Rhys H Thomas

List of Publications by Year
in descending order

99 papers	4,614 citations	147566 31 h-index	114278 63 g-index
105 all docs	105 docs citations	105 times ranked	9202 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	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	3.7	123
8	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
9	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. Journal of Neuroscience, 2010, 30, 9612-9620.	1.7	112
10	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.5	97
11	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	4.7	97
12	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
13	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology, 2015, 84, 951-958.	1.5	79
14	COVID-19 and psychosis risk: Real or delusional concern?. Neuroscience Letters, 2021, 741, 135491.	1.0	76
15	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	2.7	74
16	Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. Brain, 2013, 136, 3085-3095.	3.7	66
17	Novel missense mutations in the glycine receptor Î² subunit gene (GLRB) in startle disease. Neurobiology of Disease, 2013, 52, 137-149.	2.1	54
18	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 927-940.	1.4	50

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19	Weight change associated with antiepileptic drugs. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 796-799.	0.9	48
20	The glycinergic system in human startle disease: a genetic screening approach. Frontiers in Molecular Neuroscience, 2010, 3, 8.	1.4	47
21	The <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	1.9	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	Spectrum, risk factors and outcomes of neurological and psychiatric complications of COVID-19: a UK-wide cross-sectional surveillance study. Brain Communications, 2021, 3, fcab168.	1.5	33
34	Complement system biomarkers in epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 60, 1-7.	0.9	32
35	A comprehensive neuropsychological description of cognition in drug-refractory juvenile myoclonic epilepsy. Epilepsy and Behavior, 2014, 36, 124-129.	0.9	31
36	Epilepsy and bipolar disorder. Epilepsy and Behavior, 2015, 52, 267-274.	0.9	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	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	3.7	25
41	Artificial intelligence for classification of temporal lobe epilepsy with ROI-level MRI data: A worldwide ENIGMA-Epilepsy study. NeuroImage: Clinical, 2021, 31, 102765.	1.4	25
42	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
43	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
44	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	1.8	22
45	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
46	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
47	Epilepsy, an orphan disorder within the neurodevelopmental family. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1245-1247.	0.9	19
48	Generalized Epilepsy and Myoclonic Seizures in 22q11.2 Deletion Syndrome. Molecular Syndromology, 2016, 7, 239-246.	0.3	18
49	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	3.7	18
50	Cannabis and epilepsy. Practical Neurology, 2018, 18, 465-471.	0.5	15
51	Expert opinion: use of valproate in girls and women of childbearing potential with epilepsy: recommendations and alternatives based on a review of the literature and clinical experienceâ€”a European perspective. Journal of Neurology, 2020, 268, 2735-2748.	1.8	15
52	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
53	Valproate: life-saving, life-changing. Clinical Medicine, 2018, 18, s1-s8.	0.8	13
54	Clinicians embracing social media: Potential and pitfalls. Epilepsy and Behavior, 2021, 115, 106462.	0.9	12

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55	Juvenile myoclonic epilepsy. BMJ: British Medical Journal, 2012, 344, e360-e360.	2.4	11
56	Educational attainment of children born to mothers with epilepsy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 736-740.	0.9	11
57	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.5	11
58	Characterising neuropsychiatric disorders in patients with COVID-19 â€“ Authors' reply. Lancet Psychiatry,the, 2020, 7, 934-935.	3.7	10
59	Early-onset genetic epilepsies reaching adult clinics. Brain, 2020, 143, e19-e19.	3.7	10
60	Computational analysis of neurodevelopmental phenotypes: Harmonization empowers clinical discovery. Human Mutation, 2022, 43, 1642-1658.	1.1	10
61	Sleepwalking and Sleep Paralysis: Prevalence in Colombian Families With Genetic Generalized Epilepsy. Journal of Child Neurology, 2019, 34, 491-498.	0.7	9
62	A tiered strategy for investigating status epilepticus. Seizure: the Journal of the British Epilepsy Association, 2020, 75, 165-173.	0.9	8
63	The evidence for switching dibenzazepines in people with epilepsy. Acta Neurologica Scandinavica, 2020, 142, 121-130.	1.0	8
64	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	2.6	8
65	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
66	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
67	COVID-19â€“Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.5	7
68	General paralysis of the insane. Practical Neurology, 2011, 11, 366-369.	0.5	6
69	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 26, 241-246.	0.9	6
70	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
71	Niemann-Pick type C: contemporary diagnosis and treatment of a classical disorder. Practical Neurology, 2019, 19, 420-423.	0.5	5
72	Paediatric sudden unexpected death in epilepsy: A parental report cohort. Acta Neurologica Scandinavica, 2021, 143, 509-513.	1.0	5

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73	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
74	Research priorities for mitochondrial disorders: Current landscape and patient and professional views. Journal of Inherited Metabolic Disease, 2022, 45, 796-803.	1.7	5
75	Volumetric and structural connectivity abnormalities co-localise in TLE. NeuroImage: Clinical, 2022, 35, 103105.	1.4	5
76	Partial wound closure of the Cincinnati incision in clubfoot correction. European Journal of Orthopaedic Surgery and Traumatology, 2005, 15, 28-31.	0.6	4
77	Epilepsy: creative sparks. Practical Neurology, 2010, 10, 219-226.	0.5	4
78	Hyperekplexia: overexcitable and underdiagnosed. Developmental Medicine and Child Neurology, 2015, 57, 313-313.	1.1	4
79	On the road again: assessing driving ability in patients with neurological conditions. Practical Neurology, 2017, 17, 203-206.	0.5	4
80	Is overnight tube feeding associated with hypoxia in stroke?. Age and Ageing, 2006, 35, 627-629.	0.7	3
81	Reversible grasp reflexes in normal pressure hydrocephalus. Clinical Neurology and Neurosurgery, 2009, 111, 387-389.	0.6	3
82	Learnings from deaths â€“ the Epilepsy Deaths Register. Epilepsy and Behavior, 2020, 103, 106454.	0.9	3
83	The prevalence of genetically diagnosable epilepsies in young adulthood: How many should we be looking for?. Epilepsia, 2020, 61, 2053-2054.	2.6	3
84	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
85	Epilepsy is different. Journal of the Royal Society of Medicine, 2011, 104, 141-143.	1.1	2
86	The consequences of valproate exposure in utero. Journal of Neurology, 2016, 263, 1887-1889.	1.8	2
87	Epilepsy treatment priorities: answering the questions that matter. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 999-1001.	0.9	2
88	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. Neurology, 2020, 94, 994-999.	1.5	2
89	Defining Causality in Neurological & Neuropsychiatric COVID-19 Vaccine Complications: What Have We Learnt from Current and Previous Vaccination Campaigns?. SSRN Electronic Journal, 0, , .	0.4	2
90	The view of the clinician and the scientist on the family experience of sudden epilepsy deaths. Epilepsy and Behavior, 2020, 103, 106679.	0.9	1

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91	Novel auto-antibody syndromes. Journal of Neurology, 2014, 261, 2043-2045.	1.8	0
92	Hyperekplexia: Stiffness, startle and syncope. Journal of Pediatric Neurology, 2015, 08, 011-014.	0.0	0
93	What can rare variant genetics tell us about cognition and intellectual difficulties?. Journal of Neurology, 2016, 263, 2565-2566.	1.8	0
94	PO054â€…Ten years of studying familial epilepsy in wales. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, A26.1-A26.	0.9	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	The neurodevelopmental spectrum seen with CHD2 variants. Pediatric Investigation, 2022, 6, 147-148.	0.6	0