Anna C Jansen

List of Publications by Year
in descending order

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133 papers	7,547 citations	81900 39 h-index	58581 82 g-index
135 all docs	135 docs citations	135 times ranked	8958 citing authors

#	Article	IF	Citations
1	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 243-254.	2.1	1,185
2	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 255-265.	2.1	693
3	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	14.5	539
4	KCNQ2 encephalopathy: Emerging phenotype of a neonatal epileptic encephalopathy. Annals of Neurology, 2012, 71, 15-25.	5.3	427
5	Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. Pediatric Neurology, 2021, 123, 50-66.	2.1	230
6	Tuberous Sclerosis Associated Neuropsychiatric Disorders (TAND) and the TAND Checklist. Pediatric Neurology, 2015, 52, 25-35.	2.1	229
7	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain, 2010, 133, 1415-1427.	7.6	215
8	TuberOus SClerosis registry to increase disease Awareness (TOSCA) â€“ baseline data on 2093 patients. Orphanet Journal of Rare Diseases, 2017, 12, 2.	2.7	166
9	Management of epilepsy associated with tuberous sclerosis complex: Updated clinical recommendations. European Journal of Paediatric Neurology, 2018, 22, 738-748.	1.6	151
10	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
11	Prevention of Epilepsy in Infants with Tuberous Sclerosis Complex in the <scp>EPISTOP</scp> Trial. Annals of Neurology, 2021, 89, 304-314.	5.3	137
12	Genetics of the polymicrogyria syndromes. Journal of Medical Genetics, 2005, 42, 369-378.	3.2	132
13	Epilepsy in tuberous sclerosis complex: Findings from the <scp>TOSCA</scp> Study. Epilepsia Open, 2019, 4, 73-84.	2.4	125
14	Inborn errors in RNA polymerase III underlie severe varicella zoster virus infections. Journal of Clinical Investigation, 2017, 127, 3543-3556.	8.2	125
15	Cognitive deficits and developmental language disorders in patients with malformations of cortical development. Epilepsia, 2010, 51, 70-71.	5.1	117
16	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	2.5	114
17	TSC-associated neuropsychiatric disorders (TAND): findings from the TOSCA natural history study. Orphanet Journal of Rare Diseases, 2018, 13, 157.	2.7	106
18	Polymicrogyria: pathology, fetal origins and mechanisms. Acta Neuropathologica Communications, 2014, 2, 80.	5.2	91

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19	Unusually mild tuberous sclerosis phenotype is associated withTSC2R905Q mutation. Annals of Neurology, 2006, 60, 528-539.	5.3	82
20	Steroids in intractable childhood epilepsy: Clinical experience and review of the literature. Seizure: the Journal of the British Epilepsy Association, 2005, 14, 412-421.	2.0	79
21	Periventricular nodular heterotopia with overlying polymicrogyria. Brain, 2005, 128, 2811-2821.	7.6	73
22	Pilot Validation of the Tuberous Sclerosis-Associated Neuropsychiatric Disorders (TAND) Checklist. Pediatric Neurology, 2015, 52, 16-24.	2.1	73
23	TUBA1A mutations. Neurology, 2011, 76, 988-992.	1.1	70
24	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. Brain, 2016, 139, 2420-2430.	7.6	70
25	Clinical and genetic aspects of PCDH19-related epilepsy syndromes and the possible role of PCDH19 mutations in males with autism spectrum disorders. Neurogenetics, 2013, 14, 23-34.	1.4	69
26	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
27	The histopathology of polymicrogyria: a series of 71 brain autopsy studies. Developmental Medicine and Child Neurology, 2016, 58, 39-48.	2.1	67
28	The Role of mTOR Inhibitors in the Treatment of Patients with Tuberous Sclerosis Complex: Evidence-based and Expert Opinions. Drugs, 2016, 76, 551-565.	10.9	66
29	Familial Temporal Lobe Epilepsy as a Presenting Feature of Choreoacanthocytosis. Epilepsia, 2005, 46, 1256-1263.	5.1	62
30	TOSCA â€“ first international registry to address knowledge gaps in the natural history and management of tuberous sclerosis complex. Orphanet Journal of Rare Diseases, 2014, 9, 182.	2.7	62
31	Encephalopathy and bilateral cataract in a boy with an interstitial deletion of Xp22 comprising the CDKL5 and NHS genes. American Journal of Medical Genetics, Part A, 2007, 143A, 364-369.	1.2	61
32	Reduction of seizure frequency after epilepsy surgery in a patient with <scp>STXBP1</scp> encephalopathy and clinical description of six novel mutation carriers. Epilepsia, 2013, 54, e74-80.	5.1	59
33	Renal angiomyolipoma in patients with tuberous sclerosis complex: findings from the TuberOus SClerosis registry to increase disease Awareness. Nephrology Dialysis Transplantation, 2019, 34, 502-508.	0.7	55
34	Short-term safety of mTOR inhibitors in infants and very young children with tuberous sclerosis complex (TSC): Multicentre clinical experience. European Journal of Paediatric Neurology, 2018, 22, 1066-1073.	1.6	54
35	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
36	TSC2 pathogenic variants are predictive of severe clinical manifestations in TSC infants: results of the EPISTOP study. Genetics in Medicine, 2020, 22, 1489-1497.	2.4	51

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37	Early onset epileptic encephalopathy or genetically determined encephalopathy with early onset epilepsy? Lessons learned from TSC. European Journal of Paediatric Neurology, 2016, 20, 203-211.	1.6	49
38	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
39	Coding and small non-coding transcriptional landscape of tuberous sclerosis complex cortical tubers: implications for pathophysiology and treatment. Scientific Reports, 2017, 7, 8089.	3.3	47
40	Hearing thresholds in children with a congenital CMV infection: A prospective study. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 712-717.	1.0	45
41	Early Clinical Predictors of Autism Spectrum Disorder in Infants with Tuberous Sclerosis Complex: Results from the EPISTOP Study. Journal of Clinical Medicine, 2019, 8, 788.	2.4	42
42	Epilepsy and Neurodevelopmental Comorbidities in Tuberous Sclerosis Complex: A Natural History Study. Pediatric Neurology, 2020, 106, 10-16.	2.1	37
43	Identification of a VPS13A founder mutation in French Canadian families with chorea-acanthocytosis. Neurogenetics, 2005, 6, 151-158.	1.4	36
44	Primary maternal cytomegalovirus infections: accuracy ofÂfetal ultrasound for predicting sequelae in offspring. American Journal of Obstetrics and Gynecology, 2016, 215, 638.e1-638.e8.	1.3	36
45	A step-wise approach for establishing a multidisciplinary team for the management of tuberous sclerosis complex: a Delphi consensus report. Orphanet Journal of Rare Diseases, 2019, 14, 91.	2.7	36
46	Bi-allelic variants inCOL3A1encoding the ligand to GPR56 are associated with cobblestone-like cortical malformation, white matter changes and cerebellar cysts. Journal of Medical Genetics, 2017, 54, 432-440.	3.2	34
47	SCN4A variants and Brugada syndrome: phenotypic and genotypic overlap between cardiac and skeletal muscle sodium channelopathies. European Journal of Human Genetics, 2016, 24, 400-407.	2.8	33
48	Polymicrogyria with dysmorphic basal ganglia? Think tubulin!. Clinical Genetics, 2014, 85, 178-183.	2.0	30
49	Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. European Journal of Human Genetics, 2018, 26, 1132-1142.	2.8	30
50	Sanfilippo Syndrome Type D. Archives of Neurology, 2007, 64, 1629.	4.5	29
51	mTOR-related neuropathology in mutant tsc2 zebrafish: Phenotypic, transcriptomic and pharmacological analysis. Neurobiology of Disease, 2017, 108, 225-237.	4.4	29
52	Central nervous system gadolinium accumulation in patients undergoing periodical contrast MRI screening for hereditary tumor syndromes. Hereditary Cancer in Clinical Practice, 2018, 16, 2.	1.5	29
53	Renal progression factors in young patients with tuberous sclerosis complex: a retrospective cohort study. Pediatric Nephrology, 2018, 33, 2085-2093.	1.7	29
54	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29

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55	PRRT2 mutations: exploring the phenotypical boundaries. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 462-465.	1.9	27
56	Renal Manifestations of Tuberous Sclerosis Complex: Key Findings From the Final Analysis of the TOSCA Study Focussing Mainly on Renal Angiomyolipomas. Frontiers in Neurology, 2020, 11, 972.	2.4	27
57	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. American Journal of Human Genetics, 2019, 105, 1126-1147.	6.2	25
58	De novo MECP2 duplications in two females with intellectual disability and unfavorable complete skewed X-inactivation. Human Genetics, 2014, 133, 1359-1367.	3.8	24
59	Is autism driven by epilepsy in infants with Tuberous Sclerosis Complex?. Annals of Clinical and Translational Neurology, 2020, 7, 1371-1381.	3.7	23
60	Clinical Characteristics of Subependymal Giant Cell Astrocytoma in Tuberous Sclerosis Complex. Frontiers in Neurology, 2019, 10, 705.	2.4	22
61	Heterogeneous clinical phenotypes and cerebral malformations reflected by rotatin cellular dynamics. Brain, 2019, 142, 867-884.	7.6	22
62	Genetic heterogeneity of polymicrogyria: study of 123 patients using deep sequencing. Brain Communications, 2021, 3, fcaa221.	3.3	22
63	Abnormal development of the human cerebral cortex. Journal of Anatomy, 2010, 217, 312-323.	1.5	21
64	Myelin Pathology Beyond White Matter in Tuberous Sclerosis Complex (TSC) Cortical Tubers. Journal of Neuropathology and Experimental Neurology, 2020, 79, 1054-1064.	1.7	21
65	Burden of Illness and Quality of Life in Tuberous Sclerosis Complex: Findings From the TOSCA Study. Frontiers in Neurology, 2020, 11, 904.	2.4	20
66	Prediction of Neurodevelopment in Infants With Tuberous Sclerosis Complex Using Early EEG Characteristics. Frontiers in Neurology, 2020, 11, 582891.	2.4	19
67	Recommendations for the treatment of epilepsy in adult and pediatric patients in Belgium: 2020 update. Acta Neurologica Belgica, 2021, 121, 241-257.	1.1	19
68	Early epileptiform EEG activity in infants with tuberous sclerosis complex predicts epilepsy and neurodevelopmental outcomes. Epilepsia, 2021, 62, 1208-1219.	5.1	19
69	Cognitive functioning in bilateral perisylvian polymicrogyria (BPP): clinical and radiological correlations. Epilepsy and Behavior, 2005, 6, 393-404.	1.7	18
70	Newly Diagnosed and Growing Subependymal Giant Cell Astrocytoma in Adults With Tuberous Sclerosis Complex: Results From the International TOSCA Study. Frontiers in Neurology, 2019, 10, 821.	2.4	18
71	Clinical implementation of gene panel testing for lysosomal storage diseases. Molecular Genetics & Genomic Medicine, 2019, 7, e00527.	1.2	18
72	Recommendations for the treatment of epilepsy in adult patients in general practice in Belgium: an update. Acta Neurologica Belgica, 2012, 112, 119-131.	1.1	16

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73	Natural clusters of tuberous sclerosis complex (TSC)-associated neuropsychiatric disorders (TAND): new findings from the TOSCA TAND research project. Journal of Neurodevelopmental Disorders, 2020, 12, 24.	3.1	16
74	Leucoencephalopathy with vanishing white matter may cause progressive myoclonus epilepsy. Epilepsia, 2008, 49, 910-913.	5.1	15
75	Rare manifestations and malignancies in tuberous sclerosis complex: findings from the TuberOus SClerosis registry to increAse disease awareness (TOSCA). Orphanet Journal of Rare Diseases, 2021, 16, 301.	2.7	15
76	Striking intrafamilial phenotypic variability and spastic paraplegia in the presence of similar homozygous expansions of theFRDA1gene. Movement Disorders, 2004, 19, 1424-1431.	3.9	14
77	Early treatment of a child with NAGS deficiency using N-carbamyl glutamate results in a normal neurological outcome. European Journal of Pediatrics, 2014, 173, 1635-1638.	2.7	14
78	Attending school after treatment for a brain tumor: Experiences of children and key figures. Journal of Health Psychology, 2019, 24, 1436-1447.	2.3	14
79	The spectrum of brain malformations and disruptions in twins. American Journal of Medical Genetics, Part A, 2021, 185, 2690-2718.	1.2	13
80	Historical Patterns of Diagnosis, Treatments, and Outcome of Epilepsy Associated With Tuberous Sclerosis Complex: Results From TOSCA Registry. Frontiers in Neurology, 2021, 12, 697467.	2.4	13
81	Elaborating the phenotypic spectrum associated with mutations in ARFGEF2: Case study and literature review. European Journal of Paediatric Neurology, 2013, 17, 666-670.	1.6	12
82	Expanding the clinical spectrum of biallelic ZNF335 variants. Clinical Genetics, 2018, 94, 246-251.	2.0	12
83	Recurrent NEDD4L Variant in Periventricular Nodular Heterotopia, Polymicrogyria and Syndactyly. Frontiers in Genetics, 2020, 11, 26.	2.3	12
84	Biallelic mutations in RTTN are associated with microcephaly, short stature and a wide range of brain malformations. European Journal of Medical Genetics, 2018, 61, 733-737.	1.3	11
85	Treatment Patterns and Use of Resources in Patients With Tuberous Sclerosis Complex: Insights From the TOSCA Registry. Frontiers in Neurology, 2019, 10, 1144.	2.4	11
86	Pregabalin adjunctive therapy for focal onset seizures in children 1 month to <4 years of age: A doubleâ€blind, placeboâ€controlled, videoâ€electroencephalographic trial. Epilepsia, 2020, 61, 617-626.	5.1	11
87	Defining the phenotypical spectrum associated with variants in TUBB2A. Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11
88	Thin genu of the corpus callosum points to mutation in FOXG1 in a child with acquired microcephaly, trigonocephaly, and intellectual developmental disorder: A case report and review of literature. European Journal of Paediatric Neurology, 2014, 18, 420-426.	1.6	10
89	Mutated zinc finger protein of the cerebellum 1 leads to microcephaly, cortical malformation, callosal agenesis, cerebellar dysplasia, tethered cord and scoliosis. European Journal of Medical Genetics, 2018, 61, 783-789.	1.3	10
90	A focused 35-minute whole body MRI screening protocol for patients with von Hippel-Lindau disease. Hereditary Cancer in Clinical Practice, 2019, 17, 22.	1.5	10

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91	Reintegration into school of childhood brain tumor survivors: a qualitative study using the International Classification of Functioning, Disability and Health â€“ Children and Youth framework. Disability and Rehabilitation, 2021, 43, 2610-2620.	1.8	10
92	Phenotypic spectrum of the <scp>RBM10</scp>â€mediated intellectual disability and congenital malformation syndrome beyond classic <scp>TARP</scp> syndrome features. Clinical Genetics, 2021, 99, 449-456.	2.0	10
93	TuberOus SClerosis registry to increase disease awareness (TOSCA) Post-Authorisation Safety Study of Everolimus in Patients With Tuberous Sclerosis Complex. Frontiers in Neurology, 2021, 12, 630378.	2.4	10
94	Multivariate data analysis identifies natural clusters of Tuberous Sclerosis Complex Associated Neuropsychiatric Disorders (TAND). Orphanet Journal of Rare Diseases, 2021, 16, 447.	2.7	10
95	Stakeholdersâ€™ Perspectives on Communication and Collaboration Following School Reintegration of a Seriously Ill Child: A Literature Review. Child and Youth Care Forum, 2018, 47, 583-612.	1.6	9
96	Speech and language in bilateral perisylvian polymicrogyria: a systematic review. Developmental Medicine and Child Neurology, 2019, 61, 1145-1152.	2.1	9
97	Chudley-McCullough Syndrome: A Recognizable Clinical Entity Characterized by Deafness and Typical Brain Malformations. Journal of Child Neurology, 2021, 36, 152-158.	1.4	9
98	Neuropathology of genetically defined malformations of cortical developmentâ€”A systematic literature review. Neuropathology and Applied Neurobiology, 2021, 47, 585-602.	3.2	9
99	The research landscape of tuberous sclerosis complexâ€“associated neuropsychiatric disorders (TAND)â€”a comprehensive scoping review. Journal of Neurodevelopmental Disorders, 2022, 14, 13.	3.1	9
100	First line management of prolonged convulsive seizures in children and adults: good practice points. Acta Neurologica Belgica, 2013, 113, 375-380.	1.1	8
101	Short-term perspectives of parents and teachers on school reintegration of childhood brain tumour survivors. Developmental Neurorehabilitation, 2019, 22, 321-328.	1.1	8
102	Association of Early MRI Characteristics With Subsequent Epilepsy and Neurodevelopmental Outcomes in Children With Tuberous Sclerosis Complex. Neurology, 2022, 98, .	1.1	8
103	Infants dying suddenly and unexpectedly share demographic features with infants who die with retinal and dural bleeding: a review of neural mechanisms. Developmental Medicine and Child Neurology, 2016, 58, 1223-1234.	2.1	7
104	Fetal and neonatal neurogenetics. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2019, 162, 105-132.	1.8	7
105	Results of quantitative EEG analysis are associated with autism spectrum disorder and development abnormalities in infants with tuberous sclerosis complex. Biomedical Signal Processing and Control, 2021, 68, 102658.	5.7	7
106	Tuberous Sclerosis Complex-Associated Neuropsychiatric Disorders (TAND): New Findings on Age, Sex, and Genotype in Relation to Intellectual Phenotype. Frontiers in Neurology, 2020, 11, 603.	2.4	7
107	I-PV: a CIRCOS module for interactive protein sequence visualization. Bioinformatics, 2016, 32, 447-449.	4.1	6
108	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	1.2	6

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109	Reintegration Into School After Treatment for a Brain Tumor: The Childâ€™s Perspective. Global Pediatric Health, 2019, 6, 2333794X1986065.	0.7	5
110	MicroRNAâ€34a activation in tuberous sclerosis complex during early brain development may lead to impaired corticogenesis. Neuropathology and Applied Neurobiology, 2021, 47, 796-811.	3.2	5
111	Empowering Families Through Technology: A Mobile-Health Project to Reduce the TAND Identification and Treatment Gap (TANDem). Frontiers in Psychiatry, 2022, 13, 834628.	2.6	5
112	Multinodular and vacuolating neuronal tumor of the cerebrum (MVNT): do not touch. Acta Neurologica Belgica, 2020, 120, 747-748.	1.1	4
113	Down-regulation of the brain-specific cell-adhesion molecule contactin-3 in tuberous sclerosis complex during the early postnatal period. Journal of Neurodevelopmental Disorders, 2022, 14, 8.	3.1	4
114	Impaired catabolism of free oligosaccharides due to MAN2C1 variants causes a neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 345-360.	6.2	4
115	Early impact of Xâ€•and Yâ€chromosome variations (<scp>XXX</scp> , <scp>XXY</scp> , <scp>XYY</scp>) on social communication and social emotional development in 1â€“2â€yearâ€old children. American Journal of Medical Genetics, Part A, 2022, , .	1.2	4
116	The TOSCA Registry for Tuberous Sclerosisâ€”Lessons Learnt for Future Registry Development in Rare and Complex Diseases. Frontiers in Neurology, 2019, 10, 1182.	2.4	3
117	Organizational perspectives and diagnostic evaluations for children with neurodevelopmental disorders. Developmental Medicine and Child Neurology, 2022, 64, 88-94.	2.1	3
118	Evolution of electroencephalogram in infants with tuberous sclerosis complex and neurodevelopmental outcome: a prospective cohort study. Developmental Medicine and Child Neurology, 2022, 64, 495-501.	2.1	3
119	Careful clinical observation continues to improve understanding of the phenotype in individuals with tuberous sclerosis complex. Developmental Medicine and Child Neurology, 2014, 56, 1134-1135.	2.1	2
120	Convert your favorite protein modeling program into a mutation predictor: â€œMODICTâ€• BMC Bioinformatics, 2016, 17, 425.	2.6	2
121	Best use of the tuberous sclerosis complexâ€associated neuropsychiatric disorders (TAND) checklist. Developmental Medicine and Child Neurology, 2019, 61, 112-113.	2.1	2
122	Biparental Inheritance in Idiopathic Generalized Epilepsy. Epilepsia, 2004, 45, 1294-1295.	5.1	1
123	Uncommon Cause of Psychotic Behavior in a 9-Year-Old Girl: A Case Report. Case Reports in Medicine, 2012, 2012, 1-4.	0.7	1
124	Malformations of cerebral development and clues from the peripheral nervous system: A systematic literature review. European Journal of Paediatric Neurology, 2022, 37, 155-164.	1.6	1
125	Can clinical characteristics be criteria to perform chromosomal microarray analysis in children and adolescents with autism spectrum disorders?. Minerva Pediatrics, 2018, 70, 225-232.	0.4	1
126	Overlapping cortical malformations in patients with pathogenic variants inGRIN1andGRIN2B. Journal of Medical Genetics, 2023, 60, 183-192.	3.2	1

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127	Satellite Symposium Abstract. Epilepsia, 2012, 53, 246-250.	5.1	0
128	SP887LONG-TERM RENAL OUTCOME OF A LARGE COHORT OF PATIENTS WITH TUBEROUS SCLEROSIS COMPLEX. Nephrology Dialysis Transplantation, 2015, 30, iii669-iii669.	0.7	0
129	746: Primary maternal cytomegalovirus infections: how accurate is fetal ultrasound to predict sequelae in the offspring?. American Journal of Obstetrics and Gynecology, 2015, 212, S363-S364.	1.3	0
130	Novel Variant in COL4A1 Causes Extensive Prenatal Intracranial Hemorrhage and Porencephaly. Journal of Neuropathology and Experimental Neurology, 2021, 80, 807-810.	1.7	0
131	A clinician's approach to brain malformations. Journal of International Child Neurology Association, 0, , .	0.0	0
132	First Results of the EPISTOP Study. , 2019, 50, .		0
133	Care for the caregiver! A call for action. European Journal of Paediatric Neurology, 2021, , .	1.6	0