Charlotte L Alston

List of Publications by Year
in descending order

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92	5,712	81743	88477
papers	citations	39 h-index	70 g-index

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

#	Article	IF	Citations
1	Natural History of Leigh Syndrome: A Study of Disease Burden and Progression. Annals of Neurology, 2022, 91, 117-130.	2.8	17
2	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	3.6	85
3	The genetics of mitochondrial disease: dissecting mitochondrial pathology using multiâ€omic pipelines. Journal of Pathology, 2021, 254, 430-442.	2.1	33
4	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	1.1	8
5	Recent advances in understanding the molecular genetic basis of mitochondrial disease. Journal of Inherited Metabolic Disease, 2020, 43, 36-50.	1.7	113
6	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	1.7	43
7	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	2.6	39
8	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.	0.7	8
9	The genetic basis of isolated mitochondrial complex II deficiency. Molecular Genetics and Metabolism, 2020, 131, 53-65.	0.5	22
10	Ultrasensitive deletion detection links mitochondrial DNA replication, disease, and aging. Genome Biology, 2020, 21, 248.	3.8	48
11	The m.15043GÂ>ÂA MT-CYB variant is not a pathogenic mtDNA variant. Journal of the Neurological Sciences, 2020, 417, 116950.	0.3	1
12	Biâ€allelic pathogenic variants in NDUFC2 cause earlyâ€onset Leigh syndrome and stalled biogenesis of complex I. EMBO Molecular Medicine, 2020, 12, e12619.	3.3	17
13	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	1.4	19
14	Understanding mitochondrial DNA maintenance disorders at the single muscle fibre level. Nucleic Acids Research, 2019, 47, 7430-7443.	6.5	16
15	Pathogenic variants in MTâ€ATP6: A United Kingdomâ€“based mitochondrial disease cohort study. Annals of Neurology, 2019, 86, 310-315.	2.8	33
16	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
17	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.	1.7	17
18	MT-ND5 Mutation Exhibits Highly Variable Neurological Manifestations at Low Mutant Load. EBioMedicine, 2018, 30, 86-93.	2.7	47

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19	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.	0.7	36
20	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.	1.1	21
21	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.	2.6	41
22	Variants in PTCD1 cause combined respiratory chain deficiency and mitoribosomal instability associated with infantile cardiomyopathy. Neuromuscular Disorders, 2018, 28, S32.	0.3	0
23	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
24	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.	2.6	34
25	mt <scp>DNA</scp> heteroplasmy level and copy number indicate disease burden in m.3243A>G mitochondrial disease. EMBO Molecular Medicine, 2018, 10, .	3.3	199
26	Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurology, 2017, 74, 686.	4.5	41
27	Recent Advances in Mitochondrial Disease. Annual Review of Genomics and Human Genetics, 2017, 18, 257-275.	2.5	217
28	De novo mtDNA point mutations are common and have a low recurrence risk. Journal of Medical Genetics, 2017, 54, 73-83.	1.5	54
29	De novo CTBP1 variant is associated with decreased mitochondrial respiratory chain activities. Neurology: Genetics, 2017, 3, e187.	0.9	11
30	Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics, 2017, 18, 227-235.	0.7	10
31	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.	2.6	58
32	Decreased male reproductive success in association with mitochondrial dysfunction. European Journal of Human Genetics, 2017, 25, 1162-1164.	1.4	18
33	Using a quantitative quadruple immunofluorescent assay to diagnose isolated mitochondrial Complex I deficiency. Scientific Reports, 2017, 7, 15676.	1.6	20
34	The genetics and pathology of mitochondrial disease. Journal of Pathology, 2017, 241, 236-250.	2.1	329
35	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	2.6	57
36	Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions. Neurology: Genetics, 2016, 2, e113.	0.9	12

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37	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	1.5	35
38	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.	0.1	14
39	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.	0.0	0
40	A leaky splicing mutation in NFU1 is associated with a particular biochemical phenotype. Consequences for the diagnosis. Mitochondrion, 2016, 26, 72-80.	1.6	19
41	Three families with â€˜de novoâ€™ m.3243A>G mutation. BBA Clinical, 2016, 6, 19-24.	4.1	22
42	Clinical, Genetic, and Radiological Features of Extrapyramidal Movement Disorders in Mitochondrial Disease. JAMA Neurology, 2016, 73, 668.	4.5	69
43	Dysferlin mutations and mitochondrial dysfunction. Neuromuscular Disorders, 2016, 26, 782-788.	0.3	28
44	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.	2.6	93
45	Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Molecular Cell, 2016, 63, 621-632.	4.5	241
46	Mitochondrial dysfunction in myofibrillar myopathy. Neuromuscular Disorders, 2016, 26, 691-701.	0.3	32
47	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	2.6	48
48	Pseudoâ€obstruction, stroke, and mitochondrial dysfunction: A lethal combination. Annals of Neurology, 2016, 80, 686-692.	2.8	40
49	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.	1.5	31
50	Recessive Mutations in TRMT10C Cause Defects in Mitochondrial RNA Processing and Multiple Respiratory Chain Deficiencies. American Journal of Human Genetics, 2016, 98, 993-1000.	2.6	89
51	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	1.4	53
52	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.	1.0	53
53	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	1.7	90
54	Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Frontiers in Genetics, 2015, 06, 123.	1.1	81

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55	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.	1.1	22
56	A recessive homozygous p.Asp92Gly SDHD mutation causes prenatal cardiomyopathy and a severe mitochondrial complex II deficiency. Human Genetics, 2015, 134, 869-879.	1.8	49
57	Prevalence of nuclear and mitochondrial <scp>DNA</scp> mutations related to adult mitochondrial disease. Annals of Neurology, 2015, 77, 753-759.	2.8	706
58	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	3.7	81
59	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.	0.9	31
60	A novel de novo STXBP1 mutation is associated with mitochondrial complex I deficiency and late-onset juvenile-onset parkinsonism. Neurogenetics, 2015, 16, 65-67.	0.7	34
61	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.	2.6	83
62	A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. European Journal of Human Genetics, 2015, 23, 935-939.	1.4	32
63	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	2.6	41
64	SANDO syndrome in a cohort of 107 patients with CPEO and mitochondrial DNA deletions. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 630-634.	0.9	113
65	Accurate mitochondrial DNA sequencing using off-target reads provides a single test to identify pathogenic point mutations. Genetics in Medicine, 2014, 16, 962-971.	1.1	64
66	A national perspective on prenatal testing for mitochondrial disease. European Journal of Human Genetics, 2014, 22, 1255-1259.	1.4	57
67	A GENETIC WEAKNESSâ€"PHOENICIAN LEGACY OR CELTIC HERITAGE?. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, e4.89-e4.	0.9	0
68	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.	3.8	304
69	Distal weakness with respiratory insufficiency caused by the m.8344A>G â€œMERRFâ€•mutation. Neuromuscular Disorders, 2014, 24, 533-536.	0.3	26
70	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.	1.8	61
71	Mutations in the SPG7 gene cause chronic progressive external ophthalmoplegia through disordered mitochondrial DNA maintenance. Brain, 2014, 137, 1323-1336.	3.7	151
72	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.	0.3	7

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73	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.	1.1	62
74	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.	0.8	43
75	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.	4.5	14
76	Late-onset respiratory failure due to TK2 mutations causing multiple mtDNA deletions. Neurology, 2013, 81, 2051-2053.	1.5	23
77	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.	0.9	17
78	Adult-onset spinocerebellar ataxia syndromes due toMTATP6mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 883-886.	0.9	42
79	Adults with RRM2B-related mitochondrial disease have distinct clinical and molecular characteristics. Brain, 2012, 135, 3392-3403.	3.7	70
80	Recessive germlineSDHAandSDHBmutations causing leukodystrophy and isolated mitochondrial complex II deficiency. Journal of Medical Genetics, 2012, 49, 569-577.	1.5	100
81	Cytochrome c oxidase-intermediate fibres: Importance in understanding the pathogenesis and treatment of mitochondrial myopathy. Neuromuscular Disorders, 2012, 22, 690-698.	0.3	39
82	The clinical spectrum of the m.10191T>C mutation in complex Iâ€deficient Leigh syndrome. Developmental Medicine and Child Neurology, 2012, 54, 500-506.	1.1	35
83	Mitochondrial respiratory chain disease in children undergoing cardiac transplantation: A prospective study. International Journal of Cardiology, 2012, 155, 305-306.	0.8	14
84	A comparative analysis approach to determining the pathogenicity of mitochondrial tRNA mutations. Human Mutation, 2011, 32, 1319-1325.	1.1	159
85	Maternally inherited mitochondrial DNA disease in consanguineous families. European Journal of Human Genetics, 2011, 19, 1226-1229.	1.4	20
86	RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Neurology, 2011, 76, 2032-2034.	1.5	59
87	The p.M292T NDUFS2 mutation causes complex I-deficient Leigh syndrome in multiple families. Brain, 2010, 133, 2952-2963.	3.7	69
88	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.	0.3	12
89	The investigation and diagnosis of pathogenic mitochondrial DNA mutations in human urothelial cells. Biochemical and Biophysical Research Communications, 2010, 393, 740-745.	1.0	25
90	A novel mitochondrial MTND5 frameshift mutation causing isolated complex I deficiency, renal failure and myopathy. Neuromuscular Disorders, 2010, 20, 131-135.	0.3	34

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91	The pathogenic m.3243A>T mitochondrial DNA mutation is associated with a variable neurological phenotype. Neuromuscular Disorders, 2010, 20, 403-406.	0.3	7
92	URINE HETEROPLASMY IS THE BEST PREDICTOR OF CLINICAL OUTCOME IN THE m.3243A>G mtDNA MUTATION. Neurology, 2009, 72, 568-569.	1.5	95