Marc C Patterson

List of Publications by Year
in descending order

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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	Current directions in tau research: Highlights from Tau 2020. Alzheimer's and Dementia, 2022, 18, 988-1007.	0.8	42
2	Hematopoietic cell transplantation for sialidosis type I. Molecular Genetics and Metabolism Reports, 2022, 30, 100832.	1.1	2
3	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
4	Quo vadis now: Beyond genomics to an era of personalised medicine. Journal of Inherited Metabolic Disease, 2022, 45, 129-131.	3.6	0
5	Lost in translationâ€"Challenges in drug development for inherited metabolic diseases. Journal of Inherited Metabolic Disease, 2022, 45, 381-382.	3.6	2
6	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
7	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	3.6	146
8	Developments in evidence creation for treatments of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2021, 44, 88-98.	3.6	13
9	Secondary Narcolepsy in Children. Journal of Child Neurology, 2021, 36, 123-127.	1.4	9
10	Teaching NeuroImages: Neuroimaging Findings in Inosine Triphosphate Pyrophosphohydrolase Deficiency. Neurology, 2021, 97, e109-e110.	1.1	6
11	Validation of the 5-domain Niemann-Pick type C Clinical Severity Scale. Orphanet Journal of Rare Diseases, 2021, 16, 79.	2.7	17
12	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
13	Free sialic acid storage disorder: Progress and promise. Neuroscience Letters, 2021, 755, 135896.	2.1	12
14	Acetylation turns leucine into a drug by membrane transporter switching. Scientific Reports, 2021, 11, 15812.	3.3	16
15	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
16	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
17	Impacts and Burden of Niemann pick Type-C: a patient and caregiver perspective. Orphanet Journal of Rare Diseases, 2021, 16, 493.	2.7	3
18	Congenital disorders of N-linked glycosylation. , 2020, , 877-895.		0

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19	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
20	Expanded genetic insight and clinical experience of DNMT1-complex disorder. Neurology: Genetics, 2020, 6, e456.	1.9	7
21	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
22	Neurological Complications of Measles (Rubeola). Current Neurology and Neuroscience Reports, 2020, 20, 2.	4.2	20
23	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
24	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
25	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
26	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
27	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
28	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
29	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
30	Clinical, Radiologic, and Prognostic Features of Myelitis Associated With Myelin Oligodendrocyte Glycoprotein Autoantibody. JAMA Neurology, 2019, 76, 301.	9.0	243
31	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
32	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
33	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
34	The More Things Changeâ€¦ Child Neurology in the Age of Next-Generation Sequencing. Seminars in Pediatric Neurology, 2018, 26, 37-38.	2.0	0
35	Jeavons Syndrome: Clinical Features and Response to Treatment. Pediatric Neurology, 2018, 86, 46-51.	2.1	40
36	Consensus clinical management guidelines for Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2018, 13, 50.	2.7	200

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37	Miglustat in Niemann-Pick disease type C patients: a review. Orphanet Journal of Rare Diseases, 2018, 13, 140.	2.7	125
38	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
39	Roscoe Owen Brady, MD: Remembrances of co-investigators and colleagues. Molecular Genetics and Metabolism, 2017, 120, 1-7.	1.1	3
40	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
41	Niemann-Pick disease, type C and Roscoe Brady. Molecular Genetics and Metabolism, 2017, 120, 34-37.	1.1	26
42	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	1.6	119
43	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
44	Disorders of Glycosylation. , 2017, , 317-322.		1
45	Oculomotor and Vestibular Findings in Gaucher Disease Type 3 and Their Correlation with Neurological Findings. Frontiers in Neurology, 2017, 8, 711.	2.4	20
46	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
47	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
48	Peer review fraudâ€”itâ€™s not big and itâ€™s not clever. Journal of Inherited Metabolic Disease, 2016, 39, 1-2.	3.6	12
49	Expanding Phenotypic Spectrum ofNKX2-1â€“Related Disordersâ€”Mitochondrial and Immunologic Dysfunction. JAMA Neurology, 2016, 73, 237.	9.0	2
50	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
51	Clinical features of neuromyelitis optica in children. Neurology, 2016, 86, 245-252.	1.1	100
52	Acute Encephalopathy With Biphasic Seizures and Late Restricted Diffusion. Pediatric Neurology, 2016, 55, 74-75.	2.1	4
53	Too Old for a Diaper! A Child With Diaper Changingâ€“InducedÂSeizures. Pediatric Neurology, 2016, 54, 91-92.	2.1	3
54	Quo vadis: the reâ€definition of â€œinborn metabolic diseasesâ€•. Journal of Inherited Metabolic Disease, 2015, 38, 1003-1006.	3.6	48

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55	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
56	Congenital Disorders of N-linked Glycosylation. , 2015, , 673-686.		1
57	Disease specific therapies in leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 527-536.	1.1	45
58	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	1.1	185
59	A clinical approach to the diagnosis of patients with leukodystrophies and genetic leukoencephelopathies. Molecular Genetics and Metabolism, 2015, 114, 501-515.	1.1	163
60	Consensus statement on preventive and symptomatic care of leukodystrophy patients. Molecular Genetics and Metabolism, 2015, 114, 516-526.	1.1	29
61	The US Network of Pediatric Multiple Sclerosis Centers. Journal of Child Neurology, 2015, 30, 1381-1387.	1.4	21
62	Peripheral Neuropathy in Inherited Metabolic Disease. , 2015, , 353-378.		0
63	Epidural Hematoma in a Patient on Pegylated-L-Asparginase Therapy. Journal of Child Neurology, 2015, 30, 636-636.	1.4	3
64	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
65	Neurological Aspects of Human Glycosylation Disorders. Annual Review of Neuroscience, 2015, 38, 105-125.	10.7	302
66	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
67	Protective environmental factors for neuromyelitis optica. Neurology, 2014, 83, 1923-1929.	1.1	23
68	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
69	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
70	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
71	Brave New World. Child Neurology Open, 2014, 1, 2329048X1454239.	1.1	1
72	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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73	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
74	Posterior reversible encephalopathy syndrome and hemorrhage associated with tacrolimus in a pediatric heart transplantation recipient. Pediatric Transplantation, 2013, 17, E67-70.	1.0	25
75	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
76	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
77	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
78	Views of Recently First-Certified US Child Neurologists on Their Residency Training. Journal of Child Neurology, 2013, 28, 332-339.	1.4	8
79	Gangliosidoses. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1707-1708.	1.8	35
80	Cognitive Impairment Occurs in Children and Adolescents With Multiple Sclerosis. Journal of Child Neurology, 2013, 28, 102-107.	1.4	121
81	Teaching NeuroImages: Call it as you see it: Evolution of bilateral striatal necrosis. Neurology, 2012, 78, e123-e123.	1.1	3
82	Epilepsy in Childrenâ€"When Should We Think Neurometabolic Disease?. Journal of Child Neurology, 2012, 27, 663-671.	1.4	13
83	Neurotransmitter abnormalities and response to supplementation in SPG11. Molecular Genetics and Metabolism, 2012, 107, 229-233.	1.1	17
84	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
85	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
86	Neurology of inherited glycosylation disorders. Lancet Neurology, The, 2012, 11, 453-466.	10.2	179
87	Inborn Errors of Metabolism for Child Neurology Residents. Seminars in Pediatric Neurology, 2011, 18, 95-97.	2.0	3
88	Co-morbidity of Sanfilippo Syndrome type C and d-2-hydroxyglutaric aciduria. Journal of Neurology, 2011, 258, 1564-1565.	3.6	7
89	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
90	Measuring to improve. Neurology, 2011, 77, 1779-1780.	1.1	0

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91	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
92	Movers and shakers. Neurology, 2010, 75, 15-17.	1.1	3
93	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
94	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
95	Long-Term Miglustat Therapy in Children With Niemann-Pick Disease Type C. Journal of Child Neurology, 2010, 25, 300-305.	1.4	141
96	From stargazing chicks to seizing infants. Neurology, 2009, 73, 824-825.	1.1	4
97	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
98	Spiral analysis in Niemannâ€Pick disease type C. Movement Disorders, 2009, 24, 1984-1990.	3.9	28
99	IGF system in children with congenital disorders of glycosylation. Clinical Endocrinology, 2009, 70, 892-897.	2.4	31
100	Limbic encephalitis associated with antiâ€GAD antibody and common variable immune deficiency. Developmental Medicine and Child Neurology, 2009, 51, 563-567.	2.1	59
101	Subclinical seizures in children diagnosed with localization-related epilepsy: Clinical and EEG characteristics. Epilepsy and Behavior, 2009, 16, 86-98.	1.7	14
102	Recommendations on the diagnosis and management of Niemann-Pick disease type C. Molecular Genetics and Metabolism, 2009, 98, 152-165.	1.1	210
103	Newborn Screening for Krabbe Disease: the New York State Model. Pediatric Neurology, 2009, 40, 245-252.	2.1	174
104	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
105	Long-Term Outcome of Symptomatic Infantile Spasms Established by Video-Electroencephalography (EEG) Monitoring. Journal of Child Neurology, 2008, 23, 1288-1292.	1.4	4
106	Scurvy and Rickets Masked by Chronic Neurologic Illness: Revisiting "Psychologic Malnutrition". Pediatrics, 2007, 119, e783-e790.	2.1	46
107	Miglustat for treatment of Niemann-Pick C disease: a randomised controlled study. Lancet Neurology, The, 2007, 6, 765-772.	10.2	609
108	Pompe disease diagnosis and management guideline. Genetics in Medicine, 2006, 8, 267-288.	2.4	473

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109	Manuel Rodriguez Gomez, MD. Pediatric Neurology, 2006, 35, 47-48.	2.1	2
110	Metabolic Mimics: The Disorders of N-Linked Glycosylation. Seminars in Pediatric Neurology, 2005, 12, 144-151.	2.0	26
111	Therapy of Niemannâ€“Pick disease, type C. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2004, 1685, 77-82.	2.4	60
112	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
113	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
114	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
115	A Riddle Wrapped in a Mystery: Understanding Niemann-Pick Disease, Type C. Neurologist, 2003, 9, 301-310.	0.7	75
116	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
117	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
118	Violent recurrent ballism associated with infections in two children with static encephalopathy. Movement Disorders, 2000, 15, 570-574.	3.9	12
119	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
120	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
121	Screening for "Prelysosomal Disorders": Carbohydrate-Deficient Glycoprotein Syndromes. Journal of Child Neurology, 1999, 14, S16-S22.	1.4	18
122	Broad screening test for sphingolipid-storage diseases. Lancet, The, 1999, 354, 901-905.	13.7	131
123	Severe hypoglycemia as a presenting symptom of carbohydrate-deficient glycoprotein syndrome. Journal of Pediatrics, 1999, 135, 775-781.	1.8	89
124	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
125	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
126	Niemann-Pick C1 Disease Gene: Homology to Mediators of Cholesterol Homeostasis. Science, 1997, 277, 228-231.	12.6	1,373

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127	Angelman's syndrome: clinical and electroencephalographic findings. Electroencephalography and Clinical Neurophysiology, 1997, 102, 299-302.	0.3	39
128	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
129	Severe Hajdu-Cheney syndrome with upper airway obstruction. , 1997, 70, 261-266.		21
130	Niemann-Pick; Type C. Neurology, 1996, 46, 1785-1786.	1.1	7
131	Diagnosis of Niemann-Pick disease type C. Journal of Pediatrics, 1994, 124, 655-656.	1.8	5