Elizabeth E Palmer

List of Publications by Year
in descending order

54 papers	1,653 citations	279487 23 h-index	329751 37 g-index
59 all docs	59 docs citations	59 times ranked	2793 citing authors

#	Article	IF	Citations
1	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
2	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	13.5	103
3	DNM1 encephalopathy. Neurology, 2017, 89, 385-394.	1.5	87
4	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.	0.6	83
5	THOC2 Mutations Implicate mRNA-Export Pathway in X-Linked Intellectual Disability. American Journal of Human Genetics, 2015, 97, 302-310.	2.6	82
6	Agenesis of the corpus callosum: A clinical approach to diagnosis. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 184-197.	0.7	81
7	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.	2.6	59
8	Diagnostic Yield of Whole Genome Sequencing After Nondiagnostic Exome Sequencing or Gene Panel in Developmental and Epileptic Encephalopathies. Neurology, 2021, 96, e1770-e1782.	1.5	53
9	New insights into Brunner syndrome and potential for targeted therapy. Clinical Genetics, 2016, 89, 120-127.	1.0	52
10	Asparagine Synthetase Deficiency causes reduced proliferation of cells under conditions of limited asparagine. Molecular Genetics and Metabolism, 2015, 116, 178-186.	0.5	47
11	GaborPDNet: Gabor Transformation and Deep Neural Network for Parkinsonâ€™s Disease Detection Using EEG Signals. Electronics (Switzerland), 2021, 10, 1740.	1.8	47
12	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	2.6	46
13	Eight further individuals with intellectual disability and epilepsy carrying bi-allelicCNTNAP2aberrations allow delineation of the mutational and phenotypic spectrum. Journal of Medical Genetics, 2016, 53, 820-827.	1.5	45
14	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.	4.1	45
15	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	1.1	45
16	Automated detection of ADHD: Current trends and future perspective. Computers in Biology and Medicine, 2022, 146, 105525.	3.9	45
17	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.	1.2	42
18	Application of Deep Learning Models for Automated Identification of Parkinsonâ€™s Disease: A Review (2011â€“2021). Sensors, 2021, 21, 7034.	2.1	42

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19	A De Novo Mutation in the Sodium-Activated Potassium Channel KCNT2 Alters Ion Selectivity and Causes Epileptic Encephalopathy. Cell Reports, 2017, 21, 926-933.	2.9	40
20	Changing interpretation of chromosomal microarray over time in a community cohort with intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 377-385.	0.7	37
21	STXBP1 encephalopathy. Neurology, 2019, 93, 114-123.	1.5	37
22	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	3.7	35
23	Severe neurocognitive and growth disorders due to variation in THOC2 , an essential component of nuclear mRNA export machinery. Human Mutation, 2018, 39, 1126-1138.	1.1	28
24	Neuronal deficiency ofARV1causes an autosomal recessive epileptic encephalopathy. Human Molecular Genetics, 2016, 25, ddw157.	1.4	23
25	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.	2.6	23
26	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	1.1	23
27	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.5	23
28	The information needs of parents of children with early-onset epilepsy: A systematic review. Epilepsy and Behavior, 2020, 112, 107382.	0.9	22
29	Natural History Studies and Clinical Trial Readiness for Genetic Developmental and Epileptic Encephalopathies. Neurotherapeutics, 2021, 18, 1432-1444.	2.1	22
30	Expanding the spectrum of PEX16 mutations and novel insights into disease mechanisms. Molecular Genetics and Metabolism Reports, 2018, 16, 46-51.	0.4	21
31	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	1.1	20
32	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.	2.6	19
33	Psychosocial impact of genetic testing on parents of children with developmental and epileptic encephalopathy. Developmental Medicine and Child Neurology, 2022, 64, 95-104.	1.1	18
34	Missense variants in TAF1 and developmental phenotypes: Challenges of determining pathogenicity. Human Mutation, 2020, 41, 449-464.	1.1	17
35	Chromosome microarray in Australia: A guide for paediatricians. Journal of Paediatrics and Child Health, 2012, 48, E59-67.	0.4	16
36	Significantly Elevated FMR1 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, 3907.	1.8	12

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37	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. Frontiers in Molecular Neuroscience, 2020, 13, 12.	1.4	12
38	EPSPatNet86: eight-pointed star pattern learning network for detection ADHD disorder using EEG signals. Physiological Measurement, 2022, 43, 035002.	1.2	12
39	Exome sequencing for patients with developmental and epileptic encephalopathies in clinical practice. Developmental Medicine and Child Neurology, 2023, 65, 50-57.	1.1	11
40	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.	1.2	9
41	Brunner syndrome associated MAOA mutations result in NMDAR hyperfunction and increased network activity in human dopaminergic neurons. Neurobiology of Disease, 2022, 163, 105587.	2.1	8
42	Quantitative neurogenetics: applications in understanding disease. Biochemical Society Transactions, 2021, 49, 1621-1631.	1.6	7
43	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.	2.6	6
44	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	1.0	6
45	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	1.1	5
46	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.	0.4	4
47	<scp>CHEDDA</scp> syndrome is an underrecognized neurodevelopmental disorder with a highly restricted <scp>ATN1</scp> mutation spectrum. Clinical Genetics, 2021, 100, 468-477.	1.0	4
48	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	4
49	Preâ€genetics clinic resource evaluation for adults with intellectual disability: The preâ€genetics clinic aid. Journal of Genetic Counseling, 2020, 29, 668-677.	0.9	3
50	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.	0.7	3
51	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.	0.9	2
52	Dissecting the clinical outcome and cause of abnormalities of the corpus callosum. Developmental Medicine and Child Neurology, 2016, 58, 430-431.	1.1	1
53	Potassium Channel Mutations in Epilepsy. , 0, , 144-197.		1
54	Different types of diseaseâ€causing noncoding variants revealed by genomic and gene expression analyses in families with Xâ€linked intellectual disability. Human Mutation, 2021, 42, 835-847.	1.1	0