Charlotte L Alston

List of Publications by Year
in descending order

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92	5,712	81900	88630
papers	citations	39 h-index	70 g-index

98	98	98	7326
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	5.3	706
2	The genetics and pathology of mitochondrial disease. Journal of Pathology, 2017, 241, 236-250.	4.5	329
3	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
4	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	9.7	241
5	Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275.	6.2	217
6	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	6.9	199
7	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	2.5	159
8	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	7.6	151
9	SANDO syndrome in a cohort of 107 patients with CPEO and mitochondrial DNA deletions. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 630-634.	1.9	113
10	Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50.	3.6	113
11	Recessive germlineSDHAandSDHBmutations causing leukodystrophy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2012, 49, 569-577.	3.2	100
12	URINE HETEROPLASMY IS THE BEST PREDICTOR OF CLINICAL OUTCOME IN THE m.3243A>G mtDNA MUTATION. Neurology, 2009, 72, 568-569.	1.1	95
13	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
14	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	3.7	90
15	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
16	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
17	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
18	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	2.3	81

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19	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	7.6	81
20	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	7.6	70
21	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. Brain, 2010, 133, 2952-2963.	7.6	69
22	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	9.0	69
23	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
24	Pathogenic Mitochondrial t <scp>RNA</scp> Point Mutations: Nine Novel Mutations Affirm Their Importance as a Cause of Mitochondrial Disease. Human Mutation, 2013, 34, 1260-1268.	2.5	62
25	Mutation of the human mitochondrial phenylalanine-tRNA synthetase causes infantile-onset epilepsy and cytochrome c oxidase deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 56-64.	3.8	61
26	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
27	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Neurology, 2011, 76, 2032-2034.	1.1	59
28	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	6.2	58
29	A national perspective on prenatal testing for mitochondrial disease. European Journal of Human Genetics, 2014, 22, 1255-1259.	2.8	57
30	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	6.2	57
31	De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83.	3.2	54
32	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	2.9	53
33	Sudden adult death syndrome in m.3243A>G-related mitochondrial disease: an unrecognized clinical entity in young, asymptomatic adults. European Heart Journal, 2016, 37, 2552-2559.	2.2	53
34	A recessive homozygous p.Asp92Gly SDHD mutation causes prenatal cardiomyopathy and a severe mitochondrial complex II deficiency. Human Genetics, 2015, 134, 869-879.	3.8	49
35	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
36	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	8.8	48

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37	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	6.1	47
38	Defining cardiac adaptations and safety of endurance training in patients with m.3243A>G-related mitochondrial disease. International Journal of Cardiology, 2013, 168, 3599-3608.	1.7	43
39	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
40	Adult-onset spinocerebellar ataxia syndromes due toMTATP6mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 883-886.	1.9	42
41	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	5.2	41
42	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	9.0	41
43	Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. American Journal of Human Genetics, 2018, 103, 592-601.	6.2	41
44	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	5.3	40
45	Cytochrome c oxidase-intermediate fibres: Importance in understanding the pathogenesis and treatment of mitochondrial myopathy. Neuromuscular Disorders, 2012, 22, 690-698.	0.6	39
46	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
47	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
48	The clinical spectrum of the m.10191T>C mutation in complex Iâ€deficient Leigh syndrome. Developmental Medicine and Child Neurology, 2012, 54, 500-506.	2.1	35
49	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	3.2	35
50	A novel mitochondrial MTND5 frameshift mutation causing isolated complex I deficiency, renal failure and myopathy. Neuromuscular Disorders, 2010, 20, 131-135.	0.6	34
51	A novel de novo STXBP1 mutation is associated with mitochondrial complex I deficiency and late-onset juvenile-onset parkinsonism. Neurogenetics, 2015, 16, 65-67.	1.4	34
52	Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. American Journal of Human Genetics, 2018, 103, 100-114.	6.2	34
53	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	5.3	33
54	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442.	4.5	33

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55	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. European Journal of Human Genetics, 2015, 23, 935-939.	2.8	32
56	Mitochondrial dysfunction in myofibrillar myopathy. Neuromuscular Disorders, 2016, 26, 691-701.	0.6	32
57	Neuropathologic Characterization of Pontocerebellar Hypoplasia Type 6 Associated With Cardiomyopathy and Hydrops Fetalis and Severe Multisystem Respiratory Chain Deficiency due to NovelRARS2Mutations. Journal of Neuropathology and Experimental Neurology, 2015, 74, 688-703.	1.7	31
58	A recurrent mitochondrial p.Trp22ArgNDUFB3variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. Journal of Medical Genetics, 2016, 53, 634-641.	3.2	31
59	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
60	Dysferlin mutations and mitochondrial dysfunction. Neuromuscular Disorders, 2016, 26, 782-788.	0.6	28
61	Distal weakness with respiratory insufficiency caused by the m.8344A>G â€œMERRFâ€•mutation. Neuromuscular Disorders, 2014, 24, 533-536.	0.6	26
62	The investigation and diagnosis of pathogenic mitochondrial DNA mutations in human urothelial cells. Biochemical and Biophysical Research Communications, 2010, 393, 740-745.	2.1	25
63	Late-onset respiratory failure due to TK2 mutations causing multiple mtDNA deletions. Neurology, 2013, 81, 2051-2053.	1.1	23
64	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
65	Three families with â€˜de novoâ€™ m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24.	4.1	22
66	The genetic basis of isolated mitochondrial complex II deficiency. Molecular Genetics and Metabolism, 2020, 131, 53-65.	1.1	22
67	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
68	Maternally inherited mitochondrial DNA disease in consanguineous families. European Journal of Human Genetics, 2011, 19, 1226-1229.	2.8	20
69	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. Scientific Reports, 2017, 7, 15676.	3.3	20
70	A leaky splicing mutation in NFU1 is associated with a particular biochemical phenotype. Consequences for the diagnosis. Mitochondrion, 2016, 26, 72-80.	3.4	19
71	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	2.9	19
72	Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164.	2.8	18

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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. Journal of Neuropathology and Experimental Neurology, 2013, 72, 164-175.	1.7	17
74	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	3.7	17
75	Biâ€allelic pathogenic variants in NDUFC2 cause earlyâ€onset Leigh syndrome and stalled biogenesis of complex I. EMBO Molecular Medicine, 2020, 12, e12619.	6.9	17
76	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130.	5.3	17
77	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. Nucleic Acids Research, 2019, 47, 7430-7443.	14.5	16
78	Mitochondrial respiratory chain disease in children undergoing cardiac transplantation: A prospective study. International Journal of Cardiology, 2012, 155, 305-306.	1.7	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. JAMA Neurology, 2013, 70, 1552-5.	9.0	14
80	The frequency of the m.1555Aâ€‰>â€‰G (MTRNR1) variant in UK patients with suspected mitochondrial deafness. Hearing, Balance and Communication, 2016, 14, 101-102.	0.4	14
81	A novel mitochondrial tRNAGlu (MTTE) gene mutation causing chronic progressive external ophthalmoplegia at low levels of heteroplasmy in muscle. Journal of the Neurological Sciences, 2010, 298, 140-144.	0.6	12
82	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. Neurology: Genetics, 2016, 2, e113.	1.9	12
83	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	1.9	11
84	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	1.4	10
85	Earlyâ€onset coenzyme Q10 deficiency associated with ataxia and respiratory chain dysfunction due to novel pathogenic COQ8A variants, including a large intragenic deletion. JIMD Reports, 2020, 54, 45-53.	1.5	8
86	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	2.4	8
87	The pathogenic m.3243A>T mitochondrial DNA mutation is associated with a variable neurological phenotype. Neuromuscular Disorders, 2010, 20, 403-406.	0.6	7
88	The m.3291T>C mt-tRNALeu(UUR) mutation is definitely pathogenic and causes multisystem mitochondrial disease. Journal of the Neurological Sciences, 2013, 325, 165-169.	0.6	7
89	The m.15043GÂ>ÂA MT-CYB variant is not a pathogenic mtDNA variant. Journal of the Neurological Sciences, 2020, 417, 116950.	0.6	1
90	A GENETIC WEAKNESSâ€”PHOENICIAN LEGACY OR CELTIC HERITAGE?. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.89-e4.	1.9	0

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91	Cystic Leukoencephalopathy due to NDUFV1 mutationâ€"A Report of the Phenotype and Its Rare Co-occurrence with Primary Hyperoxaluria. Journal of Pediatric Neurology, 2016, 14, 126-132.	0.2	0
92	Variants in PTCD1 cause combined respiratory chain deficiency and mitoribosomal instability associated with infantile cardiomyopathy. Neuromuscular Disorders, 2018, 28, S32.	0.6	0