Guido Rubboli

List of Publications by Year
in descending order

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116 papers	4,997 citations	66343 42 h-index	106344 65 g-index
122 all docs	122 docs citations	122 times ranked	5433 citing authors

#	Article	IF	Citations
1	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 2017, 140, 1316-1336.	7.6	426
2	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Annals of Neurology, 2016, 79, 120-131.	5.3	190
3	Eyelid myoclonia with absences (Jeavons syndrome): A wellâ€defined idiopathic generalized epilepsy syndrome or a spectrum of photosensitive conditions?. Epilepsia, 2009, 50, 15-19.	5.1	156
4	EEG Diagnostic Procedures and Special Investigations in the Assessment of Photosensitivity. Epilepsia, 2004, 45, 35-39.	5.1	148
5	Consensus on diagnosis and management of JME: From founder's observations to current trends. Epilepsy and Behavior, 2013, 28, S87-S90.	1.7	142
6	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137
7	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	1.9	125
8	Cognition and Paroxysmal EEG Activities: From a Single Spike to Electrical Status Epilepticus during Sleep. Epilepsia, 2006, 47, 40-43.	5.1	119
9	Clinical spectrum and genotypeâ€“phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	7.6	117
10	The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124.	1.1	114
11	Phenotypic spectrum of GABRA1. Neurology, 2016, 87, 1140-1151.	1.1	113
12	Gene Panel Testing in Epileptic Encephalopathies and Familial Epilepsies. Molecular Syndromology, 2016, 7, 210-219.	0.8	103
13	Seizure outcome of epilepsy surgery in focal epilepsies associated with temporomesial glioneuronal tumors: lesionectomy compared with tailored resection. Journal of Neurosurgery, 2009, 111, 1275-1282.	1.6	101
14	MicroRNA profiles in hippocampal granule cells and plasma of rats with pilocarpine-induced epilepsy â€“ comparison with human epileptic samples. Scientific Reports, 2015, 5, 14143.	3.3	101
15	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	5.1	99
16	Standardized Computerâ€based Organized Reporting of <scp>EEG</scp>:<scp> SCORE</scp>. Epilepsia, 2013, 54, 1112-1124.	5.1	97
17	Mutations in GABRB3. Neurology, 2017, 88, 483-492.	1.1	87
18	Standardized computer-based organized reporting of EEG: SCORE â€“ Second version. Clinical Neurophysiology, 2017, 128, 2334-2346.	1.5	82

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19	The role of EEG in the diagnosis and classification of the epilepsy syndromes: a tool for clinical practice by the ILAE Neurophysiology Task Force (Part 1). Epileptic Disorders, 2017, 19, 233-298.	1.3	79
20	Electromagnetic source imaging in presurgical workup of patients with epilepsy. Neurology, 2019, 92, e576-e586.	1.1	71
21	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	5.1	70
22	Current standards of neuropsychological assessment in epilepsy surgery centers across Europe. Epilepsia, 2017, 58, 343-355.	5.1	69
23	Myoclonus epilepsy and ataxia due to <scp>KCNC</scp>1 mutation: Analysis of 20 cases and <scp>K</scp>⁺ channel properties. Annals of Neurology, 2017, 81, 677-689.	5.3	69
24	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Brain, 2022, 145, 2991-3009.	7.6	69
25	Current practices in long-term video-EEG monitoring services: A survey among partners of the E-PILEPSY pilot network of reference for refractory epilepsy and epilepsy surgery. Seizure: the Journal of the British Epilepsy Association, 2016, 38, 38-45.	2.0	67
26	Clinical and neurophysiologic features of progressive myoclonus epilepsy without renal failure caused bySCARB2mutations. Epilepsia, 2011, 52, 2356-2363.	5.1	63
27	Treatment Responsiveness in KCNT1-Related Epilepsy. Neurotherapeutics, 2019, 16, 848-857.	4.4	60
28	Utility of genetic testing for therapeutic decisionâ€making in adults with epilepsy. Epilepsia, 2020, 61, 1234-1239.	5.1	60
29	Seizure outcome in surgically treated drug-resistant mesial temporal lobe epilepsy based on the recent histopathological classifications. Journal of Neurosurgery, 2013, 119, 37-47.	1.6	59
30	Epilepsy associated tumors: Review article. World Journal of Clinical Cases, 2014, 2, 623.	0.8	58
31	Neurophysiology of juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 28, S30-S39.	1.7	53
32	Negative myoclonus induced by cortical electrical stimulation in epileptic patients. Brain, 2006, 129, 65-81.	7.6	52
33	Testing patients during seizures: A European consensus procedure developed by a joint taskforce of the <scp>ILAE</scp> â€“ Commission on European Affairs and the European Epilepsy Monitoring Unit Association. Epilepsia, 2016, 57, 1363-1368.	5.1	51
34	Diagnostic added value of electrical source imaging in presurgical evaluation of patients with epilepsy: A prospective study. Clinical Neurophysiology, 2020, 131, 324-329.	1.5	51
35	Seizure outcome of surgical treatment of focal epilepsy associated with low-grade tumors in children. Journal of Neurosurgery: Pediatrics, 2013, 11, 214-223.	1.3	50
36	The role of EEG in the diagnosis and classification of the epilepsy syndromes: a tool for clinical practice by the ILAE Neurophysiology Task Force (Part 2). Epileptic Disorders, 2017, 19, 385-437.	1.3	48

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37	Early mortality in SCN8A -related epilepsies. Epilepsy Research, 2018, 143, 79-81.	1.6	48
38	Automated EEG source imaging: A retrospective, blinded clinical validation study. Clinical Neurophysiology, 2018, 129, 2403-2410.	1.5	48
39	Remission of encephalopathy with status epilepticus (ESES) during sleep renormalizes regulation of slow wave sleep. Epilepsia, 2017, 58, 1892-1901.	5.1	47
40	Transient Global Amnesia as a Postictal State from Recurrent Partial Seizures. Epilepsia, 1991, 32, 882-885.	5.1	46
41	Novel congenital disorder of O-linked glycosylation caused by GALNT2 loss of function. Brain, 2020, 143, 1114-1126.	7.6	46
42	Overview of presurgical assessment and surgical treatment of epilepsy from the Italian League Against Epilepsy. Epilepsia, 2013, 54, 35-48.	5.1	45
43	A European survey on current practices in epilepsy monitoring units and implications for patients' safety. Epilepsy and Behavior, 2015, 44, 179-184.	1.7	45
44	Epilepsy surgery of â€œlow grade epilepsy associated neuroepithelial tumorsâ€• A retrospective nationwide Italian study. Epilepsia, 2017, 58, 1832-1841.	5.1	41
45	Photic Reflex Myoclonus: A Neurophysiological Study in Progressive Myoclonus Epilepsies. Epilepsia, 1999, 40, 50-58.	5.1	40
46	4-Aminopyridine is a promising treatment option for patients with gain-of-function KCNA2 -encephalopathy. Science Translational Medicine, 2021, 13, eaaz4957.	12.4	40
47	From next-generation sequencing to targeted treatment of non-acquired epilepsies. Expert Review of Molecular Diagnostics, 2019, 19, 217-228.	3.1	38
48	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. Genes, 2021, 12, 1051.	2.4	36
49	Identification of miRNAs Differentially Expressed in Human Epilepsy with or without Granule Cell Pathology. PLoS ONE, 2014, 9, e105521.	2.5	36
50	Neurophysiology of myoclonus and progressive myoclonus epilepsies. Epileptic Disorders, 2016, 18, 11-27.	1.3	35
51	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. Brain, 2021, 144, 3635-3650.	7.6	34
52	Mutant <scp>BRAF</scp> in lowâ€grade epilepsyâ€associated tumors and focal cortical dysplasia. Annals of Clinical and Translational Neurology, 2014, 1, 130-134.	3.7	33
53	Spinal muscular atrophy associated with progressive myoclonic epilepsy: A rare condition caused by mutations in <scp>ASAH</scp>1. Epilepsia, 2015, 56, 692-698.	5.1	33
54	GOSR2: a progressive myoclonus epilepsy gene. Epileptic Disorders, 2016, 18, 111-114.	1.3	32

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55	A pragmatic algorithm to select appropriate antiseizure medications in patients with epilepsy. Epilepsia, 2020, 61, 1668-1677.	5.1	32
56	Perampanel as add-on treatment in refractory focal epilepsy. The Dianalund experience. Acta Neurologica Scandinavica, 2016, 134, 374-377.	2.1	31
57	Epileptiform discharge propagation: Analyzing spikes from the onset to the peak. Clinical Neurophysiology, 2016, 127, 2127-2133.	1.5	31
58	Temporal lobe epilepsy and emotion recognition without amygdala: a case study of Urbachâ€Wiethe disease and review of the literature. Epileptic Disorders, 2014, 16, 518-527.	1.3	29
59	Prevalence of Nocturnal Frontal Lobe Epilepsy in the Adult Population of Bologna and Modena, Emilia-Romagna Region, Italy. Sleep, 2015, 38, 479-485.	1.1	27
60	Increasing volume and complexity of pediatric epilepsy surgery with stable seizure outcome between 2008 and 2014: A nationwide multicenter study. Epilepsy and Behavior, 2017, 75, 151-157.	1.7	27
61	SCARB2/LIMP2 deficiency in action myoclonus-renal failure syndrome. Epileptic Disorders, 2016, 18, 63-72.	1.3	26
62	Mild malformations of cortical development in sleepâ€related hypermotor epilepsy due to KCNT1 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 386-391.	3.7	25
63	How to diagnose and classify idiopathic (genetic) generalized epilepsies. Epileptic Disorders, 2020, 22, 399-420.	1.3	23
64	Trait impulsivity in Juvenile Myoclonic Epilepsy. Annals of Clinical and Translational Neurology, 2021, 8, 138-152.	3.7	21
65	Refining Genotypes and Phenotypes in KCNA2-Related Neurological Disorders. International Journal of Molecular Sciences, 2021, 22, 2824.	4.1	20
66	Patterns and prognostic markers for treatment response in generalized epilepsies. Neurology, 2020, 95, e2519-e2528.	1.1	19
67	Sex-specific disease modifiers in juvenile myoclonic epilepsy. Scientific Reports, 2022, 12, 2785.	3.3	19
68	Biallelic inherited SCN8A variants, a rare cause of SCN8A â€related developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 2277-2285.	5.1	18
69	Encephalopathy related to Status Epilepticus during slow Sleep: current concepts and future directions. Epileptic Disorders, 2019, 21, 82-87.	1.3	18
70	EEG features in Encephalopathy related to Status Epilepticus during slow Sleep. Epileptic Disorders, 2019, 21, 22-30.	1.3	17
71	BRAF V600E mutation in neocortical posterior temporal epileptogenic gangliogliomas. Journal of Clinical Neuroscience, 2015, 22, 1250-1253.	1.5	16
72	Idiopathic encephalopathy related to status epilepticus during slow sleep (ESES) as a â€œpureâ€•model of epileptic encephalopathy. An electroclinical, genetic, and follow-up study. Epilepsy and Behavior, 2019, 97, 244-252.	1.7	16

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73	Focal ESES as a selective focal brain dysfunction: a challenge for clinicians, an opportunity for cognitive neuroscientists. Epileptic Disorders, 2015, 17, 345-347.	1.3	15
74	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	1.6	15
75	Encephalopathy related to Status Epilepticus during slow Sleep: a link with sleep homeostasis?. Epileptic Disorders, 2019, 21, 62-70.	1.3	15
76	PURA- Related Developmental and Epileptic Encephalopathy. Neurology: Genetics, 2021, 7, e613.	1.9	15
77	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsiesâ€”a Study in a Tertiary Epilepsy Center. Neurotherapeutics, 2022, 19, 1353-1367.	4.4	14
78	Standard procedures for the diagnostic pathway of sleepâ€related epilepsies and comorbid sleep disorders: A European Academy of Neurology, European Sleep Research Society and International League against Epilepsyâ€Europe consensus review. Journal of Sleep Research, 2020, 29, e13184.	3.2	13
79	Optimal choice of antiseizure medication: Agreement among experts and validation of a webâ€based decision support application. Epilepsia, 2021, 62, 220-227.	5.1	13
80	Focal cortical dysplasias in temporal lobe epilepsy surgery: Challenge in defining unusual variants according to the last ILAE classification. Epilepsy and Behavior, 2015, 45, 212-216.	1.7	11
81	Is autopsy tissue a valid control for epilepsy surgery tissue in micro<scp>RNA</scp> studies?. Epilepsia Open, 2017, 2, 90-95.	2.4	11
82	Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES). Clinical Neurophysiology, 2020, 131, 1030-1039.	1.5	11
83	Automated ictal EEG source imaging: A retrospective, blinded clinical validation study. Clinical Neurophysiology, 2022, 141, 119-125.	1.5	10
84	Genetic paroxysmal neurological disorders featuring episodic ataxia and epilepsy. European Journal of Medical Genetics, 2022, 65, 104450.	1.3	10
85	Encephalopathy related to status epilepticus during sleep due to a de novo KCNA1 variant in the Kvâ€specific Proâ€Valâ€Pro motif: phenotypic description and remarkable electroclinical response to ACTH. Epileptic Disorders, 2020, 22, 802-806.	1.3	9
86	Encephalopathy with status epilepticus during sleep (ESES) induced by oxcarbazepine in idiopathic focal epilepsy in childhood. Functional Neurology, 2015, 30, 139-41.	1.3	8
87	Pathology-Based Approach to Seizure Outcome After Surgery for Pharmacoresistant Medial Temporal Lobe Epilepsy. World Neurosurgery, 2016, 90, 448-453.	1.3	8
88	The new <scp>ILAE</scp> seizure classification: 63 seizure types?. Epilepsia, 2017, 58, 1298-1300.	5.1	8
89	Risk factors of paradoxical reactions to anti-seizure medication in genetic generalized epilepsy. Epilepsy Research, 2021, 170, 106547.	1.6	8
90	Pseudoresistance in idiopathic/genetic generalized epilepsies â€“ Definitions, risk factors, and outcome. Epilepsy and Behavior, 2022, 130, 108633.	1.7	8

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91	A webâ€based algorithm to rapidly classify seizures for the purpose of drug selection. Epilepsia, 2021, 62, 2474-2484.	5.1	7
92	Linking epilepsy, sleep disruption and cognitive impairment in Encephalopathy related to Status Epilepticus during slow Sleep (ESES). Epileptic Disorders, 2019, 21, 1-2.	1.3	7
93	Webâ€based decision support system for patientâ€tailored selection of antiseizure medication in adolescents and adults: An external validation study. European Journal of Neurology, 2022, 29, 382-389.	3.3	7
94	Absence-to-bilateral-tonic-clonic seizure. Neurology, 2020, 95, e2009-e2015.	1.1	6
95	The clinical spectrum of familial and sporadic idiopathic generalized epilepsy. Epilepsy Research, 2020, 165, 106374.	1.6	6
96	Deep-Phenotyping the Less Severe Spectrum of PIGT Deficiency and Linking the Gene to Myoclonic Atonic Seizures. Frontiers in Genetics, 2021, 12, 663643.	2.3	6
97	The EpiPick algorithm to select appropriate antiseizure medications in patients with epilepsy: Validation studies and updates. Epilepsia, 2022, 63, 254-255.	5.1	6
98	Prevalence of Sleep-Related Hypermotor Epilepsyâ€”Formerly Named Nocturnal Frontal Lobe Epilepsyâ€”in the Adult Population of the Emilia-Romagna Region, Italy. Sleep, 2017, 40, .	1.1	5
99	Alternating hemiplegia of childhood and a pathogenic variant of ATP1A3: a case report and pathophysiological considerations. Epileptic Disorders, 2017, 19, 226-230.	1.3	5
100	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	1.9	5
101	PRICKLE2 revisitedâ€”further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244.	2.8	5
102	Magnetic evoked potential polyphasia in idiopathic/genetic generalized epilepsy: An endophenotype not associated with treatment response. Clinical Neurophysiology, 2021, 132, 1499-1504.	1.5	4
103	Management of Antiepileptic Treatment After Epilepsy Surgery - Practices and Problems. Current Pharmaceutical Design, 2018, 23, 5749-5759.	1.9	4
104	A commentary on Encephalopathy related to Status Epilepticus during slow Sleep: from concepts to terminology. Epileptic Disorders, 2019, 21, 13-14.	1.3	4
105	Non-age-Related Focal Epilepsies. , 2019, , 445-460.		3
106	Perampanel in refractory epilepsies: what realâ€life experience tells us. Developmental Medicine and Child Neurology, 2017, 59, 352-353.	2.1	2
107	A European questionnaire survey on epilepsy monitoring units' current practice for postoperative psychogenic nonepileptic seizures' detection. Epilepsy and Behavior, 2020, 112, 107355.	1.7	2
108	Genetic testing in adult epilepsy patients: A call to action for clinicians. Epilepsia, 2020, 61, 2055-2056.	5.1	2

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109	Reader response: Generalized polyspike train: An EEG biomarker of drug-resistant idiopathic generalized epilepsy. Neurology, 2019, 93, 562-563.	1.1	1
110	Encephalopathy related to Status Epilepticus during slow Sleep: an historical introduction. Epileptic Disorders, 2019, 21, 3-4.	1.3	1
111	Expanding the phenotype of PURAâ€related developmental epileptic encephalopathy. Epileptic Disorders, 2022, 24, 445-446.	1.3	1
112	Looking at the muscle to find out what is happening in the brain. Clinical Neurophysiology, 2016, 127, 2898-2899.	1.5	0
113	Polygraphic Investigations and Back-Averaging Techniques in theÂStudy of Epileptic Motor Phenomena. , 2019, , 281-296.		0
114	Use of fitness trackers to identify and document epileptic seizures. Epileptic Disorders, 2021, 23, 432-434.	1.3	0
115	Motor Manifestations in Epileptic Photosensitivity: Clinical Features and Pathophysiological Insights. , 2021, , 185-197.		0
116	Trisomy 20p/monosomy 18p associated with congenital bilateral perisylvian syndrome. Epileptic Disorders, 2022, 24, 577-582.	1.3	0