

Charlotte L Alston

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/8184231/publications.pdf>

Version: 2024-02-01

92
papers

5,712
citations

81743

39
h-index

88477

70
g-index

98
all docs

98
docs citations

98
times ranked

7326
citing authors

#	ARTICLE	IF	CITATIONS
1	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. <i>Annals of Neurology</i> , 2015, 77, 753-759.	2.8	706
2	The genetics and pathology of mitochondrial disease. <i>Journal of Pathology</i> , 2017, 241, 236-250.	2.1	329
3	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. <i>JAMA - Journal of the American Medical Association</i> , 2014, 312, 68.	3.8	304
4	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. <i>Molecular Cell</i> , 2016, 63, 621-632.	4.5	241
5	Recent Advances in Mitochondrial Disease. <i>Annual Review of Genomics and Human Genetics</i> , 2017, 18, 257-275.	2.5	217
6	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A<gt;G mitochondrial disease. <i>EMBO Molecular Medicine</i> , 2018, 10, .	3.3	199
7	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. <i>Human Mutation</i> , 2011, 32, 1319-1325.	1.1	159
8	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. <i>Brain</i> , 2014, 137, 1323-1336.	3.7	151
9	SANDO syndrome in a cohort of 107 patients with CPEO and mitochondrial DNA deletions. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 630-634.	0.9	113
10	Recent advances in understanding the molecular genetic basis of mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 36-50.	1.7	113
11	Recessive germline <i>SDHA</i> and <i>SDHB</i> mutations causing leukodystrophy and isolated mitochondrial complex II deficiency. <i>Journal of Medical Genetics</i> , 2012, 49, 569-577.	1.5	100
12	URINE HETEROPLASMY IS THE BEST PREDICTOR OF CLINICAL OUTCOME IN THE m.3243A<gt;G mtDNA MUTATION. <i>Neurology</i> , 2009, 72, 568-569.	1.5	95
13	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. <i>American Journal of Human Genetics</i> , 2016, 99, 860-876.	2.6	93
14	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 492-509.	1.7	90
15	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2016, 98, 993-1000.	2.6	89
16	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. <i>Genome Medicine</i> , 2022, 14, 38.	3.6	85
17	TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2015, 97, 319-328.	2.6	83
18	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. <i>Frontiers in Genetics</i> , 2015, 06, 123.	1.1	81

#	ARTICLE	IF	CITATIONS
19	<i>LRPPRC</i> mutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. <i>Brain</i> , 2015, 138, 3503-3519.	3.7	81
20	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. <i>Brain</i> , 2012, 135, 3392-3403.	3.7	70
21	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. <i>Brain</i> , 2010, 133, 2952-2963.	3.7	69
22	Clinical, Genetic, and Radiological Features of Extrapyrarnidal Movement Disorders in Mitochondrial Disease. <i>JAMA Neurology</i> , 2016, 73, 668.	4.5	69
23	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. <i>Genetics in Medicine</i> , 2014, 16, 962-971.	1.1	64
24	Pathogenic Mitochondrial tRNA Point Mutations: Nine Novel Mutations Affirm Their Importance as a Cause of Mitochondrial Disease. <i>Human Mutation</i> , 2013, 34, 1260-1268.	1.1	62
25	Mutation of the human mitochondrial phenylalanine-tRNA synthetase causes infantile-onset epilepsy and cytochrome c oxidase deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2014, 1842, 56-64.	1.8	61
26	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 120.	1.2	61
27	<i>RRM2B</i> mutations are frequent in familial PEO with multiple mtDNA deletions. <i>Neurology</i> , 2011, 76, 2032-2034.	1.5	59
28	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. <i>American Journal of Human Genetics</i> , 2017, 101, 525-538.	2.6	58
29	A national perspective on prenatal testing for mitochondrial disease. <i>European Journal of Human Genetics</i> , 2014, 22, 1255-1259.	1.4	57
30	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 217-227.	2.6	57
31	De novo mtDNA point mutations are common and have a low recurrence risk. <i>Journal of Medical Genetics</i> , 2017, 54, 73-83.	1.5	54
32	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. <i>Human Molecular Genetics</i> , 2016, 25, 1031-1041.	1.4	53
33	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. <i>European Heart Journal</i> , 2016, 37, 2552-2559.	1.0	53
34	A recessive homozygous p.Asp92Gly SDHD mutation causes prenatal cardiomyopathy and a severe mitochondrial complex II deficiency. <i>Human Genetics</i> , 2015, 134, 869-879.	1.8	49
35	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. <i>American Journal of Human Genetics</i> , 2016, 99, 674-682.	2.6	48
36	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. <i>Genome Biology</i> , 2020, 21, 248.	3.8	48

#	ARTICLE	IF	CITATIONS
37	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. <i>EBioMedicine</i> , 2018, 30, 86-93.	2.7	47
38	Defining cardiac adaptations and safety of endurance training in patients with m.3243A>G-related mitochondrial disease. <i>International Journal of Cardiology</i> , 2013, 168, 3599-3608.	0.8	43
39	Clinical presentation and proteomic signature of patients with <i>TANGO2</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 297-308.	1.7	43
40	Adult-onset spinocerebellar ataxia syndromes due to<i>MTATP6</i>mutations. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 883-886.	0.9	42
41	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. <i>Kidney International</i> , 2015, 87, 610-622.	2.6	41
42	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in <i>YARS2</i>-Related Mitochondrial Myopathy. <i>JAMA Neurology</i> , 2017, 74, 686.	4.5	41
43	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2018, 103, 592-601.	2.6	41
44	Pseudoâ€œobstruction, stroke, and mitochondrial dysfunction: A lethal combination. <i>Annals of Neurology</i> , 2016, 80, 686-692.	2.8	40
45	Cytochrome c oxidase-intermediate fibres: Importance in understanding the pathogenesis and treatment of mitochondrial myopathy. <i>Neuromuscular Disorders</i> , 2012, 22, 690-698.	0.3	39
46	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. <i>American Journal of Human Genetics</i> , 2020, 106, 92-101.	2.6	39
47	Clinical, biochemical, and genetic features of four patients with shortâ€œchain enoylâ€œCoA hydratase (ECHS1) deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1115-1127.	0.7	36
48	The clinical spectrum of the m.10191T>C mutation in complex Iâ€œdeficient Leigh syndrome. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 500-506.	1.1	35
49	The clinical, biochemical and genetic features associated with<i>RMND1</i>-related mitochondrial disease. <i>Journal of Medical Genetics</i> , 2016, 53, 768-775.	1.5	35
50	A novel mitochondrial MTND5 frameshift mutation causing isolated complex I deficiency, renal failure and myopathy. <i>Neuromuscular Disorders</i> , 2010, 20, 131-135.	0.3	34
51	A novel de novo STXBP1 mutation is associated with mitochondrial complex I deficiency and late-onset juvenile-onset parkinsonism. <i>Neurogenetics</i> , 2015, 16, 65-67.	0.7	34
52	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. <i>American Journal of Human Genetics</i> , 2018, 103, 100-114.	2.6	34
53	Pathogenic variants in <i>MTâ€œATP6</i>: A United Kingdomâ€œbased mitochondrial disease cohort study. <i>Annals of Neurology</i> , 2019, 86, 310-315.	2.8	33
54	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€œomic pipelines. <i>Journal of Pathology</i> , 2021, 254, 430-442.	2.1	33

#	ARTICLE	IF	CITATIONS
55	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. <i>European Journal of Human Genetics</i> , 2015, 23, 935-939.	1.4	32
56	Mitochondrial dysfunction in myofibrillar myopathy. <i>Neuromuscular Disorders</i> , 2016, 26, 691-701.	0.3	32
57	Neuropathologic Characterization of Pontocerebellar Hypoplasia Type 6 Associated With Cardiomyopathy and Hydrops Fetalis and Severe Multisystem Respiratory Chain Deficiency due to Novel <i>RARS2</i> Mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , 2015, 74, 688-703.	0.9	31
58	A recurrent mitochondrial p.Trp22ArgNDUFB3 variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. <i>Journal of Medical Genetics</i> , 2016, 53, 634-641.	1.5	31
59	Mutations in <i>ELAC2</i> associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3' end processing. <i>Human Mutation</i> , 2019, 40, 1731-1748.	1.1	31
60	Dysferlin mutations and mitochondrial dysfunction. <i>Neuromuscular Disorders</i> , 2016, 26, 782-788.	0.3	28
61	Distal weakness with respiratory insufficiency caused by the m.8344A>G MERRF mutation. <i>Neuromuscular Disorders</i> , 2014, 24, 533-536.	0.3	26
62	The investigation and diagnosis of pathogenic mitochondrial DNA mutations in human urothelial cells. <i>Biochemical and Biophysical Research Communications</i> , 2010, 393, 740-745.	1.0	25
63	Late-onset respiratory failure due to <i>TK2</i> mutations causing multiple mtDNA deletions. <i>Neurology</i> , 2013, 81, 2051-2053.	1.5	23
64	Adult Onset Leigh Syndrome in the Intensive Care Setting: A Novel Presentation of a C12orf65 Related Mitochondrial Disease. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 409-419.	1.1	22
65	Three families with <i>de novo</i> m.3243A>G mutation. <i>BBA Clinical</i> , 2016, 6, 19-24.	4.1	22
66	The genetic basis of isolated mitochondrial complex II deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 53-65.	0.5	22
67	Loss-of-function mutations in <i>ISCA2</i> disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. <i>Human Mutation</i> , 2018, 39, 537-549.	1.1	21
68	Maternally inherited mitochondrial DNA disease in consanguineous families. <i>European Journal of Human Genetics</i> , 2011, 19, 1226-1229.	1.4	20
69	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. <i>Scientific Reports</i> , 2017, 7, 15676.	1.6	20
70	A leaky splicing mutation in <i>NFU1</i> is associated with a particular biochemical phenotype. Consequences for the diagnosis. <i>Mitochondrion</i> , 2016, 26, 72-80.	1.6	19
71	Molecular genetic investigations identify new clinical phenotypes associated with <i>BCS1L</i> -related mitochondrial disease. <i>Human Molecular Genetics</i> , 2019, 28, 3766-3776.	1.4	19
72	Decreased male reproductive success in association with mitochondrial dysfunction. <i>European Journal of Human Genetics</i> , 2017, 25, 1162-1164.	1.4	18

#	ARTICLE	IF	CITATIONS
73	Early-Onset Cataracts, Spastic Paraparesis, and Ataxia Caused by a Novel Mitochondrial tRNAGlu(MT-TE) Gene Mutation Causing Severe Complex I Deficiency: A Clinical, Molecular, and Neuropathologic Study. <i>Journal of Neuropathology and Experimental Neurology</i> , 2013, 72, 164-175.	0.9	17
74	Leigh syndrome caused by mutations in <i>MTFMT</i> is associated with a better prognosis. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 515-524.	1.7	17
75	Biallelic pathogenic variants in <i>NDUFC2</i> cause early-onset Leigh syndrome and stalled biogenesis of complex I. <i>EMBO Molecular Medicine</i> , 2020, 12, e12619.	3.3	17
76	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. <i>Annals of Neurology</i> , 2022, 91, 117-130.	2.8	17
77	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. <i>Nucleic Acids Research</i> , 2019, 47, 7430-7443.	6.5	16
78	Mitochondrial respiratory chain disease in children undergoing cardiac transplantation: A prospective study. <i>International Journal of Cardiology</i> , 2012, 155, 305-306.	0.8	14
79	Near-Identical Segregation of mtDNA Heteroplasmy in Blood, Muscle, Urinary Epithelium, and Hair Follicles in Twins With Optic Atrophy, Ptosis, and Intractable Epilepsy. <i>JAMA Neurology</i> , 2013, 70, 1552-5.	4.5	14
80	The frequency of the m.1555A>G (MTRNR1) variant in UK patients with suspected mitochondrial deafness. <i>Hearing, Balance and Communication</i> , 2016, 14, 101-102.	0.1	14
81	A novel mitochondrial tRNAGlu (MTTE) gene mutation causing chronic progressive external ophthalmoplegia at low levels of heteroplasmy in muscle. <i>Journal of the Neurological Sciences</i> , 2010, 298, 140-144.	0.3	12
82	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. <i>Neurology: Genetics</i> , 2016, 2, e113.	0.9	12
83	De novo <i>CTBP1</i> variant is associated with decreased mitochondrial respiratory chain activities. <i>Neurology: Genetics</i> , 2017, 3, e187.	0.9	11
84	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. <i>Neurogenetics</i> , 2017, 18, 227-235.	0.7	10
85	Early-onset coenzyme Q10 deficiency associated with ataxia and respiratory chain dysfunction due to novel pathogenic <i>COQ8A</i> variants, including a large intragenic deletion. <i>JIMD Reports</i> , 2020, 54, 45-53.	0.7	8
86	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. <i>Genetics in Medicine</i> , 2021, 23, 2415-2425.	1.1	8
87	The pathogenic m.3243A>T mitochondrial DNA mutation is associated with a variable neurological phenotype. <i>Neuromuscular Disorders</i> , 2010, 20, 403-406.	0.3	7
88	The m.3291T>C mt-tRNA ^{Leu} (UUR) mutation is definitely pathogenic and causes multisystem mitochondrial disease. <i>Journal of the Neurological Sciences</i> , 2013, 325, 165-169.	0.3	7
89	The m.15043G>A MT-CYB variant is not a pathogenic mtDNA variant. <i>Journal of the Neurological Sciences</i> , 2020, 417, 116950.	0.3	1
90	A GENETIC WEAKNESS—PHOENICIAN LEGACY OR CELTIC HERITAGE?. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, e4.89-e4.	0.9	0

#	ARTICLE	IF	CITATIONS
91	Cystic Leukoencephalopathy due to NDUFV1 mutationâ€”A Report of the Phenotype and Its Rare Co-occurrence with Primary Hyperoxaluria. <i>Journal of Pediatric Neurology</i> , 2016, 14, 126-132.	0.0	0
92	Variants in PTC1 cause combined respiratory chain deficiency and mitoribosomal instability associated with infantile cardiomyopathy. <i>Neuromuscular Disorders</i> , 2018, 28, S32.	0.3	0