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List of Publications by Year
in descending order

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64 papers	8,250 citations	101496 36 h-index	102432 66 g-index
69 all docs	69 docs citations	69 times ranked	13365 citing authors

#	Article	IF	Citations
1	Mutalyzer 2: next generation HGVS nomenclature checker. Bioinformatics, 2021, 37, 2811-2817.	1.8	44
2	Human Variome Project Quality Assessment Criteria for Variation Databases. Human Mutation, 2016, 37, 549-558.	1.1	18
3	HGVS Recommendations for the Description of Sequence Variants: 2016 Update. Human Mutation, 2016, 37, 564-569.	1.1	1,194
4	A germline chromothripsis event stably segregating in 11 individuals through three generations. Genetics in Medicine, 2016, 18, 494-500.	1.1	48
5	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
6	An efficient algorithm for the extraction of HGVS variant descriptions from sequences. Bioinformatics, 2015, 31, 3751-3757.	1.8	25
7	Recommendations for Analyzing and ReportingTP53Gene Variants in the High-Throughput Sequencing Era. Human Mutation, 2014, 35, 766-778.	1.1	29
8	Preserving sequence annotations across reference sequences. Journal of Biomedical Semantics, 2014, 5, S6.	0.9	3
9	In search of triallelism in Bardetâ€"Biedl syndrome. European Journal of Human Genetics, 2012, 20, 420-427.	1.4	111
10	VarioML framework for comprehensive variation data representation and exchange. BMC Bioinformatics, 2012, 13, 254.	1.2	17
11	Curating gene variant databases (LSDBs): Toward a universal standard. Human Mutation, 2012, 33, 291-297.	1.1	41
12	A formalized description of the standard human variant nomenclature in Extended Backus-Naur Form. BMC Bioinformatics, 2011, 12, S5.	1.2	18
13	Describing structural changes by extending HGVS sequence variation nomenclature. Human Mutation, 2011, 32, 507-511.	1.1	57
14	LOVD v.2.0: the next generation in gene variant databases. Human Mutation, 2011, 32, 557-563.	1.1	854
15	Locus Reference Genomic sequences: an improved basis for describing human DNA variants. Genome Medicine, 2010, 2, 24.	3.6	100
16	Sdhd and Sdhd/H19 Knockout Mice Do Not Develop Paraganglioma or Pheochromocytoma. PLoS ONE, 2009, 4, e7987.	1.1	49
17	Deep sequencing to reveal new variants in pooled DNA samples. Human Mutation, 2009, 30, 1703-1712.	1.1	71
18	Improving sequence variant descriptions in mutation databases and literature using the Mutalyzer sequence variation nomenclature checker. Human Mutation, 2008, 29, 6-13.	1.1	383

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19	Caenorhabditis elegans as a model for lysosomal storage disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2008, 1782, 433-446.	1.8	40
20	Characterizing pathogenic processes in Batten disease: Use of small eukaryotic model systems. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 906-919.	1.8	26
21	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	112
22	Polymorphism of Î¼-Opioid Receptor Gene (OPRM1:c.118A>GÂ) Does Not Protect Against Opioid-induced Respiratory Depression despite Reduced Analgesic Response. Anesthesiology, 2005, 102, 522-530.	1.3	146
23	LOVD: Easy creation of a locus-specific sequence variation database using an â€œLSDB-in-a-boxâ€•approach. Human Mutation, 2005, 26, 63-68.	1.1	235
24	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, 1065-1080.	1.7	15
25	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	164
26	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.	1.9	39
27	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, 71-76.	0.8	8
28	Somatic loss of maternal chromosome 11 causes parent-of-origin-dependent inheritance in SDHD-linked paraganglioma and phaeochromocytoma families. Oncogene, 2004, 23, 4076-4083.	2.6	146
29	Pharmacokinetic-Pharmacodynamic Modeling of Morphine-6-glucuronide-induced Analgesia in Healthy Volunteers. Anesthesiology, 2004, 100, 120-133.	1.3	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. Journal of Pathology, 2003, 201, 480-486.	2.1	83
31	Hereditary Paraganglioma Due to the SDHD M1I Mutation in a Second Chinese Family: A Founder Effect?. Laryngoscope, 2003, 113, 1055-1058.	1.1	20
32	Phenotypic dichotomy in mitochondrial complex II genetic disorders. Journal of Molecular Medicine, 2001, 79, 495-503.	1.7	67
33	Novel mutations in theSDHD gene in pedigrees with familial carotid body paraganglioma and sensorineural hearing loss. Genes Chromosomes and Cancer, 2001, 31, 255-263.	1.5	70
34	Nearly all hereditary paragangliomas in The Netherlands are caused by two founder mutations in theSDHDgene. Genes Chromosomes and Cancer, 2001, 31, 274-281.	1.5	149
35	Adult neuronal ceroid lipofuscinosis with palmitoyl-protein thioesterase deficiency: First adult-onset patients of a childhood disease. Annals of Neurology, 2001, 50, 269-272.	2.8	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. European Journal of Human Genetics, 2001, 9, 121-129.	1.4	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.	0.7	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.	0.7	29
39	Visual disorders in children with brain lesions:. European Journal of Paediatric Neurology, 2001, 5, 115-119.	0.7	70
40	Mutations in SDHD, a Mitochondrial Complex II Gene, in Hereditary Paraganglioma. Science, 2000, 287, 848-851.	6.0	1,554
41	Repositioning the hereditary paraganglioma critical region on chromosome band 11q23. Human Genetics, 1999, 104, 219-225.	1.8	52
42	First-trimester diagnosis of infantile neuronal ceroid lipofuscinosis (INCL) using PPT enzyme assay andCLN1 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.	2.6	129
44	Genetic Heterogeneity of Neuronal Ceroid Lipofuscinosis in the Netherlands. Molecular Genetics and Metabolism, 1999, 66, 339-343.	0.5	8
45	A Murine Model for Juvenile NCL: Gene Targeting of MouseCln3. Molecular Genetics and Metabolism, 1999, 66, 309-313.	0.5	31
46	The Molecular Basis of GROD-Storing Neuronal Ceroid Lipofuscinoses in Scotland. Molecular Genetics and Metabolism, 1999, 66, 245-247.	0.5	10
47	Targeted Disruption of the Cln3 Gene Provides a Mouse Model for Batten Disease. Neurobiology of Disease, 1999, 6, 321-334.	2.1	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.	1.4	122
49	Genomic Structure and Complete Nucleotide Sequence of the Batten Disease Gene,CLN3. Genomics, 1997, 40, 346-350.	1.3	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.	1.3	159
51	Spectrum of Mutations in the Batten Disease Gene, CLN3. American Journal of Human Genetics, 1997, 61, 310-316.	2.6	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.	0.9	63
53	Mutations in the geneâ€“encoding SERCA1, the fastâ€“twitch skeletal muscle sarcoplasmic reticulum Ca2+ ATPase, are associated with Brody disease. Nature Genetics, 1996, 14, 191-194.	9.4	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. American Journal of Medical Genetics Part A, 1995, 57, 312-315.	2.4	16
56	Physical map of the region containing the gene for Batten disease (CLN3). American Journal of Medical Genetics Part A, 1995, 57, 316-319.	2.4	10
57	Isolation of genes from the Batten candidate region using exon amplification. American Journal of Medical Genetics Part A, 1995, 57, 320-323.	2.4	5
58	Carrier detection of Batten disease (Juvenile neuronal ceroid-lipofuscinosis). American Journal of Medical Genetics Part A, 1995, 57, 333-337.	2.4	13
59	Application of chromosome 16 markers in the differential diagnosis of neuronal ceroid-lipofuscinosis. American Journal of Medical Genetics Part A, 1995, 57, 338-343.	2.4	3
60	Replicase-Mediated Resistance to Alfalfa Mosaic Virus. Virology, 1995, 207, 467-474.	1.1	59
61	YAC and Cosmid Contigs Spanning the Batten Disease (CLN3) Region at 16p12.1â€“p11.2. Genomics, 1995, 29, 478-489.	1.3	8
62	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, 269-276.	1.1	27
63	Genetic Mapping of the Batten Disease Locus (CLN3) to the Interval D16S288-D16S383 by Analysis of Haplotypes and Allelic Association. Genomics, 1994, 22, 465-468.	1.3	33
64	Replication of an incomplete alfalfa mosaic virus genome in plants transformed with viral replicase genes. Virology, 1991, 181, 445-450.	1.1	102