Alfons Macaya RuÃ-z

List of Publications by Year
in descending order

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	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
2	Herpes simplex virus encephalitis is a trigger of brain autoimmunity. Annals of Neurology, 2014, 75, 317-323.	2.8	372
3	Genome-wide association analysis identifies susceptibility loci for migraine without aura. Nature Genetics, 2012, 44, 777-782.	9.4	294
4	The mutation at nt 8993 of mitochondrial DNA is a common cause of Leigh's syndrome. Annals of Neurology, 1993, 34, 827-834.	2.8	252
5	Clinical and biochemical spectrum of D-bifunctional protein deficiency. Annals of Neurology, 2006, 59, 92-104.	2.8	175
6	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
7	TuberOus SClerosis registry to increase disease Awareness (TOSCA) â€“ baseline data on 2093 patients. Orphanet Journal of Rare Diseases, 2017, 12, 2.	1.2	166
8	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
9	Evidence of Nuclear DNA Fragmentation Following Hypoxiaâ€Ischemia in the Infant Rat Brain, and Transient Forebrain Ischemia in the Adult Gerbil. Brain Pathology, 1994, 4, 115-122.	2.1	150
10	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
11	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	1.5	133
12	Apoptosis in substantia nigra following developmental striatal excitotoxic injury.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 8117-8121.	3.3	127
13	Epilepsy in tuberous sclerosis complex: Findings from the <scp>TOSCA</scp> Study. Epilepsia Open, 2019, 4, 73-84.	1.3	125
14	TSC-associated neuropsychiatric disorders (TAND): findings from the TOSCA natural history study. Orphanet Journal of Rare Diseases, 2018, 13, 157.	1.2	106
15	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
16	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	2.6	88
17	Neonatal hypoxic-ischemic or excitotoxic striatal injury results in a decreased adult number of substantia nigra neurons. Neuroscience, 1992, 50, 559-569.	1.1	85
18	Naturally occurring cell death in the developing cerebral cortex of the rat. Evidence of apoptosis-associated internucleosomal DNA fragmentation. Neuroscience Letters, 1994, 182, 77-79.	1.0	84

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19	Both apoptosis and necrosis occur following intrastriatal administration of excitotoxins. Acta Neuropathologica, 1995, 90, 504-510.	3.9	82
20	Biallelic SZT2 Mutations Cause Infantile Encephalopathy with Epilepsy and Dysmorphic Corpus Callosum. American Journal of Human Genetics, 2013, 93, 524-529.	2.6	81
21	SNP variants within the vanilloid TRPV1 and TRPV3 receptor genes are associated with migraine in the Spanish population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 94-103.	1.1	71
22	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	1.6	70
23	The Role of mTOR Inhibitors in the Treatment of Patients with Tuberous Sclerosis Complex: Evidence-based and Expert Opinions. Drugs, 2016, 76, 551-565.	4.9	66
24	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. Neuron, 2018, 98, 743-753.e4.	3.8	63
25	TOSCA â€“ first international registry to address knowledge gaps in the natural history and management of tuberous sclerosis complex. Orphanet Journal of Rare Diseases, 2014, 9, 182.	1.2	62
26	Chiari Malformation Type I: A Case-Control Association Study of 58 Developmental Genes. PLoS ONE, 2013, 8, e57241.	1.1	61
27	Disorders of Movement in Leigh Syndrome. Neuropediatrics, 1993, 24, 60-67.	0.3	58
28	Genetic Analysis of 27 Spanish Patients with Hemiplegic Migraine, Basilar-Type Migraine and Childhood Periodic Syndromes. Cephalalgia, 2008, 28, 1039-1047.	1.8	57
29	MRIâ€based Morphometric Analysis of Posterior Cranial Fossa in the Diagnosis of Chiari Malformation Type I. Journal of Neuroimaging, 2014, 24, 250-256.	1.0	57
30	Renal angiomyolipoma in patients with tuberous sclerosis complex: findings from the TuberOus SClerosis registry to increase disease Awareness. Nephrology Dialysis Transplantation, 2019, 34, 502-508.	0.4	55
31	Headache, comorbidities and lifestyle in an adolescent population (The TEENs Study). Cephalalgia, 2019, 39, 91-99.	1.8	54
32	Paroxysmal exercise-induced dyskinesia, writer's cramp, migraine with aura and absence epilepsy in twin brothers with a novel SLC2A1 missense mutation. Journal of the Neurological Sciences, 2010, 295, 110-113.	0.3	52
33	A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488.	2.2	52
34	Increased expression of bcl-2 immunoreactivity in the developing cerebral cortex of the rat. Neuroscience Letters, 1994, 179, 13-16.	1.0	51
35	AZATAX: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2â€CDG). Annals of Neurology, 2019, 85, 740-751.	2.8	50
36	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. Orphanet Journal of Rare Diseases, 2015, 10, 138.	1.2	49

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37	Stroke-Like Episodes and Cerebellar Syndrome in Phosphomannomutase Deficiency (PMM2-CDG): Evidence for Hypoglycosylation-Driven Channelopathy. International Journal of Molecular Sciences, 2018, 19, 619.	1.8	40
38	Cortical Thickness and Behavior Abnormalities in Children Born Preterm. PLoS ONE, 2012, 7, e42148.	1.1	38
39	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. Pflugers Archiv European Journal of Physiology, 2009, 458, 489-502.	1.3	36
40	Late-onset episodic ataxia type 2 associated with a novel loss-of-function mutation in the CACNA1A gene. Journal of the Neurological Sciences, 2009, 280, 10-14.	0.3	36
41	A loss-of-function CACNA1A mutation causing benign paroxysmal torticollis of infancy. European Journal of Paediatric Neurology, 2014, 18, 430-433.	0.7	36
42	GNAO1 encephalopathy: further delineation of a severe neurodevelopmental syndrome affecting females. Orphanet Journal of Rare Diseases, 2016, 11, 38.	1.2	36
43	Mutation Spectrum in the CACNA1A Gene in 49 Patients with Episodic Ataxia. Scientific Reports, 2017, 7, 2514.	1.6	36
44	Screening of <scp>CACNA1A</scp> and <scp>ATP1A2</scp> genes in hemiplegic migraine: clinical, genetic, and functional studies. Molecular Genetics & Genomic Medicine, 2013, 1, 206-222.	0.6	35
45	Childhood onset progressive myoclonic dystonia due to a de novo KCTD17 splicing mutation. Parkinsonism and Related Disorders, 2019, 61, 7-9.	1.1	34
46	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
47	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
48	Acute neurologic dysfunction associated with destructive lesions of the basal ganglia: A benign form of infantile bilateral striatal necrosis. Journal of Pediatrics, 1990, 117, 578-581.	0.9	31
49	Recommendations for the radiological diagnosis and follow-up of neuropathological abnormalities associated with tuberous sclerosis complex. Journal of Neuro-Oncology, 2014, 118, 205-223.	1.4	31
50	Molybdenum Cofactor Deficiency Presenting as Neonatal Hyperekplexia: A Clinical, Biochemical and Genetic Study. Neuropediatrics, 2005, 36, 389-394.	0.3	30
51	Neonatal Alexander Disease: MR Imaging Prenatal Diagnosis. American Journal of Neuroradiology, 2008, 29, 1973-1975.	1.2	30
52	Replication study of previous migraine genome-wide association study findings in a Spanish sample of migraine with aura. Cephalalgia, 2015, 35, 776-782.	1.8	30
53	The clinical spectrum of the congenital myasthenic syndrome resulting from COL13A1 mutations. Brain, 2019, 142, 1547-1560.	3.7	30
54	Bilateral striatal lesions in childhood. Pediatric Neurology, 1993, 9, 349-358.	1.0	29

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55	Thalamic changes in a preterm sample with periventricular leukomalacia: correlation with white-matter integrity and cognitive outcome at school age. Pediatric Research, 2012, 71, 354-360.	1.1	29
56	Two-stage case-control association study of dopamine-related genes and migraine. BMC Medical Genetics, 2009, 10, 95.	2.1	28
57	Cognitive functioning in dyskinetic cerebral palsy: Its relation to motor function, communication and epilepsy. European Journal of Paediatric Neurology, 2018, 22, 102-112.	0.7	28
58	Paroxysmal Kinesigenic Dyskinesia and Generalized Seizures: Clinical and Genetic Analysis in a Spanish Pedigree. Neuropediatrics, 2002, 33, 288-293.	0.3	27
59	Whole-brain structural connectivity in dyskinetic cerebral palsy and its association with motor and cognitive function. Human Brain Mapping, 2017, 38, 4594-4612.	1.9	27
60	Renal Manifestations of Tuberous Sclerosis Complex: Key Findings From the Final Analysis of the TOSCA Study Focussing Mainly on Renal Angiomyolipomas. Frontiers in Neurology, 2020, 11, 972.	1.1	27
61	The genetic etiology in cerebral palsy mimics: The results from a Greek tertiary care center. European Journal of Paediatric Neurology, 2019, 23, 427-437.	0.7	26
62	A novel mutation in the gene encoding TIMM8a, a component of the mitochondrial protein translocase complexes, in a Spanish familial case of deafness-dystonia (Mohrâ€“Tranebjaerg) syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 392-397.	0.7	25
63	Gray Matter Volume Decrements in Preterm Children With Periventricular Leukomalacia. Pediatric Research, 2011, 69, 554-560.	1.1	25
64	Lack of association of hormone receptor polymorphisms with migraine. European Journal of Neurology, 2009, 16, 413-415.	1.7	24
65	A mutation in the first intracellular loop of CACNA1A prevents P/Q channel modulation by SNARE proteins and lowers exocytosis. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 1672-1677.	3.3	23
66	Paroxysmal non-kinesigenic dyskinesia due to a PNKD recurrent mutation: Report of two Southern European families. European Journal of Paediatric Neurology, 2012, 16, 86-89.	0.7	23
67	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. Scientific Reports, 2017, 7, 12288.	1.6	23
68	Homomeric Kv7.2 current suppression is a common feature in <scp>KCNQ</scp>2 epileptic encephalopathy. Epilepsia, 2019, 60, 139-148.	2.6	23
69	Delineating the neurological phenotype in children with defects in the <scp>ECHS1</scp> or <scp>HIBCH</scp> gene. Journal of Inherited Metabolic Disease, 2021, 44, 401-414.	1.7	23
70	Cell death and associated c-jun induction in perinatal hypoxiaâ€“ischemia. Effect of the neuroprotective drug dexamethasone. Molecular Brain Research, 1998, 56, 29-37.	2.5	22
71	A Single Amino Acid Deletion (Î”F1502) in the S6 Segment of CaV2.1 Domain III Associated with Congenital Ataxia Increases Channel Activity and Promotes Ca2+ Influx. PLoS ONE, 2015, 10, e0146035.	1.1	22
72	Clinical Characteristics of Subependymal Giant Cell Astrocytoma in Tuberous Sclerosis Complex. Frontiers in Neurology, 2019, 10, 705.	1.1	22

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73	Epileptic encephalopathy after HHV6 post-transplant acute limbic encephalitis in children: Confirmation of a new epilepsy syndrome. Epilepsy Research, 2013, 105, 419-422.	0.8	21
74	Measuring intellectual ability in cerebral palsy: The comparison of three tests and their neuroimaging correlates. Research in Developmental Disabilities, 2016, 56, 83-98.	1.2	21
75	White matter integrity in dyskinetic cerebral palsy: Relationship with intelligence quotient and executive function. NeuroImage: Clinical, 2017, 15, 789-800.	1.4	21
76	Machine learning applied to neuroimaging for diagnosis of adult classic Chiari malformation: role of the basion as a key morphometric indicator. Journal of Neurosurgery, 2018, 129, 779-791.	0.9	21
77	Impaired proteasome activity and neurodegeneration with brain iron accumulation in FBXO7 defect. Annals of Clinical and Translational Neurology, 2020, 7, 1436-1442.	1.7	21
78	Progressive vacuolating glycine leukoencephalopathy with pulmonary hypertension. Annals of Neurology, 2006, 60, 148-152.	2.8	20
79	Candidate-gene association study searching for genetic factors involved in migraine chronification. Cephalalgia, 2015, 35, 500-507.	1.8	20
80	Burden of Illness and Quality of Life in Tuberous Sclerosis Complex: Findings From the TOSCA Study. Frontiers in Neurology, 2020, 11, 904.	1.1	20
81	Clinical and genetic analysis in alternating hemiplegia of childhood: Ten new patients from Southern Europe. Journal of the Neurological Sciences, 2014, 344, 37-42.	0.3	19
82	Proxy-reported quality of life in adolescents and adults with dyskinetic cerebral palsy is associated with executive functions and cortical thickness. Quality of Life Research, 2017, 26, 1209-1222.	1.5	19
83	eDiVAâ€"Classification and prioritization of pathogenic variants for clinical diagnostics. Human Mutation, 2019, 40, 865-878.	1.1	19
84	From gestalt to gene: early predictive dysmorphic features of PMM2-CDG. Journal of Medical Genetics, 2019, 56, 236-245.	1.5	19
85	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	3.7	19
86	Newly Diagnosed and Growing Subependymal Giant Cell Astrocytoma in Adults With Tuberous Sclerosis Complex: Results From the International TOSCA Study. Frontiers in Neurology, 2019, 10, 821.	1.1	18
87	Dominantâ€negative mutation p.Arg324Thr in KCNA1 impairs Kv1.1 channel function in episodic ataxia. Movement Disorders, 2016, 31, 1743-1748.	2.2	17
88	Rare and de novo coding variants in chromodomain genes in Chiari I malformation. American Journal of Human Genetics, 2021, 108, 100-114.	2.6	17
89	Effect of Striatal Lesion with Quinolinate on the Development of Substantia Nigra Dopaminergic Neurons: A Quantitative Morphological Analysis. Developmental Neuroscience, 1992, 14, 362-368.	1.0	16
90	Painful ophthalmoplegia with reversible carotid stenosis in a child. Pediatric Neurology, 2001, 24, 317-319.	1.0	16

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91	A quantitative assessment of the evolution of cerebellar syndrome in children with phosphomannomutase-deficiency (PMM2-CDG). Orphanet Journal of Rare Diseases, 2017, 12, 155.	1.2	16
92	Natural clusters of tuberous sclerosis complex (TSC)-associated neuropsychiatric disorders (TAND): new findings from the TOSCA TAND research project. Journal of Neurodevelopmental Disorders, 2020, 12, 24.	1.5	16
93	Acute Striatal Necrosis in Hemiplegic Migraine With de Novo CACNA1A Mutation. Headache, 2011, 51, 1542-1546.	1.8	15
94	Rare manifestations and malignancies in tuberous sclerosis complex: findings from the TuberOus SClerosis registry to increAse disease awareness (TOSCA). Orphanet Journal of Rare Diseases, 2021, 16, 301.	1.2	15
95	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
96	Familial hemiplegic migraine: linkage to chromosome 14q32 in a Spanish kindred. Neurogenetics, 2009, 10, 191-198.	0.7	14
97	Congenital myasthenic syndrome caused by novel COL13A1 mutations. Journal of Neurology, 2019, 266, 1107-1112.	1.8	14
98	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	1.2	14
99	Microcephaly with simplified gyral pattern, epilepsy and permanent neonatal diabetes syndrome (MEDS). A new patient and review of the literature. European Journal of Medical Genetics, 2017, 60, 517-520.	0.7	13
100	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. NeuroImage: Clinical, 2018, 19, 892-900.	1.4	13
101	Executive function and general intellectual functioning in dyskinetic cerebral palsy: Comparison with spastic cerebral palsy and typically developing controls. European Journal of Paediatric Neurology, 2019, 23, 546-559.	0.7	13
102	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	0.7	13
103	Historical Patterns of Diagnosis, Treatments, and Outcome of Epilepsy Associated With Tuberous Sclerosis Complex: Results From TOSCA Registry. Frontiers in Neurology, 2021, 12, 697467.	1.1	13
104	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. Neurology, 2022, , 10.1212/WNL.0000000000013278.	1.5	13
105	Pseudotumor cerebri, spinal and radicular pain, and hyporeflexia: A clinical variant of the Guillain-BarrÃ© syndrome?. Pediatric Neurology, 1988, 4, 120-121.	1.0	12
106	Cephalometric oropharynx and oral cavity analysis in Chiari malformation Type I: a retrospective case-control study. Journal of Neurosurgery, 2017, 126, 626-633.	0.9	12
107	Rare functional genetic variants in COL7A1, COL6A5, COL1A2 and COL5A2 frequently occur in Chiari Malformation Type 1. PLoS ONE, 2021, 16, e0251289.	1.1	12
108	Contribution of syntaxin 1A to the genetic susceptibility to migraine: A caseâ€“control association study in the Spanish population. Neuroscience Letters, 2009, 455, 105-109.	1.0	11

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109	A replication study of a GWAS finding in migraine does not identify association in a Spanish case-control sample. Cephalalgia, 2012, 32, 1076-1080.	1.8	11
110	Treatment Patterns and Use of Resources in Patients With Tuberous Sclerosis Complex: Insights From the TOSCA Registry. Frontiers in Neurology, 2019, 10, 1144.	1.1	11
111	TuberOus SClerosis registry to increAse disease awareness (TOSCA) Post-Authorisation Safety Study of Everolimus in Patients With Tuberous Sclerosis Complex. Frontiers in Neurology, 2021, 12, 630378.	1.1	10
112	Neonatal Rigid-Akinetic Syndrome and Dentato-Olivary Dysplasia. Pediatric Neurology, 2006, 34, 132-134.	1.0	9
113	Central Hypoventilation and Brainstem Dysgenesis. Pediatric Neurology, 2012, 46, 257-259.	1.0	9
114	Naturally occurring cell death during postnatal development of rat skeletal muscle. Muscle and Nerve, 2002, 26, 777-783.	1.0	8
115	Head Circumference Growth Function as a Marker of Neurological Impairment in a Cohort of Microcephalic Infants and Children. Neuropediatrics, 2012, 43, 271-274.	0.3	8
116	Reduced hippocampal subfield volumes and memory performance in preterm children with and without germinal matrix-intraventricular hemorrhage. Scientific Reports, 2021, 11, 2420.	1.6	8
117	Real-World Evidence Study on the Long-Term Safety of Everolimus in Patients With Tuberous Sclerosis Complex: Final Analysis Results. Frontiers in Pharmacology, 2022, 13, 802334.	1.6	8
118	Delineating the motor phenotype of SGCE-myoclonus dystonia syndrome. Parkinsonism and Related Disorders, 2020, 80, 165-174.	1.1	7
119	Îµ-Sarcoglycan: Unraveling the Myoclonus-Dystonia Gene. Molecular Neurobiology, 2021, 58, 3938-3952.	1.9	7
120	Tuberous Sclerosis Complex-Associated Neuropsychiatric Disorders (TAND): New Findings on Age, Sex, and Genotype in Relation to Intellectual Phenotype. Frontiers in Neurology, 2020, 11, 603.	1.1	7
121	Impact of Puberty in Pediatric Migraine: A Pilot Prospective Study. Journal of Clinical Neurology		

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127	Brainstem dysgenesis during the neonatal period: diagnosis and management. Journal of Perinatal Medicine, 2013, 41, 445-453.	0.6	3
128	CLCN1 Myotonia congenita mutation with a variable pattern of inheritance suggests a novel mechanism of dominant myotonia. Muscle and Nerve, 2018, 58, 157-160.	1.0	3
129	The TOSCA Registry for Tuberous Sclerosisâ€"Lessons Learnt for Future Registry Development in Rare and Complex Diseases. Frontiers in Neurology, 2019, 10, 1182.	1.1	3
130	Muscarinic acetylcholine receptor M1 mutations causing neurodevelopmental disorder and epilepsy. Human Mutation, 2021, 42, 1215-1220.	1.1	3
131	Early-onset eyelid stereotypies are a frequent and distinctive feature in Dravet syndrome. Seizure: the Journal of the British Epilepsy Association, 2021, 92, 155-157.	0.9	3
132	Recommendations for the multidisciplinary management of tuberous sclerosis complex. Medicina ClÃnica (English Edition), 2016, 147, 211-216.	0.1	2
133	Novel GCH1 Compound Heterozygosity Mutation in Infancy-Onset Generalized Dystonia. Neuropediatrics, 2018, 49, 296-297.	0.3	2
134	Clinical Assessment of Dysarthria in Children with Cerebellar Syndrome Associated with PMM2-CDG. Neuropediatrics, 2018, 49, 408-413.	0.3	2
135	Primary endovascular treatment for acute ischemic stroke in teenage patients: a short case series. Neuroradiology, 2020, 62, 851-860.	1.1	2
136	Response to Letter to the Editor. Neuropediatrics, 2018, 49, 355-355.	0.3	0