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	Prevalence of lysosomal storage disorders in Australia from 2009 to 2020. The Lancet Regional Health - Western Pacific, 2022, 19, 100344.	2.9	12
2	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
3	Autologous, lentivirusâ€modified, Tâ€rapa cell â€œmicropharmaciesâ€•for lysosomal storage disorders. EMBO Molecular Medicine, 2022, 14, e14297.	6.9	5
4	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
5	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
6	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
7	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
8	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
9	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
10	Aberrant splicing and transcriptional activity of TPP1 result in CLN2-like disorder. European Journal of Medical Genetics, 2021, 64, 104259.	1.3	2
11	Experience with the Urinary Tetrasaccharide Metabolite for Pompe Disease in the Diagnostic Laboratory. Metabolites, 2021, 11, 446.	2.9	5
12	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
13	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
14	Sphingolipid dyshomeostasis in the brain of the mouse model of mucopolysaccharidosis type IIIA. Molecular Genetics and Metabolism, 2020, 129, 111-116.	1.1	10
15	Expanding the clinical utility of glucosylsphingosine for Gaucher disease. Journal of Inherited Metabolic Disease, 2020, 43, 558-563.	3.6	26
16	Increased monohexosylceramide levels in the serum of established rheumatoid arthritis patients. Rheumatology, 2020, 59, 2085-2089.	1.9	21
17	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
18	Chondroitin sulfate disaccharide is a specific and sensitive biomarker for mucopolysaccharidosis type <scp>IVA</scp>. JIMD Reports, 2020, 55, 68-74.	1.5	6

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19	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
20	Fabry cardiomyopathy: missing links from genotype to phenotype. Heart, 2020, 106, 553-554.	2.9	5
21	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
22	Metabolomics to Improve the Diagnostic Efficiency of Inborn Errors of Metabolism. International Journal of Molecular Sciences, 2020, 21, 1195.	4.1	30
23	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
24	Laboratory Diagnosis of Lysosomal Diseases: Newborn Screening to Treatment. , 2020, 41, 53-66.		8
25	Disease and subtype specific signatures enable precise diagnosis of the mucopolysaccharidoses. Genetics in Medicine, 2019, 21, 753-757.	2.4	39
26	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
27	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
28	Evaluation of biomarkers for Sanfilippo syndrome. Molecular Genetics and Metabolism, 2019, 128, 68-74.	1.1	13
29	Globoid cell leukodystrophy (Krabbe disease) in a Merino sheep. Journal of Veterinary Diagnostic Investigation, 2019, 31, 118-121.	1.1	6
30	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
31	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
32	The brain lipidome in neurodegenerative lysosomal storage disorders. Biochemical and Biophysical Research Communications, 2018, 504, 623-628.	2.1	20
33	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
34	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
35	Reduced cerebral vascularization in experimental neuronopathic Gaucher disease. Journal of Pathology, 2018, 244, 120-128.	4.5	17
36	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

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37	Quantification of plasma sulfatides by mass spectrometry: Utility for metachromatic leukodystrophy. Analytica Chimica Acta, 2017, 955, 79-85.	5.4	15
38	A simple method for quantification of plasma globotriaosylsphingosine: Utility for Fabry disease. Molecular Genetics and Metabolism, 2017, 122, 121-125.	1.1	24
39	Is it Fabry disease?. Genetics in Medicine, 2016, 18, 1181-1185.	2.4	70
40	Drug induced exocytosis of glycogen in Pompe disease. Biochemical and Biophysical Research Communications, 2016, 479, 721-727.	2.1	6
41	Selective normalisation of regional brain bis(monoacylglycero)phosphate in the mucopolysaccharidosis 1 (Hurler) mouse. Experimental Neurology, 2016, 277, 68-75.	4.1	8
42	Stearoyl-CoA Desaturase 1 Is a Key Determinant of Membrane Lipid Composition in 3T3-L1 Adipocytes. PLoS ONE, 2016, 11, e0162047.	2.5	17
43	Glycogen Exocytosis from Cultured Pompe Skin Fibroblasts. Translational Biomedicine, 2015, 6, .	0.1	5
44	Absence of Î±-galactosidase cross-correction in Fabry heterozygote cultured skin fibroblasts. Molecular Genetics and Metabolism, 2015, 114, 268-273.	1.1	19
45	Rapid, single-phase extraction of glucosylsphingosine from plasma: A universal screening and monitoring tool. Clinica Chimica Acta, 2015, 450, 6-10.	1.1	25
46	Distribution of Heparan Sulfate Oligosaccharides in Murine Mucopolysaccharidosis Type IIIA. Metabolites, 2014, 4, 1088-1100.	2.9	6
47	Risk of Death in Heart Disease is Associated With Elevated Urinary Globotriaosylceramide. Journal of the American Heart Association, 2014, 3, e000394.	3.7	22
48	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
49	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
50	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
51	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
52	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
53	Gaucherâ€™s disease in the lipidomics era. Clinical Lipidology, 2012, 7, 431-441.	0.4	0
54	Mass spectrometric quantification of glycogen to assess primary substrate accumulation in the Pompe mouse. Analytical Biochemistry, 2012, 421, 759-763.	2.4	12

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55	Screening patients referred to a metabolic clinic for lysosomal storage disorders. Journal of Medical Genetics, 2011, 48, 422-425.	3.2	43
56	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
57	Sphingolipids: the nexus between Gaucher disease and insulin resistance. Lipids in Health and Disease, 2010, 9, 113.	3.0	41
58	Examination of intravenous and intraâ€CSF protein delivery for treatment of neurological disease. European Journal of Neuroscience, 2009, 29, 1197-1214.	2.6	61
59	Identification and Characterization of Ambroxol as an Enzyme Enhancement Agent for Gaucher Disease. Journal of Biological Chemistry, 2009, 284, 23502-23516.	3.4	260
60	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
61	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
62	Minimum substrate requirements of endoglycosidase activities toward dermatan sulfate by electrospray ionization-tandem mass spectrometry. Glycobiology, 2008, 18, 1119-1128.	2.5	8
63	Lipid composition of microdomains is altered in a cell model of Gaucher disease. Journal of Lipid Research, 2008, 49, 1725-1734.	4.2	54
64	Effect of lysosomal storage on bis(monoacylglycero)phosphate. Biochemical Journal, 2008, 411, 71-78.	3.7	80
65	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
66	Secondary sphingolipid accumulation in a macrophage model of Gaucher disease. Molecular Genetics and Metabolism, 2007, 92, 336-345.	1.1	55
67	Characterization of Sulfated Oligosaccharides in Mucopolysaccharidosis Type IIIA by Electrospray Ionization Mass Spectrometry. Analytical Chemistry, 2006, 78, 4534-4542.	6.5	36
68	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
69	Newborn screening for lysosomal storage disorders. Molecular Genetics and Metabolism, 2006, 88, 307-314.	1.1	145
70	Characterization of a C57BL/6 congenic mouse strain of mucopolysaccharidosis type IIIA. Brain Research, 2006, 1104, 1-17.	2.2	89
71	A defect in exodegradative pathways provides insight into endodegradation of heparan and dermatan sulfates. Glycobiology, 2006, 16, 318-325.	2.5	21
72	Immunochemistry of Lysosomal Storage Disorders. Clinical Chemistry, 2006, 52, 1660-1668.	3.2	28

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73	Immunoquantification of Î²-Glucosidase: Diagnosis and Prediction of Severity in Gaucher Disease. Clinical Chemistry, 2005, 51, 2200-2202.	3.2	9
74	Urinary Lipid Profiling for the Identification of Fabry Hemizygotes and Heterozygotes. Clinical Chemistry, 2005, 51, 688-694.	3.2	56
75	Prediction of neuropathology in mucopolysaccharidosis I patients. Molecular Genetics and Metabolism, 2005, 84, 18-24.	1.1	46
76	Lysosomal Degradation of Heparin and Heparan Sulfate. , 2005, , 285-311.		3
77	Laronidase Treatment of Mucopolysaccharidosis I. BioDrugs, 2005, 19, 1-7.	4.6	41
78	Disease-Specific Markers for the Mucopolysaccharidoses. Pediatric Research, 2004, 56, 733-738.	2.3	76
79	Glycosaminoglycan degradation fragments in mucopolysaccharidosis I. Glycobiology, 2004, 14, 443-450.	2.5	47
80	Immunoquantification of Î±-Galactosidase: Evaluation for the Diagnosis of Fabry Disease. Clinical Chemistry, 2004, 50, 1979-1985.	3.2	54
81	Newborn Screening for Lysosomal Storage Disorders: Clinical Evaluation of a Two-Tier Strategy. Pediatrics, 2004, 114, 909-916.	2.1	102
82	Isolation and Characterisation of a Recombinant, Precursor form of Lysosomal Acid alpha-Glucosidase. FEBS Journal, 1995, 234, 903-909.	0.2	42