

GRIN2A mutations in acquired epileptic aphasia and related encephalopathies with speech and language dysfunction

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Glycine transporters as novel therapeutic targets in schizophrenia, alcohol dependence and pain. <i>Nature Reviews Drug Discovery</i> , 2013, 12, 866-885.	21.5	175
2	GRIN2A, a green semaphore on the lumping route to idiopathic focal epilepsy in childhood. <i>Revue Neurologique</i> , 2013, 169, 921-922.	0.6	1
3	GRIN2A mutations identified as key genetic drivers of epilepsyâ€“aphasia spectrum disorders. <i>Nature Reviews Neurology</i> , 2013, 9, 541-541.	4.9	8
4	Harnessing Gene Expression Networks to Prioritize Candidate Epileptic Encephalopathy Genes. <i>PLoS ONE</i> , 2014, 9, e102079.	1.1	25
5	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. <i>Human Molecular Genetics</i> , 2014, 23, 6069-6080.	1.4	61
7	Functional analysis of a de novo GRIN2A missense mutation associated with early-onset epileptic encephalopathy. <i>Nature Communications</i> , 2014, 5, 3251.	5.8	128
8	Generalized Epilepsies: Immunologic and Inflammatory Mechanisms. <i>Seminars in Pediatric Neurology</i> , 2014, 21, 214-220.	1.0	6
9	Somatic Mutation of GRIN2A in Malignant Melanoma Results in Loss of Tumor Suppressor Activity via Aberrant NMDAR Complex Formation. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2390-2398.	0.3	26
10	<i>DEPDC5</i> does it all: Shared genetics for diverse epilepsy syndromes. <i>Annals of Neurology</i> , 2014, 75, 631-633.	2.8	20
11	Wholeâ€“exome sequencing in an individual with severe global developmental delay and intractable epilepsy identifies a novel, de novo <i>GRIN2A</i> mutation. <i>Epilepsia</i> , 2014, 55, e75-9.	2.6	36
12	<i>GRIN2A</i> mutation and earlyâ€“onset epileptic encephalopathy: personalized therapy with memantine. <i>Annals of Clinical and Translational Neurology</i> , 2014, 1, 190-198.	1.7	248
13	Continuous spikes and waves during slow sleep in a child with karyotype 47, XYY. <i>Epileptic Disorders</i> , 2014, 16, 223-226.	0.7	2
14	Syndromes at risk of status epilepticus in children: genetic and pathophysiological issues. <i>Epileptic Disorders</i> , 2014, 16, S89-95.	0.7	3
15	A subset of genomic alterations detected in rolandic epilepsies contains candidate or known epilepsy genes including <i>GRIN2A</i> and <i>PRRT2</i>. <i>Epilepsia</i> , 2014, 55, 370-378.	2.6	69
16	Genetic Forms of Epilepsies and Other Paroxysmal Disorders. <i>Seminars in Neurology</i> , 2014, 34, 266-279.	0.5	19
17	Past and Present Definitions of Epileptogenesis and Its Biomarkers. <i>Neurotherapeutics</i> , 2014, 11, 231-241.	2.1	198
18	Genetic Epilepsy Syndromes Without Structural Brain Abnormalities: Clinical Features and Experimental Models. <i>Neurotherapeutics</i> , 2014, 11, 269-285.	2.1	51
19	Towards the identification of a genetic basis for <i>L</i>-andauâ€“<i>K</i>-leffner <i>s</i>-syndrome. <i>Epilepsia</i> , 2014, 55, 858-865.	2.6	44

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20	The hidden genetics of epilepsyâ€”a clinically important new paradigm. <i>Nature Reviews Neurology</i> , 2014, 10, 283-292.	4.9	232
21	<i>GRIN2B</i> mutations in west syndrome and intellectual disability with focal epilepsy. <i>Annals of Neurology</i> , 2014, 75, 147-154.	2.8	195
22	The genetics of common epilepsies: common or distinct?. <i>Lancet Neurology</i> , The, 2014, 13, 859-860.	4.9	5
23	Epileptic Encephalopathies: New Genes and New Pathways. <i>Neurotherapeutics</i> , 2014, 11, 796-806.	2.1	96
24	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. <i>Journal of Medical Genetics</i> , 2014, 51, 724-736.	1.5	229
25	Evaluation of levetiracetam and valproic acid as low-dose monotherapies for children with typical benign childhood epilepsy with centrotemporal spikes (BECTS). <i>Seizure: the Journal of the British Epilepsy Association</i> , 2014, 23, 756-761.	0.9	30
26	De Novo Mutations in Synaptic Transmission Genes Including DNMT1 Cause Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2014, 95, 360-370.	2.6	388
27	CNVs in Epilepsy. <i>Current Genetic Medicine Reports</i> , 2014, 2, 162-167.	1.9	28
28	New technologies in molecular genetics. <i>Progress in Brain Research</i> , 2014, 213, 253-278.	0.9	6
29	Epilepsy phenotypes and genotype determinants. <i>Neurology</i> , 2014, 83, 1038-1039.	1.5	0
33	Epilepsy: Old Syndromes, New Genes. <i>Current Neurology and Neuroscience Reports</i> , 2014, 14, 447.	2.0	17
34	Epileptic encephalopathy with continuous spikeâ€”waves during sleep: The need for transition from childhood to adulthood medical care appears to be related to etiology. <i>Epilepsia</i> , 2014, 55, 21-23.	2.6	8
35	Analysis of <i>ELP4</i>, <i>SRPX2</i>, and interacting genes in typical and atypical rolandic epilepsy. <i>Epilepsia</i> , 2014, 55, e89-93.	2.6	50
36	Temporoparietal Resection in a Patient With Landau-Kleffner Syndrome. <i>Seminars in Pediatric Neurology</i> , 2014, 21, 96-100.	1.0	7
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38	Mechanical coupling maintains the fidelity of NMDA receptorâ€”mediated currents. <i>Nature Neuroscience</i> , 2014, 17, 914-922.	7.1	96
39	Epileptic encephalopathy with continuous spikes and waves in the occipitoâ€”temporal region during slowâ€”wave sleep in two patients with acquired Kanji dysgraphia. <i>Epileptic Disorders</i> , 2014, 16, 540-545.	0.7	11
40	White matter development in children with benign childhood epilepsy with centro-temporal spikes. <i>Brain</i> , 2014, 137, 1095-1106.	3.7	81

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41	Comparison of two next-generation sequencing kits for diagnosis of epileptic disorders with a user-friendly tool for displaying gene coverage, DeCovA. Applied & Translational Genomics, 2015, 7, 19-25.	2.1	19
43	A novel approach identifies the first transcriptome networks in bats: a new genetic model for vocal communication. BMC Genomics, 2015, 16, 836.	1.2	18
44	Should epileptiform discharges be treated?. Epilepsia, 2015, 56, 1492-1504.	2.6	60
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52	Insights into the Genetic Foundations of Human Communication. Neuropsychology Review, 2015, 25, 3-26.	2.5	33
53	Defective Auditory Processing in a Child With Temporal Epileptic Focus. Journal of Child Neurology, 2015, 30, 513-516.	0.7	1
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56	The contribution of next generation sequencing to epilepsy genetics. Expert Review of Molecular Diagnostics, 2015, 15, 1531-1538.	1.5	68
57	Investigation of GRIN2A in common epilepsy phenotypes. Epilepsy Research, 2015, 115, 95-99.	0.8	44
58	Advancing epilepsy genetics in the genomic era. Genome Medicine, 2015, 7, 91.	3.6	173
59	Impaired sleep-related consolidation of declarative memories in idiopathic focal epilepsies of childhood. Epilepsy and Behavior, 2015, 43, 16-23.	0.9	36

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61	Clinical and molecular delineation of a 16p13.2p13.13 microduplication. <i>European Journal of Medical Genetics</i> , 2015, 58, 194-198.	0.7	6
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63	Monogenic and chromosomal causes of isolated speech and language impairment. <i>Journal of Medical Genetics</i> , 2015, 52, 719-729.	1.5	17
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68	Global Developmental Delay and Intellectual Disability. , 2015, , 151-161.		4
69	Autism spectrum disorder and epilepsy: Disorders with a shared biology. <i>Epilepsy and Behavior</i> , 2015, 47, 191-201.	0.9	129
70	The brain's code and its canonical computational motifs. From sensory cortex to the default mode network: A multi-scale model of brain function in health and disease. <i>Neuroscience and Biobehavioral Reviews</i> , 2015, 55, 211-222.	2.9	48
71	Genetic screening and diagnosis in epilepsy?. <i>Current Opinion in Neurology</i> , 2015, 28, 136-142.	1.8	10
72	The Genetics of Neuropsychiatric Diseases: Looking In and Beyond the Exome. <i>Annual Review of Neuroscience</i> , 2015, 38, 47-68.	5.0	27
73	Epileptic Encephalopathies in Childhood: The Role of Genetic Testing. <i>Seminars in Neurology</i> , 2015, 35, 310-322.	0.5	7
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75	Autism Spectrum Disorder and Epilepsy. <i>Journal of Child Neurology</i> , 2015, 30, 1963-1971.	0.7	118
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79	Whole-exome sequencing broadens the phenotypic spectrum of rare pediatric epilepsy: a retrospective study. <i>Clinical Genetics</i> , 2015, 88, 34-40.	1.0	79
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84	Progress in autoimmune epileptic encephalitis. <i>Current Opinion in Neurology</i> , 2016, 29, 151-157.	1.8	21
85	<i>N</i> -methyl-D-aspartate (NMDA) receptor antibodies encephalitis mimicking an autistic regression. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 1092-1094.	1.1	34
86	Genetics Studies in Idiopathic Focal Epilepsies in Childhood. <i>Journal of Pediatric Epilepsy</i> , 2016, 05, 139-141.	0.1	0
87	A microRNA-328 binding site in <i>PAX6</i> is associated with centrotemporal spikes of rolandic epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2016, 3, 512-522.	1.7	27
89	Sleep-Related Epilepsy. <i>Current Treatment Options in Neurology</i> , 2016, 18, 23.	0.7	27
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93	GRIN2D Recurrent De Novo Dominant Mutation Causes a Severe Epileptic Encephalopathy Treatable with NMDA Receptor Channel Blockers. <i>American Journal of Human Genetics</i> , 2016, 99, 802-816.	2.6	138
94	Current Treatment Options for Early-Onset Pediatric Epileptic Encephalopathies. <i>Current Treatment Options in Neurology</i> , 2016, 18, 44.	0.7	5
95	Idiopathic focal epilepsies: the "lost tribe". <i>Epileptic Disorders</i> , 2016, 18, 252-288.	0.7	65
96	Structural brain abnormalities in a single gene disorder associated with epilepsy, language impairment and intellectual disability. <i>NeuroImage: Clinical</i> , 2016, 12, 655-665.	1.4	22

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97	Structural biology of glutamate receptor ion channel complexes. <i>Current Opinion in Structural Biology</i> , 2016, 41, 119-127.	2.6	45
98	The Expanding Clinical Spectrum of Genetic Pediatric Epileptic Encephalopathies. <i>Seminars in Pediatric Neurology</i> , 2016, 23, 134-142.	1.0	33
99	Structural basis of kainate subtype glutamate receptor desensitization. <i>Nature</i> , 2016, 537, 567-571.	13.7	78
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103	Response to immunotherapy in a patient with Landau-Kleffner syndrome and <i>GRIN2A</i> mutation. <i>Epileptic Disorders</i> , 2016, 18, 97-100.	0.7	49
104	Molecular Architecture and Neurobiology of the Epilepsies. , 2016, , 601-617.		0
105	Mechanistic Insight into NMDA Receptor Dysregulation by Rare Variants in the GluN2A and GluN2B Agonist Binding Domains. <i>American Journal of Human Genetics</i> , 2016, 99, 1261-1280.	2.6	158
106	Atypical Evolutions of Idiopathic Focal Epilepsies in Childhood. <i>Journal of Pediatric Epilepsy</i> , 2016, 05, 122-132.	0.1	0
107	Pediatric Epileptic Encephalopathies: Pathophysiology and Animal Models. <i>Seminars in Pediatric Neurology</i> , 2016, 23, 98-107.	1.0	16
108	Atypical benign partial epilepsy of childhood with acquired neurocognitive, lexical semantic, and autistic spectrum disorder. <i>Epilepsy & Behavior Case Reports</i> , 2016, 6, 42-48.	1.5	10
109	Altered zinc sensitivity of NMDA receptors harboring clinically-relevant mutations. <i>Neuropharmacology</i> , 2016, 109, 196-204.	2.0	50
110	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. <i>European Journal of Human Genetics</i> , 2016, 24, 1761-1770.	1.4	36
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112	The genetic landscape of the epileptic encephalopathies of infancy and childhood. <i>Lancet Neurology</i> , The, 2016, 15, 304-316.	4.9	474
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114	An Integrated Approach for Screening and Identification of Positive Allosteric Modulators of N-Methyl-D-Aspartate Receptors. <i>Journal of Biomolecular Screening</i> , 2016, 21, 468-479.	2.6	14

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116	Epilepsy in patients with GRIN2A alterations: Genetics, neurodevelopment, epileptic phenotype and response to anticonvulsive drugs. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 530-541.	0.7	37
117	Novel homozygous missense variant of GRIN1 in two sibs with intellectual disability and autistic features without epilepsy. <i>European Journal of Human Genetics</i> , 2017, 25, 376-380.	1.4	30
118	Functional Evaluation of a De Novo <i>GRIN2A</i> Mutation Identified in a Patient with Profound Global Developmental Delay and Refractory Epilepsy. <i>Molecular Pharmacology</i> , 2017, 91, 317-330.	1.0	66
119	Frequency of <i>CNKSR2</i> mutation in the X-linked epilepsy-aphasia spectrum. <i>Epilepsia</i> , 2017, 58, e40-e43.	2.6	23
120	Epileptic Encephalopathies—Clinical Syndromes and Pathophysiological Concepts. <i>Current Neurology and Neuroscience Reports</i> , 2017, 17, 10.	2.0	23
121	GRIN1 mutation associated with intellectual disability alters NMDA receptor trafficking and function. <i>Journal of Human Genetics</i> , 2017, 62, 589-597.	1.1	81
122	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by whole-exome sequencing. <i>Clinical Genetics</i> , 2017, 92, 281-289.	1.0	92
123	Decreased functional connectivity within a language subnetwork in benign epilepsy with centrotemporal spikes. <i>Epilepsia Open</i> , 2017, 2, 214-225.	1.3	19
124	DEPDC5 as a potential therapeutic target for epilepsy. <i>Expert Opinion on Therapeutic Targets</i> , 2017, 21, 591-600.	1.5	29
125	Ion Channel Genes and Epilepsy: Functional Alteration, Pathogenic Potential, and Mechanism of Epilepsy. <i>Neuroscience Bulletin</i> , 2017, 33, 455-477.	1.5	94
126	Next-generation DNA sequencing identifies novel gene variants and pathways involved in specific language impairment. <i>Scientific Reports</i> , 2017, 7, 46105.	1.6	79
127	Single-Dose Memantine Improves Cortical Oscillatory Response Dynamics in Patients with Schizophrenia. <i>Neuropsychopharmacology</i> , 2017, 42, 2633-2639.	2.8	55
128	Epilepsy-associated GRIN2A mutations reduce NMDA receptor trafficking and agonist potency—molecular profiling and functional rescue. <i>Scientific Reports</i> , 2017, 7, 66.	1.6	74
129	Progress in unraveling the genetic etiology of rolandic epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 47, 99-104.	0.9	30
130	Advances in epilepsy gene discovery and implications for epilepsy diagnosis and treatment. <i>Current Opinion in Neurology</i> , 2017, 30, 193-199.	1.8	72
131	<i>GRIN2B</i> encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. <i>Journal of Medical Genetics</i> , 2017, 54, 460-470.	1.5	190
132	De novo GRIN1 mutations: An emerging cause of severe early infantile encephalopathy. <i>European Journal of Medical Genetics</i> , 2017, 60, 317-320.	0.7	32

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134	16p13 microduplication without CREBBP involvement: Moving toward a phenotype delineation. <i>European Journal of Medical Genetics</i> , 2017, 60, 159-162.	0.7	7
135	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	2.6	136
136	What is the Relationship Between Autism Spectrum Disorders and Epilepsy?. <i>Seminars in Pediatric Neurology</i> , 2017, 24, 292-300.	1.0	37
137	NMDA Receptors in the Central Nervous System. <i>Methods in Molecular Biology</i> , 2017, 1677, 1-80.	0.4	105
138	Heterogeneous contribution of microdeletions in the development of common generalised and focal epilepsies. <i>Journal of Medical Genetics</i> , 2017, 54, 598-606.	1.5	22
139	Catastrophic Epilepsies of Childhood. <i>Annual Review of Neuroscience</i> , 2017, 40, 149-166.	5.0	23
140	Clinical spectrum and genotype-phenotype associations of KCNA2-related encephalopathies. <i>Brain</i> , 2017, 140, 2337-2354.	3.7	117
141	Speech and Language: Translating the Genome. <i>Trends in Genetics</i> , 2017, 33, 642-656.	2.9	57
142	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. <i>American Journal of Human Genetics</i> , 2017, 101, 1013-1020.	2.6	53
143	Acquired epileptic opercular syndrome related to a heterozygous deleterious substitution in <i>GRIN2A</i> . <i>Epileptic Disorders</i> , 2017, 19, 345-350.	0.7	4
144	Autism spectrum disorder and epileptic encephalopathy: common causes, many questions. <i>Journal of Neurodevelopmental Disorders</i> , 2017, 9, 23.	1.5	44
145	Diagnostic Targeted Resequencing in 349 Patients with Drug-Resistant Pediatric Epilepsies Identifies Causative Mutations in 30 Different Genes. <i>Human Mutation</i> , 2017, 38, 216-225.	1.1	152
146	Precision medicine in genetic epilepsies: break of dawn?. <i>Expert Review of Neurotherapeutics</i> , 2017, 17, 381-392.	1.4	57
147	Synaptic Zn ²⁺ and febrile seizure susceptibility. <i>British Journal of Pharmacology</i> , 2017, 174, 119-125.	2.7	18
148	Diverse modes of NMDA receptor positive allosteric modulation: Mechanisms and consequences. <i>Neuropharmacology</i> , 2017, 112, 34-45.	2.0	76
149	Topiramate in childhood epileptic encephalopathy with continuous spike-waves during sleep: A retrospective study of 21 cases. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 305-311.	0.7	11
150	Phenotypic analysis of 303 multiplex families with common epilepsies. <i>Brain</i> , 2017, 140, 2144-2156.	3.7	23

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152	A de novo loss-of-function GRIN2A mutation associated with childhood focal epilepsy and acquired epileptic aphasia. <i>PLoS ONE</i> , 2017, 12, e0170818.	1.1	51
153	Molecular Mechanism of Disease-Associated Mutations in the Pre-M1 Helix of NMDA Receptors and Potential Rescue Pharmacology. <i>PLoS Genetics</i> , 2017, 13, e1006536.	1.5	117
154	Current Status of Treatments for Children with Electrical Status in Slow-Wave Sleep (ESES/CSWS). <i>Epilepsy Currents</i> , 2017, 17, 214-216.	0.4	17
155	Genetics and Genomics of Acute Neurologic Disorders. <i>AACN Advanced Critical Care</i> , 2018, 29, 57-75.	0.6	0
156	Novel West syndrome candidate genes in a Chinese cohort. <i>CNS Neuroscience and Therapeutics</i> , 2018, 24, 1196-1206.	1.9	60
157	Diagnostic outcomes for genetic testing of 70 genes in 8565 patients with epilepsy and neurodevelopmental disorders. <i>Epilepsia</i> , 2018, 59, 1062-1071.	2.6	218
158	Genetic polymorphisms of GRIN2A and GRIN2B modify the neurobehavioral effects of low-level lead exposure in children. <i>Environmental Research</i> , 2018, 165, 1-10.	3.7	22
159	Sushi repeat-containing protein X-linked 2: A novel phylogenetically conserved hypothalamo-pituitary protein. <i>Journal of Comparative Neurology</i> , 2018, 526, 1806-1819.	0.9	4
160	Characterization of two familial cases presenting with a syndromic specific learning disorder and carrying (17q;21q) unbalanced translocations. <i>Clinical Case Reports (discontinued)</i> , 2018, 6, 827-834.	0.2	4
161	A novel missense mutation in <i>GRIN2A</i> causes a nonepileptic neurodevelopmental disorder. <i>Movement Disorders</i> , 2018, 33, 992-999.	2.2	26
162	Mutations in <i>MICAL1</i> cause autosomal dominant lateral temporal epilepsy. <i>Annals of Neurology</i> , 2018, 83, 483-493.	2.8	25
163	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. <i>Brain</i> , 2018, 141, 698-712.	3.7	72
164	Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. <i>Pharmacological Reviews</i> , 2018, 70, 142-173.	7.1	215
165	Genetics of human epilepsies: Continuing progress. <i>Presse Medicale</i> , 2018, 47, 218-226.	0.8	9
166	De novo mutations and rare variants occurring in NMDA receptors. <i>Current Opinion in Physiology</i> , 2018, 2, 27-35.	0.9	97
167	The Bioactive Protein-Ligand Conformation of GluN2C-Selective Positive Allosteric Modulators Bound to the NMDA Receptor. <i>Molecular Pharmacology</i> , 2018, 93, 141-156.	1.0	18
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