

Alfons Macaya RuÃ-z

List of Publications by Year in descending order

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Version: 2024-02-01

136
papers

6,711
citations

94269

37
h-index

74018

75
g-index

141
all docs

141
docs citations

141
times ranked

11346
citing authors

#	ARTICLE	IF	CITATIONS
1	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. <i>Neurology</i> , 2022, , 10.1212/WNL.0000000000013278.	1.5	13
2	Elucidating the relationship between migraine risk and brain structure using genetic data. <i>Brain</i> , 2022, 145, 3214-3224.	3.7	7
3	Real-World Evidence Study on the Long-Term Safety of Everolimus in Patients With Tuberous Sclerosis Complex: Final Analysis Results. <i>Frontiers in Pharmacology</i> , 2022, 13, 802334.	1.6	8
4	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 529-542.	1.2	6
5	Delineating the neurological phenotype in children with defects in the <i>ECHS1</i> or <i>HIBCH</i> gene. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 401-414.	1.7	23
6	Clinical phenotypes of infantile onset CACNA1A-related disorder. <i>European Journal of Paediatric Neurology</i> , 2021, 30, 144-154.	0.7	13
7	Reduced hippocampal subfield volumes and memory performance in preterm children with and without germinal matrix-intraventricular hemorrhage. <i>Scientific Reports</i> , 2021, 11, 2420.	1.6	8
8	Tuberous Sclerosis registry to increase disease awareness (TOSCA) Post-Authorisation Safety Study of Everolimus in Patients With Tuberous Sclerosis Complex. <i>Frontiers in Neurology</i> , 2021, 12, 630378.	1.1	10
9	Îµ-Sarcoglycan: Unraveling the Myoclonus-Dystonia Gene. <i>Molecular Neurobiology</i> , 2021, 58, 3938-3952.	1.9	7
10	Rare functional genetic variants in COL7A1, COL6A5, COL1A2 and COL5A2 frequently occur in Chiari Malformation Type 1. <i>PLoS ONE</i> , 2021, 16, e0251289.	1.1	12
11	Rare manifestations and malignancies in tuberous sclerosis complex: findings from the Tuberous Sclerosis registry to increase disease awareness (TOSCA). <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 301.	1.2	15
12	Muscarinic acetylcholine receptor M1 mutations causing neurodevelopmental disorder and epilepsy. <i>Human Mutation</i> , 2021, 42, 1215-1220.	1.1	3
13	Biallelic <i>PI4KA</i> variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. <i>Brain</i> , 2021, 144, 2659-2669.	3.7	19
14	Historical Patterns of Diagnosis, Treatments, and Outcome of Epilepsy Associated With Tuberous Sclerosis Complex: Results From TOSCA Registry. <i>Frontiers in Neurology</i> , 2021, 12, 697467.	1.1	13
15	Early-onset eyelid stereotypies are a frequent and distinctive feature in Dravet syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 92, 155-157.	0.9	3
16	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. <i>American Journal of Human Genetics</i> , 2021, 108, 100-114.	2.6	17
17	Delineating the motor phenotype of SGCE-myoclonus dystonia syndrome. <i>Parkinsonism and Related Disorders</i> , 2020, 80, 165-174.	1.1	7
18	Renal Manifestations of Tuberous Sclerosis Complex: Key Findings From the Final Analysis of the TOSCA Study Focussing Mainly on Renal Angiomyolipomas. <i>Frontiers in Neurology</i> , 2020, 11, 972.	1.1	27

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19	Impaired proteasome activity and neurodegeneration with brain iron accumulation in <i>FBXO7</i> defect. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1436-1442.	1.7	21
20	Natural clusters of tuberous sclerosis complex (TSC)-associated neuropsychiatric disorders (TAND): new findings from the TOSCA TAND research project. <i>Journal of Neurodevelopmental Disorders</i> , 2020, 12, 24.	1.5	16
21	Burden of Illness and Quality of Life in Tuberous Sclerosis Complex: Findings From the TOSCA Study. <i>Frontiers in Neurology</i> , 2020, 11, 904.	1.1	20
22	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1205-1215.	1.2	14
23	Primary endovascular treatment for acute ischemic stroke in teenage patients: a short case series. <i>Neuroradiology</i> , 2020, 62, 851-860.	1.1	2
24	Tuberous Sclerosis Complex-Associated Neuropsychiatric Disorders (TAND): New Findings on Age, Sex, and Genotype in Relation to Intellectual Phenotype. <i>Frontiers in Neurology</i> , 2020, 11, 603.	1.1	7
25	Impact of Puberty in Pediatric Migraine: A Pilot Prospective Study. <i>Journal of Clinical Neurology</i>		

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37	The genetic etiology in cerebral palsy mimics: The results from a Greek tertiary care center. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 427-437.	0.7	26
38	Congenital myasthenic syndrome caused by novel COL13A1 mutations. <i>Journal of Neurology</i> , 2019, 266, 1107-1112.	1.8	14
39	Galloping tongue syndrome in a <i>PRRT2</i> mutation carrier. <i>Neurology: Genetics</i> , 2019, 5, e377.	0.9	4
40	Homomeric Kv7.2 current suppression is a common feature in <i>KCNQ2</i> epileptic encephalopathy. <i>Epilepsia</i> , 2019, 60, 139-148.	2.6	23
41	From gestalt to gene: early predictive dysmorphic features of PMM2-CDG. <i>Journal of Medical Genetics</i> , 2019, 56, 236-245.	1.5	19
42	Childhood onset progressive myoclonic dystonia due to a de novo KCTD17 splicing mutation. <i>Parkinsonism and Related Disorders</i> , 2019, 61, 7-9.	1.1	34
43	Epilepsy in tuberous sclerosis complex: Findings from the TOSCA Study. <i>Epilepsia Open</i> , 2019, 4, 73-84.	1.3	125
44	Renal angiomyolipoma in patients with tuberous sclerosis complex: findings from the Tuberous Sclerosis registry to increase disease Awareness. <i>Nephrology Dialysis Transplantation</i> , 2019, 34, 502-508.	0.4	55
45	Novel GCH1 Compound Heterozygosity Mutation in Infancy-Onset Generalized Dystonia. <i>Neuropediatrics</i> , 2018, 49, 296-297.	0.3	2
46	A homozygous loss-of-function mutation in <i>PDE2A</i> associated to early-onset hereditary chorea. <i>Movement Disorders</i> , 2018, 33, 482-488.	2.2	52
47	<i>CLCN1</i> Myotonia congenita mutation with a variable pattern of inheritance suggests a novel mechanism of dominant myotonia. <i>Muscle and Nerve</i> , 2018, 58, 157-160.	1.0	3
48	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. <i>Neuron</i> , 2018, 98, 743-753.e4.	3.8	63
49	Machine learning applied to neuroimaging for diagnosis of adult classic Chiari malformation: role of the basion as a key morphometric indicator. <i>Journal of Neurosurgery</i> , 2018, 129, 779-791.	0.9	21
50	Cognitive functioning in dyskinetic cerebral palsy: Its relation to motor function, communication and epilepsy. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 102-112.	0.7	28
51	Response to Letter to the Editor. <i>Neuropediatrics</i> , 2018, 49, 355-355.	0.3	0
52	Clinical Assessment of Dysarthria in Children with Cerebellar Syndrome Associated with PMM2-CDG. <i>Neuropediatrics</i> , 2018, 49, 408-413.	0.3	2
53	TSC-associated neuropsychiatric disorders (TAND): findings from the TOSCA natural history study. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 157.	1.2	106
54	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085

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55	Stroke-Like Episodes and Cerebellar Syndrome in Phosphomannomutase Deficiency (PMM2-CDG): Evidence for Hypoglycosylation-Driven Channelopathy. <i>International Journal of Molecular Sciences</i> , 2018, 19, 619.	1.8	40
56	Brain lesion scores obtained using a simple semi-quantitative scale from MR imaging are associated with motor function, communication and cognition in dyskinetic cerebral palsy. <i>NeuroImage: Clinical</i> , 2018, 19, 892-900.	1.4	13
57	Cephalometric oropharynx and oral cavity analysis in Chiari malformation Type I: a retrospective case-control study. <i>Journal of Neurosurgery</i> , 2017, 126, 626-633.	0.9	12
58	Tuberous Sclerosis registry to increase disease Awareness (TOSCA) – baseline data on 2093 patients. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 2.	1.2	166
59	Mutation Spectrum in the CACNA1A Gene in 49 Patients with Episodic Ataxia. <i>Scientific Reports</i> , 2017, 7, 2514.	1.6	36
60	White matter integrity in dyskinetic cerebral palsy: Relationship with intelligence quotient and executive function. <i>NeuroImage: Clinical</i> , 2017, 15, 789-800.	1.4	21
61	Whole-brain structural connectivity in dyskinetic cerebral palsy and its association with motor and cognitive function. <i>Human Brain Mapping</i> , 2017, 38, 4594-4612.	1.9	27
62	Enterovirus and neurological complications. <i>Anales De Pediatr�a (English Edition)</i> , 2017, 86, 107-109.	0.1	4
63	Transcriptomic Changes in Rat Cortex and Brainstem After Cortical Spreading Depression With or Without Pretreatment With Migraine Prophylactic Drugs. <i>Journal of Pain</i> , 2017, 18, 366-375.	0.7	5
64	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. <i>Scientific Reports</i> , 2017, 7, 12288.	1.6	23
65	Microcephaly with simplified gyral pattern, epilepsy and permanent neonatal diabetes syndrome (MEDS). A new patient and review of the literature. <i>European Journal of Medical Genetics</i> , 2017, 60, 517-520.	0.7	13
66	Proxy-reported quality of life in adolescents and adults with dyskinetic cerebral palsy is associated with executive functions and cortical thickness. <i>Quality of Life Research</i> , 2017, 26, 1209-1222.	1.5	19
67	A quantitative assessment of the evolution of cerebellar syndrome in children with phosphomannomutase-deficiency (PMM2-CDG). <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 155.	1.2	16
68	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. <i>Mitochondrion</i> , 2016, 30, 51-58.	1.6	70
69	Clinical course of sly syndrome (mucopolysaccharidosis type VII). <i>Journal of Medical Genetics</i> , 2016, 53, 403-418.	1.5	133
70	The Role of mTOR Inhibitors in the Treatment of Patients with Tuberous Sclerosis Complex: Evidence-based and Expert Opinions. <i>Drugs</i> , 2016, 76, 551-565.	4.9	66
71	Dominant-negative mutation p.Arg324Thr in <i>KCNA1</i> impairs Kv1.1 channel function in episodic ataxia. <i>Movement Disorders</i> , 2016, 31, 1743-1748.	2.2	17
72	Recommendations for the multidisciplinary management of tuberous sclerosis complex. <i>Medicina Cl�nica (English Edition)</i> , 2016, 147, 211-216.	0.1	2

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73	Measuring intellectual ability in cerebral palsy: The comparison of three tests and their neuroimaging correlates. <i>Research in Developmental Disabilities</i> , 2016, 56, 83-98.	1.2	21
74	GNAO1 encephalopathy: further delineation of a severe neurodevelopmental syndrome affecting females. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 38.	1.2	36
75	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 138.	1.2	49
76	A Single Amino Acid Deletion (F1502) in the S6 Segment of CaV2.1 Domain III Associated with Congenital Ataxia Increases Channel Activity and Promotes Ca ²⁺ Influx. <i>PLoS ONE</i> , 2015, 10, e0146035.	1.1	22
77	Candidate-gene association study searching for genetic factors involved in migraine chronification. <i>Cephalalgia</i> , 2015, 35, 500-507.	1.8	20
78	Replication study of previous migraine genome-wide association study findings in a Spanish sample of migraine with aura. <i>Cephalalgia</i> , 2015, 35, 776-782.	1.8	30
79	TOSCA – first international registry to address knowledge gaps in the natural history and management of tuberous sclerosis complex. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 182.	1.2	62
80	Herpes simplex virus encephalitis is a trigger of brain autoimmunity. <i>Annals of Neurology</i> , 2014, 75, 317-323.	2.8	372
81	Clinical and genetic analysis in alternating hemiplegia of childhood: Ten new patients from Southern Europe. <i>Journal of the Neurological Sciences</i> , 2014, 344, 37-42.	0.3	19
82	Recommendations for the radiological diagnosis and follow-up of neuropathological abnormalities associated with tuberous sclerosis complex. <i>Journal of Neuro-Oncology</i> , 2014, 118, 205-223.	1.4	31
83	A loss-of-function CACNA1A mutation causing benign paroxysmal torticollis of infancy. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 430-433.	0.7	36
84	MRI-based Morphometric Analysis of Posterior Cranial Fossa in the Diagnosis of Chiari Malformation Type I. <i>Journal of Neuroimaging</i> , 2014, 24, 250-256.	1.0	57
85	Biallelic SZT2 Mutations Cause Infantile Encephalopathy with Epilepsy and Dysmorphic Corpus Callosum. <i>American Journal of Human Genetics</i> , 2013, 93, 524-529.	2.6	81
86	Epileptic encephalopathy after HHV6 post-transplant acute limbic encephalitis in children: Confirmation of a new epilepsy syndrome. <i>Epilepsy Research</i> , 2013, 105, 419-422.	0.8	21
87	Brainstem dysgenesis during the neonatal period: diagnosis and management. <i>Journal of Perinatal Medicine</i> , 2013, 41, 445-453.	0.6	3
88	Screening of CACNA1A and ATP1A2 genes in hemiplegic migraine: clinical, genetic, and functional studies. <i>Molecular Genetics & Genomic Medicine</i> , 2013, 1, 206-222.	0.6	35
89	Chiari Malformation Type I: A Case-Control Association Study of 58 Developmental Genes. <i>PLoS ONE</i> , 2013, 8, e57241.	1.1	61
90	A replication study of a GWAS finding in migraine does not identify association in a Spanish case-control sample. <i>Cephalalgia</i> , 2012, 32, 1076-1080.	1.8	11

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91	Thalamic changes in a preterm sample with periventricular leukomalacia: correlation with white-matter integrity and cognitive outcome at school age. <i>Pediatric Research</i> , 2012, 71, 354-360.	1.1	29
92	Head Circumference Growth Function as a Marker of Neurological Impairment in a Cohort of Microcephalic Infants and Children. <i>Neuropediatrics</i> , 2012, 43, 271-274.	0.3	8
93	Central Hypoventilation and Brainstem Dysgenesis. <i>Pediatric Neurology</i> , 2012, 46, 257-259.	1.0	9
94	Cortical Thickness and Behavior Abnormalities in Children Born Preterm. <i>PLoS ONE</i> , 2012, 7, e42148.	1.1	38
95	Genome-wide association analysis identifies susceptibility loci for migraine without aura. <i>Nature Genetics</i> , 2012, 44, 777-782.	9.4	294
96	Paroxysmal non-kinesigenic dyskinesia due to a PNKD recurrent mutation: Report of two Southern European families. <i>European Journal of Paediatric Neurology</i> , 2012, 16, 86-89.	0.7	23
97	SNP variants within the vanilloid <i>TRPV1</i> and <i>TRPV3</i> receptor genes are associated with migraine in the Spanish population. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012, 159B, 94-103.	1.1	71
98	Acute Striatal Necrosis in Hemiplegic Migraine With de Novo CACNA1A Mutation. <i>Headache</i> , 2011, 51, 1542-1546.	1.8	15
99	Gray Matter Volume Decrements in Preterm Children With Periventricular Leukomalacia. <i>Pediatric Research</i> , 2011, 69, 554-560.	1.1	25
100	A mutation in the first intracellular loop of CACNA1A prevents P/Q channel modulation by SNARE proteins and lowers exocytosis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2010, 107, 1672-1677.	3.3	23
101	Paroxysmal exercise-induced dyskinesia, writer's cramp, migraine with aura and absence epilepsy in twin brothers with a novel SLC2A1 missense mutation. <i>Journal of the Neurological Sciences</i> , 2010, 295, 110-113.	0.3	52
102	The hemiplegic migraine-associated Y1245C mutation in CACNA1A results in a gain of channel function due to its effect on the voltage sensor and G-protein-mediated inhibition. <i>Pflügers Archiv European Journal of Physiology</i> , 2009, 458, 489-502.	1.3	36
103	Familial hemiplegic migraine: linkage to chromosome 14q32 in a Spanish kindred. <i>Neurogenetics</i> , 2009, 10, 191-198.	0.7	14
104	Two-stage case-control association study of dopamine-related genes and migraine. <i>BMC Medical Genetics</i> , 2009, 10, 95.	2.1	28
105	Lack of association of hormone receptor polymorphisms with migraine. <i>European Journal of Neurology</i> , 2009, 16, 413-415.	1.7	24
106	Contribution of syntaxin 1A to the genetic susceptibility to migraine: A case-control association study in the Spanish population. <i>Neuroscience Letters</i> , 2009, 455, 105-109.	1.0	11
107	Late-onset episodic ataxia type 2 associated with a novel loss-of-function mutation in the CACNA1A gene. <i>Journal of the Neurological Sciences</i> , 2009, 280, 10-14.	0.3	36
108	Genetic Analysis of 27 Spanish Patients with Hemiplegic Migraine, Basilar-Type Migraine and Childhood Periodic Syndromes. <i>Cephalalgia</i> , 2008, 28, 1039-1047.	1.8	57

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109	Neonatal Alexander Disease: MR Imaging Prenatal Diagnosis. American Journal of Neuroradiology, 2008, 29, 1973-1975.	1.2	30
110	Neonatal Rigid-Akinetic Syndrome and Dentato-Olivary Dysplasia. Pediatric Neurology, 2006, 34, 132-134.	1.0	9
111	A novel mutation in the gene encoding TIMM8a, a component of the mitochondrial protein translocase complexes, in a Spanish familial case of deafness-dystonia (Mohr-Tranebjaerg) syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 392-397.	0.7	25
112	Clinical and biochemical spectrum of D-bifunctional protein deficiency. Annals of Neurology, 2006, 59, 92-104.	2.8	175
113	Progressive vacuolating glycine leukoencephalopathy with pulmonary hypertension. Annals of Neurology, 2006, 60, 148-152.	2.8	20
114	Molybdenum Cofactor Deficiency Presenting as Neonatal Hyperekplexia: A Clinical, Biochemical and Genetic Study. Neuropediatrics, 2005, 36, 389-394.	0.3	30
115	Brainstem dysgenesis: report of five patients with congenital hypotonia, multiple cranial nerve involvement, and ocular motor apraxia. Developmental Medicine and Child Neurology, 2003, 45, 489-493.	1.1	33
116	Brainstem dysgenesis: report of five patients with congenital hypotonia, multiple cranial nerve involvement, and ocular motor apraxia. Developmental Medicine and Child Neurology, 2003, 45, 489-93.	1.1	15
117	Paroxysmal Kinesigenic Dyskinesia and Generalized Seizures: Clinical and Genetic Analysis in a Spanish Pedigree. Neuropediatrics, 2002, 33, 288-293.	0.3	27
118	Naturally occurring cell death during postnatal development of rat skeletal muscle. Muscle and Nerve, 2002, 26, 777-783.	1.0	8
119	Painful ophthalmoplegia with reversible carotid stenosis in a child. Pediatric Neurology, 2001, 24, 317-319.	1.0	16
120	Fusion of the Human Gene for the Polyubiquitination Coeffector UEV1 with Kua, a Newly Identified Gene. Genome Research, 2000, 10, 1743-1756.	2.4	91
121	Congenital Insensitivity to Pain with Anhidrosis: Novel Mutations in the TRKA (NTRK1) Gene Encoding a High-Affinity Receptor for Nerve Growth Factor. American Journal of Human Genetics, 1999, 64, 1570-1579.	2.6	166
122	Cell death and associated c-jun induction in perinatal hypoxia-ischemia. Effect of the neuroprotective drug dexamethasone. Molecular Brain Research, 1998, 56, 29-37.	2.5	22
123	Identification of necrotic cell death by the TUNEL assay in the hypoxic-ischemic neonatal rat brain. Neuroscience Letters, 1997, 230, 1-4.	1.0	161
124	Both apoptosis and necrosis occur following intrastratial administration of excitotoxins. Acta Neuropathologica, 1995, 90, 504-510.	3.9	82
125	Evidence of internucleosomal DNA fragmentation and identification of dying cells in X-ray-induced cell death in the developing brain. International Journal of Developmental Neuroscience, 1995, 13, 21-28.	0.7	32
126	Evidence of Nuclear DNA Fragmentation Following Hypoxia-Ischemia in the Infant Rat Brain, and Transient Forebrain Ischemia in the Adult Gerbil. Brain Pathology, 1994, 4, 115-122.	2.1	150

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127	Naturally occurring cell death in the developing cerebral cortex of the rat. Evidence of apoptosis-associated internucleosomal DNA fragmentation. <i>Neuroscience Letters</i> , 1994, 182, 77-79.	1.0	84
128	Increased expression of bcl-2 immunoreactivity in the developing cerebral cortex of the rat. <i>Neuroscience Letters</i> , 1994, 179, 13-16.	1.0	51
129	Apoptosis in substantia nigra following developmental striatal excitotoxic injury.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1994, 91, 8117-8121.	3.3	127
130	The mutation at nt 8993 of mitochondrial DNA is a common cause of Leigh's syndrome. <i>Annals of Neurology</i> , 1993, 34, 827-834.	2.8	252
131	Bilateral striatal lesions in childhood. <i>Pediatric Neurology</i> , 1993, 9, 349-358.	1.0	29
132	Disorders of Movement in Leigh Syndrome. <i>Neuropediatrics</i> , 1993, 24, 60-67.	0.3	58
133	Neonatal hypoxic-ischemic or excitotoxic striatal injury results in a decreased adult number of substantia nigra neurons. <i>Neuroscience</i> , 1992, 50, 559-569.	1.1	85
134	Effect of Striatal Lesion with Quinolate on the Development of Substantia Nigra Dopaminergic Neurons: A Quantitative Morphological Analysis. <i>Developmental Neuroscience</i> , 1992, 14, 362-368.	1.0	16
135	Acute neurologic dysfunction associated with destructive lesions of the basal ganglia: A benign form of infantile bilateral striatal necrosis. <i>Journal of Pediatrics</i> , 1990, 117, 578-581.	0.9	31
136	Pseudotumor cerebri, spinal and radicular pain, and hyporeflexia: A clinical variant of the Guillain-Barré syndrome?. <i>Pediatric Neurology</i> , 1988, 4, 120-121.	1.0	12