Rhys H Thomas

List of Publications by Year
in descending order

103	4,625	147566	114278
papers	citations	31 h-index	63 g-index

108	108	108	9425
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Neurological and neuropsychiatric complications of COVID-19 in 153 patients: a UK-wide surveillance study. Lancet Psychiatry,the, 2020, 7, 875-882.	3.7	1,005
2	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	3.7	352
3	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
4	The hidden genetics of epilepsyâ€"a clinically important new paradigm. Nature Reviews Neurology, 2014, 10, 283-292.	4.9	232
5	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	4.9	190
6	Cognitive Performance Among Carriers of Pathogenic Copy Number Variants: Analysis of 152,000 UK Biobank Subjects. Biological Psychiatry, 2017, 82, 103-110.	0.7	168
7	Cerebral venous thrombosis after vaccination against COVID-19 in the UK: a multicentre cohort study. Lancet, The, 2021, 398, 1147-1156.	6.3	141
8	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	3.7	123
9	Neurological manifestations of SARS-CoV-2 infection in hospitalised children and adolescents in the UK: a prospective national cohort study. The Lancet Child and Adolescent Health, 2021, 5, 631-641.	2.7	114
10	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. Journal of Neuroscience, 2010, 30, 9612-9620.	1.7	112
11	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.5	97
12	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	4.7	97
13	Mutations in the GlyT2 Gene (SLC6A5) Are a Second Major Cause of Startle Disease. Journal of Biological Chemistry, 2012, 287, 28975-28985.	1.6	84
14	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology, 2015, 84, 951-958.	1.5	79
15	COVID-19 and psychosis risk: Real or delusional concern?. Neuroscience Letters, 2021, 741, 135491.	1.0	76
16	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74
17	Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. Brain, 2013, 136, 3085-3095.	3.7	66
18	Novel missense mutations in the glycine receptor Î² subunit gene (GLRB) in startle disease. Neurobiology of Disease, 2013, 52, 137-149.	2.1	54

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19	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 927-940.	1.4	50
20	Weight change associated with antiepileptic drugs. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 796-799.	0.9	48
21	The glycinergic system in human startle disease: a genetic screening approach. Frontiers in Molecular Neuroscience, 2010, 3, 8.	1.4	47
22	Epilepsy and deprivation, a data linkage study. Epilepsia, 2015, 56, 585-591.	2.6	45
23	Trends in the first antiepileptic drug prescribed for epilepsy between 2000 and 2010. Seizure: the Journal of the British Epilepsy Association, 2014, 23, 77-80.	0.9	44
24	Sudden death in epilepsy: Insights from the last 25 years. Seizure: the Journal of the British Epilepsy Association, 2017, 44, 232-236.	0.9	43
25	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.5	43
26	A Novel Dominant Hyperekplexia Mutation Y705C Alters Trafficking and Biochemical Properties of the Presynaptic Glycine Transporter GlyT2. Journal of Biological Chemistry, 2012, 287, 28986-29002.	1.6	42
27	The mitochondrial epilepsies. European Journal of Paediatric Neurology, 2020, 24, 47-52.	0.7	40
28	A Novel GABRG2 mutation, p.R136*, in a family with GEFS+ and extended phenotypes. Neurobiology of Disease, 2014, 64, 131-141.	2.1	39
29	Impulsive and episodic disorders of aggressive behaviour following traumatic brain injury. Brain Injury, 2013, 27, 253-261.	0.6	37
30	Epilepsy and seizures in young people with 22q11.2 deletion syndrome: Prevalence and links with other neurodevelopmental disorders. Epilepsia, 2019, 60, 818-829.	2.6	37
31	Considerations for causality assessment of neurological and neuropsychiatric complications of SARS-CoV-2 vaccines: from cerebral venous sinus thrombosis to functional neurological disorder. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 1144-1151.	0.9	37
32	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. Journal of Biological Chemistry, 2013, 288, 33745-33759.	1.6	35
33	Complement system biomarkers in epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 60, 1-7.	0.9	32
34	A comprehensive neuropsychological description of cognition in drug-refractory juvenile myoclonic epilepsy. Epilepsy and Behavior, 2014, 36, 124-129.	0.9	31
35	Epilepsy and bipolar disorder. Epilepsy and Behavior, 2015, 52, 267-274.	0.9	31
36	Cluster of atypical adult Guillain-BarrÃ© syndrome temporally associated with neurological illness due to EV-D68 in children, South Wales, United Kingdom, October 2015 to January 2016. Eurosurveillance, 2016, 21, .	3.9	31

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37	Epilepsy prevalence and socioeconomic deprivation in England. Epilepsia, 2014, 55, 1634-1641.	2.6	30
38	Concepts and controversies of juvenile myoclonic epilepsy: still an enigmatic epilepsy. Expert Review of Neurotherapeutics, 2014, 14, 819-831.	1.4	30
39	Executive functions and psychiatric symptoms in drug-refractory juvenile myoclonic epilepsy. Epilepsy and Behavior, 2014, 35, 72-77.	0.9	29
40	Characterisation of irradiation enhanced strain localisation in a zirconium alloy. Materialia, 2019, 5, 100248.	1.3	26
41	Pathogenic copy number variants and SCN1A mutations in patients with intellectual disability and childhood-onset epilepsy. BMC Medical Genetics, 2016, 17, 34.	2.1	23
42	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2020, 107, 683-697.	2.6	23
43	Next Generation Sequencing in the Clinical Domain: Clinical Advantages, Practical, and Ethical Challenges. Advances in Protein Chemistry and Structural Biology, 2012, 89, 27-63.	1.0	21
44	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	1.1	21
45	Epilepsy, an orphan disorder within the neurodevelopmental family. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1245-1247.	0.9	19
46	Generalized Epilepsy and Myoclonic Seizures in 22q11.2 Deletion Syndrome. Molecular Syndromology, 2016, 7, 239-246.	0.3	18
47	A comparative study of bone shortening and bone loss with use of saw blades versus burr in hallux valgus surgery. Foot and Ankle Surgery, 2012, 18, 195-197.	0.8	15
48	Cannabis and epilepsy. Practical Neurology, 2018, 18, 465-471.	0.5	15
49	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	0.7	15
50	Modeling seizures in the Human Phenotype Ontology according to contemporary ILAE concepts makes big phenotypic data tractable. Epilepsia, 2021, 62, 1293-1305.	2.6	15
51	Valproate: life-saving, life-changing. Clinical Medicine, 2018, 18, s1-s8.	0.8	13
52	Clinicians embracing social media: Potential and pitfalls. Epilepsy and Behavior, 2021, 115, 106462.	0.9	12
53	Juvenile myoclonic epilepsy. BMJ: British Medical Journal, 2012, 344, e360-e360.	2.4	11
54	Educational attainment of children born to mothers with epilepsy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 736-740.	0.9	11

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55	Characterising neuropsychiatric disorders in patients with COVID-19 â€“ Authors' reply. Lancet Psychiatry,the, 2020, 7, 934-935.	3.7	10
56	Early-onset genetic epilepsies reaching adult clinics. Brain, 2020, 143, e19-e19.	3.7	10
57	Sleepwalking and Sleep Paralysis: Prevalence in Colombian Families With Genetic Generalized Epilepsy. Journal of Child Neurology, 2019, 34, 491-498.	0.7	9
58	A tiered strategy for investigating status epilepticus. Seizure: the Journal of the British Epilepsy Association, 2020, 75, 165-173.	0.9	8
59	The evidence for switching dibenzazepines in people with epilepsy. Acta Neurologica Scandinavica, 2020, 142, 121-130.	1.0	8
60	How do doctors choose their specialty: first love, arranged marriage or second time around? And how may an affair with MMC change this?. Clinical Medicine, 2008, 8, 490-492.	0.8	7
61	Implications for families of advances in understanding the genetic basis of epilepsy. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 675-679.	0.9	7
62	COVID-19â€“Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.5	7
63	General paralysis of the insane. Practical Neurology, 2011, 11, 366-369.	0.5	6
64	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 26, 241-246.	0.9	6
65	The importance of the experiences of initial diagnosis and treatment failure when switching antiepileptic drugs. Epilepsy and Behavior, 2013, 29, 492-496.	0.9	5
66	Niemann-Pick type C: contemporary diagnosis and treatment of a classical disorder. Practical Neurology, 2019, 19, 420-423.	0.5	5
67	Paediatric sudden unexpected death in epilepsy: A parental report cohort. Acta Neurologica Scandinavica, 2021, 143, 509-513.	1.0	5
68	Fluctuations in <scp>EEG</scp> band power at subjectâ€specific timescales over minutes to days explain changes in seizure evolutions. Human Brain Mapping, 2022, 43, 2460-2477.	1.9	5
69	Research priorities for mitochondrial disorders: Current landscape and patient and professional views. Journal of Inherited Metabolic Disease, 2022, 45, 796-803.	1.7	5
70	Epilepsy: creative sparks. Practical Neurology, 2010, 10, 219-226.	0.5	4
71	Hyperekplexia: overexcitable and underdiagnosed. Developmental Medicine and Child Neurology, 2015, 57, 313-313.	1.1	4
72	On the road again: assessing driving ability in patients with neurological conditions. Practical Neurology, 2017, 17, 203-206.	0.5	4

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73	Is overnight tube feeding associated with hypoxia in stroke?. Age and Ageing, 2006, 35, 627-629.	0.7	3
74	Reversible grasp reflexes in normal pressure hydrocephalus. Clinical Neurology and Neurosurgery, 2009, 111, 387-389.	0.6	3
75	Learnings from deaths â€“ the Epilepsy Deaths Register. Epilepsy and Behavior, 2020, 103, 106454.	0.9	3
76	The prevalence of genetically diagnosable epilepsies in young adulthood: How many should we be looking for?. Epilepsia, 2020, 61, 2053-2054.	2.6	3
77	Self-driving cars: a qualitative study into the opportunities, challenges and perceived acceptability for people with epilepsy. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 781-782.	0.9	3
78	House calls: The case of the entertaining case. BMJ: British Medical Journal, 2009, 339, b5256-b5256.	2.4	3
79	Encephalitis guidelines: a recipe for success?. Clinical Medicine, 2009, 9, 210-211.	0.8	2
80	Epilepsy is different. Journal of the Royal Society of Medicine, 2011, 104, 141-143.	1.1	2
81	The consequences of valproate exposure in utero. Journal of Neurology, 2016, 263, 1887-1889.	1.8	2
82	Epilepsy treatment priorities: answering the questions that matter. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 999-1001.	0.9	2
83	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. Neurology, 2020, 94, 994-999.	1.5	2
84	The Effect of Loading Direction on Slip and Twinning in an Irradiated Zirconium Alloy. , 2021, , 233-261.		2
85	Injudicious antibiotic use leading to fulminating Clostridium difficile infection: a case report. Cases Journal, 2009, 2, 7978.	0.4	1
86	Invitation to participate in a prospective caseâ€“control study of sudden unexpected death in epilepsy. Epilepsia, 2021, 62, 1280-1281.	2.6	1
87	A neurological presentation of intravascular B-cell lymphoma. BMJ Case Reports, 2012, 2012, bcr2012006439-bcr2012006439.	0.2	1
88	Careers advice. BMJ: British Medical Journal, 2007, 334, 58.3-58.	2.4	0
89	The dark night. Practical Neurology, 2010, 10, 290-294.	0.5	0
90	Novel auto-antibody syndromes. Journal of Neurology, 2014, 261, 2043-2045.	1.8	0

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91	Hyperekplexia: Stiffness, startle and syncope. Journal of Pediatric Neurology, 2015, 08, 011-014.	0.0	0
92	Etiology of Epilepsy. Seminars in Neurology, 2015, 35, 191-192.	0.5	0
93	The future of medicine will be dark without international collaboration. Seizure: the Journal of the British Epilepsy Association, 2016, 41, 42.	0.9	0
94	What can rare variant genetics tell us about cognition and intellectual difficulties?. Journal of Neurology, 2016, 263, 2565-2566.	1.8	0
95	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	2.6	0
96	Genetic chameleons: remember the relapsing disorders. Practical Neurology, 2019, 19, 282-283.	0.5	0
97	Molecular genetic management of epilepsy. , 2020, , 289-308.		0
98	Response to letter to editor: â€œKnowing when and how to use epilepsy screening questionnairesâ€• Epilepsia, 2020, 61, 826-827.	2.6	0
99	Multi-Dimensional Study of the Effect of Early Slip Activity on Fatigue Crack Initiation in a Near-Î± Titanium Alloy. SSRN Electronic Journal, 0, , .	0.4	0
100	Understanding strain localisation behaviour in a near-Î± Ti-alloy during initial loading below the yield stress. MATEC Web of Conferences, 2020, 321, 11039.	0.1	0
101	Early slip activity and fatigue crack initiation of a near alpha titanium alloy. MATEC Web of Conferences, 2020, 321, 11040.	0.1	0
102	Multi-Dimensional Study of the Early Slip Activity on Crack Initiation in a Near Î± Titanium Alloy. SSRN Electronic Journal, 0, , .	0.4	0
103	The neurodevelopmental spectrum seen with CHD2 variants. Pediatric Investigation, 2022, 6, 147-148.	0.6	0