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	Biallelic <scp>ZNFX1</scp> variants are associated with a spectrum of immunoâ€hematological abnormalities. Clinical Genetics, 2022, 101, 247-254.	2.0	12
2	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
3	Recommendations for whole genome sequencing in diagnostics for rare diseases. European Journal of Human Genetics, 2022, 30, 1017-1021.	2.8	48
4	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
5	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
6	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
7	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
8	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
9	Genetic characterization of the Albanian Gaucher disease patient population. JIMD Reports, 2021, 57, 52-57.	1.5	2
10	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
11	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
12	<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
13	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
14	Biallelic lossâ€ofâ€function HACD1 variants are a bona fide cause of congenital myopathy. Clinical Genetics, 2021, 99, 513-518.	2.0	5
15	Reply letter to Battke et al.. European Journal of Human Genetics, 2021, 29, 724-725.	2.8	1
16	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
17	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
18	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

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19	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
20	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
21	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
22	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
23	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
24	Loss of MTX2 causes mandibuloacral dysplasia and links mitochondrial dysfunction to altered nuclear morphology. Nature Communications, 2020, 11, 4589.	12.8	30
25	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
26	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
27	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
28	Genetic basis of neurodevelopmental disorders in 103 Jordanian families. Clinical Genetics, 2020, 97, 621-627.	2.0	19
29	ADAMTS19 â€associated heart valve defects: Novel genetic variants consolidating a recognizable cardiac phenotype. Clinical Genetics, 2020, 98, 56-63.	2.0	11
30	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
31	A novel homozygous variant in the TRAPPC9 gene causing intellectual disability and autism Spectrum disorder. Meta Gene, 2020, 26, 100783.	0.6	2
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	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
34	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
35	Determination of the Pathological Features of NPC1 Variants in a Cellular Complementation Test. International Journal of Molecular Sciences, 2019, 20, 5185.	4.1	5
36	Prediction of Survival With Longâ€Term Disease Progression in Most Common Spinocerebellar Ataxia. Movement Disorders, 2019, 34, 1220-1227.	3.9	14

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37	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25
38	Homozygous stop mutation in AHR causes autosomal recessive foveal hypoplasia and infantile nystagmus. Brain, 2019, 142, 1528-1534.	7.6	41
39	FAHN/SPG35: a narrow phenotypic spectrum across disease classifications. Brain, 2019, 142, 1561-1572.	7.6	70
40	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
41	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
42	A previously identified missense mutation in STYXL1 is likely benign. European Journal of Medical Genetics, 2019, 62, 103582.	1.3	2
43	Understanding the role of genetic variability in LRRK2 in Indian population. Movement Disorders, 2019, 34, 496-505.	3.9	14
44	MPV17 mutations in juvenileâ€•and adultâ€onset axonal sensorimotor polyneuropathy. Clinical Genetics, 2019, 95, 182-186.	2.0	16
45	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
46	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
47	Expanding the clinical and genetic spectra of NKX6â€2â€related disorder. Clinical Genetics, 2018, 93, 1087-1092.	2.0	10
48	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
49	Long-term evolution of patient-reported outcome measures in spinocerebellar ataxias. Journal of Neurology, 2018, 265, 2040-2051.	3.6	34
50	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
51	A founder nonsense variant in NUDT2 causes a recessive neurodevelopmental disorder in Saudi Arab children. Clinical Genetics, 2018, 94, 393-395.	2.0	6
52	GPT2 mutations cause developmental encephalopathy with microcephaly and features of complicated hereditary spastic paraplegia. Clinical Genetics, 2018, 94, 356-361.	2.0	14
53	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
54	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

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55	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
56	CNTNAP1 mutations cause CNS hypomyelination and neuropathy with or without arthrogryposis. Neurology: Genetics, 2017, 3, e144.	1.9	24
57	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45
58	Capturing schizophrenia-like prodromal symptoms in a spinocerebellar ataxia-17 transgenic rat. Journal of Psychopharmacology, 2017, 31, 461-473.	4.0	5
59	Functional Characterization of Rare RAB12 Variants and Their Role in Musicianâ€™s and Other Dystonias. Genes, 2017, 8, 276.	2.4	7
60	Hereditary spastic paraplegia: Clinicogenetic lessons from 608 patients. Annals of Neurology, 2016, 79, 646-658.	5.3	218
61	Individual changes in preclinical spinocerebellar ataxia identified via increased motor complexity. Movement Disorders, 2016, 31, 1891-1900.	3.9	54
62	MultisystemicSYNE1ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. Brain, 2016, 139, e46-e46.	7.6	40
63	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
64	Uniparental disomy of chromosome 16 unmasks recessive mutations of FA2H /SPG35 in 4 families. Neurology, 2016, 87, 186-191.	1.1	27
65	Epileptic Encephalopathy Caused by Mutations in the Guanine Nucleotide Exchange Factor DENND5A. American Journal of Human Genetics, 2016, 99, 1359-1367.	6.2	30
66	Identifying Niemannâ€“Pick type C in early-onset ataxia: two quick clinical screening tools. Journal of Neurology, 2016, 263, 1911-1918.	3.6	16
67	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
68	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
69	Guidelines for diagnostic next-generation sequencing. European Journal of Human Genetics, 2016, 24, 2-5.	2.8	389
70	HBOC multi-gene panel testing: comparison of two sequencing centers. Breast Cancer Research and Treatment, 2015, 152, 129-136.	2.5	38
71	Autosomal Recessive Cerebellar Ataxia 3 Due to Homozygote c.132dupA Mutation Within theANO10Gene. JAMA Neurology, 2015, 72, 238.	9.0	5
72	Next-generation sequencing in X-linked intellectual disability. European Journal of Human Genetics, 2015, 23, 1513-1518.	2.8	112

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73	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
74	Heterozygote carriers for CNVs inPARK2are at increased risk of Parkinson's disease. Human Molecular Genetics, 2015, 24, 5637-5643.	2.9	51
75	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
76	<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
77	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
78	NPC1 is enriched in unexplained early onset ataxia: a targeted high-throughput screening. Journal of Neurology, 2015, 262, 2557-2563.	3.6	25
79	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
80	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
81	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
82	Modulation of the age at onset in spinocerebellar ataxia by CAG tracts in various genes. Brain, 2014, 137, 2444-2455.	7.6	144
83	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
84	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
85	A Revised Timescale for Human Evolution Based on Ancient Mitochondrial Genomes. Current Biology, 2013, 23, 553-559.	3.9	540
86	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
87	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
88	Mutation in the AP4B1 gene cause hereditary spastic paraplegia type 47 (SPG47). Neurogenetics, 2012, 13, 73-76.	1.4	52
89	The natural history of spinocerebellar ataxia type 1, 2, 3, and 6. Neurology, 2011, 77, 1035-1041.	1.1	259
90	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

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91	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
92	Spinocerebellar ataxia types 1, 2, 3, and 6. Neurology, 2008, 71, 982-989.	1.1	235
93	SCA3: Neurological features, pathogenesis and animal models. Cerebellum, 2008, 7, 1-13.	2.5	1
94	Spectrin mutations in spinocerebellar ataxia (SCA). BioEssays, 2006, 28, 785-787.	2.5	15
95	Mutations inTITF1 are not relevant to sporadic and familial chorea of unknown cause. Movement Disorders, 2006, 21, 1734-1737.	3.9	15
96	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
97	Full Recovery After A Chloroquine Suicide Attempt. Journal of Toxicology: Clinical Toxicology, 1991, 29, 23-30.	1.5	8