

Mark I Rees

List of Publications by Year in descending order

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136
papers

7,043
citations

53794

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docs citations

137
times ranked

9324
citing authors

#	ARTICLE	IF	CITATIONS
1	De Novo Mutations in SLC1A2 and CACNA1A Are Important Causes of Epileptic Encephalopathies. American Journal of Human Genetics, 2016, 99, 287-298.	6.2	247
2	The GDP-GTP Exchange Factor Collybistin: An Essential Determinant of Neuronal Gephyrin Clustering. Journal of Neuroscience, 2004, 24, 5816-5826.	3.6	239
3	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
4	Mutations in the gene encoding GlyT2 (SLC6A5) define a presynaptic component of human startle disease. Nature Genetics, 2006, 38, 801-806.	21.4	232
5	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
6	The genetics of hyperekplexia: more than startle!. Trends in Genetics, 2008, 24, 439-447.	6.7	187
7	TUBA1A mutations cause wide spectrum lissencephaly (smooth brain) and suggest that multiple neuronal migration pathways converge on alpha tubulins. Human Molecular Genetics, 2010, 19, 2817-2827.	2.9	176
8	Additional support for schizophrenia linkage on chromosomes 6 and 8: A multicenter study. , 1996, 67, 580-594.		166
9	An ovine transgenic Huntington's disease model. Human Molecular Genetics, 2010, 19, 1873-1882.	2.9	166
10	Immunohistochemical staining of post-mortem adult human brain sections. Nature Protocols, 2006, 1, 2719-2732.	12.0	155
11	Dynamic changes in myelin aberrations and oligodendrocyte generation in chronic amyloidosis in mice and men. Glia, 2013, 61, 273-286.	4.9	155
12	Evidence for recessive as well as dominant forms of startle disease (hyperekplexia) caused by mutations in the $\alpha 1$ subunit of the inhibitory glycine receptor. Human Molecular Genetics, 1994, 3, 2175-2179.	2.9	151
13	Hyperekplexia associated with compound heterozygote mutations in the beta-subunit of the human inhibitory glycine receptor (GLRB). Human Molecular Genetics, 2002, 11, 853-860.	2.9	151
14	Association studies of bipolar disorder at the human serotonin transporter gene (hSERT; $\alpha 5$ HTT). Molecular Psychiatry, 1997, 2, 398-402.	7.9	145
15	A Two-Stage Genome Scan for Schizophrenia Susceptibility Genes in 196 Affected Sibling Pairs. Human Molecular Genetics, 1999, 8, 1729-1739.	2.9	136
16	Overlapping cortical malformations and mutations in TUBB2B and TUBA1A. Brain, 2013, 136, 536-548.	7.6	133
17	Functional variants of antioxidant genes in smokers with COPD and in those with normal lung function. Thorax, 2006, 61, 394-399.	5.6	120
18	Isoform Heterogeneity of the Human Gephyrin Gene (GPHN), Binding Domains to the Glycine Receptor, and Mutation Analysis in Hyperekplexia. Journal of Biological Chemistry, 2003, 278, 24688-24696.	3.4	113

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19	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. <i>Journal of Neuroscience</i> , 2010, 30, 9612-9620.	3.6	112
20	Misdiagnosis of Long QT Syndrome as Epilepsy at First Presentation. <i>Annals of Emergency Medicine</i> , 2009, 54, 26-32.	0.6	103
21	Complement is activated in progressive multiple sclerosis cortical grey matter lesions. <i>Journal of Neuroinflammation</i> , 2016, 13, 161.	7.2	101
22	De Novo Mutations in the Beta-Tubulin Gene TUBB2A Cause Simplified Gyral Patterning and Infantile-Onset Epilepsy. <i>American Journal of Human Genetics</i> , 2014, 94, 634-641.	6.2	99
23	Meningeal inflammation and cortical demyelination in acute multiple sclerosis. <i>Annals of Neurology</i> , 2018, 84, 829-842.	5.3	96
24	How did the domestication of Fertile Crescent grain crops increase their yields?. <i>Functional Ecology</i> , 2017, 31, 387-397.	3.6	93
25	Tubulin genes and malformations of cortical development. <i>European Journal of Medical Genetics</i> , 2018, 61, 744-754.	1.3	93
26	Polygenic burden in focal and generalized epilepsies. <i>Brain</i> , 2019, 142, 3473-3481.	7.6	90
27	Mutations in the GlyT2 Gene (SLC6A5) Are a Second Major Cause of Startle Disease. <i>Journal of Biological Chemistry</i> , 2012, 287, 28975-28985.	3.4	84
28	Identification of large gene deletions and duplications in KCNQ1 and KCNH2 in patients with long QT syndrome. <i>Heart Rhythm</i> , 2008, 5, 1275-1281.	0.7	79
29	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. <i>Human Molecular Genetics</i> , 2015, 24, 5313-5325.	2.9	77
30	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. <i>EBioMedicine</i> , 2015, 2, 1063-1070.	6.1	74
31	Compound heterozygosity and nonsense mutations in the $\alpha 1$ -subunit of the inhibitory glycine receptor in hyperekplexia. <i>Human Genetics</i> , 2001, 109, 267-270.	3.8	72
32	Near-miss SIDS due to Brugada syndrome. <i>Archives of Disease in Childhood</i> , 2005, 90, 528-529.	1.9	72
33	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. <i>Brain</i> , 2018, 141, 698-712.	7.6	72
34	Further evidence of autosomal dominant congenital zonular pulverulent cataracts linked to 13q11 (CZP3) and a novel mutation in connexin 46 (GJA3). <i>Human Genetics</i> , 2000, 106, 206-209.	3.8	68
35	Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. <i>Brain</i> , 2013, 136, 3085-3095.	7.6	66
36	Phospho-STAT5 and phospho-Akt expression in chronic myeloproliferative neoplasms. <i>British Journal of Haematology</i> , 2009, 147, 495-506.	2.5	65

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37	No evidence for allelic association between bipolar disorder and monoamine oxidase a gene polymorphisms. <i>American Journal of Medical Genetics Part A</i> , 1995, 60, 322-324.	2.4	64
38	Brugada Syndrome Masquerading as Febrile Seizures. <i>Pediatrics</i> , 2007, 119, e1206-e1211.	2.1	64
39	A genomewide linkage study of age at onset in schizophrenia. <i>American Journal of Medical Genetics Part A</i> , 2001, 105, 439-445.	2.4	63
40	Posthumous diagnosis of long QT syndrome from neonatal screening cards. <i>Heart Rhythm</i> , 2010, 7, 481-486.	0.7	56
41	Localisation of glycine receptors in the human forebrain, brainstem, and cervical spinal cord: an immunohistochemical review. <i>Frontiers in Molecular Neuroscience</i> , 2009, 2, 25.	2.9	54
42	Not All hERG Pore Domain Mutations Have a Severe Phenotype: G584S Has an Inactivation Gating Defect with Mild Phenotype Compared to G572S, Which Has a Dominant Negative Trafficking Defect and a Severe Phenotype. <i>Journal of Cardiovascular Electrophysiology</i> , 2009, 20, 923-930.	1.7	54
43	Novel missense mutations in the glycine receptor $\hat{1}^2$ subunit gene (GLRB) in startle disease. <i>Neurobiology of Disease</i> , 2013, 52, 137-149.	4.4	54
44	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. <i>Brain</i> , 2018, 141, 651-661.	7.6	52
45	GLRB is the third major gene of effect in hyperekplexia. <i>Human Molecular Genetics</i> , 2013, 22, 927-940.	2.9	50
46	Differential localization of $\hat{1}^3$ $\hat{1}^2$ aminobutyric acid type a and glycine receptor subunits and gephyrin in the human pons, medulla oblongata and uppermost cervical segment of the spinal cord: An immunohistochemical study. <i>Journal of Comparative Neurology</i> , 2010, 518, 305-328.	1.6	48
47	Weight change associated with antiepileptic drugs. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 796-799.	1.9	48
48	Validating epilepsy diagnoses in routinely collected data. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 52, 195-198.	2.0	48
49	The glycinergic system in human startle disease: a genetic screening approach. <i>Frontiers in Molecular Neuroscience</i> , 2010, 3, 8.	2.9	47
50	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17%458 subjects. <i>Brain</i> , 2020, 143, 2106-2118.	7.6	47
51	Epilepsy and deprivation, a data linkage study. <i>Epilepsia</i> , 2015, 56, 585-591.	5.1	45
52	Trends in the first antiepileptic drug prescribed for epilepsy between 2000 and 2010. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2014, 23, 77-80.	2.0	44
53	Biophysical Properties of 9 <i>KCNQ1</i> Mutations Associated With Long-QT Syndrome. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2009, 2, 417-426.	4.8	43
54	Startle disease in Irish wolfhounds associated with a microdeletion in the glycine transporter GlyT2 gene. <i>Neurobiology of Disease</i> , 2011, 43, 184-189.	4.4	43

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55	Clinical spectrum of <i>STX1B</i> -related epileptic disorders. <i>Neurology</i> , 2019, 92, e1238-e1249.	1.1	43
56	A Novel Dominant Hyperekplexia Mutation Y705C Alters Trafficking and Biochemical Properties of the Presynaptic Glycine Transporter GlyT2. <i>Journal of Biological Chemistry</i> , 2012, 287, 28986-29002.	3.4	42
57	Incidence, Prevalence, and Health Care Outcomes in Idiopathic Intracranial Hypertension. <i>Neurology</i> , 2021, 96, .	1.1	42
58	Bipolar disorder and the serotonin transporter gene: a family-based association study. <i>Psychological Medicine</i> , 1999, 29, 1249-1254.	4.5	41
59	Long QT and Brugada syndrome gene mutations in New Zealand. <i>Heart Rhythm</i> , 2007, 4, 1306-1314.	0.7	41
60	Chrelin inhibits LPS-induced release of IL-6 from mouse dopaminergic neurones. <i>Journal of Neuroinflammation</i> , 2013, 10, 40.	7.2	41
61	Functional Traits Differ between Cereal Crop Progenitors and Other Wild Grasses Gathered in the Neolithic Fertile Crescent. <i>PLoS ONE</i> , 2014, 9, e87586.	2.5	41
62	Glycine receptors in the striatum, globus pallidus, and substantia nigra of the human brain: An immunohistochemical study. <i>Journal of Comparative Neurology</i> , 2007, 502, 1012-1029.	1.6	40
63	A Novel GABRG2 mutation, p.R136*, in a family with GEFS+ and extended phenotypes. <i>Neurobiology of Disease</i> , 2014, 64, 131-141.	4.4	39
64	Using natural language processing to extract structured epilepsy data from unstructured clinic letters: development and validation of the ExECT (extraction of epilepsy clinical text) system. <i>BMJ Open</i> , 2019, 9, e023232.	1.9	39
65	QT interval prolongation associated with sibutramine treatment. <i>British Journal of Clinical Pharmacology</i> , 2006, 61, 464-469.	2.4	38
66	A critical role for glycine transporters in hyperexcitability disorders. <i>Frontiers in Molecular Neuroscience</i> , 2008, 1, 1.	2.9	37
67	Characterisation, mutation detection, and association analysis of alternative promoters and 5â€² UTRs of the human dopamine D3 receptor gene in schizophrenia. <i>Molecular Psychiatry</i> , 2002, 7, 493-502.	7.9	35
68	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. <i>Journal of Biological Chemistry</i> , 2013, 288, 33745-33759.	3.4	35
69	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021, 108, 965-982.	6.2	35
70	Association of gephyrin and glycine receptors in the human brainstem and spinal cord: an immunohistochemical analysis. <i>Neuroscience</i> , 2003, 122, 773-784.	2.3	33
71	Sox-2 is expressed by glial and progenitor cells and Pax-6 is expressed by neuroblasts in the human subventricular zone. <i>Experimental Neurology</i> , 2007, 204, 828-831.	4.1	33
72	PICK1 interacts with Î±7 neuronal nicotinic acetylcholine receptors and controls their clustering. <i>Molecular and Cellular Neurosciences</i> , 2007, 35, 339-355.	2.2	32

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73	Molecular investigation of TBP allele length:. <i>Neurobiology of Disease</i> , 2003, 13, 37-45.	4.4	31
74	Differential localization of GABA _A receptor subunits within the substantia nigra of the human brain: An immunohistochemical study. <i>Journal of Comparative Neurology</i> , 2008, 506, 912-929.	1.6	31
75	A comprehensive neuropsychological description of cognition in drug-refractory juvenile myoclonic epilepsy. <i>Epilepsy and Behavior</i> , 2014, 36, 124-129.	1.7	31
76	The genetics of epilepsyâ€”The past, the present and future. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2010, 19, 680-683.	2.0	30
77	Optimized sample preparation for high-resolution AFM characterization of fixed human cells. <i>Journal of Microscopy</i> , 2010, 240, 111-121.	1.8	29
78	Executive functions and psychiatric symptoms in drug-refractory juvenile myoclonic epilepsy. <i>Epilepsy and Behavior</i> , 2014, 35, 72-77.	1.7	29
79	Symptoms and Signs Associated with Syncope in Young People with Primary Cardiac Arrhythmias. <i>Heart Lung and Circulation</i> , 2011, 20, 593-598.	0.4	27
80	Epilepsy, antiepileptic drugs, and the risk of major cardiovascular events. <i>Epilepsia</i> , 2021, 62, 1604-1616.	5.1	27
81	Elevated serum gastrin levels in Jervell and Lange-Nielsen syndrome: A marker of severe KCNQ1 dysfunction?. <i>Heart Rhythm</i> , 2011, 8, 551-554.	0.7	26
82	Were Fertile Crescent crop progenitors higher yielding than other wild species that were never domesticated?. <i>New Phytologist</i> , 2015, 207, 905-913.	7.3	26
83	Harris syndrome - a geographic perspective. <i>Journal of Thrombosis and Haemostasis</i> , 2005, 3, 2581-2582.	3.8	24
84	Pathogenic copy number variants and SCN1A mutations in patients with intellectual disability and childhood-onset epilepsy. <i>BMC Medical Genetics</i> , 2016, 17, 34.	2.1	23
85	Phenotypic analysis of 303 multiplex families with common epilepsies. <i>Brain</i> , 2017, 140, 2144-2156.	7.6	23
86	Distribution of gephyrin in the human brain: an immunohistochemical analysis. <i>Neuroscience</i> , 2003, 116, 145-156.	2.3	22
87	Identifying and prioritising epilepsy treatment uncertainties. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 918-921.	1.9	22
88	TBP, a polyglutamine tract containing protein, accumulates in Alzheimer's disease. <i>Molecular Brain Research</i> , 2004, 125, 120-128.	2.3	21
89	Next Generation Sequencing Methodologies - An Overview. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 1-26.	2.3	21
90	Next Generation Sequencing in the Clinical Domain: Clinical Advantages, Practical, and Ethical Challenges. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 27-63.	2.3	21

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91	Trait impulsivity in Juvenile Myoclonic Epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 138-152.	3.7	21
92	Autosome search for schizophrenia susceptibility genes in multiply affected families. <i>Molecular Psychiatry</i> , 1999, 4, 353-359.	7.9	20
93	Missense variants in the N-terminal domain of the A isoform of FHF2/FGF13 cause an X-linked developmental and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 176-185.	6.2	20
94	Structural Modelling Pipelines in Next Generation Sequencing Projects. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 117-167.	2.3	19
95	Sex-specific disease modifiers in juvenile myoclonic epilepsy. <i>Scientific Reports</i> , 2022, 12, 2785.	3.3	19
96	GABAA receptor subunit and gephyrin protein changes differ in the globus pallidus in Huntington's diseased brain. <i>Brain Research</i> , 2003, 994, 265-270.	2.2	18
97	Coinheritance of long QT syndrome and Kearns-Sayre syndrome. <i>Heart Rhythm</i> , 2007, 4, 1568-1572.	0.7	15
98	Flow-induced ATP release in patient-specific arterial geometries – a comparative study of computational models. <i>International Journal for Numerical Methods in Biomedical Engineering</i> , 2013, 29, 1038-1056.	2.1	13
99	Chromosome 2 interstitial deletion (del(2)(q14.1q21)) associated with connective tissue laxity and an attention deficit disorder. <i>Journal of Medical Genetics</i> , 2001, 38, 493-496.	3.2	13
100	Red cell dimorphism in a young man with a constitutional chromosomal translocation t(11;22)(p15.5;q11.21). <i>British Journal of Haematology</i> , 1994, 87, 386-395.	2.5	11
101	Hyperekplexia: Report on phenotype and genotype of 16 Jordanian patients. <i>Brain and Development</i> , 2017, 39, 306-311.	1.1	11
102	Cereal progenitors differ in stand harvest characteristics from related wild grasses. <i>Journal of Ecology</i> , 2018, 106, 1286-1297.	4.0	11
103	Educational attainment of children born to mothers with epilepsy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 736-740.	1.9	11
104	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. <i>Brain</i> , 2018, 141, e55-e55.	7.6	11
105	Epilepsy in families: Age at onset is a familial trait, independent of syndrome. <i>Annals of Neurology</i> , 2019, 86, 91-98.	5.3	11
106	Fine architecture and mutation mapping of human brain inhibitory system ligand gated ion channels by high-throughput homology modeling. <i>Advances in Protein Chemistry and Structural Biology</i> , 2010, 80, 117-152.	2.3	10
107	Development of a cardiac inherited disease service and clinical registry: A 15-year perspective. <i>American Heart Journal</i> , 2019, 209, 126-130.	2.7	10
108	Opportunities and Challenges for Genome Sequencing in the Clinic. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 65-83.	2.3	9

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109	Ethnicity can predict GLRA1 genotypes in hyperekplexia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 341-343.	1.9	9
110	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. <i>Annals of Neurology</i> , 2020, 87, 897-906.	5.3	9
111	Genetic epilepsy with febrile seizures plus: definite and borderline phenotypes. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 336-338.	1.9	8
112	Association of ultra-rare coding variants with genetic generalized epilepsy: A case-control whole exome sequencing study. <i>Epilepsia</i> , 2022, 63, 723-735.	5.1	8
113	An unusual case of hyperekplexia. <i>European Journal of Paediatric Neurology</i> , 2000, 4, 77-80.	1.6	7
114	Implications for families of advances in understanding the genetic basis of epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2010, 19, 675-679.	2.0	7
115	Fertile Crescent crop progenitors gained a competitive advantage from large seedlings. <i>Ecology and Evolution</i> , 2021, 11, 3300-3312.	1.9	7
116	Epilepsy mortality in Wales during COVID-19. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2022, 94, 39-42.	2.0	7
117	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. <i>Epilepsy and Behavior</i> , 2013, 26, 241-246.	1.7	6
118	The phenotype of bilateral hippocampal sclerosis and its management in "cereal life" clinical settings. <i>Epilepsia</i> , 2018, 59, 1410-1420.	5.1	6
119	Epilepsy genetics: clinical beginnings and social consequences. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2009, 102, 497-499.	0.5	4
120	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. <i>New England Journal of Medicine</i> , 2019, 380, e24.	27.0	4
121	Linkage analysis in an autosomal dominant "zonular nuclear pulverulent" congenital cataract, mapped to chromosome 13q11-13. <i>Eye</i> , 2000, 14, 172-175.	2.1	3
122	The conundrum of complexity in epilepsy. <i>Lancet Neurology</i> , The, 2007, 6, 943-944.	10.2	3
123	Substantial subpial cortical demyelination in progressive multiple sclerosis: have we underestimated the extent of cortical pathology?. <i>Neuroimmunology and Neuroinflammation</i> , 2020, , .	1.4	3
124	Polymorphisms in Neuropsychiatric and Neuroinflammatory Disorders and the Role of Next Generation Sequencing in Early Diagnosis and Treatment. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, 85-116.	2.3	2
125	Neonatal hyperekplexia with homozygous p.R392H mutation in GLRA1. <i>Epileptic Disorders</i> , 2014, 16, 354-357.	1.3	2
126	The "maternal effect" on epilepsy risk: Analysis of familial epilepsies and reassessment of prior evidence. <i>Annals of Neurology</i> , 2020, 87, 132-138.	5.3	2

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127	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. <i>Brain</i> , 2018, 141, e50-e50.	7.6	1
128	Long-term outcomes after epilepsy surgery, a retrospective cohort study linking patient-reported outcomes and routine healthcare data. <i>Epilepsy and Behavior</i> , 2020, 111, 107196.	1.7	1
129	Preface. <i>Advances in Protein Chemistry and Structural Biology</i> , 2012, 89, vii-ix.	2.3	0
130	GLRB is the third major gene of effect in hyperekplexia. <i>Human Molecular Genetics</i> , 2013, 22, 2552-2552.	2.9	0
131	TUBULINOPATHIES IN MALFORMATIONS OF THE CEREBRAL CORTEX. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, e4.139-e4.	1.9	0
132	MECHANISMS OF DISEASE IN THE HYPEREKPLEXIAS. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, e4.117-e4.	1.9	0
133	Hyperekplexia: Stiffness, startle and syncope. <i>Journal of Pediatric Neurology</i> , 2015, 08, 011-014.	0.2	0
134	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. <i>Epilepsia</i> , 2019, 60, 2194-2203.	5.1	0
135	The Cellular Localisation of GABAA and Glycine Receptors in the Human Basal Ganglia. <i>Advances in Behavioral Biology</i> , 2009, , 225-237.	0.2	0
136	204â€¦ Idiopathic intracranial hypertension in Wales: population characterisation, epidemiological trends and healthcare utilisation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, A72.1-A72.	1.9	0