Katherine L Helbig

List of Publications by Year in Descending Order

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	High-throughput evaluation of epilepsy-associated KCNQ2 variants reveals functional and pharmacological heterogeneity.. *JCI Insight*, **2022**,	9.9	4
81	Related Developmental and Epileptic Encephalopathy: Phenotypic and Genotypic Spectrum. *Neurology: Genetics*, **2021**, 7, e613	3.8	1
80	Computational analysis of 10,860 phenotypic annotations in individuals with SCN2A-related disorders. *Genetics in Medicine*, **2021**, 23, 1263-1272	8.1	8
79	Phenotypic homogeneity in childhood epilepsies evolves in gene-specific patterns across 3251 patient-years of clinical data. *European Journal of Human Genetics*, **2021**, 29, 1690-1700	5.3	1
78	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4
77	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	11	6
76	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. *American Journal of Human Genetics*, **2021**, 108, 1138-1150	11	2
75	FBXO28 causes developmental and epileptic encephalopathy with profound intellectual disability. *Epilepsia*, **2021**, 62, e13-e21	6.4	1
74	Assessing the landscape of STXBP1-related disorders in 534 individuals.. *Brain*, **2021**,	11.2	4
73	Association of Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. *Neurology*, **2021**, 96, e2251-e2260	6.5	3
72	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. *Brain*, **2021**,	11.2	8
71	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. *Brain*, **2020**, 143, 2106-2118	11.2	14
70	SCN3A-Related Neurodevelopmental Disorder: A Spectrum of Epilepsy and Brain Malformation. *Annals of Neurology*, **2020**, 88, 348-362	9.4	19
69	Gene family information facilitates variant interpretation and identification of disease-associated genes in neurodevelopmental disorders. *Genome Medicine*, **2020**, 12, 28	14.4	13
68	Damaging de novo missense variants in EEF1A2 lead to a developmental and degenerative epileptic-dyskinetic encephalopathy. *Human Mutation*, **2020**, 41, 1263-1279	4.7	10
67	RARS1-related hypomyelinating leukodystrophy: Expanding the spectrum. *Annals of Clinical and Translational Neurology*, **2020**, 7, 83-93	5.3	10
66	Novel Missense Mutations Associated with Infantile-Onset Developmental and Epileptic Encephalopathy. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	0

65	Whole-exome and HLA sequencing in Febrile infection-related epilepsy syndrome. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1429-1435	5.3	6
64	Current knowledge of SLC6A1-related neurodevelopmental disorders. *Brain Communications*, **2020**, 2, fcaa170	4.5	11
63	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. *Brain*, **2020**, 143, 3564-3573	11.2	7
62	Semantic Similarity Analysis Reveals Robust Gene-Disease Relationships in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2020**, 107, 683-697	11	8
61	A longitudinal footprint of genetic epilepsies using automated electronic medical record interpretation. *Genetics in Medicine*, **2020**, 22, 2060-2070	8.1	7
60	Heterogeneous clinical and functional features of GRIN2D-related developmental and epileptic encephalopathy. *Brain*, **2019**, 142, 3009-3027	11.2	25
59	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. *American Journal of Human Genetics*, **2019**, 104, 1060-1072	11	39
58	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
57	Clinical spectrum of -related epileptic disorders. *Neurology*, **2019**, 92, e1238-e1249	6.5	25
56	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	11	104
55	Biallelic VARS variants cause developmental encephalopathy with microcephaly that is recapitulated in vars knockout zebrafish. *Nature Communications*, **2019**, 10, 708	17.4	27
54	Defining and expanding the phenotype of -associated developmental epileptic encephalopathy. *Neurology: Genetics*, **2019**, 5, e373	3.8	3
53	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. *Genetics in Medicine*, **2019**, 21, 837-849	8.1	32
52	GRIN2A-related disorders: genotype and functional consequence predict phenotype. *Brain*, **2019**, 142, 80-92	11.2	66
51	Neuronal mechanisms of mutations in SCN8A causing epilepsy or intellectual disability. *Brain*, **2019**, 142, 376-390	11.2	53
50	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. *Genetics in Medicine*, **2019**, 21, 601-607	8.1	18
49	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. *European Journal of Human Genetics*, **2018**, 26, 695-708	5.3	14
48	Mutations in SCN3A cause early infantile epileptic encephalopathy. *Annals of Neurology*, **2018**, 83, 703-717	9.4	51

47	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. *Brain*, **2018**, 141, 698-712	11.2	46
46	Genetics of Epilepsy in the Era of Precision Medicine: Implications for Testing, Treatment, and Genetic Counseling. *Current Genetic Medicine Reports*, **2018**, 6, 73-82	2.2	2
45	Mutations in PMPCB Encoding the Catalytic Subunit of the Mitochondrial Presequence Protease Cause Neurodegeneration in Early Childhood. *American Journal of Human Genetics*, **2018**, 102, 557-573	11	44
44	FOXG1 syndrome: genotype-phenotype association in 83 patients with FOXG1 variants. *Genetics in Medicine*, **2018**, 20, 98-108	8.1	41
43	De novo ITPR1 variants are a recurrent cause of early-onset ataxia, acting via loss of channel function. *European Journal of Human Genetics*, **2018**, 26, 1623-1634	5.3	22
42	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2018**, 102, 1195-1203	11	24
41	Diagnostic exome sequencing identifies haploinsufficiency and chromosome 20 uniparental disomy in a patient with developmental anomalies. *Clinical Case Reports (discontinued)*, **2018**, 6, 1208-1213	0.7	1
40	The ClinGen Epilepsy Gene Curation Expert Panel-Bridging the divide between clinical domain knowledge and formal gene curation criteria. *Human Mutation*, **2018**, 39, 1476-1484	4.7	21
39	HCN1 mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. *Brain*, **2018**, 141, 3160-3178	11.2	48
38	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. *American Journal of Human Genetics*, **2018**, 103, 666-678	11	44
37	De novo variants in neurodevelopmental disorders with epilepsy. *Nature Genetics*, **2018**, 50, 1048-1053	36.3	139
36	Autosomal recessive mutations in THOC6 cause intellectual disability: syndrome delineation requiring forward and reverse phenotyping. *Clinical Genetics*, **2017**, 91, 92-99	4	18
35	Classification of Genes: Standardized Clinical Validity Assessment of Gene-Disease Associations Aids Diagnostic Exome Analysis and Reclassifications. *Human Mutation*, **2017**, 38, 600-608	4.7	55
34	Biallelic loss-of-function variants in DOCK3 cause muscle hypotonia, ataxia, and intellectual disability. *Clinical Genetics*, **2017**, 92, 430-433	4	12
33	The role of genetic testing in epilepsy diagnosis and management. *Expert Review of Molecular Diagnostics*, **2017**, 17, 739-750	3.8	31
32	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. *Brain*, **2017**, 140, 1316-1336	11.2	285
31	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
30	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2017**, 101, 716-724	11	38

29	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. *American Journal of Human Genetics*, **2017**, 101, 768-788	11	81
28	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. *American Journal of Human Genetics*, **2017**, 101, 516-524	11	29
27	Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. *Human Molecular Genetics*, **2017**, 26, 4937-4950	5.6	22
26	Genetic epilepsy with febrile seizures plus: Refining the spectrum. *Neurology*, **2017**, 89, 1210-1219	6.5	68
25	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. *Brain*, **2017**, 140, 2322-2336	11.2	44
24	Reply. *Annals of Neurology*, **2017**, 81, 328-329	9.4	
23	De Novo Variants in GRIA4 Lead to Intellectual Disability with or without Seizures and Gait Abnormalities. *American Journal of Human Genetics*, **2017**, 101, 1013-1020	11	26
22	encephalopathy: A new disease of vesicle fission. *Neurology*, **2017**, 89, 385-394	6.5	46
21	Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. *Genetics in Medicine*, **2017**, 19, 224-235	8.1	38
20	Phenotypic spectrum of GABRA1: From generalized epilepsies to severe epileptic encephalopathies. *Neurology*, **2016**, 87, 1140-51	6.5	78
19	A recurrent mutation in KCNA2 as a novel cause of hereditary spastic paraplegia and ataxia. *Annals of Neurology*, **2016**, 80,	9.4	31
18	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. *Brain*, **2016**, 139, 2420-30	11.2	49
17	De novo mutations of KIAA2022 in females cause intellectual disability and intractable epilepsy. *Journal of Medical Genetics*, **2016**, 53, 850-858	5.8	32
16	Loss of function of the retinoid-related nuclear receptor (RORB) gene and epilepsy. *European Journal of Human Genetics*, **2016**, 24, 1761-1770	5.3	23
15	Diagnostic exome sequencing provides a molecular diagnosis for a significant proportion of patients with epilepsy. *Genetics in Medicine*, **2016**, 18, 898-905	8.1	223
14	Multiplex families with epilepsy: Success of clinical and molecular genetic characterization. *Neurology*, **2016**, 86, 713-22	6.5	22
13	STXBP1 encephalopathy: A neurodevelopmental disorder including epilepsy. *Neurology*, **2016**, 86, 954-62	6.5	159
12	Three cases of Troyer syndrome in two families of Filipino descent. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1780-5	2.5	12

11	Delineating the GRIN1 phenotypic spectrum: A distinct genetic NMDA receptor encephalopathy. *Neurology*, **2016**, 86, 2171-8	6.5	108
10	WAC loss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. *Journal of Medical Genetics*, **2015**, 52, 754-61	5.8	30
9	A case-only study of gene-environment interaction between genetic susceptibility variants in NOD2 and cigarette smoking in CrohnŦs disease aetiology. *BMC Medical Genetics*, **2012**, 13, 14	2.1	17
8	Rare protein sequence variation in SV2A gene does not affect response to levetiracetam. *Epilepsy Research*, **2012**, 101, 277-9	3	10
7	Genetic risk perception and reproductive decision making among people with epilepsy. *Epilepsia*, **2010**, 51, 1874-7	6.4	26
6	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. *Nature Genetics*, **2009**, 41, 160-2	36.3	454
5	Founder effect with variable age at onset in Arab families with Lafora disease and EPM2A mutation. *Epilepsia*, **2007**, 48, 1011-4	6.4	15
4	Distinguishing sleep disorders from seizures: diagnosing bumps in the night. *Archives of Neurology*, **2006**, 63, 705-9		178
3	Exploration of the genetic architecture of idiopathic generalized epilepsies. *Epilepsia*, **2006**, 47, 1682-90	6.4	42
2	De novo Variants in Neurodevelopmental Disorders with Epilepsy		3
1	Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications		2