Daniele Ghezzi ScD

List of Publications by Year
in descending order

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102 papers	5,685 citations	71102 41 h-index	85541 71 g-index
120 all docs	120 docs citations	120 times ranked	7868 citing authors

#	Article	IF	Citations
1	Genetic diagnosis of Mendelian disorders via RNA sequencing. Nature Communications, 2017, 8, 15824.	12.8	432
2	SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. Nature Genetics, 2009, 41, 654-656.	21.4	233
3	Severe X-Linked Mitochondrial Encephalomyopathy Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2010, 86, 639-649.	6.2	199
4	Mitochondrial DNA haplogroup K is associated with a lower risk of Parkinson's disease in Italians. European Journal of Human Genetics, 2005, 13, 748-752.	2.8	197
5	Leukoencephalopathy with thalamus and brainstem involvement and high lactate â€˜LTBLâ€™ caused by EARS2 mutations. Brain, 2012, 135, 1387-1394.	7.6	187
6	Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology, 2014, 82, 2063-2071.	1.1	172
7	Mutations of the Mitochondrial-tRNA Modifier MTO1 Cause Hypertrophic Cardiomyopathy and Lactic Acidosis. American Journal of Human Genetics, 2012, 90, 1079-1087.	6.2	164
8	Mutations in TTC19 cause mitochondrial complex III deficiency and neurological impairment in humans and flies. Nature Genetics, 2011, 43, 259-263.	21.4	148
9	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
10	Cowchock Syndrome Is Associated with a Mutation in Apoptosis-Inducing Factor. American Journal of Human Genetics, 2012, 91, 1095-1102.	6.2	134
11	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
12	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
13	The Mitochondrial Aminoacyl tRNA Synthetases: Genes and Syndromes. International Journal of Cell Biology, 2014, 2014, 1-11.	2.5	117
14	Assembly Factors of Human Mitochondrial Respiratory Chain Complexes: Physiology and Pathophysiology. Advances in Experimental Medicine and Biology, 2012, 748, 65-106.	1.6	116
15	FASTKD2 Nonsense Mutation in an Infantile Mitochondrial Encephalomyopathy Associated with Cytochrome C Oxidase Deficiency. American Journal of Human Genetics, 2008, 83, 415-423.	6.2	107
16	RNASEH1 Mutations Impair mtDNA Replication and Cause Adult-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2015, 97, 186-193.	6.2	91
17	Disease-Causing SDHAF1 Mutations Impair Transfer of Fe-S Clusters to SDHB. Cell Metabolism, 2016, 23, 292-302.	16.2	89
18	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89

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19	New genes and pathomechanisms in mitochondrial disorders unraveled by NGS technologies. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 1326-1335.	1.0	87
20	VARS2andTARS2Mutations in Patients with Mitochondrial Encephalomyopathies. Human Mutation, 2014, 35, 983-989.	2.5	86
21	COQ4 Mutations Cause a Broad Spectrum of Mitochondrial Disorders Associated with CoQ10 Deficiency. American Journal of Human Genetics, 2015, 96, 309-317.	6.2	86
22	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
23	Mitochondrial dysfunction in Parkinson disease: evidence in mutant PARK2 fibroblasts. Frontiers in Genetics, 2015, 6, 78.	2.3	77
24	Loss of apoptosis-inducing factor critically affects MIA40 function. Cell Death and Disease, 2015, 6, e1814-e1814.	6.3	77
25	Myoclonusâ€“dystonia syndrome: Clinical presentation, disease course, and genetic features in 11 families. Movement Disorders, 2008, 23, 28-34.	3.9	75
26	Human diseases associated with defects in assembly of OXPHOS complexes. Essays in Biochemistry, 2018, 62, 271-286.	4.7	75
27	PINK1heterozygous rare variants: prevalence, significance and phenotypic spectrum. Human Mutation, 2008, 29, 565-565.	2.5	74
28	Paroxysmal non-kinesigenic dyskinesia is caused by mutations of the MR-1 mitochondrial targeting sequence. Human Molecular Genetics, 2009, 18, 1058-1064.	2.9	70
29	Sym1, the yeast ortholog of the MPV17 human disease protein, is a stress-induced bioenergetic and morphogenetic mitochondrial modulator. Human Molecular Genetics, 2010, 19, 1098-1107.	2.9	69
30	MTO1 Mutations are Associated with Hypertrophic Cardiomyopathy and Lactic Acidosis and Cause Respiratory Chain Deficiency in Humans and Yeast. Human Mutation, 2013, 34, 1501-1509.	2.5	67
31	Mutations in APOPT1, Encoding a Mitochondrial Protein, Cause Cavitating Leukoencephalopathy with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 2014, 95, 315-325.	6.2	64
32	Biallelic Mutations in DNM1L are Associated with a Slowly Progressive Infantile Encephalopathy. Human Mutation, 2016, 37, 898-903.	2.5	64
33	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
34	A Homozygous Mutation in<scp>LYRM</scp>7/<scp>MZM</scp>1<scp>L</scp>Associated with Early Onset Encephalopathy, Lactic Acidosis, and Severe Reduction of Mitochondrial Complex<scp>III</scp>Activity. Human Mutation, 2013, 34, 1619-1622.	2.5	60
35	SURF1 deficiency causes demyelinating Charcot-Marie-Tooth disease. Neurology, 2013, 81, 1523-1530.	1.1	53
36	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	7.6	51

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37	A novel AIFM1 mutation expands the phenotype to an infantile motor neuron disease. European Journal of Human Genetics, 2016, 24, 463-466.	2.8	51
38	Cavitating leukoencephalopathy with multiple mitochondrial dysfunction syndrome and NFU1 mutations. Frontiers in Genetics, 2014, 5, 412.	2.3	49
39	A slowly progressive mitochondrial encephalomyopathy widens the spectrum of AIFM1 disorders. Neurology, 2015, 84, 2193-2195.	1.1	47
40	POLG1 in idiopathic Parkinson disease. Neurology, 2006, 67, 1698-1700.	1.1	46
41	Recessive mutations in MSTO1 cause mitochondrial dynamics impairment, leading to myopathy and ataxia. Human Mutation, 2017, 38, 970-977.	2.5	44
42	The isolated carboxyâ€terminal domain of human mitochondrial leucylâ€<scp>tRNA</scp> synthetase rescues the pathological phenotype of mitochondrial <scp>tRNA</scp> mutations in human cells. EMBO Molecular Medicine, 2014, 6, 169-182.	6.9	43
43	Parkin analysis in early onset Parkinson's disease. Parkinsonism and Related Disorders, 2008, 14, 326-333.	2.2	42
44	<scp>ADCK</scp>3 mutations with epilepsy, strokeâ€like episodes and ataxia: a <scp>POLG</scp> mimic?. European Journal of Neurology, 2016, 23, 1188-1194.	3.3	42
45	High frequency stimulation of the subthalamic nucleus is efficacious in Parkin disease. Journal of Neurology, 2005, 252, 208-211.	3.6	40
46	COA7(C1orf163/RESA1) mutations associated with mitochondrial leukoencephalopathy and cytochrome c oxidase deficiency. Journal of Medical Genetics, 2016, 53, 846-849.	3.2	40
47	Not only dominant, not only optic atrophy: expanding the clinical spectrum associated with OPA1 mutations. Orphanet Journal of Rare Diseases, 2017, 12, 89.	2.7	39
48	A novel homozygous mutation in SUCLA2 gene identified by exome sequencing. Molecular Genetics and Metabolism, 2012, 107, 403-408.	1.1	38
49	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. Journal of Neurology, 2017, 264, 102-111.	3.6	38
50	SLC25A10 biallelic mutations in intractable epileptic encephalopathy with complex I deficiency. Human Molecular Genetics, 2018, 27, 499-504.	2.9	37
51	Infantile mitochondrial encephalopathy. Seminars in Fetal and Neonatal Medicine, 2011, 16, 205-215.	2.3	36
52	Clinical and Biochemical Features in a Patient With Mitochondrial Fission Factor Gene Alteration. Frontiers in Genetics, 2018, 9, 625.	2.3	34
53	The impairment of HCCS leads to MLS syndrome by activating a nonâ€canonical cell death pathway in the brain and eyes. EMBO Molecular Medicine, 2013, 5, 280-293.	6.9	33
54	KARS-related diseases: progressive leukoencephalopathy with brainstem and spinal cord calcifications as new phenotype and a review of literature. Orphanet Journal of Rare Diseases, 2018, 13, 45.	2.7	32

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55	Concurrent AFG3L2 and SPG7 mutations associated with syndromic parkinsonism and optic atrophy with aberrant OPA1 processing and mitochondrial network fragmentation. Human Mutation, 2018, 39, 2060-2071.	2.5	32
56	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
57	Clinical-genetic features and peculiar muscle histopathology in infantileDNM1L-related mitochondrial epileptic encephalopathy. Human Mutation, 2019, 40, 601-618.	2.5	31
58	ATPase Domain <scp>AFG3L2</scp> Mutations Alter <scp>OPA1</scp> Processing and Cause Optic Neuropathy. Annals of Neurology, 2020, 88, 18-32.	5.3	31
59	A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. Neurobiology of Disease, 2020, 141, 104880.	4.4	29
60	Frequency and phenotypes of LRRK2 G2019S mutation in Italian patients with Parkinson's disease. Movement Disorders, 2006, 21, 1232-1235.	3.9	28
61	GTP-cyclohydrolase I gene mutations in patients with autosomal dominant and recessive GTP-CH1 deficiency: Identification and functional characterization of four novel mutations. Journal of Inherited Metabolic Disease, 2004, 27, 455-463.	3.6	27
62	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
63	A novel de novo dominant mutation inISCUassociated with mitochondrial myopathy. Journal of Medical Genetics, 2017, 54, 815-824.	3.2	25
64	A nonsense mutation of human <scp>XRCC</scp> 4 is associated with adultâ€onset progressive encephalocardiomyopathy. EMBO Molecular Medicine, 2015, 7, 918-929.	6.9	24
65	Clinical findings in a patient with FARS2 mutations and earlyâ€infantileâ€encephalopathy with epilepsy. American Journal of Medical Genetics, Part A, 2016, 170, 3004-3007.	1.2	24
66	Common and Novel TMEM70 Mutations in a Cohort of Italian Patients with Mitochondrial Encephalocardiomyopathy. JIMD Reports, 2014, 15, 71-8.	1.5	23
67	Mutations in TIMM50 compromise cell survival in OxPhosâ€dependent metabolic conditions. EMBO Molecular Medicine, 2018, 10, .	6.9	23
68	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
69	Adult-onset Alexander disease, associated with a mutation in an alternative GFAP transcript, may be phenotypically modulated by a non-neutral HDAC6 variant. Orphanet Journal of Rare Diseases, 2013, 8, 66.	2.7	21
70	The zebrafish orthologue of the human hepatocerebral disease geneMPV17plays pleiotropic roles in mitochondria. DMM Disease Models and Mechanisms, 2019, 12, .	2.4	21
71	Current and New Next-Generation Sequencing Approaches to Study Mitochondrial DNA. Journal of Molecular Diagnostics, 2021, 23, 732-741.	2.8	21
72	Neonatal mitochondrial leukoencephalopathy with brain and spinal involvement and high lactate: expanding the phenotype of ISCA2 gene mutations. Metabolic Brain Disease, 2018, 33, 805-812.	2.9	20

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73	Clinicopathologic and molecular spectrum of RNASEH1 -related mitochondrial disease. Neurology: Genetics, 2017, 3, e149.	1.9	19
74	APOPT 1/ COA 8 assists COX assembly and is oppositely regulated by UPS and ROS. EMBO Molecular Medicine, 2019, 11, .	6.9	19
75	MELAS-like encephalomyopathy caused by a new pathogenic mutation in the mitochondrial DNA encoded cytochrome c oxidase subunit I. Neuromuscular Disorders, 2012, 22, 990-994.	0.6	18
76	A novel mutation in TTC19 associated with isolated complex III deficiency, cerebellar hypoplasia, and bilateral basal ganglia lesions. Frontiers in Genetics, 2014, 5, 397.	2.3	17
77	Role of PITRM1 in Mitochondrial Dysfunction and Neurodegeneration. Biomedicines, 2021, 9, 833.	3.2	17
78	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
79	Mitochondrial leukoencephalopathy and complex II deficiency associated with a recessive SDHB mutation with reduced penetrance. Molecular Genetics and Metabolism Reports, 2015, 5, 51-54.	1.1	16
80	Exploiting pyocyanin to treat mitochondrial disease due to respiratory complex III dysfunction. Nature Communications, 2021, 12, 2103.	12.8	16
81	Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases. Orphanet Journal of Rare Diseases, 2021, 16, 413.	2.7	16
82	Mitochondrial Complex III Deficiency Caused by TTC19 Defects: Report of a Novel Mutation and Review of Literature. JIMD Reports, 2015, 22, 115-120.	1.5	15
83	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. Journal of Human Genetics, 2018, 63, 563-568.	2.3	15
84	Epileptic phenotypes in children with earlyâ€onset mitochondrial diseases. Acta Neurologica Scandinavica, 2019, 140, 184-193.	2.1	15
85	Homozygous mutations in C1QBP as cause of progressive external ophthalmoplegia (PEO) and mitochondrial myopathy with multiple mtDNA deletions. Human Mutation, 2020, 41, 1745-1750.	2.5	15
86	A family with paroxysmal nonkinesigenic dyskinesias (PNKD): Evidence of mitochondrial dysfunction. European Journal of Paediatric Neurology, 2015, 19, 64-68.	1.6	13
87	Leber's Hereditary Optic Neuropathy: A Report on Novel mtDNA Pathogenic Variants. Frontiers in Neurology, 2021, 12, 657317.	2.4	13
88	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	2.5	12
89	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
90	Functionally pathogenic EARS2 variants in vitro may not manifest a phenotype in vivo. Neurology: Genetics, 2017, 3, e162.	1.9	11

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91	Expanding the phenotypic spectrum of BCS1Lâ€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
92	Benign hereditary chorea and deletions outside NKX2-1: What's the role of MBIP?. European Journal of Medical Genetics, 2018, 61, 581-584.	1.3	9
93	Diagnostic Challenges in Late Onset Multiple Acyl-CoA Dehydrogenase Deficiency: Clinical, Morphological, and Genetic Aspects. Frontiers in Neurology, 2022, 13, 815523.	2.4	7
94	Myopathic mitochondrial DNA depletion syndrome associated with biallelic variants in LIG3. Brain, 2021, 144, e74-e74.	7.6	5
95	Homozygous variant in OTX2 and possible genetic modifiers identified in a patient with combined pituitary hormone deficiency, ocular involvement, myopathy, ataxia, and mitochondrial impairment. American Journal of Medical Genetics, Part A, 2019, 179, 827-831.	1.2	4
96	Biallelic Variants in ENDOG Associated with Mitochondrial Myopathy and Multiple mtDNA Deletions. Cells, 2022, 11, 974.	4.1	4
97	Cerebrospinal Fluid Monoamine Metabolite Analysis in Pediatric Movement Disorders. Journal of Child Neurology, 2015, 30, 1800-1805.	1.4	3
98	A novel homozygous MSTO1 mutation in Ashkenazi Jewish siblings with ataxia and myopathy. Journal of Human Genetics, 2021, 66, 835-840.	2.3	3
99	The impairment of HCCS leads to MLS syndrome by activating a nonâ€canonical cell death pathway in the brain and eyes. EMBO Molecular Medicine, 2014, 6, 849-849.	6.9	0
100	Response to: â€œHeterogeneous phenotypic expression of C1QBP variants is attributable to variable heteroplasmy of secondary mtDNA deletions and mtDNA copy numberâ€. Human Mutation, 2020, 41, 2014-2015.	2.5	0
101	A Clinical-Based Diagnostic Approach to Cerebellar Atrophy in Children. Applied Sciences (Switzerland), 2021, 11, 2333.	2.5	0
102	Response to: Phenotypic heterogeneity of Leigh syndrome due to NDUFA12 variants is multicausal. Human Mutation, 2022, 43, 99-100.	2.5	0