Mark I Rees

List of Publications by Year
in descending order

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136 papers	7,043 citations	53794 45 h-index	71685 76 g-index
137 all docs	137 docs citations	137 times ranked	9324 citing authors

#	Article	IF	Citations
1	Epilepsy mortality in Wales during COVID-19. Seizure: the Journal of the British Epilepsy Association, 2022, 94, 39-42.	2.0	7
2	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
3	Sex-specific disease modifiers in juvenile myoclonic epilepsy. Scientific Reports, 2022, 12, 2785.	3.3	19
4	204â€… Idiopathic intracranial hypertension in Wales: population characterisation, epidemiological trends and healthcare utilisation. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A72.1-A72.	1.9	0
5	Trait impulsivity in Juvenile Myoclonic Epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 138-152.	3.7	21
6	Incidence, Prevalence, and Health Care Outcomes in Idiopathic Intracranial Hypertension. Neurology, 2021, 96, .	1.1	42
7	Missense variants in the N-terminal domain of the A isoform of FHF2/FGF13 cause an X-linked developmental and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 176-185.	6.2	20
8	Fertile Crescent crop progenitors gained a competitive advantage from large seedlings. Ecology and Evolution, 2021, 11, 3300-3312.	1.9	7
9	Epilepsy, antiepileptic drugs, and the risk of major cardiovascular events. Epilepsia, 2021, 62, 1604-1616.	5.1	27
10	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	6.2	35
11	The â€œmaternal effectâ€•on epilepsy risk: Analysis of familial epilepsies and reassessment of prior evidence. Annals of Neurology, 2020, 87, 132-138.	5.3	2
12	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
13	Long-term outcomes after epilepsy surgery, a retrospective cohort study linking patient-reported outcomes and routine healthcare data. Epilepsy and Behavior, 2020, 111, 107196.	1.7	1
14	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. Annals of Neurology, 2020, 87, 897-906.	5.3	9
15	Substantial subpial cortical demyelination in progressive multiple sclerosis: have we underestimated the extent of cortical pathology?. Neuroimmunology and Neuroinflammation, 2020, , .	1.4	3
16	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
17	Quantitative analysis of phenotypic elements augments traditional electroclinical classification of common familial epilepsies. Epilepsia, 2019, 60, 2194-2203.	5.1	0
18	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	7.6	90

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19	Epilepsy in families: Age at onset is a familial trait, independent of syndrome. Annals of Neurology, 2019, 86, 91-98.	5.3	11
20	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2019, 380, e24.	27.0	4
21	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. BMJ Open, 2019, 9, e023232.	1.9	39
22	Clinical spectrum of STX1B -related epileptic disorders. Neurology, 2019, 92, e1238-e1249.	1.1	43
23	Development of a cardiac inherited disease service and clinical registry: A 15-year perspective. American Heart Journal, 2019, 209, 126-130.	2.7	10
24	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661.	7.6	52
25	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. Brain, 2018, 141, 698-712.	7.6	72
26	Cereal progenitors differ in stand harvest characteristics from related wild grasses. Journal of Ecology, 2018, 106, 1286-1297.	4.0	11
27	Educational attainment of children born to mothers with epilepsy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 736-740.	1.9	11
28	Meningeal inflammation and cortical demyelination in acute multiple sclerosis. Annals of Neurology, 2018, 84, 829-842.	5.3	96
29	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. Brain, 2018, 141, e55-e55.	7.6	11
30	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50.	7.6	1
31	Tubulin genes and malformations of cortical development. European Journal of Medical Genetics, 2018, 61, 744-754.	1.3	93
32	The phenotype of bilateral hippocampal sclerosis and its management in â€œreal lifeâ€•clinical settings. Epilepsia, 2018, 59, 1410-1420.	5.1	6
33	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
34	Validating epilepsy diagnoses in routinely collected data. Seizure: the Journal of the British Epilepsy Association, 2017, 52, 195-198.	2.0	48
35	Hyperekplexia: Report on phenotype and genotype of 16 Jordanian patients. Brain and Development, 2017, 39, 306-311.	1.1	11
36	How did the domestication of Fertile Crescent grain crops increase their yields?. Functional Ecology, 2017, 31, 387-397.	3.6	93

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37	Phenotypic analysis of 303 multiplex families with common epilepsies. Brain, 2017, 140, 2144-2156.	7.6	23
38	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
39	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
40	Complement is activated in progressive multiple sclerosis cortical grey matter lesions. Journal of Neuroinflammation, 2016, 13, 161.	7.2	101
41	Hyperekplexia: Stiffness, startle and syncope. Journal of Pediatric Neurology, 2015, 08, 011-014.	0.2	0
42	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
43	Ethnicity can predict GLRA1 genotypes in hyperekplexia. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 341-343.	1.9	9
44	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Human Molecular Genetics, 2015, 24, 5313-5325.	2.9	77
45	Were Fertile Crescent crop progenitors higher yielding than other wild species that were never domesticated?. New Phytologist, 2015, 207, 905-913.	7.3	26
46	Epilepsy and deprivation, a data linkage study. Epilepsia, 2015, 56, 585-591.	5.1	45
47	Neonatal hyperekplexia with homozygous p.R392H mutation in GLRA1. Epileptic Disorders, 2014, 16, 354-357.	1.3	2
48	TUBULINOPATHIES IN MALFORMATIONS OF THE CEREBRAL CORTEX. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.139-e4.	1.9	0
49	MECHANISMS OF DISEASE IN THE HYPEREKPLEXIAS. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.117-e4.	1.9	0
50	De Novo Mutations in the Beta-Tubulin Gene TUBB2A Cause Simplified Gyral Patterning and Infantile-Onset Epilepsy. American Journal of Human Genetics, 2014, 94, 634-641.	6.2	99
51	A Novel GABRG2 mutation, p.R136*, in a family with GEFS+ and extended phenotypes. Neurobiology of Disease, 2014, 64, 131-141.	4.4	39
52	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
53	Executive functions and psychiatric symptoms in drug-refractory juvenile myoclonic epilepsy. Epilepsy and Behavior, 2014, 35, 72-77.	1.7	29
54	A comprehensive neuropsychological description of cognition in drug-refractory juvenile myoclonic epilepsy. Epilepsy and Behavior, 2014, 36, 124-129.	1.7	31

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55	Functional Traits Differ between Cereal Crop Progenitors and Other Wild Grasses Gathered in the Neolithic Fertile Crescent. PLoS ONE, 2014, 9, e87586.	2.5	41
56	Ghrelin inhibits LPS-induced release of IL-6 from mouse dopaminergic neurones. Journal of Neuroinflammation, 2013, 10, 40.	7.2	41
57	Flowâ€induced ATP release in patientâ€specific arterial geometries â€“ a comparative study of computational models. International Journal for Numerical Methods in Biomedical Engineering, 2013, 29, 1038-1056.	2.1	13
58	Dynamic changes in myelin aberrations and oligodendrocyte generation in chronic amyloidosis in mice and men. Glia, 2013, 61, 273-286.	4.9	155
59	Novel missense mutations in the glycine receptor Î² subunit gene (GLRB) in startle disease. Neurobiology of Disease, 2013, 52, 137-149.	4.4	54
60	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 26, 241-246.	1.7	6
61	Weight change associated with antiepileptic drugs. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 796-799.	1.9	48
62	Overlapping cortical malformations and mutations in TUBB2B and TUBA1A. Brain, 2013, 136, 536-548.	7.6	133
63	Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. Brain, 2013, 136, 3085-3095.	7.6	66
64	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 927-940.	2.9	50
65	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. Journal of Biological Chemistry, 2013, 288, 33745-33759.	3.4	35
66	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 2552-2552.	2.9	0
67	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
68	Preface. Advances in Protein Chemistry and Structural Biology, 2012, 89, vii-ix.	2.3	0
69	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
70	Genetic epilepsy with febrile seizures plus: definite and borderline phenotypes. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 336-338.	1.9	8
71	Opportunities and Challenges for Genome Sequencing in the Clinic. Advances in Protein Chemistry and Structural Biology, 2012, 89, 65-83.	2.3	9
72	Polymorphisms in Neuropsychiatric and Neuroinflammatory Disorders and the Role of Next Generation Sequencing in Early Diagnosis and Treatment. Advances in Protein Chemistry and Structural Biology, 2012, 89, 85-116.	2.3	2

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73	Next Generation Sequencing Methodologies - An Overview. Advances in Protein Chemistry and Structural Biology, 2012, 89, 1-26.	2.3	21
74	Structural Modelling Pipelines in Next Generation Sequencing Projects. Advances in Protein Chemistry and Structural Biology, 2012, 89, 117-167.	2.3	19
75	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
76	Elevated serum gastrin levels in Jervell and Lange-Nielsen syndrome: A marker of severe KCNQ1 dysfunction?. Heart Rhythm, 2011, 8, 551-554.	0.7	26
77	Symptoms and Signs Associated with Syncope in Young People with Primary Cardiac Arrhythmias. Heart Lung and Circulation, 2011, 20, 593-598.	0.4	27
78	Startle disease in Irish wolfhounds associated with a microdeletion in the glycine transporter GlyT2 gene. Neurobiology of Disease, 2011, 43, 184-189.	4.4	43
79	Differential localization of Î³â€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. Journal of Comparative Neurology, 2010, 518, 305-328.	1.6	48
80	Optimized sample preparation for highâ€resolution AFM characterization of fixed human cells. Journal of Microscopy, 2010, 240, 111-121.	1.8	29
81	An ovine transgenic Huntington's disease model. Human Molecular Genetics, 2010, 19, 1873-1882.	2.9	166
82	Identifying and prioritising epilepsy treatment uncertainties. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 918-921.	1.9	22
83	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
84	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. Journal of Neuroscience, 2010, 30, 9612-9620.	3.6	112
85	Fine architecture and mutation mapping of human brain inhibitory system ligand gated ion channels by high-throughput homology modeling. Advances in Protein Chemistry and Structural Biology, 2010, 80, 117-152.	2.3	10
86	The glycinergic system in human startle disease: a genetic screening approach. Frontiers in Molecular Neuroscience, 2010, 3, 8.	2.9	47
87	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
88	The genetics of epilepsyâ€"The past, the present and future. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 680-683.	2.0	30
89	Posthumous diagnosis of long QT syndrome from neonatal screening cards. Heart Rhythm, 2010, 7, 481-486.	0.7	56
90	Localisation of glycine receptors in the human forebrain, brainstem, and cervical spinal cord: an immunohistochemical review. Frontiers in Molecular Neuroscience, 2009, 2, 25.	2.9	54

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91	Biophysical Properties of 9 KCNQ1 Mutations Associated With Long-QT Syndrome. Circulation: Arrhythmia and Electrophysiology, 2009, 2, 417-426.	4.8	43
92	Phosphoâ€STAT5 and phosphoâ€Akt expression in chronic myeloproliferative neoplasms. British Journal of Haematology, 2009, 147, 495-506.	2.5	65
93	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. Journal of Cardiovascular Electrophysiology, 2009, 20, 923-930.	1.7	54
94	Misdiagnosis of Long QT Syndrome as Epilepsy at First Presentation. Annals of Emergency Medicine, 2009, 54, 26-32.	0.6	103
95	Epilepsy genetics: clinical beginnings and social consequences. QJM - Monthly Journal of the Association of Physicians, 2009, 102, 497-499.	0.5	4
96	The Cellular Localisation of GABAA and Glycine Receptors in the Human Basal Ganglia. Advances in Behavioral Biology, 2009, , 225-237.	0.2	0
97	Differential localization of GABA_A receptor subunits within the substantia nigra of the human brain: An immunohistochemical study. Journal of Comparative Neurology, 2008, 506, 912-929.	1.6	31
98	The genetics of hyperekplexia: more than startle!. Trends in Genetics, 2008, 24, 439-447.	6.7	187
99	Identification of large gene deletions and duplications in KCNQ1 and KCNH2 in patients with long QT syndrome. Heart Rhythm, 2008, 5, 1275-1281.	0.7	79
100	A critical role for glycine transporters in hyperexcitability disorders. Frontiers in Molecular Neuroscience, 2008, 1, 1.	2.9	37
101	Brugada Syndrome Masquerading as Febrile Seizures. Pediatrics, 2007, 119, e1206-e1211.	2.1	64
102	Sox-2 is expressed by glial and progenitor cells and Pax-6 is expressed by neuroblasts in the human subventricular zone. Experimental Neurology, 2007, 204, 828-831.	4.1	33
103	PICK1 interacts with Î±7 neuronal nicotinic acetylcholine receptors and controls their clustering. Molecular and Cellular Neurosciences, 2007, 35, 339-355.	2.2	32
104	Coinheritance of long QT syndrome and Kearns-Sayre syndrome. Heart Rhythm, 2007, 4, 1568-1572.	0.7	15
105	Long QT and Brugada syndrome gene mutations in New Zealand. Heart Rhythm, 2007, 4, 1306-1314.	0.7	41
106	Glycine receptors in the striatum, globus pallidus, and substantia nigra of the human brain: An immunohistochemical study. Journal of Comparative Neurology, 2007, 502, 1012-1029.	1.6	40
107	The conundrum of complexity in epilepsy. Lancet Neurology, The, 2007, 6, 943-944.	10.2	3
108	QT interval prolongation associated with sibutramine treatment. British Journal of Clinical Pharmacology, 2006, 61, 464-469.	2.4	38

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109	Mutations in the gene encoding GlyT2 (SLC6A5) define a presynaptic component of human startle disease. Nature Genetics, 2006, 38, 801-806.	21.4	232
110	Immunohistochemical staining of post-mortem adult human brain sections. Nature Protocols, 2006, 1, 2719-2732.	12.0	155
111	Functional variants of antioxidant genes in smokers with COPD and in those with normal lung function. Thorax, 2006, 61, 394-399.	5.6	120
112	Harris syndrome - a geographic perspective. Journal of Thrombosis and Haemostasis, 2005, 3, 2581-2582.	3.8	24
113	Near-miss SIDS due to Brugada syndrome. Archives of Disease in Childhood, 2005, 90, 528-529.	1.9	72
114	The GDP-GTP Exchange Factor Collybistin: An Essential Determinant of Neuronal Gephyrin Clustering. Journal of Neuroscience, 2004, 24, 5816-5826.	3.6	239
115	TBP, a polyglutamine tract containing protein, accumulates in Alzheimer's disease. Molecular Brain Research, 2004, 125, 120-128.	2.3	21
116	GABAA receptor subunit and gephyrin protein changes differ in the globus pallidus in Huntington's diseased brain. Brain Research, 2003, 994, 265-270.	2.2	18
117	Distribution of gephyrin in the human brain: an immunohistochemical analysis. Neuroscience, 2003, 116, 145-156.	2.3	22
118	Association of gephyrin and glycine receptors in the human brainstem and spinal cord: an immunohistochemical analysis. Neuroscience, 2003, 122, 773-784.	2.3	33
119	Molecular investigation of TBP allele length:. Neurobiology of Disease, 2003, 13, 37-45.	4.4	31
120	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
121	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
122	Characterisation, mutation detection, and association analysis of alternative promoters and 5â€² UTRs of the human dopamine D3 receptor gene in schizophrenia. Molecular Psychiatry, 2002, 7, 493-502.	7.9	35
123	Compound heterozygosity and nonsense mutations in the Î±1-subunit of the inhibitory glycine receptor in hyperekplexia. Human Genetics, 2001, 109, 267-270.	3.8	72
124	A genomewide linkage study of age at onset in schizophrenia. American Journal of Medical Genetics Part A, 2001, 105, 439-445.	2.4	63
125	Chromosome 2 interstitial deletion (del(2)(q14.1q21)) associated with connective tissue laxity and an attention deficit disorder. Journal of Medical Genetics, 2001, 38, 493-496.	3.2	13
126	An unusual case of hyperekplexia. European Journal of Paediatric Neurology, 2000, 4, 77-80.	1.6	7

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127	Further evidence of autosomal dominant congenital zonular pulverulent cataracts linked to 13q11 (CZP3) and a novel mutation in connexin 46 (GJA 3). Human Genetics, 2000, 106, 206-209.	3.8	68
128	Linkage analysis in an autosomal dominant â€˜zonular nuclear pulverulentâ€™ congenital cataract, mapped to chromosome 13q11-13. Eye, 2000, 14, 172-175.	2.1	3
129	A Two-Stage Genome Scan for Schizophrenia Susceptibility Genes in 196 Affected Sibling Pairs. Human Molecular Genetics, 1999, 8, 1729-1739.	2.9	136
130	Autosome search for schizophrenia susceptibility genes in multiply affected families. Molecular Psychiatry, 1999, 4, 353-359.	7.9	20
131	Bipolar disorder and the serotonin transporter gene: a family-based association study. Psychological Medicine, 1999, 29, 1249-1254.	4.5	41
132	Association studies of bipolar disorder at the human serotonin transporter gene (hSERT;â€‰5HTT). Molecular Psychiatry, 1997, 2, 398-402.	7.9	145
133	Additional support for schizophrenia linkage on chromosomes 6 and 8: A multicenter study. , 1996, 67, 580-594.		166
134	No evidence for allelic association between bipolar disorder and monoamine oxidase a gene polymorphisms. American Journal of Medical Genetics Part A, 1995, 60, 322-324.	2.4	64
135	Evidence for recessive as well as dominant forms of startle disease (hyperekplexia) caused by mutations in the Î±1 subunit of the inhibitory glycine receptor. Human Molecular Genetics, 1994, 3, 2175-2179.	2.9	151
136	Red cell dimorphism in a young man with a constitutional chromosomal translocation t(11;22)(p15.5;q11.21). British Journal of Haematology, 1994, 87, 386-395.	2.5	11