Sidney M Gospe Jr

List of Publications by Year
in descending order

100 papers	3,310 citations	159585 30 h-index	175258 52 g-index
105 all docs	105 docs citations	105 times ranked	2804 citing authors

#	Article	IF	Citations
1	Syndrome of Hepatic Cirrhosis, Dystonia, Polycythemia, and Hypermanganesemia Caused by Mutations in SLC30A10 , a Manganese Transporter in Man. American Journal of Human Genetics, 2012, 90, 457-466.	6.2	321
2	Pyridoxine dependent epilepsy and antiquitin deficiency. Molecular Genetics and Metabolism, 2011, 104, 48-60.	1.1	258
3	Glial localization of antiquitin: Implications for pyridoxineâ€dependent epilepsy. Annals of Neurology, 2014, 75, 22-32.	5.3	165
4	Clinical and genetic characterization of manifesting carriers of DMD mutations. Neuromuscular Disorders, 2010, 20, 499-504.	0.6	136
5	Clinical features and the management of pyridoxine-dependent and pyridoxine-responsive seizures: review of 63 North American cases submitted to a patient registry. European Journal of Pediatrics, 2009, 168, 697-704.	2.7	114
6	Lysine restricted diet for pyridoxine-dependent epilepsy: First evidence and future trials. Molecular Genetics and Metabolism, 2012, 107, 335-344.	1.1	97
7	Nonhuman Primate Models of Intrauterine Cytomegalovirus Infection. ILAR Journal, 2006, 47, 49-64.	1.8	96
8	Current perspectives on pyridoxine-dependent seizures. Journal of Pediatrics, 1998, 132, 919-923.	1.8	90
9	Reduced GABA synthesis in pyridoxine-dependent seizures. Lancet, The, 1994, 343, 1133-1134.	13.7	80
10	Pyridoxine-dependent seizures: findings from recent studies pose new questions. Pediatric Neurology, 2002, 26, 181-185.	2.1	72
11	Pyridoxine-dependent seizures: new genetic and biochemical clues to help with diagnosis and treatment. Current Opinion in Neurology, 2006, 19, 148-153.	3.6	72
12	Prevalence of ALDH7A1 mutations in 18 North American pyridoxineâ€dependent seizure (PDS) patients. Epilepsia, 2009, 50, 1167-1175.	5.1	72
13	Pathology of inherited manganese transporter deficiency. Annals of Neurology, 2014, 75, 608-612.	5.3	60
14	Simultaneous determination of alpha-aminoadipic semialdehyde, piperideine-6-carboxylate and pipecolic acid by LCâ€“MS/MS for pyridoxine-dependent seizures and folinic acid-responsive seizures. Journal of Neuroscience Methods, 2009, 184, 136-141.	2.5	58
15	Fulminant demyelinating neuropathy mimicking cerebral death. , 1997, 20, 1595-1597.		57
16	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: A common epileptic encephalopathy. Journal of Inherited Metabolic Disease, 2019, 42, 353-361.	3.6	54
17	Paraparesis, hypermanganesaemia, and polycythaemia: a novel presentation of cirrhosis. Archives of Disease in Childhood, 2000, 83, 439-442.	1.9	53
18	VISUAL EVOKED POTENTIALS AND VISUAL PROCESSING IN STIMULANT DRUGâ€EXPOSED INFANTS. Developmental Medicine and Child Neurology, 1993, 35, 798-805.	2.1	51

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19	Effects of Environmental Tobacco Smoke Exposure in Utero and/or Postnatally on Brain Development1. Pediatric Research, 1996, 39, 494-498.	2.3	51
20	Central nervous system distribution of inhaled toluene. Fundamental and Applied Toxicology, 1988, 11, 540-545.	1.8	49
21	Neonatal vitamin-responsive epileptic encephalopathies. Chang Gung Medical Journal, 2010, 33, 1-12.	0.7	48
22	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	3.6	47
23	Prenatal Exposure to Toluene Results in Abnormal Neurogenesis and Migration in Rat Somatosensory Cortex. Pediatric Research, 2000, 47, 362-368.	2.3	47
24	Acute vincristine neurotoxicity in the presence of hereditary motor and sensory neuropathy type I. Medical and Pediatric Oncology, 1989, 17, 520-523.	1.0	46
25	Behavioral and neurochemical changes in folate-deficient mice. Physiology and Behavior, 1995, 58, 935-941.	2.1	44
26	Toluene abuse embryopathy: Longitudinal neurodevelopmental effects of prenatal exposure to toluene in rats. Reproductive Toxicology, 1998, 12, 119-126.	2.9	40
27	Preliminary investigation of the use of newborn dried blood spots for screening pyridoxine-dependent epilepsy by LC-MS/MS. Molecular Genetics and Metabolism, 2013, 110, 237-240.	1.1	39
28	Studies of dopamine pharmacology in molluscs. Life Sciences, 1983, 33, 1945-1957.	4.3	38
29	Lysine-Restricted Diet as Adjunct Therapy for Pyridoxine-Dependent Epilepsy: The PDE Consortium Consensus Recommendations. JIMD Reports, 2014, 15, 1-11.	1.5	37
30	Genetic heterogeneity for autosomal recessive pyridoxine-dependent seizures. Neurogenetics, 2005, 6, 143-149.	1.4	35
31	Intragenic deletions of ALDH7A1 in pyridoxine-dependent epilepsy caused by Alu - Alu recombination. Neurology, 2015, 85, 756-762.	1.1	34
32	Nemaline myopathy associated with hypertrophic cardiomyopathy. Pediatric Neurology, 1988, 4, 306-308.	2.1	33
33	Untargeted metabolomics and infrared ion spectroscopy identify biomarkers for pyridoxine-dependent epilepsy. Journal of Clinical Investigation, 2021, 131, .	8.2	33
34	Brain injury and protective effects of hypothermia using triphenyltetrazolium chloride in neonatal rat. Pediatric Neurology, 1993, 9, 263-267.	2.1	30
35	The Effects of High-Dose Toluene on Embryonic Development in the Rat. Pediatric Research, 1994, 36, 811-815.	2.3	30
36	Pyridoxineâ€dependent seizures and cognition in adulthood. Developmental Medicine and Child Neurology, 2003, 45, 782-785.	2.1	28

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37	Telemedicine and Child Neurology. Journal of Child Neurology, 2019, 34, 22-26.	1.4	27
38	Measurement of Spontaneous Rotational Movement (Circling) in Normal Children. Journal of Child Neurology, 1990, 5, 31-34.	1.4	25
39	Brainstem Bilirubin Toxicity in the Newborn Primate May Be Promoted and Reversed by Modulating PCO2. Pediatric Research, 1993, 34, 6-9.	2.3	25
40	Reply. Journal of Pediatrics, 1999, 134, 795-796.	1.8	25
41	Evidence-Based Decision Support for Neurological Diagnosis Reduces Errors and Unnecessary Workup. Journal of Child Neurology, 2014, 29, 487-492.	1.4	25
42	Neuromuscular hip dysplasia in Charcotâ€“Marieâ€“Tooth disease type 1A. Developmental Medicine and Child Neurology, 2009, 51, 408-411.	2.1	24
43	Editorial by concerned physicians: Unintended effect of the orphan drug act on the potential cost of 3,4-diaminopyridine. Muscle and Nerve, 2016, 53, 165-168.	2.2	24
44	Hypothalamic thermosensitivity in California quail (Lophortyx californicus). Journal of Comparative Physiology A: Neuroethology, Sensory, Neural, and Behavioral Physiology, 1977, 117, 345-357.	1.6	23
45	Dramatic Response After Lamotrigine in a Patient With Epileptic Encephalopathy and a De Novo CACNA1A Variant. Pediatric Neurology, 2016, 60, 79-82.	2.1	23
46	Development of a Rat Model of Toluene-Abuse Embryopathy. Pediatric Research, 1996, 40, 82-87.	2.3	22
47	Callosal alterations in pyridoxineâ€dependent epilepsy. Developmental Medicine and Child Neurology, 2014, 56, 1106-1110.	2.1	21
48	Comparison of Oral and Inhalation Exposures to Toluene. Journal of the American College of Toxicology, 1994, 13, 21-32.	0.2	20
49	Nonfebrile Illness Seizures: A Unique Seizure Category?. Epilepsia, 2005, 46, 952-955.	5.1	20
50	Persistent figureâ€eight and sideâ€toâ€side head shaking is a marker for rhombencephalosynapsis. Movement Disorders, 2013, 28, 2019-2023.	3.9	20
51	Choline Acetyltransferase Mutations Causing Congenital Myasthenic Syndrome: Molecular Findings and Genotype-Phenotype Correlations. Human Mutation, 2015, 36, 881-893.	2.5	20
52	Hereditary long Q-T syndrome presenting as epilepsy: Electroencephalography laboratory diagnosis. Annals of Neurology, 1989, 25, 514-516.	5.3	19
53	Epilepsy due to 20q13.33 subtelomere deletion masquerading as pyridoxineâ€dependent epilepsy. American Journal of Medical Genetics, Part A, 2012, 158A, 3190-3195.	1.2	18
54	Electroencephalography laboratory diagnosis of prolonged QT interval. Annals of Neurology, 1990, 28, 387-390.	5.3	17

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55	Transient Cortical Blindness in an Infant Exposed to Methamphetamine. Annals of Emergency Medicine, 1995, 26, 380-382.	0.6	17
56	Pyridoxine or pyridoxal-5â€²-phosphate for neonatal epilepsy. Neurology, 2014, 82, 1392-1394.	1.1	17
57	Redefining Outcome of First Seizures by Acute Illness. Pediatrics, 2010, 126, e1477-e1484.	2.1	16
58	Pyridoxine-dependent seizures and cognition in adulthood. Developmental Medicine and Child Neurology, 2003, 45, 782-5.	2.1	16
59	Cerebrospinal fluid 5-hydroxyindoleacetic acid and homovanillic acid in the pediatric opsoclonus-myoclonus syndrome. Annals of Neurology, 1995, 37, 189-197.	5.3	15
60	A novel missense mutation in POMT1 modulates the severe congenital muscular dystrophy phenotype associated with POMT1 nonsense mutations. Neuromuscular Disorders, 2014, 24, 312-320.	0.6	14
61	Biomarkers Aiding Diagnosis of Atypical Presentation of Pyridoxine-Dependent Epilepsy. Pediatric Neurology, 2011, 44, 289-291.	2.1	13
62	Truncating CLCN1 mutations in myotonia congenita: Variable patterns of inheritance. Muscle and Nerve, 2014, 49, 593-600.	2.2	13
63	Seizures, Syncope, or Breath-Holding Presenting to the Pediatric Neurologistâ€”When Is the Etiology a Life-Threatening Arrhythmia?. Seminars in Pediatric Neurology, 2005, 12, 2-9.	2.0	11
64	Pyridoxineâ€dependent epilepsy and pyridoxine phosphate oxidase deficiency: unique clinical symptoms and nonâ€specific EEG characteristics. Developmental Medicine and Child Neurology, 2010, 52, 602-603.	2.1	11
65	Geometric morphometrics reveal altered corpus callosum shape in pyridoxine-dependent epilepsy. Neurology, 2018, 91, e78-e86.	1.1	11
66	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy. Molecular Genetics and Metabolism, 2022, 135, 350-356.	1.1	11
67	Infantile Spasms Following Nearâ€Drowning: A Report of Two Cases. Epilepsia, 1987, 28, 45-48.	5.1	9
68	Exposure to environmental tobacco smoke during pregnancy in rats yields less effect on indices of brain cell number and size than does postnatal exposure. Reproductive Toxicology, 2009, 27, 22-27.	2.9	9
69	Variable presentation of nemaline myopathy: Novel mutation of alpha actin gene. Muscle and Nerve, 2007, 35, 254-258.	2.2	8
70	Drug-induced dystonia in neuronal ceroid-lipofuscinosis. Pediatric Neurology, 1986, 2, 236-237.	2.1	7
71	Life-threatening congestive heart failure as the presentation of centronuclear myopathy. Pediatric Neurology, 1987, 3, 117-120.	2.1	7
72	Delayed-Onset Movement Disorder and Encephalopathy After Oxycodone Ingestion. Seminars in Pediatric Neurology, 2014, 21, 160-165.	2.0	7

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73	Myeloradiculopathy associated with wasp sting. Pediatric Neurology, 1988, 4, 379-380.	2.1	6
74	Corpus Callosum Diffusion and Connectivity Features in High Functioning Subjects With Pyridoxine-Dependent Epilepsy. Pediatric Neurology, 2016, 54, 43-48.	2.1	6
75	The genotypic spectrum of ALDH7A1 mutations resulting in pyridoxine dependent epilepsy: a common epileptic encephalopathy. Journal of Inherited Metabolic Disease, 0, , .	3.6	6
76	Organic Solvents. , 2009, , 401-414.		5
77	Natural history of pyridoxineâ€dependent epilepsy: tools for prognostication. Developmental Medicine and Child Neurology, 2012, 54, 781-782.	2.1	5
78	Integrating neurocritical care approaches into neonatology: should all infants be treated equitably?. Journal of Perinatology, 2015, 35, 977-981.	2.0	5
79	Child Neurology Applicants Place Increasing Emphasis on Quality of Life Factors. Pediatric Neurology, 2021, 114, 42-46.	2.1	5
80	Trust but Verify: The Introduction of Plagiarism Detection Software. Pediatric Neurology, 2014, 50, 287.	2.1	4
81	Double Labeling of Proliferating Neurons with Anti-BrdU and Anti-NeuN: An Improved Immunohistochemical Technique Utilizing Microwave Irradiation. Journal of Histotechnology, 1998, 21, 201-204.	0.5	3
82	Biochemical and histochemical studies of the effect of reserpine in Aplysia californica. Comparative Biochemistry and Physiology Part C: Comparative Pharmacology, 1981, 70, 273-276.	0.2	2
83	Hospital Pharmacy and Emergency Department Availability of Parenteral Pyridoxine. Pediatric Emergency Care, 2005, 21, 586-588.	0.9	2
84	Spinal Arteriovenous Malformation Presenting as Meningitis. Developmental Medicine and Child Neurology, 1996, 38, 549-553.	2.1	2
85	Pyridoxine-dependent epilepsy. , 0, , 237-241.		2
86	SSADH deficiency in an adult. Neurology, 2015, 85, 842-843.	1.1	2
87	Adverse Effects of War and Armed Conflict on Children. Pediatric Neurology, 2022, 130, 69-70.	2.1	2
88	Burst-firing inhibition of cell R 15 in Aplysia californica: Pharmacological studies of the effects of tyramine, l²-phenethylamine and D-amphetamine. Comparative Biochemistry and Physiology Part C: Comparative Pharmacology, 1982, 71, 249-254.	0.2	1
89	Developmental outcome in pyridoxine-dependent epilepsy: Better late (onset) than early. European Journal of Paediatric Neurology, 2018, 22, 575-576.	1.6	1
90	Withholding Childhood Immunizations: A Parentâ€™s Right or a Childâ€™s Neglect?. Pediatric Neurology, 2020, 113, 80-81.	2.1	1

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91	Other Organic Chemicals. , 2009, , 415-420.		1
92	Central Nervous System Distribution of Inhaled Toluene. Toxicological Sciences, 1988, 11, 540-545.	3.1	0
93	Cardiac Causes of Sudden Death: Virtual Panel Discussion of Posed Questions. Seminars in Pediatric Neurology, 2005, 12, 67-69.	2.0	0
94	Disorders of Pyridoxine Metabolism. , 2015, , 541-555.		0
95	Pediatric Neurology 2014 Trainee Publication Award Winner: Dr. Mitchel T. Williams. Pediatric Neurology, 2015, 53, 103-104.	2.1	0
96	The Pediatric Neurology Trainee Publication Award for 2015. Pediatric Neurology, 2016, 63, 1-2.	2.1	0
97	Toward the elimination of bias in Pediatric Research. Pediatric Research, 2019, 86, 680-681.	2.3	0
98	Disorders of manganese transport. , 2019, , 643-655.		0
99	Disorders of pyridoxine metabolism. , 2020, , 711-728.		0
100	A Pediatrician's Personal Reflections on Varicella. Pediatrics, 1990, 86, 494-494.	2.1	0