Seo-Kyung Chung

List of Publications by Year
in descending order

50 papers	2,740 citations	172457 29 h-index	233421 45 g-index
51 all docs	51 docs citations	51 times ranked	4330 citing authors

#	Article	IF	Citations
1	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
2	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
3	Mutations in the gene encoding GlyT2 (SLC6A5) define a presynaptic component of human startle disease. Nature Genetics, 2006, 38, 801-806.	21.4	232
4	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
5	Overlapping cortical malformations and mutations in TUBB2B and TUBA1A. Brain, 2013, 136, 536-548.	7.6	133
6	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. Journal of Neuroscience, 2010, 30, 9612-9620.	3.6	112
7	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
8	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	7.6	90
9	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
10	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
11	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Human Molecular Genetics, 2015, 24, 5313-5325.	2.9	77
12	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
13	Near-miss SIDS due to Brugada syndrome. Archives of Disease in Childhood, 2005, 90, 528-529.	1.9	72
14	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. Brain, 2018, 141, 698-712.	7.6	72
15	Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. Brain, 2013, 136, 3085-3095.	7.6	66
16	Brugada Syndrome Masquerading as Febrile Seizures. Pediatrics, 2007, 119, e1206-e1211.	2.1	64
17	Posthumous diagnosis of long QT syndrome from neonatal screening cards. Heart Rhythm, 2010, 7, 481-486.	0.7	56
18	Novel missense mutations in the glycine receptor Î² subunit gene (GLRB) in startle disease. Neurobiology of Disease, 2013, 52, 137-149.	4.4	54

#	Article	IF	Citations
19	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661.	7.6	52
20	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 927-940.	2.9	50
21	The glycinergic system in human startle disease: a genetic screening approach. Frontiers in Molecular Neuroscience, 2010, 3, 8.	2.9	47
22	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
23	Biophysical Properties of 9 <i>KCNQ1</i> Mutations Associated With Long-QT Syndrome. Circulation: Arrhythmia and Electrophysiology, 2009, 2, 417-426.	4.8	43
24	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
25	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
26	Long QT and Brugada syndrome gene mutations in New Zealand. Heart Rhythm, 2007, 4, 1306-1314.	0.7	41
27	A Novel GABRG2 mutation, p.R136*, in a family with GEFS+ and extended phenotypes. Neurobiology of Disease, 2014, 64, 131-141.	4.4	39
28	A critical role for glycine transporters in hyperexcitability disorders. Frontiers in Molecular Neuroscience, 2008, 1, 1.	2.9	37
29	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. Journal of Biological Chemistry, 2013, 288, 33745-33759.	3.4	35
30	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
31	Symptoms and Signs Associated with Syncope in Young People with Primary Cardiac Arrhythmias. Heart Lung and Circulation, 2011, 20, 593-598.	0.4	27
32	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
33	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
34	Next Generation Sequencing Methodologies - An Overview. Advances in Protein Chemistry and Structural Biology, 2012, 89, 1-26.	2.3	21
35	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
36	Coinheritance of long QT syndrome and Kearns-Sayre syndrome. Heart Rhythm, 2007, 4, 1568-1572.	0.7	15

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37	Hyperekplexia: Report on phenotype and genotype of 16 Jordanian patients. Brain and Development, 2017, 39, 306-311.	1.1	11
38	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. Brain, 2018, 141, e55-e55.	7.6	11
39	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
40	Ethnicity can predict GLRA1 genotypes in hyperekplexia. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 341-343.	1.9	9
41	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 26, 241-246.	1.7	6
42	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2019, 380, e24.	27.0	4
43	Evaluation for Retinal Therapy for RPE65 Variation Assessed in hiPSC Retinal Pigment Epithelial Cells. Stem Cells International, 2021, 2021, 1-12.	2.5	4
44	Neonatal hyperekplexia with homozygous p.R392H mutation in GLRA1. Epileptic Disorders, 2014, 16, 354-357.	1.3	2
45	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50.	7.6	1
46	PATH42 Lineage, clinical, genetic, structural and cellular characterisation of a novel epilepsy mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, e19-e19.	1.9	0
47	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 2552-2552.	2.9	0
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	A NOVEL LGI1 VARIANT IN LATERAL TEMPORAL LOBE EPILEPSY. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, e4.154-e4.	1.9	0