

Katherine L Helbig

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

82 papers	3,483 citations	31 h-index	58 g-index
91 ext. papers	4,848 ext. citations	8.1 avg, IF	4.25 L-index

#	Paper	IF	Citations
82	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. <i>Nature Genetics</i> , 2009 , 41, 160-236	36.3	454
81	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. <i>Brain</i> , 2017 , 140, 1316-1336	11.2	285
80	Diagnostic exome sequencing provides a molecular diagnosis for a significant proportion of patients with epilepsy. <i>Genetics in Medicine</i> , 2016 , 18, 898-905	8.1	223
79	Distinguishing sleep disorders from seizures: diagnosing bumps in the night. <i>Archives of Neurology</i> , 2006 , 63, 705-9		178
78	STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy. <i>Neurology</i> , 2016 , 86, 954-625	3.5	159
77	De novo variants in neurodevelopmental disorders with epilepsy. <i>Nature Genetics</i> , 2018 , 50, 1048-1053	36.3	139
76	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. <i>Journal of Medical Genetics</i> , 2017 , 54, 460-470	5.8	109
75	Delineating the GRIN1 phenotypic spectrum: A distinct genetic NMDA receptor encephalopathy. <i>Neurology</i> , 2016 , 86, 2171-8	6.5	108
74	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019 , 105, 267-282	11	104
73	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017 , 101, 768-788	11	81
72	Phenotypic spectrum of GABRA1: From generalized epilepsies to severe epileptic encephalopathies. <i>Neurology</i> , 2016 , 87, 1140-51	6.5	78
71	Genetic epilepsy with febrile seizures plus: Refining the spectrum. <i>Neurology</i> , 2017 , 89, 1210-1219	6.5	68
70	GRIN2A-related disorders: genotype and functional consequence predict phenotype. <i>Brain</i> , 2019 , 142, 80-92	11.2	66
69	Classification of Genes: Standardized Clinical Validity Assessment of Gene-Disease Associations Aids Diagnostic Exome Analysis and Reclassifications. <i>Human Mutation</i> , 2017 , 38, 600-608	4.7	55
68	Neuronal mechanisms of mutations in SCN8A causing epilepsy or intellectual disability. <i>Brain</i> , 2019 , 142, 376-390	11.2	53
67	Mutations in SCN3A cause early infantile epileptic encephalopathy. <i>Annals of Neurology</i> , 2018 , 83, 703-714	17.4	51
66	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. <i>Brain</i> , 2016 , 139, 2420-30	11.2	49

65	HCN1 mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. <i>Brain</i> , 2018 , 141, 3160-3178	11.2	48
64	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. <i>Brain</i> , 2018 , 141, 698-712	11.2	46
63	encephalopathy: A new disease of vesicle fission. <i>Neurology</i> , 2017 , 89, 385-394	6.5	46
62	Mutations in PMPCB Encoding the Catalytic Subunit of the Mitochondrial Presequence Protease Cause Neurodegeneration in Early Childhood. <i>American Journal of Human Genetics</i> , 2018 , 102, 557-573	11	44
61	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. <i>Brain</i> , 2017 , 140, 2322-2336	11.2	44
60	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018 , 103, 666-678	11	44
59	Exploration of the genetic architecture of idiopathic generalized epilepsies. <i>Epilepsia</i> , 2006 , 47, 1682-90	6.4	42
58	FOXG1 syndrome: genotype-phenotype association in 83 patients with FOXG1 variants. <i>Genetics in Medicine</i> , 2018 , 20, 98-108	8.1	41
57	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2019 , 104, 1060-1072	11	39
56	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017 , 101, 716-724	11	38
55	Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. <i>Genetics in Medicine</i> , 2017 , 19, 224-235	8.1	38
54	De novo mutations of KIAA2022 in females cause intellectual disability and intractable epilepsy. <i>Journal of Medical Genetics</i> , 2016 , 53, 850-858	5.8	32
53	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. <i>Genetics in Medicine</i> , 2019 , 21, 837-849	8.1	32
52	The role of genetic testing in epilepsy diagnosis and management. <i>Expert Review of Molecular Diagnostics</i> , 2017 , 17, 739-750	3.8	31
51	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. <i>American Journal of Human Genetics</i> , 2019 , 104, 1210-1222	11	31
50	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. <i>Annals of Neurology</i> , 2016 , 80,	9.4	31
49	WAC loss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. <i>Journal of Medical Genetics</i> , 2015 , 52, 754-61	5.8	30
48	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , 2017 , 101, 516-524	11	29

47	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. <i>Nature Communications</i> , 2019 , 10, 708	17.4	27
46	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	11	26
45	Genetic risk perception and reproductive decision making among people with epilepsy. <i>Epilepsia</i> , 2010 , 51, 1874-7	6.4	26
44	Heterogeneous clinical and functional features of GRIN2D-related developmental and epileptic encephalopathy. <i>Brain</i> , 2019 , 142, 3009-3027	11.2	25
43	Clinical spectrum of -related epileptic disorders. <i>Neurology</i> , 2019 , 92, e1238-e1249	6.5	25
42	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018 , 102, 1195-1203	11	24
41	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. <i>European Journal of Human Genetics</i> , 2016 , 24, 1761-1770	5.3	23
40	Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. <i>Human Molecular Genetics</i> , 2017 , 26, 4937-4950	5.6	22
39	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. <i>Neurology</i> , 2016 , 86, 713-22	6.5	22
38	De novo ITPR1 variants are a recurrent cause of early-onset ataxia, acting via loss of channel function. <i>European Journal of Human Genetics</i> , 2018 , 26, 1623-1634	5.3	22
37	The ClinGen Epilepsy Gene Curation Expert Panel-Bridging the divide between clinical domain knowledge and formal gene curation criteria. <i>Human Mutation</i> , 2018 , 39, 1476-1484	4.7	21
36	SCN3A-Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. <i>Annals of Neurology</i> , 2020 , 88, 348-362	9.4	19
35	Autosomal recessive mutations in THOC6 cause intellectual disability: syndrome delineation requiring forward and reverse phenotyping. <i>Clinical Genetics</i> , 2017 , 91, 92-99	4	18
34	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. <i>Genetics in Medicine</i> , 2019 , 21, 601-607	8.1	18
33	A case-only study of gene-environment interaction between genetic susceptibility variants in NOD2 and cigarette smoking in Crohn's disease aetiology. <i>BMC Medical Genetics</i> , 2012 , 13, 14	2.1	17
32	Founder effect with variable age at onset in Arab families with Lafora disease and EPM2A mutation. <i>Epilepsia</i> , 2007 , 48, 1011-4	6.4	15
31	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. <i>Brain</i> , 2020 , 143, 2106-2118	11.2	14
30	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. <i>European Journal of Human Genetics</i> , 2018 , 26, 695-708	5.3	14

29	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. <i>Genome Medicine</i> , 2020 , 12, 28	14.4	13
28	Biallelic loss-of-function variants in DOCK3 cause muscle hypotonia, ataxia, and intellectual disability. <i>Clinical Genetics</i> , 2017 , 92, 430-433	4	12
27	Three cases of Troyer syndrome in two families of Filipino descent. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 1780-5	2.5	12
26	Current knowledge of SLC6A1-related neurodevelopmental disorders. <i>Brain Communications</i> , 2020 , 2, fcaa170	4.5	11
25	Damaging de novo missense variants in EEF1A2 lead to a developmental and degenerative epileptic-dyskinetic encephalopathy. <i>Human Mutation</i> , 2020 , 41, 1263-1279	4.7	10
24	Rare protein sequence variation in SV2A gene does not affect response to levetiracetam. <i>Epilepsy Research</i> , 2012 , 101, 277-9	3	10
23	RARS1-related hypomyelinating leukodystrophy: Expanding the spectrum. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 83-93	5.3	10
22	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2020 , 107, 683-697	11	8
21	Computational analysis of 10,860 phenotypic annotations in individuals with SCN2A-related disorders. <i>Genetics in Medicine</i> , 2021 , 23, 1263-1272	8.1	8
20	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. <i>Brain</i> , 2021 ,	11.2	8
19	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. <i>Brain</i> , 2020 , 143, 3564-3573	11.2	7
18	A longitudinal footprint of genetic epilepsies using automated electronic medical record interpretation. <i>Genetics in Medicine</i> , 2020 , 22, 2060-2070	8.1	7
17	Whole-exome and HLA sequencing in Febrile infection-related epilepsy syndrome. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 1429-1435	5.3	6
16	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021 , 108, 965-982	11	6
15	High-throughput evaluation of epilepsy-associated KCNQ2 variants reveals functional and pharmacological heterogeneity.. <i>JCI Insight</i> , 2022 ,	9.9	4
14	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. <i>Brain</i> , 2021 , 144, 1435-1450	11.2	4
13	Assessing the landscape of STXBP1-related disorders in 534 individuals.. <i>Brain</i> , 2021 ,	11.2	4
12	De novo Variants in Neurodevelopmental Disorders with Epilepsy		3

11	Defining and expanding the phenotype of -associated developmental epileptic encephalopathy. <i>Neurology: Genetics</i> , 2019 , 5, e373	3.8	3
10	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. <i>Neurology</i> , 2021 , 96, e2251-e2260	6.5	3
9	Genetics of Epilepsy in the Era of Precision Medicine: Implications for Testing, Treatment, and Genetic Counseling. <i>Current Genetic Medicine Reports</i> , 2018 , 6, 73-82	2.2	2
8	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications		2
7	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021 , 108, 1138-1150	11	2
6	Diagnostic exome sequencing identifies haploinsufficiency and chromosome 20 uniparental disomy in a patient with developmental anomalies. <i>Clinical Case Reports (discontinued)</i> , 2018 , 6, 1208-1213	0.7	1
5	Related Developmental and Epileptic Encephalopathy: Phenotypic and Genotypic Spectrum. <i>Neurology: Genetics</i> , 2021 , 7, e613	3.8	1
4	Phenotypic homogeneity in childhood epilepsies evolves in gene-specific patterns across 3251 patient-years of clinical data. <i>European Journal of Human Genetics</i> , 2021 , 29, 1690-1700	5.3	1
3	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. <i>Epilepsia</i> , 2021 , 62, e13-e21	6.4	1
2	Novel Missense Mutations Associated with Infantile-Onset Developmental and Epileptic Encephalopathy. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	0
1	Reply. <i>Annals of Neurology</i> , 2017 , 81, 328-329	9.4	