

Peter E M Taschner

List of Publications by Year in descending order

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64
papers

8,250
citations

101543

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102487

66
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69
all docs

69
docs citations

69
times ranked

13365
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutalyzer 2: next generation HGVS nomenclature checker. <i>Bioinformatics</i> , 2021, 37, 2811-2817.	4.1	44
2	Human Variome Project Quality Assessment Criteria for Variation Databases. <i>Human Mutation</i> , 2016, 37, 549-558.	2.5	18
3	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. <i>Human Mutation</i> , 2016, 37, 564-569.	2.5	1,194
4	A germline chromothripsis event stably segregating in 11 individuals through three generations. <i>Genetics in Medicine</i> , 2016, 18, 494-500.	2.4	48
5	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. <i>Human Mutation</i> , 2015, 36, 915-921.	2.5	390
6	An efficient algorithm for the extraction of HGVS variant descriptions from sequences. <i>Bioinformatics</i> , 2015, 31, 3751-3757.	4.1	25
7	Recommendations for Analyzing and Reporting TP53 Gene Variants in the High-Throughput Sequencing Era. <i>Human Mutation</i> , 2014, 35, 766-778.	2.5	29
8	Preserving sequence annotations across reference sequences. <i>Journal of Biomedical Semantics</i> , 2014, 5, S6.	1.6	3
9	In search of triallelism in Bardet-Biedl syndrome. <i>European Journal of Human Genetics</i> , 2012, 20, 420-427.	2.8	111
10	VarioML framework for comprehensive variation data representation and exchange. <i>BMC Bioinformatics</i> , 2012, 13, 254.	2.6	17
11	Curating gene variant databases (LSDBs): Toward a universal standard. <i>Human Mutation</i> , 2012, 33, 291-297.	2.5	41
12	A formalized description of the standard human variant nomenclature in Extended Backus-Naur Form. <i>BMC Bioinformatics</i> , 2011, 12, S5.	2.6	18
13	Describing structural changes by extending HGVS sequence variation nomenclature. <i>Human Mutation</i> , 2011, 32, 507-511.	2.5	57
14	LOVD v.2.0: the next generation in gene variant databases. <i>Human Mutation</i> , 2011, 32, 557-563.	2.5	854
15	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. <i>Genome Medicine</i> , 2010, 2, 24.	8.2	100
16	Sdhc and Sdhc/H19 Knockout Mice Do Not Develop Paraganglioma or Pheochromocytoma. <i>PLoS ONE</i> , 2009, 4, e7987.	2.5	49
17	Deep sequencing to reveal new variants in pooled DNA samples. <i>Human Mutation</i> , 2009, 30, 1703-1712.	2.5	71
18	Improving sequence variant descriptions in mutation databases and literature using the Mutalyzer sequence variation nomenclature checker. <i>Human Mutation</i> , 2008, 29, 6-13.	2.5	383

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19	Caenorhabditis elegans as a model for lysosomal storage disorders. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2008, 1782, 433-446.	3.8	40
20	Characterizing pathogenic processes in Batten disease: Use of small eukaryotic model systems. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 906-919.	3.8	26
21	Mutation analysis of SDHB and SDHC: novel germline mutations in sporadic head and neck paraganglioma and familial paraganglioma and/or pheochromocytoma. <i>BMC Medical Genetics</i> , 2006, 7, 1.	2.1	112
22	Polymorphism of μ -Opioid Receptor Gene (OPRM1:c.118A>G) Does Not Protect Against Opioid-induced Respiratory Depression despite Reduced Analgesic Response. <i>Anesthesiology</i> , 2005, 102, 522-530.	2.5	146
23	LOVD: Easy creation of a locus-specific sequence variation database using an "SDB-in-a-box" approach. <i>Human Mutation</i> , 2005, 26, 63-68.	2.5	235
24	Deletion of the Caenorhabditis elegans homologues of the CLN3 gene, involved in human juvenile neuronal ceroid lipofuscinosis, causes a mild progeric phenotype. <i>Journal of Inherited Metabolic Disease</i> , 2005, 28, 1065-1080.	3.6	15
25	The SDH mutation database: an online resource for succinate dehydrogenase sequence variants involved in pheochromocytoma, paraganglioma and mitochondrial complex II deficiency. <i>BMC Medical Genetics</i> , 2005, 6, 39.	2.1	164
26	Increased prevalence of catecholamine excess and phaeochromocytomas in a well-defined Dutch population with SDHD-linked head and neck paragangliomas. <i>European Journal of Endocrinology</i> , 2005, 152, 87-94.	3.7	39
27	Carotid Body Tumors in Humans Caused by a Mutation in the Gene for Succinate Dehydrogenase D (SDHD). <i>Advances in Experimental Medicine and Biology</i> , 2004, 551, 71-76.	1.6	8
28	Somatic loss of maternal chromosome 11 causes parent-of-origin-dependent inheritance in SDHD-linked paraganglioma and phaeochromocytoma families. <i>Oncogene</i> , 2004, 23, 4076-4083.	5.9	146
29	Pharmacokinetic-Pharmacodynamic Modeling of Morphine-6-glucuronide-induced Analgesia in Healthy Volunteers. <i>Anesthesiology</i> , 2004, 100, 120-133.	2.5	152
30	SDHD mutations in head and neck paragangliomas result in destabilization of complex II in the mitochondrial respiratory chain with loss of enzymatic activity and abnormal mitochondrial morphology. <i>Journal of Pathology</i> , 2003, 201, 480-486.	4.5	83
31	Hereditary Paraganglioma Due to the SDHD M1I Mutation in a Second Chinese Family: A Founder Effect?. <i>Laryngoscope</i> , 2003, 113, 1055-1058.	2.0	20
32	Phenotypic dichotomy in mitochondrial complex II genetic disorders. <i>Journal of Molecular Medicine</i> , 2001, 79, 495-503.	3.9	67
33	Novel mutations in the SDHD gene in pedigrees with familial carotid body paraganglioma and sensorineural hearing loss. <i>Genes Chromosomes and Cancer</i> , 2001, 31, 255-263.	2.8	70
34	Nearly all hereditary paragangliomas in The Netherlands are caused by two founder mutations in the SDHD gene. <i>Genes Chromosomes and Cancer</i> , 2001, 31, 274-281.	2.8	149
35	Adult neuronal ceroid lipofuscinosis with palmitoyl-protein thioesterase deficiency: First adult-onset patients of a childhood disease. <i>Annals of Neurology</i> , 2001, 50, 269-272.	5.3	113
36	A high-resolution integrated map spanning the SDHD gene at 11q23: a 1.1-Mb BAC contig, a partial transcript map and 15 new repeat polymorphisms in a tumour-suppressor region. <i>European Journal of Human Genetics</i> , 2001, 9, 121-129.	2.8	31

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37	Caenorhabditis elegans homologues of the CLN3 gene, mutated in juvenile neuronal ceroid lipofuscinosis. European Journal of Paediatric Neurology, 2001, 5, 115-120.	1.6	4
38	Why and how to assess the aetiological diagnosis of children with intellectual disability/mental retardation and other neurodevelopmental disorders: description of the Finnish approach. European Journal of Paediatric Neurology, 2001, 5, 7-13.	1.6	29
39	Visual disorders in children with brain lesions. European Journal of Paediatric Neurology, 2001, 5, 115-119.	1.6	70
40	Mutations in <i>SDHD</i> , a Mitochondrial Complex II Gene, in Hereditary Paraganglioma. Science, 2000, 287, 848-851.	12.6	1,554
41	Repositioning the hereditary paraganglioma critical region on chromosome band 11q23. Human Genetics, 1999, 104, 219-225.	3.8	52
42	First-trimester diagnosis of infantile neuronal ceroid lipofuscinosis (INCL) using PPT enzyme assay and CLN1 mutation analysis. , 1999, 19, 559-562.		15
43	Molecular Analysis of SALL1 Mutations in Townes-Brocks Syndrome. American Journal of Human Genetics, 1999, 64, 435-445.	6.2	129
44	Genetic Heterogeneity of Neuronal Ceroid Lipofuscinosis in the Netherlands. Molecular Genetics and Metabolism, 1999, 66, 339-343.	1.1	8
45	A Murine Model for Juvenile NCL: Gene Targeting of Mouse Cln3. Molecular Genetics and Metabolism, 1999, 66, 309-313.	1.1	31
46	The Molecular Basis of GROD-Storing Neuronal Ceroid Lipofuscinoses in Scotland. Molecular Genetics and Metabolism, 1999, 66, 245-247.	1.1	10
47	Targeted Disruption of the Cln3 Gene Provides a Mouse Model for Batten Disease. Neurobiology of Disease, 1999, 6, 321-334.	4.4	180
48	Mutations in the palmitoyl-protein thioesterase gene (PPT; CLN1) causing juvenile neuronal ceroid lipofuscinosis with granular osmiophilic deposits [published erratum appears in Hum Mol Genet 1998 Apr;7(4):765]. Human Molecular Genetics, 1998, 7, 291-297.	2.9	122
49	Genomic Structure and Complete Nucleotide Sequence of the Batten Disease Gene, CLN3. Genomics, 1997, 40, 346-350.	2.9	47
50	Characterization of the Gene Encoding Human Sarcolipin (SLN), a Proteolipid Associated with SERCA1: Absence of Structural Mutations in Five Patients with Brody Disease. Genomics, 1997, 45, 541-553.	2.9	159
51	Spectrum of Mutations in the Batten Disease Gene, CLN3. American Journal of Human Genetics, 1997, 61, 310-316.	6.2	181
52	Linkage of Gitelman syndrome to the thiazide-sensitive sodium-chloride cotransporter gene with identification of mutations in Dutch families. Pediatric Nephrology, 1996, 10, 403-407.	1.7	63
53	Mutations in the gene encoding SERCA1, the fast-twitch skeletal muscle sarcoplasmic reticulum Ca ²⁺ ATPase, are associated with Brody disease. Nature Genetics, 1996, 14, 191-194.	21.4	223
54	Late onset juvenile neuronal ceroid-lipofuscinosis with granular osmiophilic deposits (GROD). American Journal of Medical Genetics Part A, 1995, 57, 165-167.	2.4	18

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55	Refined localization of the Batten disease gene (CLN3) by haplotype and linkage disequilibrium mapping to D16S288-D16S383 and exclusion from this region of a variant form of Batten disease with granular osmiophilic deposits. <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 312-315.	2.4	16
56	Physical map of the region containing the gene for Batten disease (CLN3). <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 316-319.	2.4	10
57	Isolation of genes from the Batten candidate region using exon amplification. <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 320-323.	2.4	5
58	Carrier detection of Batten disease (Juvenile neuronal ceroid-lipofuscinosis). <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 333-337.	2.4	13
59	Application of chromosome 16 markers in the differential diagnosis of neuronal ceroid-lipofuscinosis. <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 338-343.	2.4	3
60	Replicase-Mediated Resistance to Alfalfa Mosaic Virus. <i>Virology</i> , 1995, 207, 467-474.	2.4	59
61	YAC and Cosmid Contigs Spanning the Batten Disease (CLN3) Region at 16p12.1â€“p11.2. <i>Genomics</i> , 1995, 29, 478-489.	2.9	8
62	Plants Transformed with a Mutant Alfalfa Mosaic Virus Coat Protein Gene Are Resistant to the Mutant but Not to Wild-Type Virus. <i>Virology</i> , 1994, 203, 269-276.	2.4	27
63	Genetic Mapping of the Batten Disease Locus (CLN3) to the Interval D16S288-D16S383 by Analysis of Haplotypes and Allelic Association. <i>Genomics</i> , 1994, 22, 465-468.	2.9	33
64	Replication of an incomplete alfalfa mosaic virus genome in plants transformed with viral replicase genes. <i>Virology</i> , 1991, 181, 445-450.	2.4	102