Seo-Kyung Chung

List of Publications by Year
in descending order

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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	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
2	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
3	Evaluation for Retinal Therapy for RPE65 Variation Assessed in hiPSC Retinal Pigment Epithelial Cells. Stem Cells International, 2021, 2021, 1-12.	2.5	4
4	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
5	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
6	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	7.6	90
7	Intestinal-Cell Kinase and Juvenile Myoclonic Epilepsy. New England Journal of Medicine, 2019, 380, e24.	27.0	4
8	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661.	7.6	52
9	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. Brain, 2018, 141, 698-712.	7.6	72
10	Expanding the phenotype of TRAK1 mutations: hyperekplexia and refractory status epilepticus. Brain, 2018, 141, e55-e55.	7.6	11
11	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50.	7.6	1
12	Ultra-rare genetic variation in common epilepsies: a case-control sequencing study. Lancet Neurology, The, 2017, 16, 135-143.	10.2	190
13	Hyperekplexia: Report on phenotype and genotype of 16 Jordanian patients. Brain and Development, 2017, 39, 306-311.	1.1	11
14	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
15	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
16	Genome-wide Polygenic Burden of Rare Deleterious Variants in Sudden Unexpected Death in Epilepsy. EBioMedicine, 2015, 2, 1063-1070.	6.1	74
17	Ethnicity can predict GLRA1 genotypes in hyperekplexia. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 341-343.	1.9	9
18	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Human Molecular Genetics, 2015, 24, 5313-5325.	2.9	77

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19	A NOVEL LGI1 VARIANT IN LATERAL TEMPORAL LOBE EPILEPSY. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, e4.154-e4.	1.9	0
20	Neonatal hyperekplexia with homozygous p.R392H mutation in GLRA1. Epileptic Disorders, 2014, 16, 354-357.	1.3	2
21	TUBULINOPATHIES IN MALFORMATIONS OF THE CEREBRAL CORTEX. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.139-e4.	1.9	0
22	MECHANISMS OF DISEASE IN THE HYPEREKPLEXIAS. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.117-e4.	1.9	0
23	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
24	A Novel GABRG2 mutation, p.R136*, in a family with GEFS+ and extended phenotypes. Neurobiology of Disease, 2014, 64, 131-141.	4.4	39
25	Novel missense mutations in the glycine receptor Î² subunit gene (GLRB) in startle disease. Neurobiology of Disease, 2013, 52, 137-149.	4.4	54
26	Translation of genetic findings to clinical practice in juvenile myoclonic epilepsy. Epilepsy and Behavior, 2013, 26, 241-246.	1.7	6
27	Overlapping cortical malformations and mutations in TUBB2B and TUBA1A. Brain, 2013, 136, 536-548.	7.6	133
28	Genotype-phenotype correlations in hyperekplexia: apnoeas, learning difficulties and speech delay. Brain, 2013, 136, 3085-3095.	7.6	66
29	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 927-940.	2.9	50
30	New Hyperekplexia Mutations Provide Insight into Glycine Receptor Assembly, Trafficking, and Activation Mechanisms. Journal of Biological Chemistry, 2013, 288, 33745-33759.	3.4	35
31	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 2552-2552.	2.9	0
32	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
33	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
34	Next Generation Sequencing Methodologies - An Overview. Advances in Protein Chemistry and Structural Biology, 2012, 89, 1-26.	2.3	21
35	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
36	Symptoms and Signs Associated with Syncope in Young People with Primary Cardiac Arrhythmias. Heart Lung and Circulation, 2011, 20, 593-598.	0.4	27

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37	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
38	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. Journal of Neuroscience, 2010, 30, 9612-9620.	3.6	112
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	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
41	The glycinergic system in human startle disease: a genetic screening approach. Frontiers in Molecular Neuroscience, 2010, 3, 8.	2.9	47
42	Posthumous diagnosis of long QT syndrome from neonatal screening cards. Heart Rhythm, 2010, 7, 481-486.	0.7	56
43	Biophysical Properties of 9 KCNQ1 Mutations Associated With Long-QT Syndrome. Circulation: Arrhythmia and Electrophysiology, 2009, 2, 417-426.	4.8	43
44	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
45	A critical role for glycine transporters in hyperexcitability disorders. Frontiers in Molecular Neuroscience, 2008, 1, 1.	2.9	37
46	Brugada Syndrome Masquerading as Febrile Seizures. Pediatrics, 2007, 119, e1206-e1211.	2.1	64
47	Coinheritance of long QT syndrome and Kearns-Sayre syndrome. Heart Rhythm, 2007, 4, 1568-1572.	0.7	15
48	Long QT and Brugada syndrome gene mutations in New Zealand. Heart Rhythm, 2007, 4, 1306-1314.	0.7	41
49	Mutations in the gene encoding GlyT2 (SLC6A5) define a presynaptic component of human startle disease. Nature Genetics, 2006, 38, 801-806.	21.4	232
50	Near-miss SIDS due to Brugada syndrome. Archives of Disease in Childhood, 2005, 90, 528-529.	1.9	72