Penelope E Bonnen

List of Publications by Year
in descending order

47 papers	6,354 citations	186265 28 h-index	214800 47 g-index
48 all docs	48 docs citations	48 times ranked	14219 citing authors

#	Article	IF	Citations
1	Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. Journal of Medical Genetics, 2022, 59, 878-887.	3.2	9
2	<scp>TAB2</scp> variants cause cardiovascular heart disease, connective tissue disorder, and developmental delay. Clinical Genetics, 2022, 101, 214-220.	2.0	7
3	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
4	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
5	LONP1 de novo dominant mutation causes mitochondrial encephalopathy with loss of LONP1 chaperone activity and excessive LONP1 proteolytic activity. Mitochondrion, 2020, 51, 68-78.	3.4	16
6	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	6.2	39
7	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	6.2	30
8	Incidence of PKAN determined by bioinformatic and population-based analysis of ~140,000 humans. Molecular Genetics and Metabolism, 2019, 128, 463-469.	1.1	20
9	From incomplete penetrance with normal telomere length to severe disease and telomere shortening in a family with monoallelic and biallelic PARN pathogenic variants. Human Mutation, 2019, 40, 2414-2429.	2.5	14
10	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. Neuron, 2019, 104, 665-679.e8.	8.1	43
11	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
12	Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. Scientific Reports, 2019, 9, 5108.	3.3	12
13	FBXL4-Related Mitochondrial DNA Depletion Syndrome 13 (MTDPS13): A Case Report With a Comprehensive Mutation Review. Frontiers in Genetics, 2019, 10, 39.	2.3	21
14	Metabolomics Profile in ABAT Deficiency Pre- and Post-treatment. JIMD Reports, 2018, 43, 13-17.	1.5	5
15	Clinical, biochemical, and genetic features of four patients with shortâ€chain enoylâ€CoA hydratase (ECHS1) deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1115-1127.	1.2	36
16	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	2.4	50
17	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
18	Loss-of-function mutations inISCA2disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. Human Mutation, 2018, 39, 537-549.	2.5	21

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19	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	6.2	40
20	<scp>OXA</scp> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, .	6.9	54
21	POLG2 deficiency causes adultâ€onset syndromic sensory neuropathy, ataxia and parkinsonism. Annals of Clinical and Translational Neurology, 2017, 4, 4-14.	3.7	13
22	Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924.	1.1	49
23	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	6.2	63
24	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American Journal of Human Genetics, 2017, 101, 833-843.	6.2	56
25	Pathogenic variants in HTRA2 cause an earlyâ€onset mitochondrial syndrome associated with 3â€methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2017, 40, 121-130.	3.6	23
26	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
27	Cerebrotendinous Xanthomatosis Presenting with Infantile Spasms and Intellectual Disability. JIMD Reports, 2016, 35, 1-5.	1.5	10
28	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
29	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	7.6	81
30	Identification of Variant-Specific Functions of PIK3CA by Rapid Phenotyping of Rare Mutations. Cancer Research, 2015, 75, 5341-5354.	0.9	130
31	Apparent underdiagnosis of Cerebrotendinous Xanthomatosis revealed by analysis of ~60,000 human exomes. Molecular Genetics and Metabolism, 2015, 116, 298-304.	1.1	79
32	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
33	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427.	16.2	119
34	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	3.6	45
35	Successful diagnosis of HIBCH deficiency from exome sequencing and positive retrospective analysis of newborn screening cards in two siblings presenting with Leigh's disease. Molecular Genetics and Metabolism, 2015, 115, 161-167.	1.1	32
36	Translational control of mGluR-dependent long-term depression and object-place learning by eIF2Î±. Nature Neuroscience, 2014, 17, 1073-1082.	14.8	159

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37	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
38	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. American Journal of Human Genetics, 2013, 93, 471-481.	6.2	137
39	Longitudinal study shows increasing obesity and hyperglycemia in micronesia. Obesity, 2013, 21, E421-7.	3.0	3
40	WDR35 mutation in siblings with Sensenbrenner syndrome: A ciliopathy with variable phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2917-2924.	1.2	40
41	European admixture on the Micronesian island of Kosrae: lessons from complete genetic information. European Journal of Human Genetics, 2010, 18, 309-316.	2.8	11
42	Integrating common and rare genetic variation in diverse human populations. Nature, 2010, 467, 52-58.	27.8	2,625
43	First Complete Genome Sequence of Two Staphylococcus epidermidis Bacteriophages. Journal of Bacteriology, 2007, 189, 2086-2100.	2.2	71
44	Evaluating potential for whole-genome studies in Kosrae, an isolated population in Micronesia. Nature Genetics, 2006, 38, 214-217.	21.4	61
45	Haplotype and Linkage Disequilibrium Architecture for Human Cancer-Associated Genes. Genome Research, 2002, 12, 1846-1853.	5.5	78
46	Haplotypes at ATM Identify Coding-Sequence Variation and Indicate a Region of Extensive Linkage Disequilibrium. American Journal of Human Genetics, 2000, 67, 1437-1451.	6.2	56
47	Autosomal dominant cerebellar ataxia (SCA6) associated with small polyglutamine expansions in the Î±1A-voltage-dependent calcium channel. Nature Genetics, 1997, 15, 62-69.	21.4	1,606