Peter Bauer

List of Publications by Year
in descending order

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97 papers	4,389 citations	186265 28 h-index	123424 61 g-index
100 all docs	100 docs citations	100 times ranked	8369 citing authors

#	Article	IF	Citations
1	A Revised Timescale for Human Evolution Based on Ancient Mitochondrial Genomes. Current Biology, 2013, 23, 553-559.	3.9	540
2	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	2.8	389
3	The natural history of spinocerebellar ataxia type 1, 2, 3, and 6. Neurology, 2011, 77, 1035-1041.	1.1	259
4	Spinocerebellar ataxia types 1, 2, 3, and 6. Neurology, 2008, 71, 982-989.	1.1	235
5	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	5.3	218
6	Long-term disease progression in spinocerebellar ataxia types 1, 2, 3, and 6: a longitudinal cohort study. Lancet Neurology, The, 2015, 14, 1101-1108.	10.2	213
7	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	7.6	144
8	The tumour suppressor gene WWOX is mutated in autosomal recessive cerebellar ataxia with epilepsy and mental retardation. Brain, 2014, 137, 411-419.	7.6	127
9	Next-generation sequencing in X-linked intellectual disability. European Journal of Human Genetics, 2015, 23, 1513-1518.	2.8	112
10	SYNE1 ataxia is a common recessive ataxia with major non-cerebellar features: a large multi-centre study. Brain, 2016, 139, 1378-1393.	7.6	87
11	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
12	Genetic screening for Niemann-Pick disease type C in adults with neurological and psychiatric symptoms: findings from the ZOOM study. Human Molecular Genetics, 2013, 22, 4349-4356.	2.9	75
13	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. Brain, 2019, 142, 1561-1572.	7.6	70
14	Survival in patients with spinocerebellar ataxia types 1, 2, 3, and 6 (EUROSCA): a longitudinal cohort study. Lancet Neurology, The, 2018, 17, 327-334.	10.2	69
15	FOXP2variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	3.2	67
16	Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. European Journal of Human Genetics, 2021, 29, 141-153.	2.8	66
17	Individual changes in preclinical spinocerebellar ataxia identified via increased motor complexity. Movement Disorders, 2016, 31, 1891-1900.	3.9	54
18	Mutation in the AP4B1 gene cause hereditary spastic paraplegia type 47 (SPG47). Neurogenetics, 2012, 13, 73-76.	1.4	52

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19	Heterozygote carriers for CNVs inPARK2are at increased risk of Parkinson's disease. Human Molecular Genetics, 2015, 24, 5637-5643.	2.9	51
20	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
21	Spinocerebellar ataxia type 11 (SCA11) is an uncommon cause of dominant ataxia among French and German kindreds. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 1229-1232.	1.9	47
22	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45
23	Homozygous stop mutation in AHR causes autosomal recessive foveal hypoplasia and infantile nystagmus. Brain, 2019, 142, 1528-1534.	7.6	41
24	MultisystemicSYNE1ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	7.6	40
25	HBOC multi-gene panel testing: comparison of two sequencing centers. Breast Cancer Research and Treatment, 2015, 152, 129-136.	2.5	38
26	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133.	6.2	37
27	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
28	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
29	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.	2.8	32
30	Epileptic Encephalopathy Caused by Mutations in the Guanine Nucleotide Exchange Factor DENND5A. American Journal of Human Genetics, 2016, 99, 1359-1367.	6.2	30
31	Epilepsy is not a mandatory feature of STXBP1 associated ataxia-tremor-retardation syndrome. European Journal of Paediatric Neurology, 2016, 20, 661-665.	1.6	30
32	Development of an evidence-based algorithm that optimizes sensitivity and specificity in ES-based diagnostics of a clinically heterogeneous patient population. Genetics in Medicine, 2019, 21, 53-61.	2.4	30
33	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	12.8	30
34	Combining exome/genome sequencing with data repository analysis reveals novel geneâ€“disease associations for a wide range of genetic disorders. Genetics in Medicine, 2021, 23, 1551-1568.	2.4	30
35	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.1	27
36	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. Molecular Psychiatry, 2019, 24, 1748-1768.	7.9	26

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37	NPC1 is enriched in unexplained early onset ataxia: a targeted high-throughput screening. Journal of Neurology, 2015, 262, 2557-2563.	3.6	25
38	The hidden Niemann-Pick type C patient: clinical niches for a rare inherited metabolic disease. Current Medical Research and Opinion, 2017, 33, 877-890.	1.9	25
39	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25
40	Loss of function of PGAP1 as a cause of severe encephalopathy identified by Whole Exome Sequencing: Lessons of the bioinformatics pipeline. Molecular and Cellular Probes, 2015, 29, 323-329.	2.1	24
41	CNTNAP1 mutations cause CNS hypomyelination and neuropathy with or without arthrogryposis. Neurology: Genetics, 2017, 3, e144.	1.9	24
42	Genomic testing in 1019 individuals from 349 Pakistani families results in high diagnostic yield and clinical utility. Npj Genomic Medicine, 2020, 5, 44.	3.8	24
43	EIF4G1is neither a strong nor a common risk factor for Parkinson's disease: evidence from large European cohorts: Table1. Journal of Medical Genetics, 2015, 52, 37-41.	3.2	23
44	Treatment Efficiency in Gaucher Patients Can Reliably Be Monitored by Quantification of Lyso-Gb1 Concentrations in Dried Blood Spots. International Journal of Molecular Sciences, 2020, 21, 4577.	4.1	23
45	Identification of a heterozygous genomic deletion in the spatacsin gene in SPG11 patients using high-resolution comparative genomic hybridization. Neurogenetics, 2009, 10, 43-48.	1.4	22
46	Biallelic inactivation of protoporphyrinogen oxidase and hydroxymethylbilane synthase is associated with liver cancer in acute porphyrias. Journal of Hepatology, 2015, 62, 734-738.	3.7	22
47	Biallelic inactivating variants in the GTPBP2 gene cause a neurodevelopmental disorder with severe intellectual disability. European Journal of Human Genetics, 2018, 26, 592-598.	2.8	22
48	First-line exome sequencing in Palestinian and Israeli Arabs with neurological disorders is efficient and facilitates disease gene discovery. European Journal of Human Genetics, 2020, 28, 1034-1043.	2.8	20
49	Genetic basis of neurodevelopmental disorders in 103 Jordanian families. Clinical Genetics, 2020, 97, 621-627.	2.0	19
50	Identifying Niemannâ€“Pick type C in early-onset ataxia: two quick clinical screening tools. Journal of Neurology, 2016, 263, 1911-1918.	3.6	16
51	MPV17 mutations in juvenileâ€•and adultâ€•onset axonal sensorimotor polyneuropathy. Clinical Genetics, 2019, 95, 182-186.	2.0	16
52	Spectrin mutations in spinocerebellar ataxia (SCA). BioEssays, 2006, 28, 785-787.	2.5	15
53	Mutations inTITF1 are not relevant to sporadic and familial chorea of unknown cause. Movement Disorders, 2006, 21, 1734-1737.	3.9	15
54	Recommendations for patient screening in ultra-rare inherited metabolic diseases: what have we learned from Niemann-Pick disease type C?. Orphanet Journal of Rare Diseases, 2019, 14, 20.	2.7	15

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55	GPT2 mutations cause developmental encephalopathy with microcephaly and features of complicated hereditary spastic paraplegia. Clinical Genetics, 2018, 94, 356-361.	2.0	14
56	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14
57	Understanding the role of genetic variability in LRRK2 in Indian population. Movement Disorders, 2019, 34, 496-505.	3.9	14
58	Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Journal of Medical Genetics, 2021, 58, 495-504.	3.2	14
59	Association of A Novel Splice Site Mutation in P/Q-Type Calcium Channels with Childhood Epilepsy and Late-Onset Slowly Progressive Non-Episodic Cerebellar Ataxia. International Journal of Molecular Sciences, 2020, 21, 3810.	4.1	14
60	An Xâ€linked syndrome with severe neurodevelopmental delay, hydrocephalus, and early lethality caused by a missense variation in the <scp>OTUD5</scp> gene. Clinical Genetics, 2021, 99, 303-308.	2.0	14
61	Spinocerebellar ataxia type 14: refining clinicogenetic diagnosis in a rare adultâ€onset disorder. Annals of Clinical and Translational Neurology, 2021, 8, 774-789.	3.7	13
62	An integrated multiomic approach as an excellent tool for the diagnosis of metabolic diseases: our first 3720 patients. European Journal of Human Genetics, 2022, 30, 1029-1035.	2.8	13
63	Biallelic <scp>ZNFX1</scp> variants are associated with a spectrum of immunoâ€hematological abnormalities. Clinical Genetics, 2022, 101, 247-254.	2.0	12
64	ADAMTS19 â€associated heart valve defects: Novel genetic variants consolidating a recognizable cardiac phenotype. Clinical Genetics, 2020, 98, 56-63.	2.0	11
65	Sanfilippo type A: new clinical manifestations and neuro-imaging findings in patients from the same family in Israel: a case report. Journal of Medical Case Reports, 2014, 8, 78.	0.8	10
66	Expanding the clinical and genetic spectra of NKX6â€2â€related disorder. Clinical Genetics, 2018, 93, 1087-1092.	2.0	10
67	Investigating the effects of additional truncating variants in DNA-repair genes on breast cancer risk in BRCA1-positive women. BMC Cancer, 2019, 19, 787.	2.6	10
68	Biallelic Pathogenic GFRA1 Variants Cause Autosomal Recessive Bilateral Renal Agenesis. Journal of the American Society of Nephrology: JASN, 2021, 32, 223-228.	6.1	10
69	Full Recovery After A Chloroquine Suicide Attempt. Journal of Toxicology: Clinical Toxicology, 1991, 29, 23-30.	1.5	8
70	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	2.8	8
71	<scp>L</scp>actotransferrin Gene (<scp>LTF</scp>) Polymorphisms and Dental Implant Loss: A Caseâ€Control Association Study. Clinical Implant Dentistry and Related Research, 2015, 17, e550-61.	3.7	7
72	Functional Characterization of Rare RAB12 Variants and Their Role in Musicianâ€™s and Other Dystonias. Genes, 2017, 8, 276.	2.4	7

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73	A novel c.671_682del NCSTN variant in a family with hidradenitis suppurativa: a pilot study. Clinical and Experimental Dermatology, 2021, 46, 1306-1308.	1.3	7
74	Whole genome sequencing (WGS), whole exome sequencing (WES) and clinical exome sequencing (CES) in patient care. Laboratoriums Medizin, 2014, 38, 221-230.	0.6	6
75	A founder nonsense variant in NUDT2 causes a recessive neurodevelopmental disorder in Saudi Arab children. Clinical Genetics, 2018, 94, 393-395.	2.0	6
76	Comprehensive clinical, biochemical and genetic screening reveals four distinct GBA genotypes as underlying variable manifestation of Gaucher disease in a single family. Molecular Genetics and Metabolism Reports, 2019, 21, 100532.	1.1	6
77	Autosomal Recessive Cerebellar Ataxia 3 Due to Homozygote c.132dupA Mutation Within theANO10Gene. JAMA Neurology, 2015, 72, 238.	9.0	5
78	Parental mosaicism in another case of Dravet syndrome caused by a novel SCN1A deletion: a case report. Journal of Medical Case Reports, 2016, 10, 67.	0.8	5
79	Capturing schizophrenia-like prodromal symptoms in a spinocerebellar ataxia-17 transgenic rat. Journal of Psychopharmacology, 2017, 31, 461-473.	4.0	5
80	Determination of the Pathological Features of NPC1 Variants in a Cellular Complementation Test. International Journal of Molecular Sciences, 2019, 20, 5185.	4.1	5
81	A homozygous frameshift variant in an alternatively spliced exon of DLG5 causes hydrocephalus and renal dysplasia. Clinical Genetics, 2019, 95, 631-633.	2.0	5
82	Biallelic lossâ€ofâ€function HACD1 variants are a bona fide cause of congenital myopathy. Clinical Genetics, 2021, 99, 513-518.	2.0	5
83	Autosomal dominant cerebellar ataxia with slow ocular saccades, neuropathy and orthostatism: A novel entity?. Parkinsonism and Related Disorders, 2014, 20, 748-754.	2.2	4
84	Intermediate phenotype of ATP13A2 mutation in two Chilean siblings: Towards a continuum between parkinsonism and hereditary spastic paraplegia. Parkinsonism and Related Disorders, 2020, 81, 45-47.	2.2	4
85	<scp>LRRK2</scp> Lossâ€ofâ€Function Variants in Patients with Rare Diseases: No Evidence for a Phenotypic Impact. Movement Disorders, 2021, 36, 1029-1031.	3.9	4
86	Whole Exome Sequencing in a Series of Patients with a Clinical Diagnosis of Tuberous Sclerosis Not Confirmed by Targeted TSC1/TSC2 Sequencing. Genes, 2021, 12, 1401.	2.4	4
87	VPS26C homozygous nonsense variant in two cousins with neurodevelopmental deficits, growth failure, skeletal abnormalities, and distinctive facial features. Clinical Genetics, 2020, 97, 644-648.	2.0	3
88	Further clinical and genetic evidence of ASC-1 complex dysfunction in congenital neuromuscular disease. European Journal of Medical Genetics, 2022, 65, 104537.	1.3	3
89	A previously identified missense mutation in STYXL1 is likely benign. European Journal of Medical Genetics, 2019, 62, 103582.	1.3	2
90	Genetic characterization of the Albanian Gaucher disease patient population. JIMD Reports, 2021, 57, 52-57.	1.5	2

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91	A novel homozygous variant in the TRAPPC9 gene causing intellectual disability and autism Spectrum disorder. Meta Gene, 2020, 26, 100783.	0.6	2
92	Unraveling the genetic cause of hereditary ophthalmic disorders in Arab societies from Israel and the Palestinian Authority. European Journal of Human Genetics, 2020, 28, 742-753.	2.8	1
93	Reply letter to Battke et al.. European Journal of Human Genetics, 2021, 29, 724-725.	2.8	1
94	SCA3: Neurological features, pathogenesis and animal models. Cerebellum, 2008, 7, 1-13.	2.5	1
95	Ciliary neurotrophic factor overexpression in neural progenitor cells (ST14A) increases proliferation, metabolic activity, and resistance to stress during differentiation. Journal of Neuroscience Research, 2004, 75, 861-861.	2.9	0
96	C10â€…Generation and characterisation of a transgenic rat model of huntington's disease-like 4 (HDL4), also called spinocerebellar ataxia type 17 (SCA17). Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, A18.2-A18.	1.9	0
97	Comorbid palmoplantar keratoderma type 1A and Loeysâ€Dietz syndrome type 3 in a patient with a chromosome 15 microdeletion. Journal of the European Academy of Dermatology and Venereology, 2022, 36, .	2.4	0