# Angela Pyle

List of Publications by Year
in descending order

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

Version: 2024-02-01

| 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 | Pathogenic SLC25A26 variants impair SAH transport activity causing mitochondrial disease. Human Molecular Genetics, 2022, 31, 2049-2062. | 2.9 | 3 |
| 2 | Interactions between nuclear and mitochondrial SNPs and Parkinsonâ€™s disease risk. Mitochondrion, 2022, 63, 85-88. | 3.4 | 6 |
| 3 | A subcellular cookie cutter for spatial genomics in human tissue. Analytical and Bioanalytical Chemistry, 2022, 414, 5483-5492. | 3.7 | 6 |
| 4 | Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. PLoS Genetics, 2022, 18, e1010068. | 3.5 | 19 |
| 5 | Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50. | 3.6 | 113 |
| 6 | Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308. | 3.6 | 43 |
| 7 | Effects of thyroid hormone on mitochondria and metabolism of human preimplantation embryos. Stem Cells, 2020, 38, 369-381. | 3.2 | 20 |
| 8 | 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 |
| 9 | Metabolic effects of bezafibrate in mitochondrial disease. EMBO Molecular Medicine, 2020, 12, e11589. | 6.9 | 45 |
| 10 | Cell-free mitochondrial DNA in progressive multiple sclerosis. Mitochondrion, 2019, 46, 307-312. | 3.4 | 32 |
| 11 | Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, . | 12.6 | 178 |
| 12 | NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58. | 7.6 | 51 |
| 13 | Defective mitochondrial protease LonP1 can cause classical mitochondrial disease. Human Molecular Genetics, 2018, 27, 1743-1753. | 2.9 | 46 |
| 14 | 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 |
| 15 | A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195. | 2.9 | 52 |
| 16 | Segregation of mitochondrial DNA heteroplasmy through a developmental genetic bottleneck in human embryos. Nature Cell Biology, 2018, 20, 144-151. | 10.3 | 182 |
| 17 | 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 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234. | 1.1 | 81 |
| 20 | Hypomorphic mutations in POLR3A are a frequent cause of sporadic and recessive spastic ataxia. Brain, 2017, 140, 1561-1578. | 7.6 | 85 |
| 21 | 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 |
| 22 | De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187. | 1.9 | 11 |
| 23 | Opening One's Eyes to Mosaicism in Progressive External Ophthalmoplegia. Neurology: Genetics, 2017, 3, e202. | 1.9 | 1 |
| 24 | Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood. Neurology: Genetics, 2016, 2, e59. | 1.9 | 86 |
| 25 | Phenotypic convergence of Menkes and Wilson disease. Neurology: Genetics, 2016, 2, e119. | 1.9 | 18 |
| 26 | A Woman With Intellectual Disability, Amenorrhoea, Seizures, and Balance Problems. JAMA Neurology, 2016, 73, 1494. | 9.0 | 1 |
| 27 | 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 |
| 28 | 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 |
| 29 | 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 |
| 30 | Clonal Expansion of Secondary Mitochondrial DNA Deletions Associated With Spinocerebellar Ataxia Type 28. JAMA Neurology, 2015, 72, 106. | 9.0 | 41 |
| 31 | Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6. | 1.9 | 23 |
| 32 | Reply: Evaluation of exome sequencing variation in undiagnosed ataxias. Brain, 2015, 138, e384-e384. | 7.6 | 2 |
| 33 | 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 |
| 34 | Extreme-Depth Re-sequencing of Mitochondrial DNA Finds No Evidence of Paternal Transmission in Humans. PLoS Genetics, 2015, 11, e1005040. | 3.5 | 62 |
| 35 | SPG7 mutations are a common cause of undiagnosed ataxia. Neurology, 2015, 84, 1174-1176. | 1.1 | 87 |
| 36 | Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521. | 0.6 | 27 |

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|---|---|---|---|
| 37 | Clinical heterogeneity of primary familial brain calcification due to a novel mutation in PDGFB. Neurology, 2015, 84, 1818-1820. | 1.1 | 14 |
| 38 | The p.Ser107Leu inBICD2is a mutation â€˜hot spotâ€™ causing distal spinal muscular atrophy. Brain, 2015, 138, e391-e391. | 7.6 | 13 |
| 39 | SCP2 mutations and neurodegeneration with brain iron accumulation. Neurology, 2015, 85, 1909-1911. | 1.1 | 39 |
| 40 | Exome sequencing in undiagnosed inherited and sporadic ataxias. Brain, 2015, 138, 276-283. | 7.6 | 120 |
| 41 | 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 |
| 42 | 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 |
| 43 | 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 |
| 44 | 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 |
| 45 | EXOSC8 mutations alter mRNA metabolism and cause hypomyelination with spinal muscular atrophy and cerebellar hypoplasia. Nature Communications, 2014, 5, 4287. | 12.8 | 120 |
| 46 | 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 |
| 47 | 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 |
| 48 | 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 |
| 49 | Prominent Sensorimotor Neuropathy Due to SACS Mutations Revealed by Whole-Exome Sequencing. Archives of Neurology, 2012, 69, 1351-4. | 4.5 | 21 |
| 50 | Fall in circulating mononuclear cell mitochondrial DNA content in human sepsis. Intensive Care Medicine, 2010, 36, 956-962. | 8.2 | 62 |