Marc C Patterson

List of Publications by Year
in descending order

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Version: 2024-02-01

131 papers	9,569 citations	53794 45 h-index	40979 93 g-index
140 all docs	140 docs citations	140 times ranked	8567 citing authors

#	Article	IF	Citations
1	Niemann-Pick C1 Disease Gene: Homology to Mediators of Cholesterol Homeostasis. Science, 1997, 277, 228-231.	12.6	1,373
2	Miglustat for treatment of Niemann-Pick C disease: a randomised controlled study. Lancet Neurology, The, 2007, 6, 765-772.	10.2	609
3	Pompe disease diagnosis and management guideline. Genetics in Medicine, 2006, 8, 267-288.	2.4	473
4	Recommendations for the diagnosis and management of Niemannâ€“Pick disease type C: An update. Molecular Genetics and Metabolism, 2012, 106, 330-344.	1.1	465
5	The Niemann-Pick C1 Protein Resides in a Vesicular Compartment Linked to Retrograde Transport of Multiple Lysosomal Cargo. Journal of Biological Chemistry, 1999, 274, 9627-9635.	3.4	347
6	Neurological Aspects of Human Glycosylation Disorders. Annual Review of Neuroscience, 2015, 38, 105-125.	10.7	302
7	Clinical, Radiologic, and Prognostic Features of Myelitis Associated With Myelin Oligodendrocyte Glycoprotein Autoantibody. JAMA Neurology, 2019, 76, 301.	9.0	243
8	Recommendations on the diagnosis and management of Niemann-Pick disease type C. Molecular Genetics and Metabolism, 2009, 98, 152-165.	1.1	210
9	Consensus clinical management guidelines for Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2018, 13, 50.	2.7	200
10	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	1.1	185
11	Miglustat in adult and juvenile patients with Niemannâ€“Pick disease type C: Long-term data from a clinical trial. Molecular Genetics and Metabolism, 2010, 99, 351-357.	1.1	184
12	Identification of 58 novel mutations in Niemann-Pick disease type C: Correlation with biochemical phenotype and importance ofPTC1-like domains inNPC1. Human Mutation, 2003, 22, 313-325.	2.5	181
13	Neurology of inherited glycosylation disorders. Lancet Neurology, The, 2012, 11, 453-466.	10.2	179
14	Newborn Screening for Krabbe Disease: the New York State Model. Pediatric Neurology, 2009, 40, 245-252.	2.1	174
15	Disease and patient characteristics in NP-C patients: findings from an international disease registry. Orphanet Journal of Rare Diseases, 2013, 8, 12.	2.7	171
16	A clinical approach to the diagnosis of patients with leukodystrophies and genetic leukoencephelopathies. Molecular Genetics and Metabolism, 2015, 114, 501-515.	1.1	163
17	Dolichol phosphate mannose synthase (DPM1) mutations define congenital disorder of glycosylation Ie (CDG-Ie). Journal of Clinical Investigation, 2000, 105, 191-198.	8.2	150
18	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	3.6	146

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19	Linear clinical progression, independent of age of onset, in Niemannâ€“Pick disease, type C. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 132-140.	1.7	145
20	Long-Term Miglustat Therapy in Children With Niemann-Pick Disease Type C. Journal of Child Neurology, 2010, 25, 300-305.	1.4	141
21	Niemann-Pick C Variant Detection by Altered Sphingolipid Trafficking and Correlation with Mutations within a Specific Domain of NPC1. American Journal of Human Genetics, 2001, 68, 1361-1372.	6.2	140
22	Broad screening test for sphingolipid-storage diseases. Lancet, The, 1999, 354, 901-905.	13.7	131
23	The pathophysiology and mechanisms of NP-C disease. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2004, 1685, 83-87.	2.4	131
24	Miglustat in Niemann-Pick disease type C patients: a review. Orphanet Journal of Rare Diseases, 2018, 13, 140.	2.7	125
25	A 19-Month-Old Girl of South Indian Parents Presented to a General Pediatric Clinic for Evaluation of Global Developmental Regression. Seminars in Pediatric Neurology, 2014, 21, 88-89.	2.0	122
26	Cognitive Impairment Occurs in Children and Adolescents With Multiple Sclerosis. Journal of Child Neurology, 2013, 28, 102-107.	1.4	121
27	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	1.6	119
28	Mosaicism of the UDP-Galactose Transporter SLC35A2 Causes a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2013, 92, 632-636.	6.2	114
29	Prospective study of neurological responses to treatment with macrophage-targeted glucocerebrosidase in patients with type 3 Gaucher's disease. Annals of Neurology, 1997, 42, 613-621.	5.3	109
30	Clinical features of neuromyelitis optica in children. Neurology, 2016, 86, 245-252.	1.1	100
31	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	3.6	91
32	Severe hypoglycemia as a presenting symptom of carbohydrate-deficient glycoprotein syndrome. Journal of Pediatrics, 1999, 135, 775-781.	1.8	89
33	Spinocerebellar ataxia type 2 (SCA 2) in an infant with extreme CAG repeat expansion. American Journal of Medical Genetics Part A, 1998, 79, 383-387.	2.4	88
34	Stable or improved neurological manifestations during miglustat therapy in patients from the international disease registry for Niemann-Pick disease type C: an observational cohort study. Orphanet Journal of Rare Diseases, 2015, 10, 65.	2.7	83
35	A Riddle Wrapped in a Mystery: Understanding Niemann-Pick Disease, Type C. Neurologist, 2003, 9, 301-310.	0.7	75
36	Genetic screening for Niemann-Pick disease type C in adults with neurological and psychiatric symptoms: findings from the ZOOM study. Human Molecular Genetics, 2013, 22, 4349-4356.	2.9	75

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37	Childhood-onset ataxia: Testing for large CAG-repeats in SCA2 and SCA7. American Journal of Medical Genetics Part A, 2002, 110, 338-345.	2.4	73
38	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
39	Dysphagia as a risk factor for mortality in Niemann-Pick disease type C: systematic literature review and evidence from studies with miglustat. Orphanet Journal of Rare Diseases, 2012, 7, 76.	2.7	66
40	Therapy of Niemannâ€“Pick disease, type C. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2004, 1685, 77-82.	2.4	60
41	Limbic encephalitis associated with antiâ€GAD antibody and common variable immune deficiency. Developmental Medicine and Child Neurology, 2009, 51, 563-567.	2.1	59
42	Treatment outcomes following continuous miglustat therapy in patients with Niemann-Pick disease Type C: a final report of the NPC Registry. Orphanet Journal of Rare Diseases, 2020, 15, 104.	2.7	51
43	Niemann-Pick type C Suspicion Index tool: analyses by age and association of manifestations. Journal of Inherited Metabolic Disease, 2014, 37, 93-101.	3.6	48
44	Quo vadis: the reâ€‹definition of â€œinborn metabolic diseasesâ€•. Journal of Inherited Metabolic Disease, 2015, 38, 1003-1006.	3.6	48
45	Scurvy and Rickets Masked by Chronic Neurologic Illness: Revisiting "Psychologic Malnutrition". Pediatrics, 2007, 119, e783-e790.	2.1	46
46	Proton Magnetic Resonance Spectroscopy as a Probe into the Pathophysiology of <scp>A</scp>utism <scp>S</scp>pectrum <scp>D</scp>isorders (<scp>ASD</scp>): A Review. Autism Research, 2013, 6, 119-133.	3.8	46
47	Disease specific therapies in leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 527-536.	1.1	45
48	Longâ€‹term survival outcomes of patients with <scp>Niemannâ€‹Pick</scp> disease type C receiving miglustat treatment: A large retrospective observational study. Journal of Inherited Metabolic Disease, 2020, 43, 1060-1069.	3.6	43
49	Current directions in tau research: Highlights from Tau 2020. Alzheimer's and Dementia, 2022, 18, 988-1007.	0.8	42
50	Jeavons Syndrome: Clinical Features and Response to Treatment. Pediatric Neurology, 2018, 86, 46-51.	2.1	40
51	Angelman's syndrome: clinical and electroencephalographic findings. Electroencephalography and Clinical Neurophysiology, 1997, 102, 299-302.	0.3	39
52	SLC35A2â€‹CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
53	Mitochondrial myopathy and complex III deficiency in a patient with a new stop-codon mutation (G339X) in the cytochrome b gene. Journal of the Neurological Sciences, 2003, 209, 61-63.	0.6	37
54	Gangliosidoses. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1707-1708.	1.8	35

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55	The Eight and a Half Year Journey of Undiagnosed AD: Gene Sequencing and Funding of Advanced Genetic Testing Has Led to Hope and New Beginnings. Frontiers in Endocrinology, 2017, 8, 107.	3.5	35
56	Whole-Exome Sequencing in the Clinic: Lessons from Six Consecutive Cases from the Clinician's Perspective. Molecular Syndromology, 2015, 6, 23-31.	0.8	33
57	IGF system in children with congenital disorders of glycosylation. Clinical Endocrinology, 2009, 70, 892-897.	2.4	31
58	Consensus statement on preventive and symptomatic care of leukodystrophy patients. Molecular Genetics and Metabolism, 2015, 114, 516-526.	1.1	29
59	The Value of Genetic Testing in Polycystic Kidney Diseases Illustrated by a Family With PKD2 and COL4A1 Mutations. American Journal of Kidney Diseases, 2018, 72, 302-308.	1.9	29
60	Spiral analysis in Niemannâ€Pick disease type C. Movement Disorders, 2009, 24, 1984-1990.	3.9	28
61	Metabolic Mimics: The Disorders of N-Linked Glycosylation. Seminars in Pediatric Neurology, 2005, 12, 144-151.	2.0	26
62	Niemann-Pick disease, type C and Roscoe Brady. Molecular Genetics and Metabolism, 2017, 120, 34-37.	1.1	26
63	Efficacy and safety of arimoclomol in <scp>Niemannâ€Pick</scp> disease type C: Results from a doubleâ€blind, randomised, placeboâ€controlled, multinational phase 2/3 trial of a novel treatment. Journal of Inherited Metabolic Disease, 2021, 44, 1463-1480.	3.6	26
64	Posterior reversible encephalopathy syndrome and hemorrhage associated with tacrolimus in a pediatric heart transplantation recipient. Pediatric Transplantation, 2013, 17, E67-70.	1.0	25
65	De Novo DNM1L Variant in a Teenager With Progressive Paroxysmal Dystonia and Lethal Super-refractory Myoclonic Status Epilepticus. Journal of Child Neurology, 2018, 33, 651-658.	1.4	25
66	Protective environmental factors for neuromyelitis optica. Neurology, 2014, 83, 1923-1929.	1.1	23
67	Severe Hajdu-Cheney syndrome with upper airway obstruction. , 1997, 70, 261-266.		21
68	The US Network of Pediatric Multiple Sclerosis Centers. Journal of Child Neurology, 2015, 30, 1381-1387.	1.4	21
69	Psychiatric and neurological symptoms in patients with Niemann-Pick disease type C (NP-C): Findings from the International NPC Registry. World Journal of Biological Psychiatry, 2019, 20, 310-319.	2.6	21
70	Oculomotor and Vestibular Findings in Gaucher Disease Type 3 and Their Correlation with Neurological Findings. Frontiers in Neurology, 2017, 8, 711.	2.4	20
71	Neurological Complications of Measles (Rubeola). Current Neurology and Neuroscience Reports, 2020, 20, 2.	4.2	20
72	Screening for "Prelysosomal Disorders": Carbohydrate-Deficient Glycoprotein Syndromes. Journal of Child Neurology, 1999, 14, S16-S22.	1.4	18

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73	The frequency of non-epileptic spells in children: Results of videoâ€“EEG monitoring in a tertiary care center. Seizure: the Journal of the British Epilepsy Association, 2008, 17, 583-587.	2.0	18
74	Whole Exome Sequencing and Heterologous Cellular Electrophysiology Studies Elucidate a Novel Loss-of-Function Mutation in the CACNA1A-Encoded Neuronal P/Q-Type Calcium Channel in a Child With Congenital Hypotonia and Developmental Delay. Pediatric Neurology, 2016, 55, 46-51.	2.1	18
75	Neurotransmitter abnormalities and response to supplementation in SPG11. Molecular Genetics and Metabolism, 2012, 107, 229-233.	1.1	17
76	Validation of the 5-domain Niemann-Pick type C Clinical Severity Scale. Orphanet Journal of Rare Diseases, 2021, 16, 79.	2.7	17
77	Acetylation turns leucine into a drug by membrane transporter switching. Scientific Reports, 2021, 11, 15812.	3.3	16
78	Recommendations for patient screening in ultra-rare inherited metabolic diseases: what have we learned from Niemann-Pick disease type C?. Orphanet Journal of Rare Diseases, 2019, 14, 20.	2.7	15
79	Safety and efficacy of (+)â€epicatechin in subjects with Friedreich's ataxia: A phase <scp>II</scp>, openâ€label, prospective study. Journal of Inherited Metabolic Disease, 2021, 44, 502-514.	3.6	15
80	Subclinical seizures in children diagnosed with localization-related epilepsy: Clinical and EEG characteristics. Epilepsy and Behavior, 2009, 16, 86-98.	1.7	14
81	Clinical disease characteristics of patients with Niemann-Pick Disease Type C: findings from the International Niemann-Pick Disease Registry (INPDR). Orphanet Journal of Rare Diseases, 2022, 17, 51.	2.7	14
82	Epilepsy in Childrenâ€”When Should We Think Neurometabolic Disease?. Journal of Child Neurology, 2012, 27, 663-671.	1.4	13
83	Developments in evidence creation for treatments of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2021, 44, 88-98.	3.6	13
84	Violent recurrent ballism associated with infections in two children with static encephalopathy. Movement Disorders, 2000, 15, 570-574.	3.9	12
85	An Unusual Presentation of Copper Metabolism Disorder and a Possible Connection With Niemann-Pick Type C. Journal of Child Neurology, 2011, 26, 518-521.	1.4	12
86	Peer review fraudâ€”itâ€™s not big and itâ€™s not clever. Journal of Inherited Metabolic Disease, 2016, 39, 1-2.	3.6	12
87	Clinical disease progression and biomarkers in Niemannâ€“Pick disease type C: a prospective cohort study. Orphanet Journal of Rare Diseases, 2020, 15, 328.	2.7	12
88	Free sialic acid storage disorder: Progress and promise. Neuroscience Letters, 2021, 755, 135896.	2.1	12
89	Biochemical and clinical response after umbilical cord blood transplant in a boy with early childhoodâ€onset betaâ€mannosidosis. Molecular Genetics & Genomic Medicine, 2019, 7, e00712.	1.2	10
90	Secondary Narcolepsy in Children. Journal of Child Neurology, 2021, 36, 123-127.	1.4	9

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91	Views of Recently First-Certified US Child Neurologists on Their Residency Training. Journal of Child Neurology, 2013, 28, 332-339.	1.4	8
92	Spontaneous improvement of carbohydrate-deficient transferrin in PMM2-CDG without mannose observed in CDG natural history study. Orphanet Journal of Rare Diseases, 2021, 16, 102.	2.7	8
93	International consensus on clinical severity scale use in evaluating Niemannâ€“Pick disease Type C in paediatric and adult patients: results from a Delphi Study. Orphanet Journal of Rare Diseases, 2021, 16, 482.	2.7	8
94	Niemann-Pick; Type C. Neurology, 1996, 46, 1785-1786.	1.1	7
95	Co-morbidity of Sanfilippo Syndrome type C and d-2-hydroxyglutaric aciduria. Journal of Neurology, 2011, 258, 1564-1565.	3.6	7
96	Expanded genetic insight and clinical experience of DNMT1-complex disorder. Neurology: Genetics, 2020, 6, e456.	1.9	7
97	Newborn screening: To <scp>WES</scp> or not to <scp>WES</scp>, that is the question. Journal of Inherited Metabolic Disease, 2020, 43, 904-905.	3.6	6
98	Teaching NeuroImages: Neuroimaging Findings in Inosine Triphosphate Pyrophosphohydrolase Deficiency. Neurology, 2021, 97, e109-e110.	1.1	6
99	Diagnosis of Niemann-Pick disease type C. Journal of Pediatrics, 1994, 124, 655-656.	1.8	5
100	Long-Term Outcome of Symptomatic Infantile Spasms Established by Video-Electroencephalography (EEG) Monitoring. Journal of Child Neurology, 2008, 23, 1288-1292.	1.4	4
101	From stargazing chicks to seizing infants. Neurology, 2009, 73, 824-825.	1.1	4
102	Slowly progressive encephalopathy with hearing loss due to a mutation in the mtDNA tRNALeu(CUN) gene. Journal of the Neurological Sciences, 2010, 290, 166-168.	0.6	4
103	Acute Encephalopathy With Biphasic Seizures and Late Restricted Diffusion. Pediatric Neurology, 2016, 55, 74-75.	2.1	4
104	Cerebral and cerebellar white matter abnormalities with magnetic resonance imaging in a child with Feingold syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2824-2827.	1.2	3
105	Movers and shakers. Neurology, 2010, 75, 15-17.	1.1	3
106	Inborn Errors of Metabolism for Child Neurology Residents. Seminars in Pediatric Neurology, 2011, 18, 95-97.	2.0	3
107	Teaching NeuroImages: Call it as you see it: Evolution of bilateral striatal necrosis. Neurology, 2012, 78, e123-e123.	1.1	3
108	Correction: Disease and patient characteristics in NP-C patients: findings from an international disease registry. Orphanet Journal of Rare Diseases, 2013, 8, 73.	2.7	3

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109	Epidural Hematoma in a Patient on Pegylated-L-Asparginase Therapy. Journal of Child Neurology, 2015, 30, 636-636.	1.4	3
110	Functional characterization of a <scp>GFAP</scp> variant of uncertain significance in an Alexander disease case within the setting of an individualized medicine clinic. Clinical Case Reports (discontinued), 2016, 4, 885-895.	0.5	3
111	Too Old for a Diaper! A Child With Diaper Changingâ€“InducedÂSeizures. Pediatric Neurology, 2016, 54, 91-92.	2.1	3
112	Roscoe Owen Brady, MD: Remembrances of co-investigators and colleagues. Molecular Genetics and Metabolism, 2017, 120, 1-7.	1.1	3
113	Spinocerebellar ataxia type 2 (SCA 2) in an infant with extreme CAG repeat expansion. American Journal of Medical Genetics Part A, 1998, 79, 383-387.	2.4	3
114	Impacts and Burden of Niemann pick Type-C: a patient and caregiver perspective. Orphanet Journal of Rare Diseases, 2021, 16, 493.	2.7	3
115	Manuel Rodriguez Gomez, MD. Pediatric Neurology, 2006, 35, 47-48.	2.1	2
116	Expanding Phenotypic Spectrum ofNKX2-1â€“Related Disordersâ€”Mitochondrial and Immunologic Dysfunction. JAMA Neurology, 2016, 73, 237.	9.0	2
117	A Patient as Art: Andrew Wyethâ€™s Portrayal of Christina Olsonâ€™s Neurologic Disorder in Christinaâ€™s World. Journal of Child Neurology, 2017, 32, 647-649.	1.4	2
118	Hematopoietic cell transplantation for sialidosis type I. Molecular Genetics and Metabolism Reports, 2022, 30, 100832.	1.1	2
119	Lost in translationâ€”Challenges in drug development for inherited metabolic diseases. Journal of Inherited Metabolic Disease, 2022, 45, 381-382.	3.6	2
120	Brave New World. Child Neurology Open, 2014, 1, 2329048X1454239.	1.1	1
121	Congenital Disorders of N-linked Glycosylation. , 2015, , 673-686.		1
122	Recommendations and guidelines in the JIMD: suggested procedures and avoidance of conflicts of interest. Journal of Inherited Metabolic Disease, 2016, 39, 327-329.	3.6	1
123	Disorders of Glycosylation. , 2017, , 317-322.		1
124	Measuring to improve. Neurology, 2011, 77, 1779-1780.	1.1	0
125	Editorial Comment: Ataxia, Ophthalmoplegia, and Impairment of Consciousness in a 19 Month-Old American Boy. Seminars in Pediatric Neurology, 2014, 21, 144.	2.0	0
126	Editorial Comment: Cerebellar Ataxia, Vertical Supranuclear Gaze Palsy, Sensorineural Deafness, Epilepsy, Dementia and Hallucinations in an Adolescent Male. Seminars in Pediatric Neurology, 2014, 21, 109-110.	2.0	0

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127	Peripheral Neuropathy in Inherited Metabolic Disease. , 2015, , 353-378.		0
128	The More Things Changeâ€¦ Child Neurology in the Age of Next-Generation Sequencing. Seminars in Pediatric Neurology, 2018, 26, 37-38.	2.0	0
129	Congenital disorders of N-linked glycosylation. , 2020, , 877-895.		0
130	Rapidly Progressive Paraplegia in an 11-Year-Old Girl: A Case of Spinal Cord Infarction and Expected Imaging Findings. Child Neurology Open, 2020, 7, 2329048X2098129.	1.1	0
131	Quo vadis now: Beyond genomics to an era of personalised medicine. Journal of Inherited Metabolic Disease, 2022, 45, 129-131.	3.6	0