Rhys H Thomas

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8168683/publications.pdf

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99 papers	4,614 citations	147801 31 h-index	114465 63 g-index
105 all docs	105 docs citations	105 times ranked	9202 citing authors

#	Article	IF	Citations
1	The <scp>ENIGMAâ€Epilepsy</scp> working group: Mapping disease from large data sets. Human Brain Mapping, 2022, 43, 113-128.	3.6	47
2	Forecasting stroke-like episodes and outcomes in mitochondrial disease. Brain, 2022, 145, 542-554.	7.6	25
3	A systemsâ€level analysis highlights microglial activation as a modifying factor in common epilepsies. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	22
4	Topographic divergence of atypical cortical asymmetry and atrophy patterns in temporal lobe epilepsy. Brain, 2022, 145, 1285-1298.	7.6	18
5	Association of ultraâ€rare coding variants with genetic generalized epilepsy: A caseâ€“control whole exome sequencing study. Epilepsia, 2022, 63, 723-735.	5.1	8
6	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.	3.6	5
7	COVID-19â€“Related Outcomes in Primary Mitochondrial Diseases. Neurology, 2022, 98, 576-582.	1.1	7
8	Computational analysis of neurodevelopmental phenotypes: Harmonization empowers clinical discovery. Human Mutation, 2022, 43, 1642-1658.	2.5	10
9	The neurodevelopmental spectrum seen with CHD2 variants. Pediatric Investigation, 2022, 6, 147-148.	1.4	0
10	Research priorities for mitochondrial disorders: Current landscape and patient and professional views. Journal of Inherited Metabolic Disease, 2022, 45, 796-803.	3.6	5
11	Volumetric and structural connectivity abnormalities co-localise in TLE. NeuroImage: Clinical, 2022, 35, 103105.	2.7	5
12	Paediatric sudden unexpected death in epilepsy: A parental report cohort. Acta Neurologica Scandinavica, 2021, 143, 509-513.	2.1	5
13	COVID-19 and psychosis risk: Real or delusional concern?. Neuroscience Letters, 2021, 741, 135491.	2.1	76
14	Clinicians embracing social media: Potential and pitfalls. Epilepsy and Behavior, 2021, 115, 106462.	1.7	12
15	Artificial intelligence for classification of temporal lobe epilepsy with ROI-level MRI data: A worldwide ENIGMA-Epilepsy study. NeuroImage: Clinical, 2021, 31, 102765.	2.7	25
16	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.	3.3	33
17	Modeling seizures in the Human Phenotype Ontology according to contemporary ILAE concepts makes big phenotypic data tractable. Epilepsia, 2021, 62, 1293-1305.	5.1	15
18	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.1	11

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19	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.	1.9	37
20	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.	5.6	114
21	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.	2.4	21
22	Learnings from deaths â€“ the Epilepsy Deaths Register. Epilepsy and Behavior, 2020, 103, 106454.	1.7	3
23	A tiered strategy for investigating status epilepticus. Seizure: the Journal of the British Epilepsy Association, 2020, 75, 165-173.	2.0	8
24	The mitochondrial epilepsies. European Journal of Paediatric Neurology, 2020, 24, 47-52.	1.6	40
25	Molecular genetic management of epilepsy. , 2020, , 289-308.		0
26	The view of the clinician and the scientist on the family experience of sudden epilepsy deaths. Epilepsy and Behavior, 2020, 103, 106679.	1.7	1
27	Characterising neuropsychiatric disorders in patients with COVID-19 â€“ Authors' reply. Lancet Psychiatry,the, 2020, 7, 934-935.	7.4	10
28	Network-based atrophy modeling in the common epilepsies: A worldwide ENIGMA study. Science Advances, 2020, 6, .	10.3	97
29	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2020, 107, 683-697.	6.2	23
30	The prevalence of genetically diagnosable epilepsies in young adulthood: How many should we be looking for?. Epilepsia, 2020, 61, 2053-2054.	5.1	3
31	Epilepsy, an orphan disorder within the neurodevelopmental family. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1245-1247.	1.9	19
32	White matter abnormalities across different epilepsy syndromes in adults: an ENIGMA-Epilepsy study. Brain, 2020, 143, 2454-2473.	7.6	123
33	Clinical Reasoning: A 25-year-old woman with recurrent episodes of collapse and loss of consciousness. Neurology, 2020, 94, 994-999.	1.1	2
34	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.	1.9	3
35	Response to letter to editor: â€œKnowing when and how to use epilepsy screening questionnairesâ€• Epilepsia, 2020, 61, 826-827.	5.1	0
36	Neurological and neuropsychiatric complications of COVID-19 in 153 patients: a UK-wide surveillance study. Lancet Psychiatry,the, 2020, 7, 875-882.	7.4	1,005

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37	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.	3.6	15
38	The evidence for switching dibenzazepines in people with epilepsy. Acta Neurologica Scandinavica, 2020, 142, 121-130.	2.1	8
39	Early-onset genetic epilepsies reaching adult clinics. Brain, 2020, 143, e19-e19.	7.6	10
40	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	5.1	0
41	Niemann-Pick type C: contemporary diagnosis and treatment of a classical disorder. Practical Neurology, 2019, 19, 420-423.	1.1	5
42	Sleepwalking and Sleep Paralysis: Prevalence in Colombian Families With Genetic Generalized Epilepsy. Journal of Child Neurology, 2019, 34, 491-498.	1.4	9
43	Epilepsy and seizures in young people with 22q11.2 deletion syndrome: Prevalence and links with other neurodevelopmental disorders. Epilepsia, 2019, 60, 818-829.	5.1	37
44	Genetic chameleons: remember the relapsing disorders. Practical Neurology, 2019, 19, 282-283.	1.1	0
45	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.1	43
46	Structural brain abnormalities in the common epilepsies assessed in a worldwide ENIGMA study. Brain, 2018, 141, 391-408.	7.6	352
47	Valproate: life-saving, life-changing. Clinical Medicine, 2018, 18, s1-s8.	1.9	13
48	Educational attainment of children born to mothers with epilepsy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 736-740.	1.9	11
49	Neurologic phenotypes associated with COL4A1 / 2 mutations. Neurology, 2018, 91, e2078-e2088.	1.1	97
50	Cannabis and epilepsy. Practical Neurology, 2018, 18, 465-471.	1.1	15
51	Complement system biomarkers in epilepsy. Seizure: the Journal of the British Epilepsy Association, 2018, 60, 1-7.	2.0	32
52	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
53	On the road again: assessing driving ability in patients with neurological conditions. Practical Neurology, 2017, 17, 203-206.	1.1	4
54	Epilepsy treatment priorities: answering the questions that matter. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 999-1001.	1.9	2

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55	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
56	Sudden death in epilepsy: Insights from the last 25 years. Seizure: the Journal of the British Epilepsy Association, 2017, 44, 232-236.	2.0	43
57	Cognitive Performance Among Carriers of Pathogenic Copy Number Variants: Analysis of 152,000 UK Biobank Subjects. Biological Psychiatry, 2017, 82, 103-110.	1.3	168
58	PO054â€...Ten years of studying familial epilepsy in wales. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, A26.1-A26.	1.9	0
59	Generalized Epilepsy and Myoclonic Seizures in 22q11.2 Deletion Syndrome. Molecular Syndromology, 2016, 7, 239-246.	0.8	18
60	The consequences of valproate exposure in utero. Journal of Neurology, 2016, 263, 1887-1889.	3.6	2
61	What can rare variant genetics tell us about cognition and intellectual difficulties?. Journal of Neurology, 2016, 263, 2565-2566.	3.6	0
62	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
63	Hyperekplexia: Stiffness, startle and syncope. Journal of Pediatric Neurology, 2015, 08, 011-014.	0.2	0
64	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
65	CHD2 myoclonic encephalopathy is frequently associated with self-induced seizures. Neurology, 2015, 84, 951-958.	1.1	79
66	Hyperekplexia: overexcitable and underdiagnosed. Developmental Medicine and Child Neurology, 2015, 57, 313-313.	2.1	4
67	Epilepsy and deprivation, a data linkage study. Epilepsia, 2015, 56, 585-591.	5.1	45
68	Epilepsy and bipolar disorder. Epilepsy and Behavior, 2015, 52, 267-274.	1.7	31
69	Epilepsy prevalence and socioeconomic deprivation in England. Epilepsia, 2014, 55, 1634-1641.	5.1	30
70	A Novel GABRG2 mutation, p.R136*, in a family with GEFS+ and extended phenotypes. Neurobiology of Disease, 2014, 64, 131-141.	4.4	39
71	The hidden genetics of epilepsyâ€"a clinically important new paradigm. Nature Reviews Neurology, 2014, 10, 283-292.	10.1	232
72	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.	2.0	44

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73	Novel auto-antibody syndromes. Journal of Neurology, 2014, 261, 2043-2045.	3.6	0
74	Concepts and controversies of juvenile myoclonic epilepsy: still an enigmatic epilepsy. Expert Review of Neurotherapeutics, 2014, 14, 819-831.	2.8	30
75	Executive functions and psychiatric symptoms in drug-refractory juvenile myoclonic epilepsy. Epilepsy and Behavior, 2014, 35, 72-77.	1.7	29
76	A comprehensive neuropsychological description of cognition in drug-refractory juvenile myoclonic epilepsy. Epilepsy and Behavior, 2014, 36, 124-129.	1.7	31
77	Impulsive and episodic disorders of aggressive behaviour following traumatic brain injury. Brain Injury, 2013, 27, 253-261.	1.2	37
78	The importance of the experiences of initial diagnosis and treatment failure when switching antiepileptic drugs. Epilepsy and Behavior, 2013, 29, 492-496.	1.7	5
79	Novel missense mutations in the glycine receptor Î² subunit gene (GLRB) in startle disease. Neurobiology of Disease, 2013, 52, 137-149.	4.4	54
80	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 26, 241-246.	1.7	6
81	Weight change associated with antiepileptic drugs. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 796-799.	1.9	48
82	Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. Brain, 2013, 136, 3085-3095.	7.6	66
83	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 927-940.	2.9	50
84	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. Journal of Biological Chemistry, 2013, 288, 33745-33759.	3.4	35
85	Mutations in the GlyT2 Gene (SLC6A5) Are a Second Major Cause of Startle Disease. Journal of Biological Chemistry, 2012, 287, 28975-28985.	3.4	84
86	Juvenile myoclonic epilepsy. BMJ: British Medical Journal, 2012, 344, e360-e360.	2.3	11
87	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.	3.4	42
88	Next Generation Sequencing in the Clinical Domain: Clinical Advantages, Practical, and Ethical Challenges. Advances in Protein Chemistry and Structural Biology, 2012, 89, 27-63.	2.3	21
89	General paralysis of the insane. Practical Neurology, 2011, 11, 366-369.	1.1	6
90	Epilepsy is different. Journal of the Royal Society of Medicine, 2011, 104, 141-143.	2.0	2

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91	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. Journal of Neuroscience, 2010, 30, 9612-9620.	3.6	112
92	The glycinergic system in human startle disease: a genetic screening approach. Frontiers in Molecular Neuroscience, 2010, 3, 8.	2.9	47
93	Epilepsy: creative sparks. Practical Neurology, 2010, 10, 219-226.	1.1	4
94	Implications for families of advances in understanding the genetic basis of epilepsy. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 675-679.	2.0	7
95	Reversible grasp reflexes in normal pressure hydrocephalus. Clinical Neurology and Neurosurgery, 2009, 111, 387-389.	1.4	3
96	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.	1.9	7
97	Is overnight tube feeding associated with hypoxia in stroke?. Age and Ageing, 2006, 35, 627-629.	1.6	3
98	Partial wound closure of the Cincinnati incision in clubfoot correction. European Journal of Orthopaedic Surgery and Traumatology, 2005, 15, 28-31.	1.4	4
99	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