

# Peter E M Taschner

## List of Publications by Citations

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64  
papers

6,753  
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35  
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69  
g-index

69  
ext. papers

7,619  
ext. citations

6  
avg, IF

5.14  
L-index

#	Paper	IF	Citations
64	Mutations in SDHD, a mitochondrial complex II gene, in hereditary paraganglioma. <i>Science</i> , <b>2000</b> , 287, 848-51	33.3	1367
63	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. <i>Human Mutation</i> , <b>2016</b> , 37, 564-9	4.7	792
62	LOVD v.2.0: the next generation in gene variant databases. <i>Human Mutation</i> , <b>2011</b> , 32, 557-63	4.7	661
61	Improving sequence variant descriptions in mutation databases and literature using the Mutalyzer sequence variation nomenclature checker. <i>Human Mutation</i> , <b>2008</b> , 29, 6-13	4.7	332
60	The Matchmaker Exchange: a platform for rare disease gene discovery. <i>Human Mutation</i> , <b>2015</b> , 36, 915-24	4.7	280
59	LOVD: easy creation of a locus-specific sequence variation database using an "LSDB-in-a-box" approach. <i>Human Mutation</i> , <b>2005</b> , 26, 63-8	4.7	202
58	Mutations in the gene-encoding SERCA1, the fast-twitch skeletal muscle sarcoplasmic reticulum Ca <sup>2+</sup> ATPase, are associated with Brody disease. <i>Nature Genetics</i> , <b>1996</b> , 14, 191-4	36.3	200
57	Targeted disruption of the Cln3 gene provides a mouse model for Batten disease. The Batten Mouse Model Consortium [corrected]. <i>Neurobiology of Disease</i> , <b>1999</b> , 6, 321-34	7.5	163
56	Spectrum of mutations in the Batten disease gene, CLN3. <i>American Journal of Human Genetics</i> , <b>1997</b> , 61, 310-6	11	155
55	Pharmacokinetic-pharmacodynamic modeling of morphine-6-glucuronide-induced analgesia in healthy volunteers: absence of sex differences. <i>Anesthesiology</i> , <b>2004</b> , 100, 120-33	4.3	146
54	Characterization of the gene encoding human sarcolipin (SLN), a proteolipid associated with SERCA1: absence of structural mutations in five patients with Brody disease. <i>Genomics</i> , <b>1997</b> , 45, 541-53	4.3	142
53	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> , <b>2005</b> , 6, 39	2.1	142
52	Polymorphism of mu-opioid receptor gene (OPRM1:c.118A>G) does not protect against opioid-induced respiratory depression despite reduced analgesic response. <i>Anesthesiology</i> , <b>2005</b> , 102, 522-30	4.3	132
51	Somatic loss of maternal chromosome 11 causes parent-of-origin-dependent inheritance in SDHD-linked paraganglioma and pheochromocytoma families. <i>Oncogene</i> , <b>2004</b> , 23, 4076-83	9.2	127
50	Nearly all hereditary paragangliomas in the Netherlands are caused by two founder mutations in the SDHD gene. <i>Genes Chromosomes and Cancer</i> , <b>2001</b> , 31, 274-81	5	119
49	Molecular analysis of SALL1 mutations in Townes-Brocks syndrome. <i>American Journal of Human Genetics</i> , <b>1999</b> , 64, 435-45	11	113
48	Mutations in the palmitoyl-protein thioesterase gene (PPT; CLN1) causing juvenile neuronal ceroid lipofuscinosis with granular osmiophilic deposits. <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 291-7	5.6	102

47	Adult neuronal ceroid lipofuscinosis with palmitoyl-protein thioesterase deficiency: first adult-onset patients of a childhood disease. <i>Annals of Neurology</i> , <b>2001</b> , 50, 269-72	9.4	96
46	In search of triallelism in Bardet-Biedl syndrome. <i>European Journal of Human Genetics</i> , <b>2012</b> , 20, 420-7	5.3	88
45	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. <i>Genome Medicine</i> , <b>2010</b> , 2, 24	14.4	86
44	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> , <b>2006</b> , 7, 1	2.1	86
43	Replication of an incomplete alfalfa mosaic virus genome in plants transformed with viral replicase genes. <i>Virology</i> , <b>1991</b> , 181, 445-50	3.6	84
42	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> , <b>2003</b> , 201, 480-6	9.4	78
41	Deep sequencing to reveal new variants in pooled DNA samples. <i>Human Mutation</i> , <b>2009</b> , 30, 1703-12	4.7	68
40	Novel mutations in the SDHD gene in pedigrees with familial carotid body paraganglioma and sensorineural hearing loss. <i>Genes Chromosomes and Cancer</i> , <b>2001</b> , 31, 255-63	5	62
39	Phenotypic dichotomy in mitochondrial complex II genetic disorders. <i>Journal of Molecular Medicine</i> , <b>2001</b> , 79, 495-503	5.5	60
38	Linkage of Gitelman syndrome to the thiazide-sensitive sodium-chloride cotransporter gene with identification of mutations in Dutch families. <i>Pediatric Nephrology</i> , <b>1996</b> , 10, 403-7	3.2	58
37	Describing structural changes by extending HGVS sequence variation nomenclature. <i>Human Mutation</i> , <b>2011</b> , 32, 507-11	4.7	54
36	Visual disorders in children with brain lesions: 2. Visual impairment associated with cerebral palsy. <i>European Journal of Paediatric Neurology</i> , <b>2001</b> , 5, 115-9	3.8	53
35	Replicase-mediated resistance to alfalfa mosaic virus. <i>Virology</i> , <b>1995</b> , 207, 467-74	3.6	51
34	Repositioning the hereditary paraganglioma critical region on chromosome band 11q23. <i>Human Genetics</i> , <b>1999</b> , 104, 219-25	6.3	48
33	A germline chromothripsis event stably segregating in 11 individuals through three generations. <i>Genetics in Medicine</i> , <b>2016</b> , 18, 494-500	8.1	42
32	Sdhc and SDHD/H19 knockout mice do not develop paraganglioma or pheochromocytoma. <i>PLoS ONE</i> , <b>2009</b> , 4, e7987	3.7	40
31	Curating gene variant databases (LSDBs): toward a universal standard. <i>Human Mutation</i> , <b>2012</b> , 33, 291-7	4.7	39
30	Genomic structure and complete nucleotide sequence of the Batten disease gene, CLN3. <i>Genomics</i> , <b>1997</b> , 40, 346-50	4.3	38

29	Increased prevalence of catecholamine excess and pheochromocytomas in a well-defined Dutch population with SDHD-linked head and neck paragangliomas. <i>European Journal of Endocrinology</i> , <b>2005</b> , 152, 87-94	6.5	35
28	<i>Caenorhabditis elegans</i> as a model for lysosomal storage disorders. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2008</b> , 1782, 433-46	6.9	32
27	Genetic mapping of the Batten disease locus (CLN3) to the interval D16S288-D16S383 by analysis of haplotypes and allelic association. <i>Genomics</i> , <b>1994</b> , 22, 465-8	4.3	29
26	A murine model for juvenile NCL: gene targeting of mouse Cln3. <i>Molecular Genetics and Metabolism</i> , <b>1999</b> , 66, 309-13	3.7	27
25	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> , <b>2001</b> , 9, 121-9	5.3	26
24	Recommendations for analyzing and reporting TP53 gene variants in the high-throughput sequencing era. <i>Human Mutation</i> , <b>2014</b> , 35, 766-78	4.7	25
23	Characterizing pathogenic processes in Batten disease: use of small eukaryotic model systems. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2006</b> , 1762, 906-19	6.9	24
22	Why and how to assess the aetiological diagnosis of children with intellectual disability/mental retardation and other neurodevelopmental disorders: description of the Finnish approach. <i>European Journal of Paediatric Neurology</i> , <b>2001</b> , 5, 7-13	3.8	22
21	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> , <b>1994</b> , 203, 269-76	3.6	21
20	Hereditary paraganglioma due to the SDHD M11 mutation in a second Chinese family: a founder effect?. <i>Laryngoscope</i> , <b>2003</b> , 113, 1055-8	3.6	17
19	Late onset juvenile neuronal ceroid-lipofuscinosis with granular osmiophilic deposits (GROD). <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 57, 165-7		17
18	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> , <b>1995</b> , 57, 312-5		16
17	A formalized description of the standard human variant nomenclature in Extended Backus-Naur Form. <i>BMC Bioinformatics</i> , <b>2011</b> , 12 Suppl 4, S5	3.6	15
16	VarioML framework for comprehensive variation data representation and exchange. <i>BMC Bioinformatics</i> , <b>2012</b> , 13, 254	3.6	14
15	Deletion of the <i>Caenorhabditis elegans</i> homologues of the CLN3 gene, involved in human juvenile neuronal ceroid lipofuscinosis, causes a mild progeric phenotype. <i>Journal of Inherited Metabolic Disease</i> , <b>2005</b> , 28, 1065-80	5.4	13
14	First-trimester diagnosis of infantile neuronal ceroid lipofuscinosis (INCL) using PPT enzyme assay and CLN1 mutation analysis <b>1999</b> , 19, 559-562		13
13	Human Variome Project Quality Assessment Criteria for Variation Databases. <i>Human Mutation</i> , <b>2016</b> , 37, 549-58	4.7	13
12	Carrier detection of Batten disease (juvenile neuronal ceroid-lipofuscinosis). <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 57, 333-7		12

11	An efficient algorithm for the extraction of HGVS variant descriptions from sequences. <i>Bioinformatics</i> , <b>2015</b> , 31, 3751-7	7.2	10
10	Physical map of the region containing the gene for Batten disease (CLN3). <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 57, 316-9		10
9	The molecular basis of GROD-storing neuronal ceroid lipofuscinoses in Scotland. <i>Molecular Genetics and Metabolism</i> , <b>1999</b> , 66, 245-7	3.7	9
8	Genetic heterogeneity of neuronal ceroid lipofuscinosis in The Netherlands. <i>Molecular Genetics and Metabolism</i> , <b>1999</b> , 66, 339-43	3.7	8
7	YAC and cosmid contigs spanning the Batten disease (CLN3) region at 16p12.1-p11.2. <i>Genomics</i> , <b>1995</b> , 29, 478-89	4.3	8
6	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> , <b>2004</b> , 551, 71-6	3.6	6
5	Isolation of genes from the Batten candidate region using exon amplification. Batten Disease Consortium. <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 57, 320-3		5
4	Caenorhabditis elegans homologues of the CLN3 gene, mutated in juvenile neuronal ceroid lipofuscinosis. <i>European Journal of Paediatric Neurology</i> , <b>2001</b> , 5 Suppl A, 115-20	3.8	4
3	Preserving sequence annotations across reference sequences. <i>Journal of Biomedical Semantics</i> , <b>2014</b> , 5, S6	2.2	3
2	Application of chromosome 16 markers in the differential diagnosis of neuronal ceroid-lipofuscinosis. <i>American Journal of Medical Genetics Part A</i> , <b>1995</b> , 57, 338-43		3
1	Next Generation HGVS Nomenclature Checker. <i>Bioinformatics</i> , <b>2021</b> ,	7.2	2