Angela Pyle

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9029355/publications.pdf

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50	2,850	186265	189892
papers	citations	28 h-index	50 g-index

52	52	52	5374
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. JAMA - Journal of the American Medical Association, 2014, 312, 68.	7.4	304
2	Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. Nature Cell Biology, 2018, 20, 144-151.	10.3	182
3	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
4	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	6.2	123
5	EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287.	12.8	120
6	Exome sequencing in undiagnosed inherited and sporadic ataxias. Brain, 2015, 138, 276-283.	7.6	120
7	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	6.2	118
8	Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50.	3.6	113
9	Defective i6A37 Modification of Mitochondrial and Cytosolic tRNAs Results from Pathogenic Mutations in TRIT1 and Its Substrate tRNA. PLoS Genetics, 2014, 10, e1004424.	3.5	112
10	Synaptotagmin 2 Mutations Cause an Autosomal-Dominant Form of Lambert-Eaton Myasthenic Syndrome and Nonprogressive Motor Neuropathy. American Journal of Human Genetics, 2014, 95, 332-339.	6.2	96
11	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	6.2	89
12	SPG7 mutations are a common cause of undiagnosed ataxia. Neurology, 2015, 84, 1174-1176.	1.1	87
13	Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59.	1.9	86
14	Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578.	7.6	85
15	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2015, 97, 319-328.	6.2	83
16	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.1	81
17	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. Genetics in Medicine, 2014, 16, 962-971.	2.4	64
18	Fall in circulating mononuclear cell mitochondrial DNA content in human sepsis. Intensive Care Medicine, 2010, 36, 956-962.	8.2	62

#	Article	IF	Citations
19	Extreme-Depth Re-sequencing of Mitochondrial DNA Finds No Evidence of Paternal Transmission in Humans. PLoS Genetics, 2015, 11, e1005040.	3.5	62
20	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	2.9	52
21	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	7.6	51
22	Defective mitochondrial protease LonP1 can cause classical mitochondrial disease. Human Molecular Genetics, 2018, 27, 1743-1753.	2.9	46
23	Exposure of Monocytic Cells to Lipopolysaccharide Induces Coordinated Endotoxin Tolerance, Mitochondrial Biogenesis, Mitophagy, and Antioxidant Defenses. Frontiers in Immunology, 2018, 9, 2217.	4.8	45
24	Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589.	6.9	45
25	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
26	Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106.	9.0	41
27	SCP2 mutations and neurodegeneration with brain iron accumulation. Neurology, 2015, 85, 1909-1911.	1.1	39
28	Cell-free mitochondrial DNA in progressive multiple sclerosis. Mitochondrion, 2019, 46, 307-312.	3.4	32
29	Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophyâ€“like disease. Genetics in Medicine, 2018, 20, 1224-1235.	2.4	31
30	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.6	27
31	Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6.	1.9	23
32	Adult Onset Leigh Syndrome in the Intensive Care Setting: A Novel Presentation of a C12orf65 Related Mitochondrial Disease. Journal of Neuromuscular Diseases, 2015, 2, 409-419.	2.6	22
33	Prominent Sensorimotor Neuropathy Due to SACS Mutations Revealed by Whole-Exome Sequencing. Archives of Neurology, 2012, 69, 1351-4.	4.5	21
34	Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. Scientific Reports, 2018, 8, 11682.	3.3	21
35	Behr's Syndrome is Typically Associated with Disturbed Mitochondrial Translation and Mutations in the C12orf65 Gene. Journal of Neuromuscular Diseases, 2014, 1, 55-63.	2.6	20
36	Effects of thyroid hormone on mitochondria and metabolism of human preimplantation embryos. Stem Cells, 2020, 38, 369-381.	3.2	20

#	Article	IF	Citations
37	Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. PLoS Genetics, 2022, 18, e1010068.	3.5	19
38	Phenotypic convergence of Menkes and Wilson disease. Neurology: Genetics, 2016, 2, e119.	1.9	18
39	Clinical heterogeneity of primary familial brain calcification due to a novel mutation in PDGFB. Neurology, 2015, 84, 1818-1820.	1.1	14
40	The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e391-e391.	7.6	13
41	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	1.9	11
42	Interactions between nuclear and mitochondrial SNPs and Parkinsonâ€™s disease risk. Mitochondrion, 2022, 63, 85-88.	3.4	6
43	A subcellular cookie cutter for spatial genomics in human tissue. Analytical and Bioanalytical Chemistry, 2022, 414, 5483-5492.	3.7	6
44	Mitochondrial DNA depletion induces innate immune dysfunction rescued by IFN-Î³. Journal of Allergy and Clinical Immunology, 2017, 140, 1461-1464.e8.	2.9	5
45	The Human Coronavirus Receptor <scp>ANPEP</scp> (<scp>CD13</scp>) Is Overexpressed in Parkinson's Disease. Movement Disorders, 2020, 35, 2134-2136.	3.9	4
46	Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062.	2.9	3
47	Reply: Evaluation of exome sequencing variation in undiagnosed ataxias. Brain, 2015, 138, e384-e384.	7.6	2
48	A Woman With Intellectual Disability, Amenorrhoea, Seizures, and Balance Problems. JAMA Neurology, 2016, 73, 1494.	9.0	1
49	Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202.	1.9	1
50	1353The Induction of Endotoxin Tolerance is Associated with the Activation of Mitochondrial Biogenesis in THP-1 Cells. Open Forum Infectious Diseases, 2014, 1, S354-S354.	0.9	0