

# Nathalie Boddaert

## List of Publications by Year in descending order

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132  
papers

3,799  
citations

159358

30  
h-index

168136

53  
g-index

144  
all docs

144  
docs citations

144  
times ranked

6466  
citing authors

#	ARTICLE	IF	CITATIONS
1	Arterial Spin Labeling for the Etiological Workup of Intracerebral Hemorrhage in Children. <i>Stroke</i> , 2022, 53, 185-193.	1.0	6
2	A malignant choroid plexus tumour with prevailing immature blastematos elements. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	1
3	Deciphering the genetic and epigenetic landscape of pediatric bithalamic tumors. <i>Brain Pathology</i> , 2022, 32, e13039.	2.1	5
4	Treatment of two infants with PIK3CA-related overgrowth spectrum by alpelisib. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	27
5	A novel LARGE1-AFF2 fusion expanding the molecular alterations associated with the methylation class of neuroepithelial tumors with PÄTZ1 fusions. <i>Acta Neuropathologica Communications</i> , 2022, 10, 15.	2.4	3
6	A partial form of inherited human USP18 deficiency underlies infection and inflammation. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	28
7	Spectrum of Neuroradiologic Findings Associated with Monogenic Interferonopathies. <i>American Journal of Neuroradiology</i> , 2022, 43, 2-10.	1.2	6
8	Feasibility and Added Value of Fetal DTI Tractography in the Evaluation of an Isolated Short Corpus Callosum: Preliminary Results. <i>American Journal of Neuroradiology</i> , 2022, 43, 132-138.	1.2	7
9	Imaging Features with Histopathologic Correlation of CNS High-Grade Neuroepithelial Tumors with a <i>BCOR</i> Internal Tandem Duplication. <i>American Journal of Neuroradiology</i> , 2022, 43, 151-156.	1.2	17
10	Suleiman-El-Hattab syndrome: a histone modification disorder caused by TASP1 deficiency. <i>Human Molecular Genetics</i> , 2022, 31, 3083-3094.	1.4	3
11	Neuroinflammatory Disease following Severe Acute Respiratory Syndrome Coronavirus 2 Infection in Children. <i>Journal of Pediatrics</i> , 2022, 247, 22-28.e2.	0.9	15
12	The genomic landscape of dysembryoplastic neuroepithelial tumours and a comprehensive analysis of recurrent cases. <i>Neuropathology and Applied Neurobiology</i> , 2022, 48, .	1.8	4
13	Imaging features of medulloblastoma: Conventional imaging, diffusion-weighted imaging, perfusion-weighted imaging, and spectroscopy: From general features to subtypes and characteristics. <i>Neurochirurgie</i> , 2021, 67, 6-13.	0.6	19
14	Cerebral blood flow and acute episodes of Leigh syndrome in neurometabolic disorders. <i>Developmental Medicine and Child Neurology</i> , 2021, 63, 705-711.	1.1	6
15	Arterial abnormalities identified in kidneys transplanted into children during the COVID-19 pandemic. <i>American Journal of Transplantation</i> , 2021, 21, 1937-1943.	2.6	3
16	Hemorrhage Expansion After Pediatric Intracerebral Hemorrhage. <i>Stroke</i> , 2021, 52, 588-594.	1.0	4
17	Fast-track virtual reality for cardiac imaging in congenital heart disease. <i>Journal of Cardiac Surgery</i> , 2021, 36, 2598-2602.	0.3	21
18	Evolution of acute myocarditis in a pediatric population: An MRI based study. <i>International Journal of Cardiology</i> , 2021, 329, 226-233.	0.8	10

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19	Acute surgical management of children with ruptured brain arteriovenous malformation. <i>Journal of Neurosurgery: Pediatrics</i> , 2021, 27, 437-445.	0.8	2
20	Topographic variability of the normal circle of Willis anatomy on a paediatric population. <i>Brain Communications</i> , 2021, 3, fcab055.	1.5	0
21	Inherited deficiency of stress granule ZNFX1 in patients with monocytosis and mycobacterial disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	47
22	Radiogenomics of diffuse intrinsic pontine gliomas (DIPGs): correlation of histological and biological characteristics with multimodal MRI features. <i>European Radiology</i> , 2021, 31, 8913-8924.	2.3	11
23	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	5.8	28
24	Pineal alveolar rhabdomyosarcoma with PAX3:NCOA2 fusion inducing OLIG2 expression, a potential pitfall in the central nervous system. <i>Histopathology</i> , 2021, 79, 437-439.	1.6	3
25	A novel case of cribriform neuroepithelial tumor: A potential diagnostic pitfall in the ventricular system. <i>Pediatric Blood and Cancer</i> , 2021, 68, e29037.	0.8	3
26	A CBF decrease in the left supplementary motor areas: New insight into postoperative pediatric cerebellar mutism syndrome using arterial spin labeling perfusion MRI. <i>Journal of Cerebral Blood Flow and Metabolism</i> , 2021, 41, 3339-3349.	2.4	10
27	Fatal encephalitis caused by Newcastle disease virus in a child. <i>Acta Neuropathologica</i> , 2021, 142, 605-608.	3.9	9
28	Brain perfusion magnetic resonance imaging using pseudocontinuous arterial spin labeling in 314 dogs and cats. <i>Journal of Veterinary Internal Medicine</i> , 2021, 35, 2327-2341.	0.6	3
29	Alternative pathways for the development of lymphoid structures in humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	11
30	Pediatric brain arteriovenous malformation recurrence: a cohort study, systematic review and meta-analysis. <i>Journal of NeuroInterventional Surgery</i> , 2021, , neurintsurg-2021-017777.	2.0	10
31	Prenatal onset of congenital neuronal ceroid lipofuscinosis with a novel CTSD mutation. <i>Birth Defects Research</i> , 2021, 113, 1324-1332.	0.8	1
32	Variants in the MIPEP gene presenting with complex neurological phenotype without cardiomyopathy, impair OXPHOS protein maturation and lead to a reduced OXPHOS abundance in patient cells. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 267-273.	0.5	4
33	Rest Functional Brain Maturation during the First Year of Life. <i>Cerebral Cortex</i> , 2021, 31, 1776-1785.	1.6	11
34	Object Detection Improves Tumour Segmentation in MR Images of Rare Brain Tumours. <i>Cancers</i> , 2021, 13, 6113.	1.7	9
35	Prevalence of Venovenous Shunting and High-Output State Quantified with 4D Flow MRI in Patients with Fontan Circulation. <i>Radiology: Cardiothoracic Imaging</i> , 2021, 3, e210161.	0.9	5
36	Myocardial involvement in children with post-COVID multisystem inflammatory syndrome: a cardiovascular magnetic resonance based multicenter international study—the CARDOVID registry. <i>Journal of Cardiovascular Magnetic Resonance</i> , 2021, 23, 140.	1.6	33

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37	Arterial Spin Labeling and Central Precocious Puberty. <i>Clinical Neuroradiology</i> , 2020, 30, 137-144.	1.0	5
38	Biometric and morphological features on magnetic resonance imaging of fetal bladder in lower urinary tract obstruction: new perspectives for fetal cystoscopy. <i>Ultrasound in Obstetrics and Gynecology</i> , 2020, 56, 86-95.	0.9	6
39	Central nervous system complications in adult cystinosis patients. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 348-356.	1.7	14
40	Scurvy: A New Old Cause of Skeletal Pain in Young Children. <i>Frontiers in Pediatrics</i> , 2020, 8, 8.	0.9	25
41	Posterior Fossa Arachnoid Cyst in a Pediatric Population is Associated with Social Perception and Rest Cerebral Blood Flow Abnormalities. <i>Cerebellum</i> , 2020, 19, 58-67.	1.4	2
42	The pediatric supratentorial MYCN-amplified high-grade gliomas methylation class presents the same radiological, histopathological and molecular features as their pontine counterparts. <i>Acta Neuropathologica Communications</i> , 2020, 8, 104.	2.4	24
43	Focal Areas of High Signal Intensity in Children with Neurofibromatosis Type 1: Expected Evolution on MRI. <i>American Journal of Neuroradiology</i> , 2020, 41, 1733-1739.	1.2	8
44	Complete hemispherotomy leads to lateralized functional organization and lower level of consciousness in the isolated hemisphere. <i>Epilepsia Open</i> , 2020, 5, 537-549.	1.3	3
45	Risk Factors for Early Brain AVM Rupture: Cohort Study of Pediatric and Adult Patients. <i>American Journal of Neuroradiology</i> , 2020, 41, 2358-2363.	1.2	16
46	Histone H3 wild-type DIPG/DMG overexpressing EZHIP extend the spectrum diffuse midline gliomas with PRC2 inhibition beyond H3-K27M mutation. <i>Acta Neuropathologica</i> , 2020, 139, 1109-1113.	3.9	104
47	Neonatal factors related to survival and intellectual and developmental outcome of patients with early-onset urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 110-117.	0.5	4
48	Loss of Function of RIMS2 Causes a Syndromic Congenital Cone-Rod Synaptic Disease with Neurodevelopmental and Pancreatic Involvement. <i>American Journal of Human Genetics</i> , 2020, 106, 859-871.	2.6	22
49	High-grade gliomas in adolescents and young adults highlight histomolecular differences from their adult and pediatric counterparts. <i>Neuro-Oncology</i> , 2020, 22, 1190-1202.	0.6	50
50	The "salt and pepper" pattern on renal ultrasound in a group of children with molecular-proven diagnosis of ciliopathy-related renal diseases. <i>Pediatric Nephrology</i> , 2020, 35, 1033-1040.	0.9	10
51	Pediatric cardiac computed tomography angiography: Expert consensus from the Filiale de Cardiologie Pédiatrique et Congénitale (FCPC) and the Société Française d'Imagerie Cardiaque et Vasculaire diagnostique et interventionnelle (SFICV). <i>Diagnostic and Interventional Imaging</i> , 2020, 101, 335-345.	1.8	5
52	Imaging features of complete congenital atresia of left coronary artery. <i>Diagnostic and Interventional Imaging</i> , 2020, 101, 421-423.	1.8	2
53	Neural basis of interindividual variability in social perception in typically developing children and adolescents using diffusion tensor imaging. <i>Scientific Reports</i> , 2020, 10, 6379.	1.6	2
54	Hydrocephalus in children with ruptured cerebral arteriovenous malformation. <i>Journal of Neurosurgery: Pediatrics</i> , 2020, 26, 283-287.	0.8	2

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55	DIPG-61. RESCUE REGIMENS AFTER BIOMEDE: POSSIBLE INFLUENCE ON OS ASSESSMENT. <i>Neuro-Oncology</i> , 2020, 22, iii299-iii299.	0.6	0
56	Mutations in the <i>MRPS28</i> gene encoding the small mitoribosomal subunit protein bS1m in a patient with intrauterine growth retardation, craniofacial dysmorphism and multisystemic involvement. <i>Human Molecular Genetics</i> , 2019, 28, 1445-1462.	1.4	19
57	Elevated thrombin generation in patients with congenital disorder of glycosylation and combined coagulation factor deficiencies. <i>Journal of Thrombosis and Haemostasis</i> , 2019, 17, 1798-1807.	1.9	18
58	Incidental Brain MRI Findings in Children: A Systematic Review and Meta-Analysis. <i>American Journal of Neuroradiology</i> , 2019, 40, 1818-1823.	1.2	25
59	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. <i>Nature Communications</i> , 2019, 10, 3967.	5.8	66
60	Neural and behavioral signature of human social perception. <i>Scientific Reports</i> , 2019, 9, 9252.	1.6	8
61	Aortic atresia and interrupted aortic arch communicating through external carotid anastomosis. <i>Cardiology in the Young</i> , 2019, 29, 699-700.	0.4	0
62	An integrative radiological, histopathological and molecular analysis of pediatric pontine histone-wildtype glioma with MYCN amplification (HGG-MYCN). <i>Acta Neuropathologica Communications</i> , 2019, 7, 87.	2.4	22
63	E-047 Vessel wall imaging and brain arteriovenous malformations: initial description of enhancement patterns. , 2019, , .		0
64	E-065 Ruptured brain arterio-venous malformations in children and adults: angioarchitectural variations at presentation across the lifespan. , 2019, , .		0
65	Corpus callosum metrics predict severity of visuospatial and neuromotor dysfunctions in ARID1B mutations with Coffin-Siris syndrome. <i>Psychiatric Genetics</i> , 2019, 29, 237-242.	0.6	8
66	TP53 Pathway Alterations Drive Radioresistance in Diffuse Intrinsic Pontine Gliomas (DIPG). <i>Clinical Cancer Research</i> , 2019, 25, 6788-6800.	3.2	66
67	CT and Multimodal MR Imaging Features of Embryonal Tumors with Multilayered Rosettes in Children. <i>American Journal of Neuroradiology</i> , 2019, 40, 732-736.	1.2	9
68	High predictive value of brain MRI imaging in primary mitochondrial respiratory chain deficiency. <i>Journal of Medical Genetics</i> , 2018, 55, 378-383.	1.5	21
69	Historadiological correlations in high-grade glioma with the histone 3.3 G34R mutation. <i>Journal of Neuroradiology</i> , 2018, 45, 316-322.	0.6	26
70	Comprehensive molecular screening strategy of <i>OCLN</i> in band-like calcification with simplified gyration and polymicrogyria. <i>Clinical Genetics</i> , 2018, 93, 228-234.	1.0	9
71	Predictors of Outcome in Patients with Pediatric Intracerebral Hemorrhage: Development and Validation of a Modified Score. <i>Radiology</i> , 2018, 286, 651-658.	3.6	31
72	Prenatally diagnosed periventricular nodular heterotopia: Further delineation of the imaging phenotype and outcome. <i>European Journal of Medical Genetics</i> , 2018, 61, 773-782.	0.7	8

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73	Reverse-Transcriptase Inhibitors in the Aicardi-Goutières Syndrome. <i>New England Journal of Medicine</i> , 2018, 379, 2275-2277.	13.9	106
74	Inhibition of mitochondrial translation in fibroblasts from a patient expressing the KARS p.(Pro228Leu) variant and presenting with sensorineural deafness, developmental delay, and lactic acidosis. <i>Human Mutation</i> , 2018, 39, 2047-2059.	1.1	14
75	Further refinement of COL4A1 and COL4A2 related cortical malformations. <i>European Journal of Medical Genetics</i> , 2018, 61, 765-772.	0.7	29
76	Challenges in managing epilepsy associated with focal cortical dysplasia in children. <i>Epilepsy Research</i> , 2018, 145, 1-17.	0.8	25
77	PLA2G6-associated neurodegeneration: Lessons from neurophysiological findings. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 854-861.	0.7	9
78	TLE1, a key player in neurogenesis, a new candidate gene for autosomal recessive postnatal microcephaly. <i>European Journal of Medical Genetics</i> , 2018, 61, 729-732.	0.7	8
79	NSRG-05. SAFETY OF ULTRASOUND-INDUCED BLOOD-BRAIN BARRIER OPENING IN PEDIATRIC PATIENTS WITH REFRACTORY SUB-TENTORIAL MALIGNANT BRAIN TUMORS BEFORE CHEMOTHERAPY ADMINISTRATION – THE SONOKID CLINICAL TRIAL. <i>Neuro-Oncology</i> , 2018, 20, i146-i146.	0.6	2
80	Recurrent RTTN mutation leading to severe microcephaly, polymicrogyria and growth restriction. <i>European Journal of Medical Genetics</i> , 2018, 61, 755-758.	0.7	9
81	Myocardial inflammation detected by cardiac MRI in Arrhythmogenic right ventricular cardiomyopathy: A paediatric case series. <i>International Journal of Cardiology</i> , 2018, 271, 81-86.	0.8	52
82	Targeted therapy in patients with PIK3CA-related overgrowth syndrome. <i>Nature</i> , 2018, 558, 540-546.	13.7	374
83	Arterial Spin-Labeling to Discriminate Pediatric Cervicofacial Soft-Tissue Vascular Anomalies. <i>American Journal of Neuroradiology</i> , 2017, 38, 633-638.	1.2	20
84	Prenatal and postnatal presentations of corpus callosum agenesis with polymicrogyria caused by <i>EGP5</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 706-711.	0.7	12
85	Moyamoya syndrome in children with neurofibromatosis type 1: Italian-French experience. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1521-1530.	0.7	36
86	Multimodal Magnetic Resonance Imaging of Treatment-Induced Changes to Diffuse Infiltrating Pontine Gliomas in Children and Correlation to Patient Progression-Free Survival. <i>International Journal of Radiation Oncology Biology Physics</i> , 2017, 99, 476-485.	0.4	18
87	Epileptic spasms in congenital disorders of glycosylation. <i>Epileptic Disorders</i> , 2017, 19, 15-23.	0.7	12
88	A novel recurrent <i>LIS1</i> splice site mutation in classic lissencephaly. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 561-564.	0.7	6
89	Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDC (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature. <i>Journal of Medical Genetics</i> , 2017, 54, 843-851.	1.5	88
90	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and <i>Drosophila</i> neural stem cells. <i>Brain</i> , 2017, 140, 2597-2609.	3.7	28

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91	Fetal MRI compared with ultrasound for the diagnosis of obstructive genital malformations. <i>Prenatal Diagnosis</i> , 2017, 37, 1138-1145.	1.1	14
92	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. <i>American Journal of Human Genetics</i> , 2017, 101, 283-290.	2.6	55
93	Severe neuroimaging anomalies are usually associated with random X inactivation in leucocytes circulating DNA in X-linked dominant Incontinentia Pigmenti. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 140-144.	0.5	11
94	Usefulness of cocaine drops in investigating infant anisocoria. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 852-857.	0.7	12
95	Neuroimaging evidence of brain abnormalities in mastocytosis. <i>Translational Psychiatry</i> , 2017, 7, e1197-e1197.	2.4	21
96	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. <i>Nature Genetics</i> , 2017, 49, 1529-1538.	9.4	164
97	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitochondrial Subunit and Leigh Syndrome. <i>American Journal of Human Genetics</i> , 2017, 101, 239-254.	2.6	83
98	Mutations in TUBB4B Cause a Distinctive Sensorineural Disease. <i>American Journal of Human Genetics</i> , 2017, 101, 1006-1012.	2.6	30
99	Neuropsychological improvement after posterior fossa arachnoid cyst drainage. <i>Child's Nervous System</i> , 2017, 33, 135-141.	0.6	17
100	Type I interferon-mediated autoinflammation due to DNase II deficiency. <i>Nature Communications</i> , 2017, 8, 2176.	5.8	164
101	HG-46 RECURRENT DIFFUSE INTRINSIC PONTINE GLIOMAS: CLINICAL, BIOLOGICAL, RADIOLOGICAL AND THERAPEUTIC FACTORS CORRELATING WITH THE SURVIVAL. <i>Neuro-Oncology</i> , 2016, 18, iii57.4-iii58.	0.6	0
102	Magnetic resonance imaging arterial spin labeling perfusion alterations in childhood migraine with atypical aura: a case-control study. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 965-969.	1.1	26
103	Cerebral Blood Flow Improvement after Indirect Revascularization for Pediatric Moyamoya Disease: A Statistical Analysis of Arterial Spin-Labeling MRI. <i>American Journal of Neuroradiology</i> , 2016, 37, 706-712.	1.2	41
104	Arterial spin labeling shows pre-epileptic tuber hyperperfusion in tuberous sclerosis complex. <i>Neurology</i> , 2016, 86, 1744-1745.	1.5	4
105	Arterial Spin Labeling to Predict Brain Tumor Grading in Children: Correlations between Histopathologic Vascular Density and Perfusion MR Imaging. <i>Radiology</i> , 2016, 281, 553-566.	3.6	82
106	Recurrent KIF5C mutation leading to frontal pachygyria without microcephaly. <i>Neurogenetics</i> , 2016, 17, 79-82.	0.7	17
107	Tuning Eye-Gaze Perception by Transitory STS Inhibition. <i>Cerebral Cortex</i> , 2016, 26, 2823-2831.	1.6	19
108	A nonsense variant in HERC1 is associated with intellectual disability, megalencephaly, thick corpus callosum and cerebellar atrophy. <i>European Journal of Human Genetics</i> , 2016, 24, 455-458.	1.4	53

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109	ISDN2014_0400: Mutations in <i>DOCK7</i> in individuals with epileptic encephalopathy and cortical blindness. <i>International Journal of Developmental Neuroscience</i> , 2015, 47, 119-120.	0.7	0
110	Choroid Plexus Neoplasms: Toward a Distinction between Carcinoma and Papilloma Using Arterial Spin-Labeling. <i>American Journal of Neuroradiology</i> , 2015, 36, 1786-1790.	1.2	31
111	Contiguous mutation syndrome in the era of high-throughput sequencing. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2015, 3, 215-220.	0.6	9
112	Mutations in <i>NONO</i> lead to syndromic intellectual disability and inhibitory synaptic defects. <i>Nature Neuroscience</i> , 2015, 18, 1731-1736.	7.1	65
113	Arterial spin labeling magnetic resonance imaging: toward noninvasive diagnosis and follow-up of pediatric brain arteriovenous malformations. <i>Journal of Neurosurgery: Pediatrics</i> , 2015, 15, 451-458.	0.8	35
114	From splitting <i>GLUT1</i> deficiency syndromes to overlapping phenotypes. <i>European Journal of Medical Genetics</i> , 2015, 58, 443-454.	0.7	52
115	Myocardial inflammation on cardiovascular magnetic resonance predicts left ventricular function recovery in children with recent dilated cardiomyopathy. <i>European Heart Journal Cardiovascular Imaging</i> , 2015, 16, 756-762.	0.5	15
116	<i>IFT81</i> , encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. <i>Journal of Medical Genetics</i> , 2015, 52, 657-665.	1.5	32
117	A neuropathological study of cerebrovascular abnormalities in a signal transducer and activator of transcription 3-deficient patient. <i>Journal of Allergy and Clinical Immunology</i> , 2015, 136, 1418-1421.e5.	1.5	5
118	Arterial Spin Labeling MRI: A step forward in non-invasive delineation of focal cortical dysplasia in children. <i>Epilepsy Research</i> , 2014, 108, 1932-1939.	0.8	46
119	Loss-of-Function Mutations in <i>WDR73</i> Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 637-648.	2.6	108
120	Long-Term Outcome of 106 Consecutive Pediatric Ruptured Brain Arteriovenous Malformations After Combined Treatment. <i>Stroke</i> , 2014, 45, 1664-1671.	1.0	86
121	Recurrent <i>Streptococcus pyogenes</i> genital infection in a woman: test and treat the partner!. <i>International Journal of Infectious Diseases</i> , 2014, 29, 37-39.	1.5	8
122	Mutations in <i>QARS</i> , Encoding Glutamyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. <i>American Journal of Human Genetics</i> , 2014, 94, 547-558.	2.6	106
123	Early epileptic encephalopathies associated with <i>STXBP1</i> mutations: Could we better delineate the phenotype?. <i>European Journal of Medical Genetics</i> , 2014, 57, 15-20.	0.7	50
124	Mutations in <i>DOCK7</i> in Individuals with Epileptic Encephalopathy and Cortical Blindness. <i>American Journal of Human Genetics</i> , 2014, 94, 891-897.	2.6	44
125	Brain imaging in mitochondrial respiratory chain deficiency: combination of brain MRI features as a useful tool for genotype/phenotype correlations. <i>Journal of Medical Genetics</i> , 2014, 51, 429-435.	1.5	40
126	Clinical and imaging diagnosis for hereditary degenerative diseases. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 111, 63-78.	1.0	124



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127	Massive and exclusive pontocerebellar damage in mitochondrial disease and <i>NUBPL</i> mutations. <i>Neurology</i> , 2012, 79, 391-391.	1.5	27
128	Mutation in PNPT1 , which Encodes a Polyribonucleotide Nucleotidyltransferase, Impairs RNA Import into Mitochondria and Causes Respiratory-Chain Deficiency. <i>American Journal of Human Genetics</i> , 2012, 91, 912-918.	2.6	81
129	Childhood gangliogliomas with ependymal differentiation. <i>Neuropathology and Applied Neurobiology</i> , 2009, 35, 437-441.	1.8	3
130	Multiple bur hole surgery for the treatment of moyamoya disease in children. <i>Journal of Neurosurgery: Pediatrics</i> , 2006, 105, 437-443.	0.8	37
131	Atypical case of hemiconvulsionsâ€chemiplegiaâ€cepilepsy syndrome revealing contralateral focal cortical dysplasia. <i>Developmental Medicine and Child Neurology</i> , 2005, 47, 830-834.	1.1	1
132	Functional neuroimaging and childhood autism. <i>Pediatric Radiology</i> , 2002, 32, 1-7.	1.1	107