Mariasavina Severino

List of Publications by Year
in descending order

192	3,917	185998 28	197535 49
papers	citations	h-index	g-index

203	203	203	6487
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	1.9	47
2	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	22
3	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	3.7	18
4	Epilepsy in KAT6A syndrome: Description of two individuals and revision of the literature. European Journal of Medical Genetics, 2022, 65, 104380.	0.7	5
5	Atlas of lesion locations and postsurgical seizure freedom in focal cortical dysplasia: A MELD study. Epilepsia, 2022, 63, 61-74.	2.6	36
6	Balamuthia mandrillaris infection: report of 1st autochthonous, fatal case in Italy. European Journal of Clinical Microbiology and Infectious Diseases, 2022, 41, 685-687.	1.3	3
7	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	1.4	5
8	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	3.7	8
9	Calcifications in diffuse leptomeningeal glioneuronal tumors: a case series. Quantitative Imaging in Medicine and Surgery, 2022, 12, 2985-2994.	1.1	3
10	De novo POLR2A p.(Ile457Thr) variant associated with early-onset encephalopathy and cerebellar atrophy: expanding the phenotypic spectrum. Brain and Development, 2022, 44, 480-485.	0.6	2
11	Guidelines for magnetic resonance imaging in pediatric head and neck pathologies: a multicentre international consensus paper. Neuroradiology, 2022, 64, 1081-1100.	1.1	12
12	A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. Frontiers in Pediatrics, 2022, 10, 847549.	0.9	3
13	Diffusion Kurtosis Imaging of Neonatal Spinal Cord in Clinical Routine. Frontiers in Radiology, 2022, 2, .	1.2	1
14	Imaging characteristics and neurosurgical outcome in subjects with agenesis of the corpus callosum and interhemispheric cysts. Neuroradiology, 2022, 64, 2163-2177.	1.1	1
15	Peculiar bony involvement of sinus pericranii in children: Extensive diploic erosion in three â€œkarsticâ€• variants. Clinical Neurology and Neurosurgery, 2022, 219, 107334.	0.6	0
16	An Atypical Case of Aphasia: Transitory Ischemic Attack in a 13-Year-Old Patient with Asymptomatic SARS-CoV-2 Infection. Children, 2022, 9, 983.	0.6	0
17	Genotype-Phenotype Correlation and Functional Insights for Two Monoallelic TREX1 Missense Variants Affecting the Catalytic Core. Genes, 2022, 13, 1179.	1.0	2
18	Role of visual evoked potentials and optical coherence tomography in the screening for optic pathway gliomas in patients with neurofibromatosis type I. European Journal of Ophthalmology, 2021, 31, 698-703.	0.7	5

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19	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.5	6
20	Characteristic Cochlear Hypoplasia in Patients with Walker-Warburg Syndrome: A Radiologic Study of the Inner Ear in Î±-Dystroglycanâ€“Related Muscular Disorders. American Journal of Neuroradiology, 2021, 42, 167-172.	1.2	9
21	Changes in appearance of cortical formation abnormalities in the foetus detected on sequential in utero MR imaging. European Radiology, 2021, 31, 1367-1377.	2.3	1
22	Homozygous missense WIPI2 variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Brain Communications, 2021, 3, fcab183.	1.5	10
23	An atypical case of post-varicella stroke in a child presenting with hemichorea followed by late-onset inflammatory focal cerebral arteriopathy. Quantitative Imaging in Medicine and Surgery, 2021, 11, 463-471.	1.1	6
24	Photoparoxysmal response in ADCK3 autosomal recessive ataxia: a case report and literature review. Epileptic Disorders, 2021, 23, 153-160.	0.7	5
25	Widening the Neuroimaging Features of Adenosine Deaminase 2 Deficiency. American Journal of Neuroradiology, 2021, 42, 975-979.	1.2	10
26	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	0.7	7
27	Ganglionic Eminence Anomalies and Coexisting Cerebral Developmental Anomalies on Fetal MR Imaging: Multicenter-Based Review of 60 Cases. American Journal of Neuroradiology, 2021, 42, 1151-1156.	1.2	7
28	Nosological Differences in the Nature of Punctate White Matter Lesions in Preterm Infants. Frontiers in Neurology, 2021, 12, 657461.	1.1	7
29	A rare triad of morning glory disc anomaly, moyamoya vasculopathy, and transsphenoidal cephalocele: pathophysiological considerations and surgical management. Neurological Sciences, 2021, 42, 5433-5439.	0.9	6
30	Biallelic variants in LIG3 cause a novel mitochondrial neurogastrointestinal encephalomyopathy. Brain, 2021, 144, 1451-1466.	3.7	28
31	Expanding the clinical and neuroimaging features of post-varicella arteriopathy of childhood. Journal of Neurology, 2021, 268, 4846-4865.	1.8	6
32	Cognitive and White Matter Microstructure Development in Congenital Hypothyroidism and Familial Thyroid Disorders. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e3990-e4006.	1.8	10
33	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	1.1	11
34	Combined medical therapy and neurosurgical revascularization preventing stroke in post-varicella angiopathy: Case report and review of literature. Brain and Development, 2021, 43, 1051-1056.	0.6	1
35	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	1.0	6
36	External ventricular drainage for posthemorrhagic ventricular dilatation in preterm infants: insights on efficacy and failure. Journal of Neurosurgery: Pediatrics, 2021, 28, 563-571.	0.8	5

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37	L1CAM variants cause two distinct imaging phenotypes on fetal MRI. Annals of Clinical and Translational Neurology, 2021, 8, 2004-2012.	1.7	8
38	Multifactorial Posterior Reversible Encephalopathy Syndrome in Children: Clinical, Laboratory, and Neuroimaging Findings. Journal of Pediatric Neurology, 2021, 19, 083-091.	0.0	2
39	Diagnostic Approach to Macrocephaly in Children. Frontiers in Pediatrics, 2021, 9, 794069.	0.9	17
40	Malformations of Cortical Development. , 2021, , 1-237.		1
41	Cognitive Profiles and Brain Volume Are Affected in Patients with Silverâ€“Russell Syndrome. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1478-e1488.	1.8	4
42	Role of diffusion weighted imaging for differentiating cerebral pilocytic astrocytoma and ganglioglioma BRAF V600E-mutant from wild type. Neuroradiology, 2020, 62, 71-80.	1.1	13
43	Prenatal magnetic resonance imaging within the 26th week of gestation may predict the fate of isolated upward rotation of the cerebellar vermis: insights from a multicentre study. European Radiology, 2020, 30, 2161-2170.	2.3	5
44	Bivalirudin anticoagulation to overcome heparin resistance in a neonate with cerebral sinovenus thrombosis. Blood Coagulation and Fibrinolysis, 2020, 31, 97-100.	0.5	5
45	Novel compound heterozygous pathogenic variants in nucleotide-binding protein like protein (NUBPL) cause leukoencephalopathy with multi-systemic involvement. Molecular Genetics and Metabolism, 2020, 129, 26-34.	0.5	9
46	Increased Childhood Peripheral Facial Palsy in the Emergency Department During COVID-19 Pandemic. Pediatric Emergency Care, 2020, 36, e595-e596.	0.5	16
47	Targeted re-sequencing in pediatric and perinatal stroke. European Journal of Medical Genetics, 2020, 63, 104030.	0.7	9
48	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	4.7	97
49	Correlation of multimodal ¹⁸F-DOPA PET and conventional MRI with treatment response and survival in children with diffuse intrinsic pontine gliomas. Theranostics, 2020, 10, 11881-11891.	4.6	14
50	Bilateral lesions of the basal ganglia and thalami (central grey matter)â€”pictorial review. Neuroradiology, 2020, 62, 1565-1605.	1.1	36
51	Listeria meningitis complicated by hydrocephalus in an immunocompetent child: case report and review of the literature. Italian Journal of Pediatrics, 2020, 46, 111.	1.0	11
52	Regional impairment of cortical and deep gray matter perfusion in preterm neonates with low-grade germinal matrix-intraventricular hemorrhage: an ASL study. Neuroradiology, 2020, 62, 1689-1699.	1.1	19
53	Definitions and classification of malformations of cortical development: practical guidelines. Brain, 2020, 143, 2874-2894.	3.7	145
54	Substrate reduction therapy with Miglustat in pediatric patients with GM1 type 2 gangliosidosis delays neurological involvement: A multicenter experience. Molecular Genetics & Genomic Medicine, 2020, 8, e1371.	0.6	18

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55	Perinatal Arterial Ischemic Stroke in Fetal Vascular Malperfusion: A Case Series and Literature Review. American Journal of Neuroradiology, 2020, 41, 2377-2383.	1.2	6
56	Neonatal Developmental Venous Anomalies: Clinicoradiologic Characterization and Follow-Up. American Journal of Neuroradiology, 2020, 41, 2370-2376.	1.2	3
57	Placental Pathology Findings and the Risk of Intraventricular and Cerebellar Hemorrhage in Preterm Neonates. Frontiers in Neurology, 2020, 11, 761.	1.1	5
58	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	3.7	123
59	Endocrine Outcomes In Central Diabetes Insipidus: the Predictive Value of Neuroimaging â€œMismatch Patternâ€. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 3562-3574.	1.8	3
60	Spatial coefficient of variation applied to arterial spin labeling MRI may contribute to predict surgical revascularization outcomes in pediatric moyamoya vasculopathy. Neuroradiology, 2020, 62, 1003-1015.	1.1	11
61	Cortical formation abnormalities on foetal MR imaging: a proposed classification system trialled on 356 cases from Italian and UK centres. European Radiology, 2020, 30, 5250-5260.	2.3	6
62	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7 â€related disorder. Birth Defects Research, 2020, 112, 1085-1092.	0.8	5
63	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	1.4	21
64	Targeted re-sequencing in malformations of cortical development: genotype-phenotype correlations. Seizure: the Journal of the British Epilepsy Association, 2020, 80, 145-152.	0.9	13
65	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	1.1	24
66	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	1.4	22
67	Schimke immuno-osseous dysplasia, two new cases with peculiar EEG pattern. Brain and Development, 2020, 42, 408-413.	0.6	2
68	White matter and cerebellar involvement in alternating hemiplegia of childhood. Journal of Neurology, 2020, 267, 1300-1311.	1.8	10
69	Adult-onset glutaric aciduria type I: rare presentation of a treatable disorder. Neurogenetics, 2020, 21, 179-186.	0.7	12
70	A further contribution to the delineation of epileptic phenotype in PACS2-related syndrome. Seizure: the Journal of the British Epilepsy Association, 2020, 79, 53-55.	0.9	15
71	Impact on rehabilitation programs during COVID-19 containment for children with pediatric and perinatal stroke. European Journal of Physical and Rehabilitation Medicine, 2020, 56, 692-694.	1.1	15
72	Arterial spin labeling perfusion in neonates. Seminars in Fetal and Neonatal Medicine, 2020, 25, 101130.	1.1	12

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73	CNNM2 homozygous mutations cause severe refractory hypomagnesemia, epileptic encephalopathy and brain malformations. European Journal of Medical Genetics, 2019, 62, 198-203.	0.7	28
74	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. Brain, 2019, 142, 2965-2978.	3.7	12
75	Cerebral blood flow in a case of typical aura without headache. Journal of Neurology, 2019, 266, 2869-2871.	1.8	1
76	Severe early-onset developmental and epileptic encephalopathy (DEE) associated with novel compound heterozygous pathogenic variants in SLC25A22: Case report and literature review. Seizure: the Journal of the British Epilepsy Association, 2019, 70, 56-58.	0.9	4
77	Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. Neurogenetics, 2019, 20, 165-172.	0.7	8
78	Posterior Fossa Malformations. Neuroimaging Clinics of North America, 2019, 29, 367-383.	0.5	18
79	Early Pain Exposure Influences Functional Brain Connectivity in Very Preterm Neonates. Frontiers in Neuroscience, 2019, 13, 899.	1.4	30
80	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	2.6	48
81	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	2.6	29
82	Punctate white matter lesions of preterm infants: Risk factor analysis. European Journal of Paediatric Neurology, 2019, 23, 733-739.	0.7	16
83	Pelizaeusâ€“Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. Neuropediatrics, 2019, 50, 268-270.	0.3	1
84	The features of the m.10197G>A mtDNA mutation. Journal of the Neurological Sciences, 2019, 400, 184-185.	0.3	0
85	Advanced MR imaging and 18F-DOPA PET characteristics of H3K27M-mutant and wild-type pediatric diffuse midline gliomas. European Journal of Nuclear Medicine and Molecular Imaging, 2019, 46, 1685-1694.	3.3	41
86	Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females. European Journal of Human Genetics, 2019, 27, 1254-1259.	1.4	41
87	Dissecting the neurological phenotype in children with callosal agenesis, interhemispheric cysts and malformations of cortical development. Journal of Neurology, 2019, 266, 1167-1181.	1.8	12
88	Clinical and neuroimaging features of the m.10197G>A mtDNA mutation: New case reports and expansion of the phenotype variability. Journal of the Neurological Sciences, 2019, 399, 69-75.	0.3	8
89	Asymmetric cavernous sinus enlargement: a novel finding in Sturgeâ€“Weber syndrome. Neuroradiology, 2019, 61, 595-602.	1.1	6
90	Pediatric Brain Tissue Segmentation from MRI using Clustering: a Preliminary Study. , 2019, 2019, 6557-6560.		0

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91	Familial ROBO1 deletion associated with ectopic posterior pituitary, duplication of the pituitary stalk and anterior pituitary hypoplasia. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 95-99.	0.4	11
92	3q29 microduplication syndrome: Description of two new cases and delineation of the minimal critical region. European Journal of Medical Genetics, 2018, 61, 428-433.	0.7	13
93	Noninvasive Assessment of Hemodynamic Stress Distribution after Indirect Revascularization for Pediatric Moyamoya Vasculopathy. American Journal of Neuroradiology, 2018, 39, 1157-1163.	1.2	4
94	Acute Communicating Hydrocephalus as Spinal Cord Surgery Complication in Patient with Lumbar Lipomyelocele. World Neurosurgery, 2018, 115, 468-472.e2.	0.7	2
95	Brain-injured Survivors of Monochorionic Twin Pregnancies Complicated by Single Intrauterine Death: MR Findings in a Multicenter Study. Radiology, 2018, 288, 582-590.	3.6	23
96	Unusual white matter involvement in EAST syndrome associated with novel KCNJ10 mutations. Journal of Neurology, 2018, 265, 1419-1425.	1.8	8
97	Role of MRI T2-DRIVE in the assessment of pituitary stalk abnormalities without gadolinium in pituitary diseases. European Journal of Endocrinology, 2018, 178, 613-622.	1.9	22
98	Pediatric astrocytic tumor grading: comparison between arterial spin labeling and dynamic susceptibility contrast MRI perfusion. Neuroradiology, 2018, 60, 437-446.	1.1	43
99	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	3.7	352
100	Spinal motor neuron involvement in a patient with homozygous PRUNE mutation. European Journal of Paediatric Neurology, 2018, 22, 541-543.	0.7	10
101	ABCC6 mutations and early onset stroke: Two cases of a typical Pseudoxanthoma Elasticum. European Journal of Paediatric Neurology, 2018, 22, 725-728.	0.7	15
102	Differences in subependymal vein anatomy may predispose preterm infants to GMHâ€“IVH. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2018, 103, F59-F65.	1.4	30
103	The effects of mild germinal matrix-intraventricular haemorrhage on the developmental white matter microstructure of preterm neonates: a DTI study. European Radiology, 2018, 28, 1157-1166.	2.3	41
104	T2*-based MR imaging (gradient echo or susceptibility-weighted imaging) in midline and off-midline intracranial germ cell tumors: a pilot study. Neuroradiology, 2018, 60, 89-99.	1.1	25
105	Clinical and Molecular Characterization of Two Patients with CNTN6 Copy Number Variations. Cytogenetic and Genome Research, 2018, 156, 144-149.	0.6	6
106	Novel CNS malformations and skeletal anomalies in a patient with Beaulieuâ€boycottâ€innes syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2835-2840.	0.7	9
107	Reversible cerebral vasoconstriction complicating cerebral atherosclerotic vascular disease in Schimke immuno-osseous dysplasia. Neuroradiology, 2018, 60, 885-888.	1.1	5
108	Quantitative susceptibility map analysis in preterm neonates with germinal matrixâ€intraventricular hemorrhage. Journal of Magnetic Resonance Imaging, 2018, 48, 1199-1207.	1.9	15

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109	Intragenic Microdeletion of <i>ULK4</i> and Partial Microduplication of <i>BRWD3</i> in Siblings with Neuropsychiatric Features and Obesity. Cytogenetic and Genome Research, 2018, 156, 14-21.	0.6	9
110	Structural Connectivity Analysis in Children with Segmental Callosal Agenesis. American Journal of Neuroradiology, 2017, 38, 639-647.	1.2	13
111	Widening the Heterogeneity of Leigh Syndrome: Clinical, Biochemical, and Neuroradiologic Features in a Patient Harboring a NDUFA10 Mutation. JIMD Reports, 2017, 37, 37-43.	0.7	13
112	Incidental findings on routine brain MRI scans in preterm infants. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2017, 102, F73-F78.	1.4	15
113	Neuroimaging Changes in Menkes Disease, Part 1. American Journal of Neuroradiology, 2017, 38, 1850-1857.	1.2	42
114	ADA2 deficiency (DADA2) as an unrecognised cause of early onset polyarteritis nodosa and stroke: a multicentre national study. Annals of the Rheumatic Diseases, 2017, 76, 1648-1656.	0.5	199
115	MR Imaging Diagnosis of Diencephalic-Mesencephalic Junction Dysplasia in Fetuses with Developmental Ventriculomegaly. American Journal of Neuroradiology, 2017, 38, 1643-1646.	1.2	16
116	Characterization of the Phenotype Associated with Microduplication Reciprocal to NF1 Microdeletion Syndrome. Cytogenetic and Genome Research, 2017, 152, 22-28.	0.6	0
117	Moyamoya Vasculopathy in PHACE Syndrome: Six New Cases and Review of the Literature. World Neurosurgery, 2017, 108, 291-302.	0.7	17
118	Teaching Neuro Images : Figure of 8. Neurology, 2017, 89, e172-e173.	1.5	5
119	Novel AMPD2 mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	0.9	22
120	Cryopyrin-associated Periodic Syndromes in Italian Patients: Evaluation of the Rate of Somatic NLRP3 Mosaicism and Phenotypic Characterization. Journal of Rheumatology, 2017, 44, 1667-1673.	1.0	28
121	Grading and outcome prediction of pediatric diffuse astrocytic tumors with diffusion and arterial spin labeling perfusion MRI in comparison with 18Fâ€“DOPA PET. European Journal of Nuclear Medicine and Molecular Imaging, 2017, 44, 2084-2093.	3.3	53
122	Early impairment of somatosensory evoked potentials in very young children with achondroplasia with foramen magnum stenosis. Developmental Medicine and Child Neurology, 2017, 59, 192-198.	1.1	11
123	Torcular pseudomass: a potential diagnostic pitfall in infants and young children. Pediatric Radiology, 2017, 47, 227-234.	1.1	7
124	Improvement in White Matter Tract Reconstruction with Constrained Spherical Deconvolution and Track Density Mapping in Low Angular Resolution Data: A Pediatric Study and Literature Review. Frontiers in Pediatrics, 2017, 5, 182.	0.9	28
125	Added value of diffusion weighted imaging in pediatric central nervous system embryonal tumors surveillance. Oncotarget, 2017, 8, 60401-60413.	0.8	16
126	Novel asymptomatic CNS findings in patients withACVR1/ALK2mutations causing fibrodysplasia ossificans progressiva. Journal of Medical Genetics, 2016, 53, 859-864.	1.5	12

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127	Interstitial de novo 18q22.3q23 deletion: clinical, neuroradiological and molecular characterization of a new case and review of the literature. Molecular Cytogenetics, 2016, 9, 78.	0.4	8
128	White matter involvement in a family with a novel PDGFB mutation. Neurology: Genetics, 2016, 2, e77.	0.9	19
129	Ability of 18F-DOPA PET/CT and fused 18F-DOPA PET/MRI to assess striatal involvement in paediatric glioma. European Journal of Nuclear Medicine and Molecular Imaging, 2016, 43, 1664-1672.	3.3	25
130	Variability of Cerebral Deep Venous System in Preterm and Term Neonates Evaluated on MR SWI Venography. American Journal of Neuroradiology, 2016, 37, 2144-2149.	1.2	17
131	Diagnostic Approach to Pediatric Spine Disorders. Magnetic Resonance Imaging Clinics of North America, 2016, 24, 621-644.	0.6	13
132	Neuroimaging of Infectious and Inflammatory Diseases of the Pediatric Cerebellum and Brainstem. Neuroimaging Clinics of North America, 2016, 26, 471-487.	0.5	20
133	Crossed Pontine Hemiatrophy Associated with Unilateral Cerebellar Hemorrhage in Premature Infants. Neuropediatrics, 2016, 47, 404-407.	0.3	3
134	Beyond spinal muscular atrophy with lower extremity dominance: cerebellar hypoplasia associated with a novel mutation in <scp>BICD</scp>2. European Journal of Neurology, 2016, 23, e19-21.	1.7	18
135	Delayed rotation of the cerebellar vermis: a pitfall in early second-trimester fetal magnetic resonance imaging. Ultrasound in Obstetrics and Gynecology, 2016, 48, 121-124.	0.9	21
136	Idiopathic Cervical Hematomyelia in an Infant: Spinal Cord Injury without Radiographic Abnormality Caused by a Trivial Trauma? Case Report and Review of the Literature. World Neurosurgery, 2016, 90, 38-44.	0.7	2
137	Expanding the spectrum of congenital anomalies of the diencephalicâ€“mesencephalic junction. Neuroradiology, 2016, 58, 33-44.	1.1	23
138	New insights into central nervous system involvement in FOP: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2015, 167, 2817-2821.	0.7	12
139	Pretransplant management of basilar artery aneurysm and moyamoya disease in a child with Alagille syndrome. Liver Transplantation, 2015, 21, 1227-1230.	1.3	12
140	Congenital aural atresia associated with agenesis of internal carotid artery in a girl with a FOXI3 deletion. American Journal of Medical Genetics, Part A, 2015, 167, 537-544.	0.7	27
141	Association of achondroplasia with sagittal synostosis and scaphocephaly in two patients, an underestimated condition?. American Journal of Medical Genetics, Part A, 2015, 167, 646-652.	0.7	15
142	A novel homozygous MCOLN1 double mutant allele leading to TRP channel domain ablation underlies Mucolipidosis IV in an Italian Child. Metabolic Brain Disease, 2015, 30, 681-686.	1.4	8
143	Reversible cerebral vasoconstriction mimicking posterior reversible encephalopathy syndrome in an infant with end-stage renal disease. Cephalalgia, 2015, 35, 1031-1033.	1.8	3
144	Accuracy of ultrasound in assessing cerebellar haemorrhages in very low birthweight babies. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2015, 100, F289-F292.	1.4	51

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145	Low-grade intraventricular hemorrhage: is ultrasound good enough?. Journal of Maternal-Fetal and Neonatal Medicine, 2015, 28, 2261-2264.	0.7	68
146	Clinico-radiological and molecular characterization of a child with ring chromosome 2 presenting growth failure, microcephaly, kidney and brain malformations. Molecular Cytogenetics, 2015, 8, 17.	0.4	7
147	Expanding the Clinical and Magnetic Resonance Spectrum of Leukoencephalopathy with Thalamus and Brainstem Involvement and High Lactate (LTBL) in a Patient Harboring a Novel EARS2 Mutation. JIMD Reports, 2015, 23, 85-89.	0.7	15
148	Intradural Extramedullary Ependymoma with Leptomeningeal Dissemination: The First Case Report in a Child and Literature Review. World Neurosurgery, 2015, 84, 865.e13-865.e19.	0.7	14
149	Sinus pericranii: diagnosis and management in 21 pediatric patients. Journal of Neurosurgery: Pediatrics, 2015, 15, 60-70.	0.8	67
150	Update on neuroimaging phenotypes of mid-hindbrain malformations. Neuroradiology, 2015, 57, 113-138.	1.1	45
151	Pituitary deficiency and congenital infiltrating lipomatosis of the face in a girl with deletion of chromosome 1q24.3q31.1. American Journal of Medical Genetics, Part A, 2014, 164, 495-499.	0.7	16
152	Further genotypeâ€“phenotype correlation emerging from two families with PLP1 exon 4 skipping. Clinical Genetics, 2014, 85, 267-272.	1.0	7
153	Congenital multifocal rhabdoid tumor: a case with peculiar biological behavior and different response to treatment according to location (central nervous system and kidney). Cancer Genetics, 2014, 207, 441-444.	0.2	3
154	Successful urgent neurosugery management with rFVIIa mega doses in a child with haemophilia A and high titre inhibitor. Blood Coagulation and Fibrinolysis, 2014, 25, 518-521.	0.5	6
155	Midbrain-Hindbrain Involvement in Septo-Optic Dysplasia. American Journal of Neuroradiology, 2014, 35, 1586-1592.	1.2	22
156	Severe growth hormone deficiency and pituitary malformation in a patient with chromosome 2p25 duplication and 2q37 deletion. Molecular Cytogenetics, 2014, 7, 41.	0.4	11
157	Novel DyneinDYNC1H1Neck and Motor Domain Mutations Link Distal Spinal Muscular Atrophy and Abnormal Cortical Development. Human Mutation, 2014, 35, 298-302.	1.1	77
158	Constitutional chromosomal events at 22q11 and 15q26 in a child with a pilocytic astrocytoma of the spinal cord. Molecular Cytogenetics, 2014, 7, 31.	0.4	2
159	Spontaneously Regressing Leukoencephalopathy With Bilateral Temporal Cysts in Congenital Rubella Infection. Pediatric Infectious Disease Journal, 2014, 33, 422-424.	1.1	5
160	Expanded spectrum of Pelizaeusâ€“Merzbacher-like disease: literature revision and description of a novel GJC2 mutation in an unusually severe form. European Journal of Human Genetics, 2013, 21, 34-39.	1.4	30
161	The Shrunken, Bright Cerebellum: A Characteristic MRI Finding in Congenital Disorders of Glycosylation Type 1a. American Journal of Neuroradiology, 2012, 33, 2062-2067.	1.2	47
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