

# Mariasavina Severino

## List of Publications by Year in descending order

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192  
papers

3,917  
citations

185998

28  
h-index

197535

49  
g-index

203  
all docs

203  
docs citations

203  
times ranked

6487  
citing authors

#	ARTICLE	IF	CITATIONS
1	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. <i>Brain</i> , 2018, 141, 391-408.	3.7	352
2	ADA2 deficiency (DADA2) as an unrecognised cause of early onset polyarteritis nodosa and stroke: a multicentre national study. <i>Annals of the Rheumatic Diseases</i> , 2017, 76, 1648-1656.	0.5	199
3	Definitions and classification of malformations of cortical development: practical guidelines. <i>Brain</i> , 2020, 143, 2874-2894.	3.7	145
4	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. <i>Brain</i> , 2020, 143, 2454-2473.	3.7	123
5	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. <i>Science Advances</i> , 2020, 6, .	4.7	97
6	Diffuse Leptomeningeal Glioneuronal Tumors: A New Entity?. <i>Brain Pathology</i> , 2010, 20, 361-366.	2.1	95
7	Congenital tumors of the central nervous system. <i>Neuroradiology</i> , 2010, 52, 531-548.	1.1	87
8	The pulvinar sign: frequency and clinical correlations in Fabry disease. <i>Journal of Neurology</i> , 2008, 255, 738-744.	1.8	82
9	Novel Dynein<i>DYNC1H1</i> Neck and Motor Domain Mutations Link Distal Spinal Muscular Atrophy and Abnormal Cortical Development. <i>Human Mutation</i> , 2014, 35, 298-302.	1.1	77
10	Optic pathway glioma: Long-term visual outcome in children without neurofibromatosis type-1. <i>Pediatric Blood and Cancer</i> , 2010, 55, 1083-1088.	0.8	70
11	Low-grade intraventricular hemorrhage: is ultrasound good enough?. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2015, 28, 2261-2264.	0.7	68
12	Sinus pericranii: diagnosis and management in 21 pediatric patients. <i>Journal of Neurosurgery: Pediatrics</i> , 2015, 15, 60-70.	0.8	67
13	New MR sequences (diffusion, perfusion, spectroscopy) in brain tumours. <i>Pediatric Radiology</i> , 2010, 40, 999-1009.	1.1	53
14	Grading and outcome prediction of pediatric diffuse astrocytic tumors with diffusion and arterial spin labeling perfusion MRI in comparison with 18Fâ€“DOPA PET. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 2017, 44, 2084-2093.	3.3	53
15	Accuracy of ultrasound in assessing cerebellar haemorrhages in very low birthweight babies. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2015, 100, F289-F292.	1.4	51
16	A Delayed Methadone Encephalopathy: Clinical and Neuroradiological Findings. <i>Journal of Child Neurology</i> , 2010, 25, 748-751.	0.7	50
17	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyposis. <i>American Journal of Human Genetics</i> , 2019, 105, 689-705.	2.6	48
18	The Shrunken, Bright Cerebellum: A Characteristic MRI Finding in Congenital Disorders of Glycosylation Type 1a. <i>American Journal of Neuroradiology</i> , 2012, 33, 2062-2067.	1.2	47

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19	The <sc>ENIGMA&Epilepsy</sc> working group: Mapping disease from large data sets. <i>Human Brain Mapping</i> , 2022, 43, 113-128.	1.9	47
20	Update on neuroimaging phenotypes of mid-hindbrain malformations. <i>Neuroradiology</i> , 2015, 57, 113-138.	1.1	45
21	Pontine tegmental cap dysplasia: developmental and cognitive outcome in three adolescent patients. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 36.	1.2	44
22	Pediatric astrocytic tumor grading: comparison between arterial spin labeling and dynamic susceptibility contrast MRI perfusion. <i>Neuroradiology</i> , 2018, 60, 437-446.	1.1	43
23	Neuroimaging Changes in Menkes Disease, Part 1. <i>American Journal of Neuroradiology</i> , 2017, 38, 1850-1857.	1.2	42
24	The effects of mild germinal matrix-intraventricular haemorrhage on the developmental white matter microstructure of preterm neonates: a DTI study. <i>European Radiology</i> , 2018, 28, 1157-1166.	2.3	41
25	Advanced MR imaging and 18F-DOPA PET characteristics of H3K27M-mutant and wild-type pediatric diffuse midline gliomas. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 2019, 46, 1685-1694.	3.3	41
26	Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females. <i>European Journal of Human Genetics</i> , 2019, 27, 1254-1259.	1.4	41
27	Bilateral lesions of the basal ganglia and thalami (central grey matter)&quot;pictorial review. <i>Neuroradiology</i> , 2020, 62, 1565-1605.	1.1	36
28	Atlas of lesion locations and postsurgical seizure freedom in focal cortical dysplasia: A MELD study. <i>Epilepsia</i> , 2022, 63, 61-74.	2.6	36
29	Neuroradiologic findings and follow&uor with magnetic resonance imaging of the genetic forms of haemophagocytic lymphohistiocytosis with CNS involvement. <i>Pediatric Blood and Cancer</i> , 2012, 58, 810-814.	0.8	32
30	Expanded spectrum of Pelizaeus&quot;Merzbacher-like disease: literature revision and description of a novel GJC2 mutation in an unusually severe form. <i>European Journal of Human Genetics</i> , 2013, 21, 34-39.	1.4	30
31	Differences in subependymal vein anatomy may predispose preterm infants to GMH&quot;IVH. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2018, 103, F59-F65.	1.4	30
32	Early Pain Exposure Influences Functional Brain Connectivity in Very Preterm Neonates. <i>Frontiers in Neuroscience</i> , 2019, 13, 899.	1.4	30
33	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. <i>American Journal of Human Genetics</i> , 2019, 105, 854-868.	2.6	29
34	Cryopyrin-associated Periodic Syndromes in Italian Patients: Evaluation of the Rate of Somatic NLRP3 Mosaicism and Phenotypic Characterization. <i>Journal of Rheumatology</i> , 2017, 44, 1667-1673.	1.0	28
35	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. <i>Frontiers in Pediatrics</i> , 2017, 5, 182.	0.9	28
36	CNNM2 homozygous mutations cause severe refractory hypomagnesemia, epileptic encephalopathy and brain malformations. <i>European Journal of Medical Genetics</i> , 2019, 62, 198-203.	0.7	28

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37	Biallelic variants in <i>LIG3</i> cause a novel mitochondrial neurogastrointestinal encephalomyopathy. <i>Brain</i> , 2021, 144, 1451-1466.	3.7	28
38	Congenital aural atresia associated with agenesis of internal carotid artery in a girl with a <i>FOXI3</i> deletion. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 537-544.	0.7	27
39	Spinal Cord Infarction Due to Fibrocartilaginous Embolization: The Role of Diffusion Weighted Imaging and Short-Tau Inversion Recovery Sequences. <i>Journal of Child Neurology</i> , 2010, 25, 1024-1028.	0.7	26
40	Skull base osteomyelitis and potential cerebrovascular complications in children. <i>Pediatric Radiology</i> , 2012, 42, 867-874.	1.1	25
41	Ability of 18F-DOPA PET/CT and fused 18F-DOPA PET/MRI to assess striatal involvement in paediatric glioma. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 2016, 43, 1664-1672.	3.3	25
42	T2*-based MR imaging (gradient echo or susceptibility-weighted imaging) in midline and off-midline intracranial germ cell tumors: a pilot study. <i>Neuroradiology</i> , 2018, 60, 89-99.	1.1	25
43	Damaging de novo missense variants in <i>EEF1A2</i> lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. <i>Human Mutation</i> , 2020, 41, 1263-1279.	1.1	24
44	Expanding the spectrum of congenital anomalies of the diencephalicâ€mesencephalic junction. <i>Neuroradiology</i> , 2016, 58, 33-44.	1.1	23
45	Brain-injured Survivors of Monochorionic Twin Pregnancies Complicated by Single Intrauterine Death: MR Findings in a Multicenter Study. <i>Radiology</i> , 2018, 288, 582-590.	3.6	23
46	Midbrain-Hindbrain Involvement in Septo-Optic Dysplasia. <i>American Journal of Neuroradiology</i> , 2014, 35, 1586-1592.	1.2	22
47	Novel <i>AMPD2</i> mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. <i>Neurology: Genetics</i> , 2017, 3, e179.	0.9	22
48	Role of MRI T2-DRIVE in the assessment of pituitary stalk abnormalities without gadolinium in pituitary diseases. <i>European Journal of Endocrinology</i> , 2018, 178, 613-622.	1.9	22
49	Loss of <i>Wwox</i> Perturbs Neuronal Migration and Impairs Early Cortical Development. <i>Frontiers in Neuroscience</i> , 2020, 14, 644.	1.4	22
50	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	22
51	Delayed rotation of the cerebellar vermis: a pitfall in early second-trimester fetal magnetic resonance imaging. <i>Ultrasound in Obstetrics and Gynecology</i> , 2016, 48, 121-124.	0.9	21
52	Biallelic <i>MFSD2A</i> variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. <i>European Journal of Human Genetics</i> , 2020, 28, 1509-1519.	1.4	21
53	Transverse dural sinuses: incidence of anatomical variants and flow artefacts with 2D time-of-flight MR venography at 1 Tesla. <i>Radiologia Medica</i> , 2010, 115, 326-338.	4.7	20
54	Neuroimaging of Infectious and Inflammatory Diseases of the Pediatric Cerebellum and Brainstem. <i>Neuroimaging Clinics of North America</i> , 2016, 26, 471-487.	0.5	20

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55	White matter involvement in a family with a novel <i>PDGFB</i> mutation. <i>Neurology: Genetics</i> , 2016, 2, e77.	0.9	19
56	Regional impairment of cortical and deep gray matter perfusion in preterm neonates with low-grade germinal matrix-intraventricular hemorrhage: an ASL study. <i>Neuroradiology</i> , 2020, 62, 1689-1699.	1.1	19
57	Intermittent-relapsing pyruvate dehydrogenase complex deficiency: a case with clinical, biochemical, and neuroradiological reversibility. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 472-476.	1.1	18
58	Beyond spinal muscular atrophy with lower extremity dominance: cerebellar hypoplasia associated with a novel mutation in <i>BICD2</i> . <i>European Journal of Neurology</i> , 2016, 23, e19-21.	1.7	18
59	Posterior Fossa Malformations. <i>Neuroimaging Clinics of North America</i> , 2019, 29, 367-383.	0.5	18
60	Substrate reduction therapy with Miglustat in pediatric patients with GM1 type 2 gangliosidosis delays neurological involvement: A multicenter experience. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1371.	0.6	18
61	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. <i>Brain</i> , 2022, 145, 1285-1298.	3.7	18
62	Variability of Cerebral Deep Venous System in Preterm and Term Neonates Evaluated on MR SWI Venography. <i>American Journal of Neuroradiology</i> , 2016, 37, 2144-2149.	1.2	17
63	Moyamoya Vasculopathy in PHACE Syndrome: Six New Cases and Review of the Literature. <i>World Neurosurgery</i> , 2017, 108, 291-302.	0.7	17
64	Diagnostic Approach to Macrocephaly in Children. <i>Frontiers in Pediatrics</i> , 2021, 9, 794069.	0.9	17
65	Pituitary deficiency and congenital infiltrating lipomatosis of the face in a girl with deletion of chromosome 1q24.3q31.1. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 495-499.	0.7	16
66	MR Imaging Diagnosis of Diencephalic-Mesencephalic Junction Dysplasia in Fetuses with Developmental Ventriculomegaly. <i>American Journal of Neuroradiology</i> , 2017, 38, 1643-1646.	1.2	16
67	Punctate white matter lesions of preterm infants: Risk factor analysis. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 733-739.	0.7	16
68	Increased Childhood Peripheral Facial Palsy in the Emergency Department During COVID-19 Pandemic. <i>Pediatric Emergency Care</i> , 2020, 36, e595-e596.	0.5	16
69	Added value of diffusion weighted imaging in pediatric central nervous system embryonal tumors surveillance. <i>Oncotarget</i> , 2017, 8, 60401-60413.	0.8	16
70	Association of achondroplasia with sagittal synostosis and scaphocephaly in two patients, an underestimated condition?. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 646-652.	0.7	15
71	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. <i>JIMD Reports</i> , 2015, 23, 85-89.	0.7	15
72	Incidental findings on routine brain MRI scans in preterm infants. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2017, 102, F73-F78.	1.4	15

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73	ABCC6 mutations and early onset stroke: Two cases of a typical Pseudoxanthoma Elasticum. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 725-728.	0.7	15
74	Quantitative susceptibility map analysis in preterm neonates with germinal matrixâ€intra ventricular hemorrhage. <i>Journal of Magnetic Resonance Imaging</i> , 2018, 48, 1199-1207.	1.9	15
75	A further contribution to the delineation of epileptic phenotype in PACS2-related syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 79, 53-55.	0.9	15
76	Impact on rehabilitation programs during COVID-19 containment for children with pediatric and perinatal stroke. <i>European Journal of Physical and Rehabilitation Medicine</i> , 2020, 56, 692-694.	1.1	15
77	Intradural Extramedullary Ependymoma with Leptomeningeal Dissemination: The First Case Report in a Child and Literature Review. <i>World Neurosurgery</i> , 2015, 84, 865.e13-865.e19.	0.7	14
78	Correlation of multimodal <sup>18</sup> F-DOPA PET and conventional MRI with treatment response and survival in children with diffuse intrinsic pontine gliomas. <i>Theranostics</i> , 2020, 10, 11881-11891.	4.6	14
79	Diagnostic Approach to Pediatric Spine Disorders. <i>Magnetic Resonance Imaging Clinics of North America</i> , 2016, 24, 621-644.	0.6	13
80	Structural Connectivity Analysis in Children with Segmental Callosal Agenesis. <i>American Journal of Neuroradiology</i> , 2017, 38, 639-647.	1.2	13
81	Widening the Heterogeneity of Leigh Syndrome: Clinical, Biochemical, and Neuroradiologic Features in a Patient Harboring a NDUFA10 Mutation. <i>JIMD Reports</i> , 2017, 37, 37-43.	0.7	13
82	3q29 microduplication syndrome: Description of two new cases and delineation of the minimal critical region. <i>European Journal of Medical Genetics</i> , 2018, 61, 428-433.	0.7	13
83	Role of diffusion weighted imaging for differentiating cerebral pilocytic astrocytoma and ganglioglioma BRAF V600E-mutant from wild type. <i>Neuroradiology</i> , 2020, 62, 71-80.	1.1	13
84	Targeted re-sequencing in malformations of cortical development: genotype-phenotype correlations. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 80, 145-152.	0.9	13
85	Anorectal malformation and spinal dysraphism: the value of diffusion-weighted imaging in detecting associated intradural (epi)dermoid cyst. <i>Journal of Pediatric Surgery</i> , 2008, 43, 1935-1938.	0.8	12
86	New insights into central nervous system involvement in FOP: Case report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2817-2821.	0.7	12
87	Pretransplant management of basilar artery aneurysm and moyamoya disease in a child with Alagille syndrome. <i>Liver Transplantation</i> , 2015, 21, 1227-1230.	1.3	12
88	Novel asymptomatic CNS findings in patients with ACVR1/ALK2 mutations causing fibrodysplasia ossificans progressiva. <i>Journal of Medical Genetics</i> , 2016, 53, 859-864.	1.5	12
89	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. <i>Brain</i> , 2019, 142, 2965-2978.	3.7	12
90	Dissecting the neurological phenotype in children with callosal agenesis, interhemispheric cysts and malformations of cortical development. <i>Journal of Neurology</i> , 2019, 266, 1167-1181.	1.8	12

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91	Adult-onset glutaric aciduria type I: rare presentation of a treatable disorder. <i>Neurogenetics</i> , 2020, 21, 179-186.	0.7	12
92	Arterial spin labeling perfusion in neonates. <i>Seminars in Fetal and Neonatal Medicine</i> , 2020, 25, 101130.	1.1	12
93	Guidelines for magnetic resonance imaging in pediatric head and neck pathologies: a multicentre international consensus paper. <i>Neuroradiology</i> , 2022, 64, 1081-1100.	1.1	12
94	Chronic cystic lesion of the sacrum: characterisation with diffusion-weighted MR imaging. <i>Radiologia Medica</i> , 2008, 113, 739-746.	4.7	11
95	Analysis of NADP+-dependent isocitrate dehydrogenase-1/2 gene mutations in pediatric brain tumors: report of a secondary anaplastic astrocytoma carrying the IDH1 mutation. <i>Journal of Neuro-Oncology</i> , 2012, 109, 477-484.	1.4	11
96	Severe growth hormone deficiency and pituitary malformation in a patient with chromosome 2p25 duplication and 2q37 deletion. <i>Molecular Cytogenetics</i> , 2014, 7, 41.	0.4	11
97	Early impairment of somatosensory evoked potentials in very young children with achondroplasia with foramen magnum stenosis. <i>Developmental Medicine and Child Neurology</i> , 2017, 59, 192-198.	1.1	11
98	Familial ROBO1 deletion associated with ectopic posterior pituitary, duplication of the pituitary stalk and anterior pituitary hypoplasia. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2019, 32, 95-99.	0.4	11
99	Listeria meningitis complicated by hydrocephalus in an immunocompetent child: case report and review of the literature. <i>Italian Journal of Pediatrics</i> , 2020, 46, 111.	1.0	11
100	Spatial coefficient of variation applied to arterial spin labeling MRI may contribute to predict surgical revascularization outcomes in pediatric moyamoya vasculopathy. <i>Neuroradiology</i> , 2020, 62, 1003-1015.	1.1	11
101	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. <i>Genetics in Medicine</i> , 2021, 23, 1933-1943.	1.1	11
102	Spinal motor neuron involvement in a patient with homozygous PRUNE mutation. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 541-543.	0.7	10
103	White matter and cerebellar involvement in alternating hemiplegia of childhood. <i>Journal of Neurology</i> , 2020, 267, 1300-1311.	1.8	10
104	Homozygous missense <i>WIPI2</i> variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. <i>Brain Communications</i> , 2021, 3, fcab183.	1.5	10
105	Widening the Neuroimaging Features of Adenosine Deaminase 2 Deficiency. <i>American Journal of Neuroradiology</i> , 2021, 42, 975-979.	1.2	10
106	Cognitive and White Matter Microstructure Development in Congenital Hypothyroidism and Familial Thyroid Disorders. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e3990-e4006.	1.8	10
107	Idiopathic intervertebral disc calcification in childhood. <i>Archives of Disease in Childhood</i> , 2009, 94, 233-234.	1.0	9
108	Novel CNS malformations and skeletal anomalies in a patient with Beaulieu-Étneux syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2835-2840.	0.7	9

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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. <i>Cytogenetic and Genome Research</i> , 2018, 156, 14-21.	0.6	9
110	Novel compound heterozygous pathogenic variants in nucleotide-binding protein like protein (NUBPL) cause leukoencephalopathy with multi-systemic involvement. <i>Molecular Genetics and Metabolism</i> , 2020, 129, 26-34.	0.5	9
111	Targeted re-sequencing in pediatric and perinatal stroke. <i>European Journal of Medical Genetics</i> , 2020, 63, 104030.	0.7	9
112	Characteristic Cochlear Hypoplasia in Patients with Walker-Warburg Syndrome: A Radiologic Study of the Inner Ear in $\pm$ -Dystroglycan-Related Muscular Disorders. <i>American Journal of Neuroradiology</i> , 2021, 42, 167-172.	1.2	9
113	A novel homozygous MCOLN1 double mutant allele leading to TRP channel domain ablation underlies Mucopolipidosis IV in an Italian Child. <i>Metabolic Brain Disease</i> , 2015, 30, 681-686.	1.4	8
114	Interstitial de novo 18q22.3q23 deletion: clinical, neuroradiological and molecular characterization of a new case and review of the literature. <i>Molecular Cytogenetics</i> , 2016, 9, 78.	0.4	8
115	Unusual white matter involvement in EAST syndrome associated with novel KCNJ10 mutations. <i>Journal of Neurology</i> , 2018, 265, 1419-1425.	1.8	8
116	Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. <i>Neurogenetics</i> , 2019, 20, 165-172.	0.7	8
117	Clinical and neuroimaging features of the m.10197G>A mtDNA mutation: New case reports and expansion of the phenotype variability. <i>Journal of the Neurological Sciences</i> , 2019, 399, 69-75.	0.3	8
118	L1CAM variants cause two distinct imaging phenotypes on fetal MRI. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2004-2012.	1.7	8
119	Biallelic <i>ADAM22</i> pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. <i>Brain</i> , 2022, 145, 2301-2312.	3.7	8
120	Further genotype-phenotype correlation emerging from two families with PLP1 exon 4 skipping. <i>Clinical Genetics</i> , 2014, 85, 267-272.	1.0	7
121	Clinico-radiological and molecular characterization of a child with ring chromosome 2 presenting growth failure, microcephaly, kidney and brain malformations. <i>Molecular Cytogenetics</i> , 2015, 8, 17.	0.4	7
122	Torcular pseudomass: a potential diagnostic pitfall in infants and young children. <i>Pediatric Radiology</i> , 2017, 47, 227-234.	1.1	7
123	CASK related disorder: Epilepsy and developmental outcome. <i>European Journal of Paediatric Neurology</i> , 2021, 31, 61-69.	0.7	7
124	Ganglionic Eminence Anomalies and Coexisting Cerebral Developmental Anomalies on Fetal MR Imaging: Multicenter-Based Review of 60 Cases. <i>American Journal of Neuroradiology</i> , 2021, 42, 1151-1156.	1.2	7
125	Nosological Differences in the Nature of Punctate White Matter Lesions in Preterm Infants. <i>Frontiers in Neurology</i> , 2021, 12, 657461.	1.1	7
126	Successful urgent neurosurgery management with rFVIIa mega doses in a child with haemophilia A and high titre inhibitor. <i>Blood Coagulation and Fibrinolysis</i> , 2014, 25, 518-521.	0.5	6



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127	Clinical and Molecular Characterization of Two Patients with CNTN6 Copy Number Variations. <i>Cytogenetic and Genome Research</i> , 2018, 156, 144-149.	0.6	6
128	Asymmetric cavernous sinus enlargement: a novel finding in Sturge-Weber syndrome. <i>Neuroradiology</i> , 2019, 61, 595-602.	1.1	6
129	Perinatal Arterial Ischemic Stroke in Fetal Vascular Malperfusion: A Case Series and Literature Review. <i>American Journal of Neuroradiology</i> , 2020, 41, 2377-2383.	1.2	6
130	Cortical formation abnormalities on foetal MR imaging: a proposed classification system trialled on 356 cases from Italian and UK centres. <i>European Radiology</i> , 2020, 30, 5250-5260.	2.3	6
131	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. <i>Neurology</i> , 2021, 96, e1319-e1333.	1.5	6
132	An atypical case of post-varicella stroke in a child presenting with hemichorea followed by late-onset inflammatory focal cerebral arteriopathy. <i>Quantitative Imaging in Medicine and Surgery</i> , 2021, 11, 463-471.	1.1	6
133	A rare triad of morning glory disc anomaly, moyamoya vasculopathy, and transsphenoidal cephalocele: pathophysiological considerations and surgical management. <i>Neurological Sciences</i> , 2021, 42, 5433-5439.	0.9	6
134	Expanding the clinical and neuroimaging features of post-varicella arteriopathy of childhood. <i>Journal of Neurology</i> , 2021, 268, 4846-4865.	1.8	6
135	Epileptic encephalopathy caused by <i>ARV1</i> deficiency: Refinement of the genotype-phenotype spectrum and functional impact on <i>GPI</i> -anchored proteins. <i>Clinical Genetics</i> , 2021, 100, 607-614.	1.0	6
136	Radiological-Pathological Comparison in a Case of Conjoined Gnatho-Thoracopagus Twins. <i>Fetal Diagnosis and Therapy</i> , 2009, 26, 223-226.	0.6	5
137	MR Imaging of Neonatal Brain Infections. <i>Magnetic Resonance Imaging Clinics of North America</i> , 2011, 19, 761-775.	0.6	5
138	Spontaneously Regressing Leukoencephalopathy With Bilateral Temporal Cysts in Congenital Rubella Infection. <i>Pediatric Infectious Disease Journal</i> , 2014, 33, 422-424.	1.1	5
139	Teaching Neuro Images : Figure of 8. <i>Neurology</i> , 2017, 89, e172-e173.	1.5	5
140	Reversible cerebral vasoconstriction complicating cerebral atherosclerotic vascular disease in Schimke immuno-osseous dysplasia. <i>Neuroradiology</i> , 2018, 60, 885-888.	1.1	5
141	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. <i>European Radiology</i> , 2020, 30, 2161-2170.	2.3	5
142	Bivalirudin anticoagulation to overcome heparin resistance in a neonate with cerebral sinovenous thrombosis. <i>Blood Coagulation and Fibrinolysis</i> , 2020, 31, 97-100.	0.5	5
143	Placental Pathology Findings and the Risk of Intraventricular and Cerebellar Hemorrhage in Preterm Neonates. <i>Frontiers in Neurology</i> , 2020, 11, 761.	1.1	5
144	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7-related disorder. <i>Birth Defects Research</i> , 2020, 112, 1085-1092.	0.8	5

#	ARTICLE	IF	CITATIONS
145	Role of visual evoked potentials and optical coherence tomography in the screening for optic pathway gliomas in patients with neurofibromatosis type I. <i>European Journal of Ophthalmology</i> , 2021, 31, 698-703.	0.7	5
146	Photoparoxysmal response in ADCK3 autosomal recessive ataxia: a case report and literature review. <i>Epileptic Disorders</i> , 2021, 23, 153-160.	0.7	5
147	External ventricular drainage for posthemorrhagic ventricular dilatation in preterm infants: insights on efficacy and failure. <i>Journal of Neurosurgery: Pediatrics</i> , 2021, 28, 563-571.	0.8	5
148	Epilepsy in KAT6A syndrome: Description of two individuals and revision of the literature. <i>European Journal of Medical Genetics</i> , 2022, 65, 104380.	0.7	5
149	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. <i>Cerebellum</i> , 2022, , 1.	1.4	5
150	Noninvasive Assessment of Hemodynamic Stress Distribution after Indirect Revascularization for Pediatric Moyamoya Vasculopathy. <i>American Journal of Neuroradiology</i> , 2018, 39, 1157-1163.	1.2	4
151	Severe early-onset developmental and epileptic encephalopathy (DEE) associated with novel compound heterozygous pathogenic variants in SLC25A22: Case report and literature review. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 70, 56-58.	0.9	4
152	Cognitive Profiles and Brain Volume Are Affected in Patients with Silver-Russell Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e1478-e1488.	1.8	4
153	Congenital multifocal rhabdoid tumor: a case with peculiar biological behavior and different response to treatment according to location (central nervous system and kidney). <i>Cancer Genetics</i> , 2014, 207, 441-444.	0.2	3
154	Reversible cerebral vasoconstriction mimicking posterior reversible encephalopathy syndrome in an infant with end-stage renal disease. <i>Cephalalgia</i> , 2015, 35, 1031-1033.	1.8	3
155	Crossed Pontine Hemiatrophy Associated with Unilateral Cerebellar Hemorrhage in Premature Infants. <i>Neuropediatrics</i> , 2016, 47, 404-407.	0.3	3
156	Neonatal Developmental Venous Anomalies: Clinicoradiologic Characterization and Follow-Up. <i>American Journal of Neuroradiology</i> , 2020, 41, 2370-2376.	1.2	3
157	Endocrine Outcomes In Central Diabetes Insipidus: the Predictive Value of Neuroimaging "Mismatch Pattern". <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 3562-3574.	1.8	3
158	Balamuthia mandrillaris infection: report of 1st autochthonous, fatal case in Italy. <i>European Journal of Clinical Microbiology and Infectious Diseases</i> , 2022, 41, 685-687.	1.3	3
159	Calcifications in diffuse leptomeningeal glioneuronal tumors: a case series. <i>Quantitative Imaging in Medicine and Surgery</i> , 2022, 12, 2985-2994.	1.1	3
160	A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. <i>Frontiers in Pediatrics</i> , 2022, 10, 847549.	0.9	3
161	A 3-YEAR-OLD BOY WITH DRUG-RESISTANT COMPLEX PARTIAL SEIZURES. <i>Brain Pathology</i> , 2012, 22, 725-728.	1.1	2
162	Constitutional chromosomal events at 22q11 and 15q26 in a child with a pilocytic astrocytoma of the spinal cord. <i>Molecular Cytogenetics</i> , 2014, 7, 31.	0.4	2

#	ARTICLE	IF	CITATIONS
163	Idiopathic Cervical Hematomyelia in an Infant: Spinal Cord Injury without Radiographic Abnormality Caused by a Trivial Trauma? Case Report and Review of the Literature. <i>World Neurosurgery</i> , 2016, 90, 38-44.	0.7	2
164	Acute Communicating Hydrocephalus as Spinal Cord Surgery Complication in Patient with Lumbar Lipomyelocele. <i>World Neurosurgery</i> , 2018, 115, 468-472.e2.	0.7	2
165	Schimke immuno-osseous dysplasia, two new cases with peculiar EEG pattern. <i>Brain and Development</i> , 2020, 42, 408-413.	0.6	2
166	Multifactorial Posterior Reversible Encephalopathy Syndrome in Children: Clinical, Laboratory, and Neuroimaging Findings. <i>Journal of Pediatric Neurology</i> , 2021, 19, 083-091.	0.0	2
167	De novo POLR2A p.(Ile457Thr) variant associated with early-onset encephalopathy and cerebellar atrophy: expanding the phenotypic spectrum. <i>Brain and Development</i> , 2022, 44, 480-485.	0.6	2
168	Genotype-Phenotype Correlation and Functional Insights for Two Monoallelic TREX1 Missense Variants Affecting the Catalytic Core. <i>Genes</i> , 2022, 13, 1179.	1.0	2
169	Hypoxic Ischemic Encephalopathy in Term Neonates. , 0, , 15-16.		1
170	Cerebral blood flow in a case of typical aura without headache. <i>Journal of Neurology</i> , 2019, 266, 2869-2871.	1.8	1
171	Pelizaeusâ€™Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. <i>Neuropediatrics</i> , 2019, 50, 268-270.	0.3	1
172	Changes in appearance of cortical formation abnormalities in the foetus detected on sequential in utero MR imaging. <i>European Radiology</i> , 2021, 31, 1367-1377.	2.3	1
173	Combined medical therapy and neurosurgical revascularization preventing stroke in post-varicella angiopathy: Case report and review of literature. <i>Brain and Development</i> , 2021, 43, 1051-1056.	0.6	1
174	Malformations of Cortical Development. , 2021, , 1-237.		1
175	Diffusion Kurtosis Imaging of Neonatal Spinal Cord in Clinical Routine. <i>Frontiers in Radiology</i> , 2022, 2, .	1.2	1
176	Imaging characteristics and neurosurgical outcome in subjects with agenesis of the corpus callosum and interhemispheric cysts. <i>Neuroradiology</i> , 2022, 64, 2163-2177.	1.1	1
177	Microcephaly. , 0, , 193-194.		0
178	Septo-Optic Dysplasia. , 0, , 147-148.		0
179	Bilateral Perisylvian Polymicrogyria (BPP). , 0, , 37-38.		0
180	X-Linked Adrenoleukodystrophy (X-ALD). , 0, , 61-62.		0

#	ARTICLE	IF	CITATIONS
181	Glutaric Aciduria Type 1. , 0, , 33-34.		0
182	Lissencephaly. , 0, , 39-40.		0
183	Megalencephalic Leukoencephalopathy with Subcortical Cysts. , 0, , 47-48.		0
184	Alexander Disease. , 0, , 67-68.		0
185	Gangliosidosis GM2. , 0, , 19-20.		0
186	Leigh Disease. , 0, , 21-22.		0
187	Active Multiple Sclerosis. , 0, , 257-258.		0
188	Characterization of the Phenotype Associated with Microduplication Reciprocal to NF1 Microdeletion Syndrome. Cytogenetic and Genome Research, 2017, 152, 22-28.	0.6	0
189	The features of the m.10197G>A mtDNA mutation. Journal of the Neurological Sciences, 2019, 400, 184-185.	0.3	0
190	Pediatric Brain Tissue Segmentation from MRI using Clustering: a Preliminary Study. , 2019, 2019, 6557-6560.		0
191	Peculiar bony involvement of sinus pericranii in children: Extensive diploic erosion in three "karstic" variants. Clinical Neurology and Neurosurgery, 2022, 219, 107334.	0.6	0
192	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