Peter E M Taschner

List of Publications by Year in Descending Order

64 papers	6,753 citations	35 h-index	69 g-index
69 ext. papers	7,619 ext. citations	6 avg, IF	5.14 L-index

#	Paper	IF	Citations
64	Next Generation HGVS Nomenclature Checker. *Bioinformatics*, **2021**,	7.2	2
63	A germline chromothripsis event stably segregating in 11 individuals through three generations. *Genetics in Medicine*, **2016**, 18, 494-500	8.1	42
62	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. *Human Mutation*, **2016**, 37, 564-9	4.7	792
61	Human Variome Project Quality Assessment Criteria for Variation Databases. *Human Mutation*, **2016**, 37, 549-58	4.7	13
60	The Matchmaker Exchange: a platform for rare disease gene discovery. *Human Mutation*, **2015**, 36, 915-21	4.7	280
59	An efficient algorithm for the extraction of HGVS variant descriptions from sequences. *Bioinformatics*, **2015**, 31, 3751-7	7.2	10
58	Recommendations for analyzing and reporting TP53 gene variants in the high-throughput sequencing era. *Human Mutation*, **2014**, 35, 766-78	4.7	25
57	Preserving sequence annotations across reference sequences. *Journal of Biomedical Semantics*, **2014**, 5, S6	2.2	3
56	Curating gene variant databases (LSDBs): toward a universal standard. *Human Mutation*, **2012**, 33, 291-7	4.7	39
55	VarioML framework for comprehensive variation data representation and exchange. *BMC Bioinformatics*, **2012**, 13, 254	3.6	14
54	In search of triallelism in Bardet-Biedl syndrome. *European Journal of Human Genetics*, **2012**, 20, 420-7	5.3	88
53	A formalized description of the standard human variant nomenclature in Extended Backus-Naur Form. *BMC Bioinformatics*, **2011**, 12 Suppl 4, S5	3.6	15
52	Describing structural changes by extending HGVS sequence variation nomenclature. *Human Mutation*, **2011**, 32, 507-11	4.7	54
51	LOVD v.2.0: the next generation in gene variant databases. *Human Mutation*, **2011**, 32, 557-63	4.7	661
50	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. *Genome Medicine*, **2010**, 2, 24	14.4	86
49	Sdhd and SDHD/H19 knockout mice do not develop paraganglioma or pheochromocytoma. *PLoS ONE*, **2009**, 4, e7987	3.7	40
48	Deep sequencing to reveal new variants in pooled DNA samples. *Human Mutation*, **2009**, 30, 1703-12	4.7	68

47	Caenorhabditis elegans as a model for lysosomal storage disorders. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2008**, 1782, 433-46	6.9	32
46	Improving sequence variant descriptions in mutation databases and literature using the Mutalyzer sequence variation nomenclature checker. *Human Mutation*, **2008**, 29, 6-13	4.7	332
45	Characterizing pathogenic processes in Batten disease: use of small eukaryotic model systems. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2006**, 1762, 906-19	6.9	24
44	Mutation analysis of SDHB and SDHC: novel germline mutations in sporadic head and neck paraganglioma and familial paraganglioma and/or pheochromocytoma. *BMC Medical Genetics*, **2006** , 7, 1	2.1	86
43	Polymorphism of mu-opioid receptor gene (OPRM1:c.118A>G) does not protect against opioid-induced respiratory depression despite reduced analgesic response. *Anesthesiology*, **2005**, 102, 522-30	4.3	132
42	LOVD: easy creation of a locus-specific sequence variation database using an "LSDB-in-a-box" approach. *Human Mutation*, **2005**, 26, 63-8	4.7	202
41	Deletion of the Caenorhabditis elegans homologues of the CLN3 gene, involved in human juvenile neuronal ceroid lipofuscinosis, causes a mild progeric phenotype. *Journal of Inherited Metabolic Disease*, **2005**, 28, 1065-80	5.4	13
40	The SDH mutation database: an online resource for succinate dehydrogenase sequence variants involved in pheochromocytoma, paraganglioma and mitochondrial complex II deficiency. *BMC Medical Genetics*, **2005**, 6, 39	2.1	142
39	Increased prevalence of catecholamine excess and phaeochromocytomas in a well-defined Dutch population with SDHD-linked head and neck paragangliomas. *European Journal of Endocrinology*, **2005**, 152, 87-94	6.5	35
38	Carotid body tumors in humans caused by a mutation in the gene for succinate dehydrogenase D (SDHD). *Advances in Experimental Medicine and Biology*, **2004**, 551, 71-6	3.6	6
37	Somatic loss of maternal chromosome 11 causes parent-of-origin-dependent inheritance in SDHD-linked paraganglioma and phaeochromocytoma families. *Oncogene*, **2004**, 23, 4076-83	9.2	127
36	Pharmacokinetic-pharmacodynamic modeling of morphine-6-glucuronide-induced analgesia in healthy volunteers: absence of sex differences. *Anesthesiology*, **2004**, 100, 120-33	4.3	146
35	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. *Journal of Pathology*, **2003**, 201, 480-6	9.4	78
34	Hereditary paraganglioma due to the SDHD M1I mutation in a second Chinese family: a founder effect?. *Laryngoscope*, **2003**, 113, 1055-8	3.6	17
33	Phenotypic dichotomy in mitochondrial complex II genetic disorders. *Journal of Molecular Medicine*, **2001**, 79, 495-503	5.5	60
32	Novel mutations in the SDHD gene in pedigrees with familial carotid body paraganglioma and sensorineural hearing loss. *Genes Chromosomes and Cancer*, **2001**, 31, 255-63	5	62
31	Nearly all hereditary paragangliomas in the Netherlands are caused by two founder mutations in the SDHD gene. *Genes Chromosomes and Cancer*, **2001**, 31, 274-81	5	119
30	Adult neuronal ceroid lipofuscinosis with palmitoyl-protein thioesterase deficiency: first adult-onset patients of a childhood disease. *Annals of Neurology*, **2001**, 50, 269-72	9.4	96

29	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. *European Journal of Human Genetics*, **2001**, 9, 121-9	5.3	26
28	Caenorhabditis elegans homologues of the CLN3 gene, mutated in juvenile neuronal ceroid lipofuscinosis. *European Journal of Paediatric Neurology*, **2001**, 5 Suppl A, 115-20	3.8	4
27	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	3.8	22
26	Visual disorders in children with brain lesions: 2. Visual impairment associated with cerebral palsy. *European Journal of Paediatric Neurology*, **2001**, 5, 115-9	3.8	53
25	Mutations in SDHD, a mitochondrial complex II gene, in hereditary paraganglioma. *Science*, **2000**, 287, 848-51	33.3	1367
24	Repositioning the hereditary paraganglioma critical region on chromosome band 11q23. *Human Genetics*, **1999**, 104, 219-25	6.3	48
23	First-trimester diagnosis of infantile neuronal ceroid lipofuscinosis (INCL) using PPT enzyme assay and CLN1 mutation analysis **1999**, 19, 559-562		13
22	Molecular analysis of SALL1 mutations in Townes-Brocks syndrome. *American Journal of Human Genetics*, **1999**, 64, 435-45	11	113
21	Genetic heterogeneity of neuronal ceroid lipofuscinosis in The Netherlands. *Molecular Genetics and Metabolism*, **1999**, 66, 339-43	3.7	8
20	A murine model for juvenile NCL: gene targeting of mouse Cln3. *Molecular Genetics and Metabolism*, **1999**, 66, 309-13	3.7	27
19	The molecular basis of GROD-storing neuronal ceroid lipofuscinoses in Scotland. *Molecular Genetics and Metabolism*, **1999**, 66, 245-7	3.7	9
18	Targeted disruption of the Cln3 gene provides a mouse model for Batten disease. The Batten Mouse Model Consortium [corrected]. *Neurobiology of Disease*, **1999**, 6, 321-34	7.5	163
17	Mutations in the palmitoyl-protein thioesterase gene (PPT; CLN1) causing juvenile neuronal ceroid lipofuscinosis with granular osmiophilic deposits. *Human Molecular Genetics*, **1998**, 7, 291-7	5.6	102
16	Genomic structure and complete nucleotide sequence of the Batten disease gene, CLN3. *Genomics*, **1997**, 40, 346-50	4.3	38
15	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, 541-53	4.3	142
14	Spectrum of mutations in the Batten disease gene, CLN3. *American Journal of Human Genetics*, **1997**, 61, 310-6	11	155
13	Linkage of Gitelman syndrome to the thiazide-sensitive sodium-chloride cotransporter gene with identification of mutations in Dutch families. *Pediatric Nephrology*, **1996**, 10, 403-7	3.2	58
12	Mutations in the gene-encoding SERCA1, the fast-twitch skeletal muscle sarcoplasmic reticulum Ca2+ ATPase, are associated with Brody disease. *Nature Genetics*, **1996**, 14, 191-4	36.3	200

11	Replicase-mediated resistance to alfalfa mosaic virus. *Virology*, **1995**, 207, 467-74	3.6	51
10	YAC and cosmid contigs spanning the Batten disease (CLN3) region at 16p12.1-p11.2. *Genomics*, **1995**, 29, 478-89	4.3	8
9	Late onset juvenile neuronal ceroid-lipofuscinosis with granular osmiophilic deposits (GROD). *American Journal of Medical Genetics Part A*, **1995**, 57, 165-7		17
8	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. *American Journal of Medical Genetics Part A*, **1995**, 57, 312-5		16
7	Physical map of the region containing the gene for Batten disease (CLN3). *American Journal of Medical Genetics Part A*, **1995**, 57, 316-9		10
6	Isolation of genes from the Batten candidate region using exon amplification. Batten Disease Consortium. *American Journal of Medical Genetics Part A*, **1995**, 57, 320-3		5
5	Carrier detection of Batten disease (juvenile neuronal ceroid-lipofuscinosis). *American Journal of Medical Genetics Part A*, **1995**, 57, 333-7		12
4	Application of chromosome 16 markers in the differential diagnosis of neuronal ceroid-lipofuscinosis. *American Journal of Medical Genetics Part A*, **1995**, 57, 338-43		3
3	Plants transformed with a mutant alfalfa mosaic virus coat protein gene are resistant to the mutant but not to wild-type virus. *Virology*, **1994**, 203, 269-76	3.6	21
2	Genetic mapping of the Batten disease locus (CLN3) to the interval D16S288-D16S383 by analysis of haplotypes and allelic association. *Genomics*, **1994**, 22, 465-8	4.3	29
1	Replication of an incomplete alfalfa mosaic virus genome in plants transformed with viral replicase genes. *Virology*, **1991**, 181, 445-50	3.6	84