IIIB Mice. PLoS ONE, 2008, 3, e2296.	2.5	114
4	Newborn Screening for Lysosomal Storage Disorders: Clinical Evaluation of a Two-Tier Strategy. Pediatrics, 2004, 114, 909-916.	2.1	102
5	Characterization of a C57BL/6 congenic mouse strain of mucopolysaccharidosis type IIIA. Brain Research, 2006, 1104, 1-17.	2.2	89
6	Effect of lysosomal storage on bis(monoacylglycero)phosphate. Biochemical Journal, 2008, 411, 71-78.	3.7	80
7	Disease-Specific Markers for the Mucopolysaccharidoses. Pediatric Research, 2004, 56, 733-738.	2.3	76
8	Is it Fabry disease?. Genetics in Medicine, 2016, 18, 1181-1185.	2.4	70
9	Heparan Sulfate Proteoglycans Containing a Glypican 5 Core and 2-O-Sulfo-iduronic Acid Function as Sonic Hedgehog Co-receptors to Promote Proliferation. Journal of Biological Chemistry, 2013, 288, 26275-26288.	3.4	64
10	Examination of intravenous and intraâ€CSF protein delivery for treatment of neurological disease. European Journal of Neuroscience, 2009, 29, 1197-1214.	2.6	61
11	Urinary Lipid Profiling for the Identification of Fabry Hemizygotes and Heterozygotes. Clinical Chemistry, 2005, 51, 688-694.	3.2	56
12	Value of Glucosylsphingosine (Lyso-Gb1) as a Biomarker in Gaucher Disease: A Systematic Literature Review. International Journal of Molecular Sciences, 2020, 21, 7159.	4.1	56
13	Secondary sphingolipid accumulation in a macrophage model of Gaucher disease. Molecular Genetics and Metabolism, 2007, 92, 336-345.	1.1	55
14	Immunoquantification of Î±-Galactosidase: Evaluation for the Diagnosis of Fabry Disease. Clinical Chemistry, 2004, 50, 1979-1985.	3.2	54
15	Lipid composition of microdomains is altered in a cell model of Gaucher disease. Journal of Lipid Research, 2008, 49, 1725-1734.	4.2	54
16	Glycosaminoglycan degradation fragments in mucopolysaccharidosis I. Glycobiology, 2004, 14, 443-450.	2.5	47
17	Prediction of neuropathology in mucopolysaccharidosis I patients. Molecular Genetics and Metabolism, 2005, 84, 18-24.	1.1	46
18	Screening patients referred to a metabolic clinic for lysosomal storage disorders. Journal of Medical Genetics, 2011, 48, 422-425.	3.2	43

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19	Isolation and Characterisation of a Recombinant, Precursor form of Lysosomal Acid alpha-Glucosidase. FEBS Journal, 1995, 234, 903-909.	0.2	42
20	Laronidase Treatment of Mucopolysaccharidosis I. BioDrugs, 2005, 19, 1-7.	4.6	41
21	Sphingolipids: the nexus between Gaucher disease and insulin resistance. Lipids in Health and Disease, 2010, 9, 113.	3.0	41
22	Migalastat HCl Reduces Globotriaosylsphingosine (Lyso-Gb3) in Fabry Transgenic Mice and in the Plasma of Fabry Patients. PLoS ONE, 2013, 8, e57631.	2.5	40
23	Maternal overnutrition by hypercaloric diets programs hypothalamic mitochondrial fusion and metabolic dysfunction in rat male offspring. Nutrition and Metabolism, 2018, 15, 38.	3.0	39
24	Disease and subtype specific signatures enable precise diagnosis of the mucopolysaccharidoses. Genetics in Medicine, 2019, 21, 753-757.	2.4	39
25	Determination of urinary oligosaccharides by high-performance liquid chromatography/electrospray ionizationâ€“tandem mass spectrometry: Application to Hunter syndrome. Analytical Biochemistry, 2010, 402, 113-120.	2.4	38
26	Glucosylceramide accumulation is not confined to the lysosome in fibroblasts from patients with Gaucher disease. Molecular Genetics and Metabolism, 2008, 93, 437-443.	1.1	37
27	Characterization of Sulfated Oligosaccharides in Mucopolysaccharidosis Type IIIA by Electrospray Ionization Mass Spectrometry. Analytical Chemistry, 2006, 78, 4534-4542.	6.5	36
28	Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation. Clinica Chimica Acta, 2018, 481, 1-8.	1.1	32
29	Lysophosphatidylcholine is a Major Component of Platelet Microvesicles Promoting Platelet Activation and Reporting Atherosclerotic Plaque Instability. Thrombosis and Haemostasis, 2019, 119, 1295-1310.	3.4	32
30	Metabolomics to Improve the Diagnostic Efficiency of Inborn Errors of Metabolism. International Journal of Molecular Sciences, 2020, 21, 1195.	4.1	30
31	Validation of a heparan sulfate-derived disaccharide as a marker of accumulation in murine mucopolysaccharidosis type IIIA. Molecular Genetics and Metabolism, 2006, 87, 107-112.	1.1	29
32	Immunochemistry of Lysosomal Storage Disorders. Clinical Chemistry, 2006, 52, 1660-1668.	3.2	28
33	Selective reduction of bis(monoacylglycero)phosphate ameliorates the storage burden in a THP-1 macrophage model of Gaucher disease. Journal of Lipid Research, 2013, 54, 1691-1697.	4.2	27
34	Plasma lipids are altered in Gaucher disease: Biochemical markers to evaluate therapeutic intervention. Blood Cells, Molecules, and Diseases, 2008, 40, 420-427.	1.4	26
35	Expanding the clinical utility of glucosylsphingosine for Gaucher disease. Journal of Inherited Metabolic Disease, 2020, 43, 558-563.	3.6	26
36	Lipid composition of membrane rafts, isolated with and without detergent, from the spleen of a mouse model of Gaucher disease. Biochemical and Biophysical Research Communications, 2013, 442, 62-67.	2.1	25

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37	Rapid, single-phase extraction of glucosylsphingosine from plasma: A universal screening and monitoring tool. Clinica Chimica Acta, 2015, 450, 6-10.	1.1	25
38	Evaluation of Multiple Methods for Quantification of Glycosaminoglycan Biomarkers in Newborn Dried Blood Spots from Patients with Severe and Attenuated Mucopolysaccharidosis-I. International Journal of Neonatal Screening, 2020, 6, 69.	3.2	25
39	A simple method for quantification of plasma globotriaosylsphingosine: Utility for Fabry disease. Molecular Genetics and Metabolism, 2017, 122, 121-125.	1.1	24
40	Risk of Death in Heart Disease is Associated With Elevated Urinary Globotriaosylceramide. Journal of the American Heart Association, 2014, 3, e000394.	3.7	22
41	A defect in exodegradative pathways provides insight into endodegradation of heparan and dermatan sulfates. Glycobiology, 2006, 16, 318-325.	2.5	21
42	Liquid chromatography/electrospray ionisationâ€“tandem mass spectrometry quantification of GM2 gangliosides in human peripheral cells and plasma. Analytical Biochemistry, 2014, 458, 20-26.	2.4	21
43	Increased monohexosylceramide levels in the serum of established rheumatoid arthritis patients. Rheumatology, 2020, 59, 2085-2089.	1.9	21
44	A multi-centre, open label, randomised, parallel-group, superiority Trial to compare the efficacy of URsodeoxycholic acid with RIFampicin in the management of women with severe early onset Intrahepatic Cholestasis of pregnancy: the TURRIFIC randomised trial. BMC Pregnancy and Childbirth, 2021, 21, 51.	2.4	21
45	The brain lipidome in neurodegenerative lysosomal storage disorders. Biochemical and Biophysical Research Communications, 2018, 504, 623-628.	2.1	20
46	Absence of Î±-galactosidase cross-correction in Fabry heterozygote cultured skin fibroblasts. Molecular Genetics and Metabolism, 2015, 114, 268-273.	1.1	19
47	Subregional brain distribution of simple and complex glycosphingolipids in the mucopolysaccharidosis type I (Hurler syndrome) mouse: impact of diet. Journal of Neurochemistry, 2017, 141, 287-295.	3.9	17
48	Glycosaminoglycan fragments as a measure of disease burden in the mucopolysaccharidosis type I mouse. Molecular Genetics and Metabolism, 2018, 123, 112-117.	1.1	17
49	Reduced cerebral vascularization in experimental neuronopathic Gaucher disease. Journal of Pathology, 2018, 244, 120-128.	4.5	17
50	Stearoyl-CoA Desaturase 1 Is a Key Determinant of Membrane Lipid Composition in 3T3-L1 Adipocytes. PLoS ONE, 2016, 11, e0162047.	2.5	17
51	Quantification of plasma sulfatides by mass spectrometry: Utility for metachromatic leukodystrophy. Analytica Chimica Acta, 2017, 955, 79-85.	5.4	15
52	Evaluation of biomarkers for Sanfilippo syndrome. Molecular Genetics and Metabolism, 2019, 128, 68-74.	1.1	13
53	Mass spectrometric quantification of glycogen to assess primary substrate accumulation in the Pompe mouse. Analytical Biochemistry, 2012, 421, 759-763.	2.4	12
54	Prevalence of lysosomal storage disorders in Australia from 2009 to 2020. The Lancet Regional Health - Western Pacific, 2022, 19, 100344.	2.9	12

#	Article	IF	Citations
55	Evaluation of Two Methods for Quantification of Glycosaminoglycan Biomarkers in Newborn Dried Blood Spots from Patients with Severe and Attenuated Mucopolysaccharidosis Type II. International Journal of Neonatal Screening, 2022, 8, 9.	3.2	12
56	Sphingolipid dyshomeostasis in the brain of the mouse model of mucopolysaccharidosis type IIIA. Molecular Genetics and Metabolism, 2020, 129, 111-116.	1.1	10
57	The long and the short of Huntingtonâ€™s disease: how the sphingolipid profile is shifted in the caudate of advanced clinical cases. Brain Communications, 2022, 4, fcab303.	3.3	10
58	Immunoquantification of Î²-Glucosidase: Diagnosis and Prediction of Severity in Gaucher Disease. Clinical Chemistry, 2005, 51, 2200-2202.	3.2	9
59	The Ckd. Qld fabRy Epidemiology (aCQuiRE) study protocol: identifying the prevalence of Fabry disease amongst patients with kidney disease in Queensland, Australia. BMC Nephrology, 2020, 21, 58.	1.8	9
60	Minimum substrate requirements of endoglycosidase activities toward dermatan sulfate by electrospray ionization-tandem mass spectrometry. Glycobiology, 2008, 18, 1119-1128.	2.5	8
61	Selective normalisation of regional brain bis(monoacylglycero)phosphate in the mucopolysaccharidosis 1 (Hurler) mouse. Experimental Neurology, 2016, 277, 68-75.	4.1	8
62	Systemic scAAV9.U1a.hSGSH Delivery Corrects Brain Biochemistry in Mucopolysaccharidosis Type IIIA at Early and Later Stages of Disease. Human Gene Therapy, 2021, 32, 420-430.	2.7	8
63	Laboratory Diagnosis of Lysosomal Diseases: Newborn Screening to Treatment. , 2020, 41, 53-66.		8
64	The prevalence of Fabry disease in a statewide chronic kidney disease cohort â€“ Outcomes of the aCQuiRE (Ckd.Qld fabRy Epidemiology) study. BMC Nephrology, 2022, 23, 169.	1.8	7
65	Distribution of Heparan Sulfate Oligosaccharides in Murine Mucopolysaccharidosis Type IIIA. Metabolites, 2014, 4, 1088-1100.	2.9	6
66	Drug induced exocytosis of glycogen in Pompe disease. Biochemical and Biophysical Research Communications, 2016, 479, 721-727.	2.1	6
67	Collection of cerebrospinal fluid from murine lateral ventricles for biomarker determination in mucopolysaccharidosis type IIIA. Journal of Neuroscience Methods, 2019, 324, 108314.	2.5	6
68	Globoid cell leukodystrophy (Krabbe disease) in a Merino sheep. Journal of Veterinary Diagnostic Investigation, 2019, 31, 118-121.	1.1	6
69	Chondroitin sulfate disaccharide is a specific and sensitive biomarker for mucopolysaccharidosis type <scp>IVA</scp>. JIMD Reports, 2020, 55, 68-74.	1.5	6
70	Glycogen Exocytosis from Cultured Pompe Skin Fibroblasts. Translational Biomedicine, 2015, 6, .	0.1	5
71	Fabry cardiomyopathy: missing links from genotype to phenotype. Heart, 2020, 106, 553-554.	2.9	5
72	Challenges in Diagnosing Intermediate Maple Syrup Urine Disease by Newborn Screening and Functional Validation of Genomic Results Imperative for Reproductive Family Planning. International Journal of Neonatal Screening, 2021, 7, 25.	3.2	5

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73	Experience with the Urinary Tetrasaccharide Metabolite for Pompe Disease in the Diagnostic Laboratory. Metabolites, 2021, 11, 446.	2.9	5
74	Autologous, lentivirusâ€modified, Tâ€rapa cell â€œmicropharmaciesâ€for lysosomal storage disorders. EMBO Molecular Medicine, 2022, 14, e14297.	6.9	5
75	The BACH project protocol: an international multicentre total Bile Acid Comparison and Harmonisation project and sub-study of the TURRIFIC randomised trial. Clinical Chemistry and Laboratory Medicine, 2021, 59, 1921-1929.	2.3	4
76	Lysosomal Degradation of Heparin and Heparan Sulfate. , 2005, , 285-311.		3
77	Aberrant splicing and transcriptional activity of TPP1 result in CLN2-like disorder. European Journal of Medical Genetics, 2021, 64, 104259.	1.3	2
78	Functional assessment of the genetic findings indicating mucopolysaccharidosis type <scp>II</scp> in the prenatal setting. JIMD Reports, 2021, 60, 10-14.	1.5	1
79	Mono-symptomatic Fabry disease in a population with mild-to-moderate left ventricular hypertrophy. Molecular Genetics and Metabolism Reports, 2020, 25, 100697.	1.1	1
80	Impaired neural differentiation of MPS IIIA patient induced pluripotent stem cell-derived neural progenitor cells. Molecular Genetics and Metabolism Reports, 2021, 29, 100811.	1.1	1
81	Gaucherâ€™s disease in the lipidomics era. Clinical Lipidology, 2012, 7, 431-441.	0.4	0
82	014â€...The diagnostic odyssey for fabry disease: ten years experience in testing. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, A7.1-A7.	1.9	0