Elizabeth E Palmer

List of Publications by Citations

52 papers	749 citations	18 h-index	26 g-index
59 ext. papers	1,234 ext. citations	6.6 avg, IF	3.6 L-index

#	Paper	IF	Citations
52	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. *Cell*, **2018**, 172, 924-936.e11	56.2	65
51	Agenesis of the corpus callosum: a clinical approach to diagnosis. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 184-97	3.1	58
50	Integrating exome sequencing into a diagnostic pathway for epileptic encephalopathy: Evidence of clinical utility and cost effectiveness. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 186-199	2.3	46
49	encephalopathy: A new disease of vesicle fission. *Neurology*, **2017**, 89, 385-394	6.5	46
48	The molecular and phenotypic spectrum of IQSEC2-related epilepsy. *Epilepsia*, **2016**, 57, 1858-1869	6.4	38
47	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. *American Journal of Human Genetics*, **2015**, 97, 302-10	11	34
46	New insights into Brunner syndrome and potential for targeted therapy. *Clinical Genetics*, **2016**, 89, 120-7	4	34
45	Changing interpretation of chromosomal microarray over time in a community cohort with intellectual disability. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 377-85	2.5	34
44	Asparagine Synthetase Deficiency causes reduced proliferation of cells under conditions of limited asparagine. *Molecular Genetics and Metabolism*, **2015**, 116, 178-86	3.7	33
43	Eight further individuals with intellectual disability and epilepsy carrying bi-allelic CNTNAP2 aberrations allow delineation of the mutational and phenotypic spectrum. *Journal of Medical Genetics*, **2016**, 53, 820-827	5.8	28
42	Spinocerebellar ataxia type 29 due to mutations in ITPR1: a case series and review of this emerging congenital ataxia. *Orphanet Journal of Rare Diseases*, **2017**, 12, 121	4.2	27
41	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. *American Journal of Human Genetics*, **2018**, 102, 985-994	11	26
40	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
39	A De Novo Mutation in the Sodium-Activated Potassium Channel KCNT2 Alters Ion Selectivity and Causes Epileptic Encephalopathy. *Cell Reports*, **2017**, 21, 926-933	10.6	20
38	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. *Molecular Psychiatry*, **2018**, 23, 222-230	15.1	20
37	Neuronal deficiency of ARV1 causes an autosomal recessive epileptic encephalopathy. *Human Molecular Genetics*, **2016**, 25, 3042-3054	5.6	20
36	encephalopathy: Connecting neurodevelopmental disorders with □synucleinopathies?. *Neurology*, **2019**, 93, 114-123	6.5	18

35	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. *American Journal of Human Genetics*, **2017**, 101, 995-1005	11	18
34	Diagnostic Yield of Whole Genome Sequencing After Nondiagnostic Exome Sequencing or Gene Panel in Developmental and Epileptic Encephalopathies. *Neurology*, **2021**, 96, e1770-e1782	6.5	16
33	Chromosome microarray in Australia: a guide for paediatricians. *Journal of Paediatrics and Child Health*, **2012**, 48, E59-67	1.3	15
32	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. *Genetics in Medicine*, **2019**, 21, 2059-2069	8.1	11
31	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. *American Journal of Human Genetics*, **2019**, 104, 542-552	11	11
30	GaborPDNet: Gabor Transformation and Deep Neural Network for Parkinson Disease Detection Using EEG Signals. *Electronics (Switzerland)*, **2021**, 10, 1740	2.6	10
29	Expanding the spectrum of mutations and novel insights into disease mechanisms. *Molecular Genetics and Metabolism Reports*, **2018**, 16, 46-51	1.8	9
28	The information needs of parents of children with early-onset epilepsy: A systematic review. *Epilepsy and Behavior*, **2020**, 112, 107382	3.2	9
27	Severe neurocognitive and growth disorders due to variation in THOC2, an essential component of nuclear mRNA export machinery. *Human Mutation*, **2018**, 39, 1126-1138	4.7	8
26	Application of Deep Learning Models for Automated Identification of Parkinson's Disease: A Review (2011-2021). *Sensors*, **2021**, 21,	3.8	8
25	Missense variants in TAF1 and developmental phenotypes: challenges of determining pathogenicity. *Human Mutation*, **2019**, 41, 449	4.7	7
24	Significantly Elevated mRNA and Mosaicism for Methylated Premutation and Full Mutation Alleles in Two Brothers with Autism Features Referred for Fragile X Testing. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	6
23	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. *Genetics in Medicine*, **2019**, 21, 2036-2042	8.1	6
22	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. *Frontiers in Molecular Neuroscience*, **2020**, 13, 12	6.1	4
21	Natural History Studies and Clinical Trial Readiness for Genetic Developmental and Epileptic Encephalopathies. *Neurotherapeutics*, **2021**, 18, 1432-1444	6.4	4
20	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4
19	Psychosocial impact of genetic testing on parents of children with developmental and epileptic encephalopathy. *Developmental Medicine and Child Neurology*,	3.3	4
18	Current use of chromosomal microarray by Australian paediatricians and implications for the implementation of next generation sequencing. *Journal of Paediatrics and Child Health*, **2017**, 53, 650-656	1.3	3

17	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants.. *Genetics in Medicine*, **2021**,	8.1	3
16	Brunner syndrome associated MAOA mutations result in NMDAR hyperfunction and increased network activity in human dopaminergic neurons.. *Neurobiology of Disease*, **2021**, 163, 105587	7.5	2
15	CHEDDA syndrome is an underrecognized neurodevelopmental disorder with a highly restricted ATN1 mutation spectrum. *Clinical Genetics*, **2021**, 100, 468-477	4	2
14	Feasibility of a mental health informed physical activity intervention for the carers of children with developmental and epileptic encephalopathy. *Epilepsy and Behavior*, **2021**, 121, 108022	3.2	2
13	RLIM Is a Candidate Dosage-Sensitive Gene for Individuals with Varying Duplications of Xq13, Intellectual Disability, and Distinct Facial Features. *American Journal of Human Genetics*, **2020**, 107, 1157-1169	11	1
12	Pre-genetics clinic resource evaluation for adults with intellectual disability: The pre-genetics clinic aid. *Journal of Genetic Counseling*, **2020**, 29, 668-677	2.5	1
11	Potassium Channel Mutations in Epilepsy **2020**,		1
10	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. *Genetics in Medicine*, **2021**, 23, 1873-1881	8.1	1
9	Quantitative neurogenetics: applications in understanding disease. *Biochemical Society Transactions* , **2021**, 49, 1621-1631	5.1	1
8	Epileptic encephalopathy caused by ARV1 deficiency: Refinement of the genotype-phenotype spectrum and functional impact on GPI-anchored proteins. *Clinical Genetics*, **2021**, 100, 607-614	4	1
7	Genetics of Epileptic Encephalopathies1-11		1
6	Automated detection of ADHD: Current trends and future perspective.. *Computers in Biology and Medicine*, **2022**, 146, 105525	7	1
5	The involvement of rare disease patient organisations in therapeutic innovation across rare paediatric neurological conditions: a narrative review.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 167	4.2	1
4	Hearing parents' voices: A priority-setting workshop to inform a suite of psychological resources for parents of children with rare genetic epilepsies. *PEC Innovation*, **2022**, 1, 100014		0
3	Dissecting the clinical outcome and cause of abnormalities of the corpus callosum. *Developmental Medicine and Child Neurology*, **2016**, 58, 430-1	3.3	0
2	Piloting positive psychology resources for caregivers of a child with a genetic developmental and epileptic encephalopathy.. *European Journal of Paediatric Neurology*, **2022**, 37, 129-138	3.8	
1	Different types of disease-causing noncoding variants revealed by genomic and gene expression analyses in families with X-linked intellectual disability. *Human Mutation*, **2021**, 42, 835-847	4.7	