

# Bronwyn Kerr

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2376728/publications.pdf>

Version: 2024-02-01

24  
papers

1,525  
citations

361045

20  
h-index

610482

24  
g-index

26  
all docs

26  
docs citations

26  
times ranked

3844  
citing authors

#	ARTICLE	IF	CITATIONS
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
2	Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis. Human Molecular Genetics, 2014, 23, 4315-4327.	1.4	114
3	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	4.9	108
4	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.	2.6	87
5	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. European Journal of Medical Genetics, 2015, 58, 455-465.	0.7	83
6	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	2.6	83
7	Neurodevelopmental Disorders Caused by De Novo Variants in <i>KCNB1</i> Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	4.5	79
8	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	0.7	70
9	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	2.4	70
10	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
11	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	2.6	59
12	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	1.4	56
13	Autism spectrum disorder and other neurobehavioural comorbidities in rare disorders of the Ras/MAPK pathway. Developmental Medicine and Child Neurology, 2017, 59, 544-549.	1.1	40
14	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	2.6	40
15	Further Clinical Delineation of the MEF2C Haploinsufficiency Syndrome: Report on New Cases and Literature Review of Severe Neurodevelopmental Disorders Presenting with Seizures, Absent Speech, and Involuntary Movements. Journal of Pediatric Genetics, 2017, 06, 129-141.	0.3	38
16	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.	2.6	37
17	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. American Journal of Human Genetics, 2016, 98, 363-372.	2.6	36
18	Protein structure and phenotypic analysis of pathogenic and population missense variants in <i>STXBP1</i> . Molecular Genetics & Genomic Medicine, 2017, 5, 495-507.	0.6	29

#	ARTICLE	IF	CITATIONS
19	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. <i>American Journal of Human Genetics</i> , 2017, 101, 995-1005.	2.6	23
20	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 103.	1.2	23
21	Leri's pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing <i>GDF6</i> and <i>SDC2</i> and provides insight into systemic sclerosis pathogenesis. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1249-1256.	0.5	22
22	Catalogue of inherited disorders found among the Irish Traveller population. <i>Journal of Medical Genetics</i> , 2018, 55, 233-239.	1.5	19
23	Ligase IV syndrome can present with microcephaly and radial ray anomalies similar to Fanconi anaemia plus fatal kidney malformations. <i>European Journal of Medical Genetics</i> , 2020, 63, 103974.	0.7	5
24	High penetrance of myeloid neoplasia with diverse clinical and cytogenetic features in three siblings with a familial GATA2 deficiency. <i>Cancer Genetics</i> , 2021, 256-257, 77-80.	0.2	0