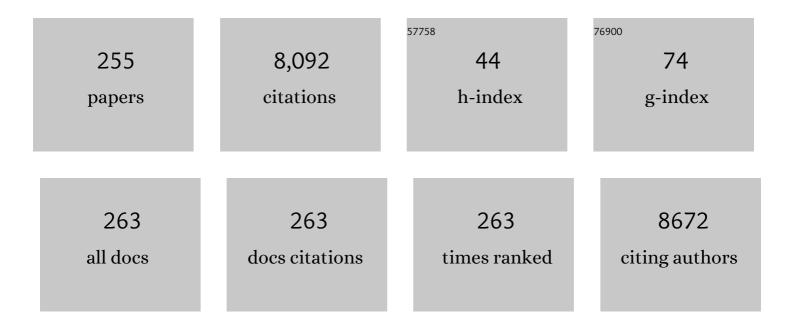
List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	<i>SUFU</i> haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	3.2	19
2	Xq22 deletions and correlation with distinct neurological disease traits in females: Further evidence for a contiguous gene syndrome. Human Mutation, 2020, 41, 150-168.	2.5	15
3	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
4	Inside Back Cover, Volume 41, Issue 1. Human Mutation, 2020, 41, ii.	2.5	0
5	The spectrum of brainstem malformations associated to mutations of the tubulin genes family: MRI and DTI analysis. European Radiology, 2019, 29, 770-782.	4.5	22
6	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	61
7	Characterization of the Basal Ganglia Using Diffusion Tensor Imaging in Children with Selfâ€Injurious Behavior and Tuberous Sclerosis Complex. Journal of Neuroimaging, 2019, 29, 506-511.	2.0	5
8	Correlation Between White Matter Injury Identified by Neonatal Diffusion Tensor Imaging and Neurodevelopmental Outcomes Following Term Neonatal Asphyxia and Therapeutic Hypothermia: An Exploratory Pilot Study. Journal of Child Neurology, 2019, 34, 556-566.	1.4	11
9	PLA2G6-associated neurodegeneration: New insights into brain abnormalities and disease progression. Parkinsonism and Related Disorders, 2019, 61, 179-186.	2.2	41
10	Persistent Extreme Hyperextension of the Fetal Neck: Clinical and Neuroimaging Findings. Journal of Neuroimaging, 2018, 28, 278-282.	2.0	3
11	Congenital Neuronal Ceroid Lipofuscinosis with a Novel CTSD Gene Mutation: A Rare Cause of Neonatal-Onset Neurodegenerative Disorder. Neuropediatrics, 2018, 49, 150-153.	0.6	15
12	Blake's pouch cyst in children: Atypical clinical presentation. Neuroradiology Journal, 2018, 31, 430-433.	1.2	6
13	White Matter Anatomy. , 2018, , 35-44.		0
14	Rhombencephalosynapsis: Fused cerebellum, confused geneticists. , 2018, 178, 432-439.		26
15	Arrested Hydrocephalus in Childhood: Case Series and Review of the Literature. Neuropediatrics, 2018, 49, 302-309.	0.6	14
16	Genetics of cerebellar disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 154, 267-286.	1.8	3
17	Congenital Malformations of the Central Nervous System. , 2018, , 857-878.e5.		4
18	Cerebellar Microstructural Organization is Altered by Complications of Premature Birth: A Case-Control Study. Journal of Pediatrics, 2017, 182, 28-33.e1.	1.8	30

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19	Neuroimaging findings of congenital Zika virus infection: a pictorial essay. Japanese Journal of Radiology, 2017, 35, 89-94.	2.4	44
20	Early-onset head titubation in a child with Poretti-Boltshauser syndrome. Neurology, 2017, 88, 1478-1479.	1.1	5
21	MR Imaging of Acute Cerebellar Involvement in Pediatric Anti-N-Methyl-D-Aspartate Receptor Encephalitis. Neuropediatrics, 2017, 48, 398-399.	0.6	0
22	Joubert syndrome: neuroimaging findings in 110 patients in correlation with cognitive function and genetic cause. Journal of Medical Genetics, 2017, 54, 521-529.	3.2	53
23	Optimizing Cerebral Autoregulation May Decrease Neonatal Regional Hypoxic-Ischemic Brain Injury. Developmental Neuroscience, 2017, 39, 248-256.	2.0	59
24	Neuropsychological phenotypes of 76 individuals with Joubert syndrome evaluated at a single center. American Journal of Medical Genetics, Part A, 2017, 173, 1796-1812.	1.2	26
25	Neuroimaging findings of postnatally acquired Zika virus infection: a pictorial essay. Japanese Journal of Radiology, 2017, 35, 341-349.	2.4	22
26	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36.	6.2	74
27	Prenatal Brainstem Disruptions: Small Lesions–Big Problems. Neuropediatrics, 2017, 48, 350-355.	0.6	1
28	ACCIDENTAL DUPLICATION: MR Imaging Findings in Children with Spasmus Nutans. Journal of AAPOS, 2017, , .	0.3	0
29	Back pain and scoliosis in children: When to image, what to consider. Neuroradiology Journal, 2017, 30, 393-404.	1.2	21
30	Acute brain injury following illicit drug abuse in adolescent and young adult patients: spectrum of neuroimaging findings. Neuroradiology Journal, 2017, 30, 144-150.	1.2	21
31	Magnetic resonance imaging to diagnose leptomeningeal spread of medulloblastoma in children: Decreasing diagnostic uncertainty, moving in new directions. Pediatric Blood and Cancer, 2017, 64, e26514.	1.5	2
32	Longitudinal volumetric and 2D assessment of cerebellar atrophy in a large cohort of children with phosphomannomutase deficiency (PMM2 DG). Journal of Inherited Metabolic Disease, 2017, 40, 709-713.	3.6	16
33	The structural connectome in children: basic concepts, how to build it, and synopsis of challenges for the developing pediatric brain. Neuroradiology, 2017, 59, 445-460.	2.2	14
34	Compound Heterozygous Variants in ROBO1 Cause a Neurodevelopmental Disorder With Absence of Transverse Pontine Fibers and Thinning of the Anterior Commissure and Corpus Callosum. Pediatric Neurology, 2017, 70, 70-74.	2.1	16
35	Magnetic resonance imaging findings in children with spasmus nutans. Journal of AAPOS, 2017, 21, 127-130.	0.3	8
36	Neuroimaging Findings in Pediatric Genetic Skeletal Disorders: A Review. Journal of Neuroimaging, 2017, 27, 162-209.	2.0	6

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37	Middle Cerebellar Peduncles T2-Hyperintense Signal in Fazio–Londe Syndrome. Neuropediatrics, 2017, 48, 396-397.	0.6	0
38	Implementing a Multidisciplinary Approach to Treating Tuberous Sclerosis Complex. Child Neurology Open, 2017, 4, 2329048X1772560.	1.1	4
39	In Reply. Obstetrics and Gynecology, 2017, 129, 389-390.	2.4	0
40	Novel Contrastâ€Enhanced Ultrasound Evaluation in Neonatal Hypoxic Ischemic Injury: Clinical Application and Future Directions. Journal of Ultrasound in Medicine, 2017, 36, 2379-2386.	1.7	42
41	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
42	Pitfalls in Diffusion-Weighted and Diffusion Tensor Imaging of the Pediatric Brain. Neuropediatrics, 2017, 48, 340-349.	0.6	8
43	Sensitivity of susceptibility-weighted imaging in detecting developmental venous anomalies and associated cavernomas and microhemorrhages in children. Neuroradiology, 2017, 59, 797-802.	2.2	21
44	Neuroimaging findings of Zika virus infection: emphasis of congenital versus acquired aspects. Japanese Journal of Radiology, 2017, 35, 41-42.	2.4	8
45	Deregulated expression of <i>EZH2</i> in congenital brainstem disconnection. Neuropathology and Applied Neurobiology, 2017, 43, 358-365.	3.2	7
46	Tumor and Tumorlike Masses in Pediatric Patients that Involve Multiple Spaces. Neuroimaging Clinics of North America, 2017, 27, 135-153.	1.0	0
47	Further evidence that <i>de novo</i> missense and truncating variants in <i><scp>ZBTB18</scp></i> cause intellectual disability with variable features. Clinical Genetics, 2017, 91, 697-707.	2.0	29
48	Diffusion Tensor Imaging (DTI)â~†. , 2017, , .		5
49	Diffusion Tensor Imaging and Fiber Tractography of Pediatric Posterior Fossa Malformations. Neurographics, 2017, 7, 243-261.	0.2	1
50	Disorders of Cerebellar and Brainstem Development. , 2017, , 199-207.		0
51	Radiological findings of abnormalities associated with congenital Zika virus infection: conclusions from World Radiology Day 2016. Revista Panamericana De Salud Publica/Pan American Journal of Public Health, 2017, 41, 1-6.	1.1	2
52	Trauma. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2016, 136, 1199-1220.	1.8	6
53	Neurocognitive Functions and Behavior in Joubert Syndrome. Pediatric Neurology Briefs, 2016, 30, 47.	0.2	4
54	Histogram Analysis of Diffusion Tensor Imaging Parameters in Pediatric Cerebellar Tumors. Journal of Neuroimaging, 2016, 26, 360-365.	2.0	19

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55	<i>BRAT1</i> mutations present with a spectrum of clinical severity. American Journal of Medical Genetics, Part A, 2016, 170, 2265-2273.	1.2	34
56	Does superior caval vein pressure impact head growth in Fontan circulation?. Cardiology in the Young, 2016, 26, 1327-1332.	0.8	1
57	Cerebellar disruptions and neurodevelopmental disabilities. Seminars in Fetal and Neonatal Medicine, 2016, 21, 339-348.	2.3	20
58	Computed Tomographic Findings in Microcephaly Associated with Zika Virus. New England Journal of Medicine, 2016, 374, 2193-2195.	27.0	218
59	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Poretti–Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267.	2.8	43
60	The genetics of cerebellar malformations. Seminars in Fetal and Neonatal Medicine, 2016, 21, 321-332.	2.3	47
61	Cerebellar Bottom-of-Fissure Dysplasia—a Novel Cerebellar Gray Matter Neuroimaging Pattern. Cerebellum, 2016, 15, 705-709.	2.5	5
62	SCN8A Epileptic Encephalopathy: Detection of Fetal Seizures Guides Multidisciplinary Approach to Diagnosis and Treatment. Pediatric Neurology, 2016, 64, 87-91.	2.1	13
63	Neuroimaging findings of Zika virus infection: a review article. Japanese Journal of Radiology, 2016, 34, 765-770.	2.4	314
64	Neuroimaging Findings in Congenital Zika Syndrome. American Journal of Neuroradiology, 2016, 37, 1764-1765.	2.4	7
65	Prenatal Cerebellar Disruptions. Neuroimaging Clinics of North America, 2016, 26, 359-372.	1.0	24
66	Cerebellar and Brainstem Malformations. Neuroimaging Clinics of North America, 2016, 26, 341-357.	1.0	30
67	Pediatric Neurocutaneous Syndromes with Cerebellar Involvement. Neuroimaging Clinics of North America, 2016, 26, 417-434.	1.0	5
68	Acute Brain Imaging in Children: Can MRI Replace CT as a Screening Tool?. Journal of Neuroimaging, 2016, 26, 68-74.	2.0	10
69	The Pediatric Cerebellum. Neuroimaging Clinics of North America, 2016, 26, xiii-xiv.	1.0	1
70	Neuroimaging findings in acute pediatric diabetic ketoacidosis. Neuroradiology Journal, 2016, 29, 317-322.	1.2	15
71	Case 236: Middle Interhemispheric Variant of Holoprosencephaly. Radiology, 2016, 281, 969-974.	7.3	2
72	Diffusion tensor imaging: A biomarker of outcome in <scp>K</scp> rabbe's disease. Journal of Neuroscience Research, 2016, 94, 1108-1115.	2.9	16

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73	Case 236. Radiology, 2016, 280, 640-642.	7.3	0
74	Mutations in <i>CEP120</i> cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	3.2	55
75	Diffusion Tractography Biomarkers of Pediatric Cerebellar Hypoplasia/Atrophy: Preliminary Results Using Constrained Spherical Deconvolution. American Journal of Neuroradiology, 2016, 37, 917-923.	2.4	8
76	Brain injury in neonatal incontinentia pigmenti: the role of multimodality neuroimaging. Developmental Medicine and Child Neurology, 2016, 58, 1000-1001.	2.1	5
77	Spinal Nerve Root Enhancement on MRI Scans in Children: A Review. Journal of Neuroimaging, 2016, 26, 169-179.	2.0	10
78	The Role of ADCâ€Based Thermometry in Measuring Brain Intraventricular Temperature in Children. Journal of Neuroimaging, 2016, 26, 315-323.	2.0	3
79	Clinical and neuroimaging features as diagnostic guides in neonatal neurology diseases with cerebellar involvement. Cerebellum and Ataxias, 2016, 3, 1.	1.9	24
80	Chiari Type 1 Deformity in Children: Pathogenetic, Clinical, Neuroimaging, and Management Aspects. Neuropediatrics, 2016, 47, 293-307.	0.6	48
81	Chiari type 1 malformation, corpus callosum agenesis and patent craniopharyngeal canal in an 11-year-old boy. Neuroradiology Journal, 2016, 29, 307-309.	1.2	3
82	Cystic cerebellar dysplasia and biallelic <i>LAMA1</i> mutations: a lamininopathy associated with tics, obsessive compulsive traits and myopia due to cell adhesion and migration defects. Journal of Medical Genetics, 2016, 53, 318-329.	3.2	25
83	Cerebral Reorganization after Hemispherectomy: A DTI Study. American Journal of Neuroradiology, 2016, 37, 924-931.	2.4	17
84	Pseudotumoral hemicerebellitis as a mimicker of Lhermitte-Duclos disease in children: does neuroimaging help to differentiate them?. Child's Nervous System, 2016, 32, 865-871.	1.1	7
85	Cystic Malformations Within the Posterior Fossa. Current Radiology Reports, 2016, 4, 1.	1.4	3
86	Value of Susceptibility-Weighted Imaging in the Evaluation of Altered Brain Perfusion in Children. Neuropediatrics, 2016, 47, 003-004.	0.6	10
87	Diffusion tensor imaging suggests extrapontine extension of pediatric diffuse intrinsic pontine gliomas. European Journal of Radiology, 2016, 85, 700-706.	2.6	10
88	Chiari Malformations and Syringohydromyelia in Children. Seminars in Ultrasound, CT and MRI, 2016, 37, 129-142.	1.5	11
89	Differential diagnosis of ventriculomegaly and brainstem kinking on fetal MRI. Brain and Development, 2016, 38, 103-108.	1.1	22
90	Transfontanellar duplex brain ultrasonography resistive indices as a prognostic tool in neonatal hypoxic-ischemic encephalopathy before and after treatment with therapeutic hypothermia. Journal of Perinatology, 2016, 36, 202-206.	2.0	28

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91	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72.	3.2	48
92	Temporal bone and cranial nerve findings in pontine tegmental cap dysplasia. Neuroradiology, 2016, 58, 179-187.	2.2	17
93	Pre- and Postnatal Neuroimaging of Congenital Cerebellar Abnormalities. Cerebellum, 2016, 15, 5-9.	2.5	27
94	Pediatric Neurovascular Imaging (CT/MRI/Ultrasound). , 2016, , 77-109.		1
95	Overview of Ataxias in Children. , 2016, , 531-538.		4
96	Brain malformations and fetal ventriculomegaly: What to look for?. Journal of Pediatric Neuroradiology, 2015, 01, 185-195.	0.1	2
97	"Tumor-like―lesions of the pediatric brain. Journal of Pediatric Neuroradiology, 2015, 01, 261-267.	0.1	0
98	Terminology in morphological anomalies of the cerebellum does matter. Cerebellum and Ataxias, 2015, 2, 8.	1.9	28
99	Impaired Growth and Abnormal Microstructure of Supratentorial Gray and White Matter Regions in a Child with Joubert Syndrome. Neurographics, 2015, 5, 209-216.	0.1	3
100	<i>KIAA0586</i> is Mutated in Joubert Syndrome. Human Mutation, 2015, 36, 831-835.	2.5	62
101	Parturitional Injury of the Head and Neck. Journal of Neuroimaging, 2015, 25, 151-166.	2.0	18
102	Cerebral sinovenous thrombosis in a child with Crohn's disease, otitis media, and meningitis. Neuroradiology Journal, 2015, 28, 274-277.	1.2	8
103	Differential Diagnosis of Cerebellar Atrophy in Childhood: An Update. Neuropediatrics, 2015, 46, 359-370.	0.6	39
104	Fetal Diagnosis of Rhombencephalosynapsis. Neuropediatrics, 2015, 46, 357-358.	0.6	11
105	T1 Hyperintensity of the Pediatric Neural Axis: What to Consider?. Journal of Pediatrics, 2015, 167, 497.497.e1.	1.8	0
106	Cerebellar Cysts in Children: a Pattern Recognition Approach. Cerebellum, 2015, 14, 308-316.	2.5	23
107	Longitudinally extensive myelopathy in children. Pediatric Radiology, 2015, 45, 244-257.	2.0	11
108	Congenital Abnormalities of the Posterior Fossa. Radiographics, 2015, 35, 200-220.	3.3	152

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109	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	3.8	30
110	Brainstem Disconnection: Two Additional Patients and Expansion of the Phenotype. Neuropediatrics, 2015, 46, 139-144.	0.6	9
111	Susceptibility-Weighted Imaging in Pediatric Arterial Ischemic Stroke: A Valuable Alternative for the Noninvasive Evaluation of Altered Cerebral Hemodynamics. American Journal of Neuroradiology, 2015, 36, 783-788.	2.4	30
112	Retinal Hemorrhages in Nonaccidental Trauma: Look at Susceptibility-Weighted Imaging on Pediatric MRI. Pediatric Neurology, 2015, 52, 464-465.	2.1	4
113	Structural connectivity analysis reveals abnormal brain connections in agenesis of the corpus callosum in children. European Radiology, 2015, 25, 1471-1478.	4.5	17
114	Neuroimaging findings in pediatric cerebral sinovenous thrombosis. Child's Nervous System, 2015, 31, 705-712.	1.1	19
115	Conventional and advanced (DTI/SWI) neuroimaging findings in pediatric oligodendroglioma. Child's Nervous System, 2015, 31, 885-891.	1.1	12
116	Cerebellar Hypoplasia and Dysmorphia in Neurofibromatosis Type 1. Cerebellum, 2015, 14, 642-649.	2.5	9
117	Prenatal Cerebellar Hemorrhage: Fetal and Postnatal Neuroimaging Findings and Postnatal Outcome. Pediatric Neurology, 2015, 52, 529-534.	2.1	19
118	Vein of Galen Aneurysmal Malformation: Prognostic Markers Depicted on Fetal MR I. Neuroradiology Journal, 2015, 28, 71-75.	1.2	28
119	Pediatric Cerebellar Tumors: Does ADC Analysis of Solid, Contrastâ€Enhancing Tumor Components Correlate Better with Tumor Grade than ADC Analysis of the Entire Tumor?. Journal of Neuroimaging, 2015, 25, 785-791.	2.0	7
120	Pediatric skull fracture diagnosis: should 3D CT reconstructions be added as routine imaging?. Journal of Neurosurgery: Pediatrics, 2015, 16, 426-431.	1.3	60
121	Cerebellar agenesis: An extreme form of cerebellar disruption in preterm neonates. Journal of Pediatric Neuroradiology, 2015, 02, 163-167.	0.1	6
122	Apparent Diffusion Coefficient Scalars Correlate with Near-Infrared Spectroscopy Markers of Cerebrovascular Autoregulation in Neonates Cooled for Perinatal Hypoxic-Ischemic Injury. American Journal of Neuroradiology, 2015, 36, 188-193.	2.4	45
123	Autism Phenotypes in Tuberous Sclerosis Complex. Journal of Child Neurology, 2015, 30, 1871-1876.	1.4	6
124	Neurometabolic diseases of childhood. Pediatric Radiology, 2015, 45, 473-484.	2.0	23
125	Neuroimaging Findings in Encephalocraniocutaneous Lipomatosis. Pediatric Neurology, 2015, 53, 462-463.	2.1	5
126	Pediatric Spinal Trauma. Journal of Neuroimaging, 2015, 25, 337-353.	2.0	33

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127	Achondroplasia in children: correlation of ventriculomegaly, size of foramen magnum and jugular foramina, and emissary vein enlargement. Child's Nervous System, 2015, 31, 129-133.	1.1	30
128	Neonatal Head Ultrasonography Today: A Powerful Imaging Tool!. Journal of Neuroimaging, 2015, 25, 31-55.	2.0	29
129	Moyamoya disease and syndrome in children: Spectrum of neuroimaging findings including differential diagnosis. Journal of Pediatric Neuroradiology, 2015, 03, 003-012.	0.1	0
130	Multi-Contrast Multi-Atlas Parcellation of Diffusion Tensor Imaging of the Human Brain. PLoS ONE, 2014, 9, e96985.	2.5	55
131	Neonatal neuroimaging findings in congenital myotonic dystrophy. Journal of Perinatology, 2014, 34, 159-160.	2.0	9
132	Christianson Syndrome: Spectrum of Neuroimaging Findings. Neuropediatrics, 2014, 45, 247-251.	0.6	22
133	Diffusion Tensor Imaging of Neurofibromatosis Bright Objects in Children with Neurofibromatosis Type 1. Neuroradiology Journal, 2014, 27, 616-626.	1.2	12
134	Hypertrophic olivary degeneration in a child following midbrain tumor resection: longitudinal diffusion tensor imaging studies. Journal of Neurosurgery: Pediatrics, 2014, 13, 408-413.	1.3	9
135	Congenital Brain Abnormalities: An Update on Malformations of Cortical Development and Infratentorial Malformations. Seminars in Neurology, 2014, 34, 239-248.	1.4	12
136	Horizontal head titubation in infants with <scp>J</scp> oubert syndrome: a new finding. Developmental Medicine and Child Neurology, 2014, 56, 1016-1020.	2.1	12
137	A Diagnostic Approach for Cerebral Palsy in the Genomic Era. NeuroMolecular Medicine, 2014, 16, 821-844.	3.4	89
138	Will a novel semiâ€quantitative scale for classification of structural brain <scp>MRI</scp> improve patient care and research in children with cerebral palsy?. Developmental Medicine and Child Neurology, 2014, 56, 801-802.	2.1	0
139	Pediatric hemiplegic migraine: Role of multiple MRI techniques in evaluation of reversible hypoperfusion. Cephalalgia, 2014, 34, 311-315.	3.9	33
140	Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease?. Cerebellum, 2014, 13, 79-88.	2.5	50
141	Neuroimaging Findings in Pediatric Propofol Infusion Syndrome. Pediatric Neurology, 2014, 50, 431-432.	2.1	5
142	Mutations in CSPP1 Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2014, 94, 62-72.	6.2	104
143	Anti-Ma2-Associated Paraneoplastic Encephalitis in a Male Adolescent With Mediastinal Seminoma. Pediatric Neurology, 2014, 50, 433-434.	2.1	14
144	Scout view in pediatric CT neuroradiological evaluation: do not underestimate!. Child's Nervous System, 2014, 30, 307-311.	1.1	10

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145	Neuroimaging findings of retroclival hemorrhage in children: a diagnostic conundrum. Child's Nervous System, 2014, 30, 835-839.	1.1	25
146	Posttraumatic Carotid Artery Dissection in Children: Not to be missed!. Journal of Neuroimaging, 2014, 24, 467-472.	2.0	22
147	Novel <i>TUBB4A</i> mutations and expansion of the neuroimaging phenotype of hypomyelination with atrophy of the basal ganglia and cerebellum (Hâ€ABC). American Journal of Medical Genetics, Part A, 2014, 164, 1802-1807.	1.2	27
148	Cerebellar hypoplasia: Differential diagnosis and diagnostic approach. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 211-226.	1.6	107
149	Pitfalls in Susceptibilityâ€Weighted Imaging of the Pediatric Brain. Journal of Neuroimaging, 2014, 24, 221-225.	2.0	21
150	Diffusion tensor imaging in pediatric <scp>C</scp> hiari type <scp>I</scp> malformation. Developmental Medicine and Child Neurology, 2014, 56, 742-748.	2.1	24
151	Diffusion Tensor Imaging for Brain Malformations. Neuroimaging Clinics of North America, 2014, 24, 619-637.	1.0	15
152	Early Predictive Value of Susceptibility Weighted Imaging (SWI) in Pediatric Hypoxicâ€Ischemic Injury. Journal of Neuroimaging, 2014, 24, 528-530.	2.0	8
153	Susceptibility-weighted imaging in pediatric neuroimaging. Journal of Magnetic Resonance Imaging, 2014, 40, 530-544.	3.4	33
154	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. American Journal of Human Genetics, 2014, 95, 227-234.	6.2	92
155	Neuroimaging findings in children with Keutel syndrome. Pediatric Radiology, 2014, 44, 73-78.	2.0	9
156	Clinicoradiological correlation of scoliosis in children with Jarcho-Levin and Escobar syndromes: associated "flat bone or wing-like―imaging findings. European Journal of Pediatrics, 2014, 173, 1377-1380.	2.7	3
157	Subthalamic nucleus involvement in children: AÂneuroimaging pattern-recognition approach. European Journal of Paediatric Neurology, 2014, 18, 249-256.	1.6	7
158	A detailed analysis of methylmalonic acid kinetics during hemodialysis and after combined liver/kidney transplantation in a patient with <i>mut</i> ⁰ methylmalonic acidemia. Journal of Inherited Metabolic Disease, 2014, 37, 899-907.	3.6	40
159	Undecussated Superior Cerebellar Peduncles and Absence of the Dorsal Transverse Pontine Fibers: a New Axonal Guidance Disorder?. Cerebellum, 2014, 13, 536-540.	2.5	7
160	Benign frontal horn cysts in a preterm neonate. Journal of Perinatology, 2014, 34, 488-488.	2.0	2
161	Evaluation of the ischemic penumbra focusing on the venous drainage: The role of susceptibility weighted imaging (SWI) in pediatric ischemic cerebral stroke. Journal of Neuroradiology, 2014, 41, 108-116.	1.1	38
162	Mucopolysaccharidoses type I and II: New neuroimaging findings in the cerebellum. European Journal of Paediatric Neurology, 2014, 18, 211-217.	1.6	26

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163	Novel diffusion tensor imaging findings in Krabbe disease. European Journal of Paediatric Neurology, 2014, 18, 150-156.	1.6	24
164	Nationwide survey of Arima syndrome: A note of doubt. Brain and Development, 2014, 36, 88.	1.1	0
165	Early Neurodevelopmental Screening in Tuberous Sclerosis Complex: A Potential Window of Opportunity. Pediatric Neurology, 2014, 51, 398-402.	2.1	7
166	Susceptibility-Weighted Imaging in Pantothenate Kinase–Associated Neurodegeneration. Journal of Pediatrics, 2014, 164, 212.	1.8	4
167	Diffusion tensor imaging of the brainstem in children with achondroplasia. Developmental Medicine and Child Neurology, 2014, 56, 1085-1092.	2.1	7
168	Susceptibility-Weighted Imaging for Calcification in Cockayne Syndrome. Journal of Pediatrics, 2014, 165, 416-416.e1.	1.8	2
169	The Molar Tooth Sign Is Pathognomonic for Joubert Syndrome!. Pediatric Neurology, 2014, 50, e15-e16.	2.1	34
170	Oropharyngeal Dermoid Cyst in an Infant with Intermittent Airway Obstruction. Neuroradiology Journal, 2014, 27, 627-631.	1.2	4
171	Neonatal neuroimaging findings in inborn errors of metabolism. Journal of Magnetic Resonance Imaging, 2013, 37, 294-312.	3.4	63
172	Diffusion tensor imaging and fiber tractography in brain malformations. Pediatric Radiology, 2013, 43, 28-54.	2.0	71
173	Diffusion Tensor Imaging in a Child with Hypertrophic Olivary Degeneration. Cerebellum, 2013, 12, 469-474.	2.5	14
174	Chiari II Malformation and Syntelencephaly in a Young Woman: Coincidence or Pathogenetic Association?. Clinical Neuroradiology, 2013, 23, 319-321.	1.9	1
175	Everolimus and intensive behavioral therapy in an adolescent with tuberous sclerosis complex and severe behavior. Epilepsy & Behavior Case Reports, 2013, 1, 122-125.	1.5	11
176	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
177	A neuroimaging patternâ€recognition approach in pediatric intracranial calcifications. Developmental Medicine and Child Neurology, 2013, 55, 7-8.	2.1	1
178	Midbrain and hindbrain malformations: advances in clinical diagnosis, imaging, and genetics. Lancet Neurology, The, 2013, 12, 381-393.	10.2	110
179	Serial Fetal MRI for the Diagnosis of Aicardi Syndrome. Neuroradiology Journal, 2013, 26, 380-384.	1.2	7
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