Rosalba Carrozzo

List of Publications by Year
in descending order

144	5,736	76326	98798
papers	citations	40 h-index	67 g-index

151	151	151	7943
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621.	6.2	504
2	Supercomplexes and subcomplexes of mitochondrial oxidative phosphorylation. Biochimica Et Biophysica Acta - Bioenergetics, 2006, 1757, 1066-1072.	1.0	189
3	SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. Brain, 2007, 130, 862-874.	7.6	180
4	Glutathione in blood of patients with Friedreich's ataxia. European Journal of Clinical Investigation, 2001, 31, 1007-1011.	3.4	154
5	Identification and Characterization of Human cDNAs Specific to BCS1, PET112, SCO1, COX15, and COX11, Five Genes Involved in the Formation and Function of the Mitochondrial Respiratory Chain. Genomics, 1998, 54, 494-504.	2.9	144
6	Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase. Human Mutation, 2008, 29, 330-331.	2.5	144
7	Actin Glutathionylation Increases in Fibroblasts of Patients with Friedreich's Ataxia. Journal of Biological Chemistry, 2003, 278, 42588-42595.	3.4	142
8	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.5	142
9	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain, 2013, 136, 872-881.	7.6	130
10	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
11	MtDNA Mutations Associated with Leberâ€²s Hereditary Optic Neuropathy: Studies on Cytoplasmic Hybrid (Cybrid) Cells. Biochemical and Biophysical Research Communications, 1995, 210, 880-888.	2.1	117
12	Functional assays in highâ€resolution clear native gels to quantify mitochondrial complexes in human biopsies and cell lines. Electrophoresis, 2007, 28, 3811-3820.	2.4	97
13	Disease-Causing SDHAF1 Mutations Impair Transfer of Fe-S Clusters to SDHB. Cell Metabolism, 2016, 23, 292-302.	16.2	89
14	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
15	Multiple mtDNA deletions features in autosomal dominant and recessive diseases suggest distinct pathogeneses. Neurology, 1998, 50, 99-106.	1.1	81
16	The T9176G mtDNA mutation severely affects ATP production and results in Leigh syndrome. Neurology, 2001, 56, 687-690.	1.1	79
17	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	3.6	79
18	Subcomplexes of human ATP synthase mark mitochondrial biosynthesis disorders. Annals of Neurology, 2006, 59, 265-275.	5.3	75

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19	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	3.6	70
20	Mutation analysis in 16 patients with mtDNA depletion. Human Mutation, 2003, 21, 453-454.	2.5	69
21	Muscle carnitine deficiency in patients with severe peripheral vascular disease.. Circulation, 1991, 84, 1490-1495.	1.6	66
22	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
23	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
24	A new simple and rapid LCâ€“ESI-MS/MS method for quantification of plasma oxysterols as dimethylaminobutyrate esters. Its successful use for the diagnosis of Niemannâ€“Pick type C disease. Clinica Chimica Acta, 2014, 437, 93-100.	1.1	62
25	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. Human Mutation, 2009, 30, E530-E540.	2.5	59
26	Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. PLoS ONE, 2019, 14, e0214250.	2.5	59
27	Nrf2-Inducers Counteract Neurodegeneration in Frataxin-Silenced Motor Neurons: Disclosing New Therapeutic Targets for Friedreichâ€™s Ataxia. International Journal of Molecular Sciences, 2017, 18, 2173.	4.1	58
28	Pyruvate dehydrogenase deficiency presenting as isolated paroxysmal exercise induced dystonia successfully reversed with thiamine supplementation. Case report and mini-review. European Journal of Paediatric Neurology, 2015, 19, 497-503.	1.6	56
29	Transcriptomic Profiling Discloses Molecular and Cellular Events Related to Neuronal Differentiation in SH-SY5Y Neuroblastoma Cells. Cellular and Molecular Neurobiology, 2017, 37, 665-682.	3.3	54
30	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	7.6	51
31	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
32	â€˜Behr syndromeâ€™ with OPA1 compound heterozygote mutations. Brain, 2015, 138, e321-e321.	7.6	50
33	Glutathione: A redox signature in monitoring EPI-743 therapy in children with mitochondrial encephalomyopathies. Molecular Genetics and Metabolism, 2013, 109, 208-214.	1.1	49
34	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
35	Correlation between clinical and molecular features in two MELAS families. Journal of the Neurological Sciences, 1992, 113, 222-229.	0.6	45
36	Variant late infantile ceroid lipofuscinoses associated with novel mutations in CLN6. Biochemical and Biophysical Research Communications, 2009, 379, 892-897.	2.1	45

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37	Dystonia and deafness due to SUCLA2 defect; Clinical course and biochemical markers in 16 children. Mitochondrion, 2009, 9, 438-442.	3.4	45
38	Understanding pyrrolineâ€5â€carboxylate synthetase deficiency: clinical, molecular, functional, and expression studies, structureâ€based analysis, and novel therapy with arginine. Journal of Inherited Metabolic Disease, 2012, 35, 761-776.	3.6	44
39	Progressive cavitating leukoencephalopathy associated with respiratory chain complex I deficiency and a novel mutation in NDUFS1. Neurogenetics, 2011, 12, 9-17.	1.4	43
40	Novel TTC19 mutation in a family with severe psychiatric manifestations and complex III deficiency. Neurogenetics, 2013, 14, 153-160.	1.4	42
41	Mitochondrial myopathy, parkinsonism, and multiple mtDNA deletions in a Sephardic Jewish family. Neurology, 2001, 56, 802-805.	1.1	41
42	Preliminary evidences on mitochondrial injury and impaired oxidative metabolism in breast cancer. Mitochondrion, 2012, 12, 363-369.	3.4	41
43	Effects of levosimendan on mitochondrial function in patients withÂseptic shock: A randomized trial. Biochimie, 2014, 102, 166-173.	2.6	41
44	The impact of biomarkers analysis in the diagnosis of Niemann-Pick C disease and acid sphingomyelinase deficiency. Clinica Chimica Acta, 2018, 486, 387-394.	1.1	41
45	The use of muscle biopsy in the diagnosis of undefined ataxia with cerebellar atrophy in children. European Journal of Paediatric Neurology, 2012, 16, 248-256.	1.6	39
46	Riboflavin responsive mitochondrial myopathy is a new phenotype of dihydrolipoamide dehydrogenase deficiency. The chaperon-like effect of vitamin B2. Mitochondrion, 2014, 18, 49-57.	3.4	39
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	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. Journal of Neurology, 2017, 264, 102-111.	3.6	38
49	Deficient Muscle Carnitine Transport in Primary Carnitine Deficiency. Pediatric Research, 1997, 42, 583-587.	2.3	38
50	A novel SURF1 mutation results in Leigh syndrome with peripheral neuropathy caused by cytochrome c oxidase deficiency. Neuromuscular Disorders, 2000, 10, 450-453.	0.6	37
51	Cytochrome c Oxidase-deficient Patients Have Distinct Subunit Assembly Profiles. Journal of Biological Chemistry, 2001, 276, 16296-16301.	3.4	36
52	Biallelic SQSTM1 mutations in early-onset, variably progressive neurodegeneration. Neurology, 2018, 91, e319-e330.	1.1	35
53	<scp>DJ</scp>â€1 modulates mitochondrial response to oxidative stress: clues from a novel diagnosis of <scp>PARK7</scp>. Clinical Genetics, 2017, 92, 18-25.	2.0	34
54	A new method for analysis of mitochondrial DNA point mutations and assess levels of heteroplasmy. Biochemical and Biophysical Research Communications, 2006, 342, 387-393.	2.1	33

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55	Peroxisomal acylâ€CoAâ€oxidase deficiency: Two new cases. American Journal of Medical Genetics, Part A, 2008, 146A, 1676-1681.	1.2	31
56	Riboflavin transporter 3 involvement in infantile Brown-Vialetto-Van Laere disease: two novel mutations. Journal of Medical Genetics, 2013, 50, 104-107.	3.2	31
57	Persistent pulmonary arterial hypertension in the newborn (PPHN): A frequent manifestation of TMEM70 defective patients. Molecular Genetics and Metabolism, 2014, 111, 353-359.	1.1	31
58	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
59	Clinical-genetic features and peculiar muscle histopathology in infantileDNM1L-related mitochondrial epileptic encephalopathy. Human Mutation, 2019, 40, 601-618.	2.5	31
60	High proportions of mtDNA duplications in patients with Kearns-Sayre syndrome occur in the heart. , 1997, 71, 443-452.		30
61	Hypertrophic cardiomyopathy and mtDNA depletion. Successful treatment with heart transplantation. Neuromuscular Disorders, 2002, 12, 56-59.	0.6	30
62	Hypertrophic cardiomyopathy, cataract, developmental delay, lactic acidosis: A novel subtype of 3-methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2006, 29, 546-550.	3.6	30
63	Atypical Leigh syndrome associated with the D393N mutation in the mitochondrial ND5 subunit. Neurology, 2003, 61, 1017-1018.	1.1	29
64	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. Neurogenetics, 2016, 17, 65-70.	1.4	29
65	A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. Neurobiology of Disease, 2020, 141, 104880.	4.4	29
66	The T9176G mutation of human mtDNA gives a fully assembled but inactive ATP synthase when modeled inEscherichia coli. FEBS Letters, 2000, 486, 297-299.	2.8	28
67	Identification of a Genetic Variation in ERAP1 Aminopeptidase that Prevents Human Cytomegalovirus miR-UL112-5p-Mediated Immunoevasion. Cell Reports, 2017, 20, 846-853.	6.4	28
68	Mitochondrial tRNACys gene mutation (A5814G): a second family with mitochondrial encephalopathy. Neuromuscular Disorders, 1997, 7, 156-159.	0.6	27
69	Missense and splice site mutations in SPG4 suggest loss-of-function in dominant spastic paraplegia. Journal of Neurology, 2002, 249, 200-205.	3.6	27
70	Human melanoma/NG2 chondroitin sulfate proteoglycan is expressed in the sarcolemma of postnatal human skeletal myofibers. Molecular and Cellular Neurosciences, 2003, 23, 219-231.	2.2	27
71	Comparative analysis of the pathogenic mechanisms associated with the G8363A and A8296G mutations in the mitochondrial tRNALys gene. Biochemical Journal, 2005, 387, 773-778.	3.7	27
72	Altered expression of the MCSP/NG2 chondroitin sulfate proteoglycan in collagen VI deficiency. Molecular and Cellular Neurosciences, 2005, 30, 408-417.	2.2	27

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73	Novel mutations in <scp>KARS</scp> cause hypertrophic cardiomyopathy and combined mitochondrial respiratory chain defect. Clinical Genetics, 2017, 91, 918-923.	2.0	27
74	An inherited large-scale rearrangement in SACS associated with spastic ataxia and hearing loss. Neurogenetics, 2009, 10, 151-155.	1.4	26
75	Assaying ATP synthesis in cultured cells: A valuable tool for the diagnosis of patients with mitochondrial disorders. Biochemical and Biophysical Research Communications, 2009, 383, 58-62.	2.1	26
76	Maternally-inherited Leigh syndrome-related mutations bolster mitochondrial-mediated apoptosis. Journal of Neurochemistry, 2004, 90, 490-501.	3.9	25
77	TMEM70: a mutational hot spot in nuclear ATP synthase deficiency with a pivotal role in complex V biogenesis. Neurogenetics, 2012, 13, 375-386.	1.4	25
78	ISCA1 mutation in a patient with infantile-onset leukodystrophy causes defects in mitochondrial [4Feâ€“4S] proteins. Human Molecular Genetics, 2018, 27, 2739-2754.	2.9	25
79	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism and Related Disorders, 2019, 68, 8-16.	2.2	25
80	Mitochondrial Dynamics: Molecular Mechanisms, Related Primary Mitochondrial Disorders and Therapeutic Approaches. Genes, 2021, 12, 247.	2.4	25
81	Ullrich myopathy phenotype with secondary ColVI defect identified by confocal imaging and electron microscopy analysis. Neuromuscular Disorders, 2007, 17, 587-596.	0.6	24
82	A novel mutation in <scp>NDUFB11</scp> unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. Clinical Genetics, 2017, 91, 441-447.	2.0	24
83	The Vici syndrome protein EPG5 regulates intracellular nucleic acid trafficking linking autophagy to innate and adaptive immunity. Autophagy, 2018, 14, 22-37.	9.1	23
84	A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Clâ¯/H+-Exchanger, Causes Early-Onset Neurodegeneration. American Journal of Human Genetics, 2020, 107, 1062-1077.	6.2	23
85	Delineating the neurological phenotype in children with defects in the <scp>ECHS1</scp> or <scp>HIBCH</scp> gene. Journal of Inherited Metabolic Disease, 2021, 44, 401-414.	3.6	23
86	Human mitochondrial pyrophosphatase: cDNA cloning and analysis of the gene in patients with mtDNA depletion syndromes. Genomics, 2006, 87, 410-416.	2.9	22
87	Frataxin Silencing Inactivates Mitochondrial Complex I in NSC34 Motoneuronal Cells and Alters Glutathione Homeostasis. International Journal of Molecular Sciences, 2014, 15, 5789-5806.	4.1	22
88	Increased NO production in lysinuric protein intolerance. Journal of Inherited Metabolic Disease, 2005, 28, 123-129.	3.6	21
89	Introducing a novel human mtDNA mutation into the Paracoccus denitrificans COX I gene explains functional deficits in a patient. Neurogenetics, 2006, 7, 51-57.	1.4	21
90	Enhancement of mitochondrial <scp>ATP</scp> production by the EscherichiaÂcoli cytotoxic necrotizing factor 1. FEBS Journal, 2014, 281, 3473-3488.	4.7	21

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91	Molecular Genetics of Niemannâ€“Pick Type C Disease in Italy: An Update on 105 Patients and Description of 18 NPC1 Novel Variants. Journal of Clinical Medicine, 2020, 9, 679.	2.4	21
92	Liver fatty acid-binding protein in two cases of human lipid storage. Molecular and Cellular Biochemistry, 1990, 98, 225-30.	3.1	20
93	Urine acylcarnitine analysis by ESIâ€“MS/MS: A new tool for the diagnosis of peroxisomal biogenesis disorders. Clinica Chimica Acta, 2008, 398, 86-89.	1.1	20
94	APOPT 1/ COA 8 assists COX assembly and is oppositely regulated by UPS and ROS. EMBO Molecular Medicine, 2019, 11, .	6.9	19
95	Involvement of the mitochondrial compartment in human NCL fibroblasts. Biochemical and Biophysical Research Communications, 2011, 416, 159-164.	2.1	18
96	Clinical, radiological, and genetic characteristics of 16 patients with ACO2 gene defects: Delineation of an emerging neurometabolic syndrome. Journal of Inherited Metabolic Disease, 2019, 42, 264-275.	3.6	18
97	Changes in skeletal muscle histology and metabolism in patients undergoing exercise deconditioning: Effect of propionyl-L-carnitine. , 1997, 20, 1115-1120.		17
98	The Networks of Genes Encoding Palmitoylated Proteins in Axonal and Synaptic Compartments Are Affected in PPT1 Overexpressing Neuronal-Like Cells. Frontiers in Molecular Neuroscience, 2017, 10, 266.	2.9	17
99	Disorders of nuclear-mitochondrial intergenomic signalling. Journal of Bioenergetics and Biomembranes, 1997, 29, 121-130.	2.3	16
100	Cellular and functional analysis of four mutations located in the mitochondrial ATPase6 gene. Journal of Cellular Biochemistry, 2009, 106, 878-886.	2.6	16
101	Co-occurring WARS2 and CHRNA6 mutations in a child with a severe form of infantile parkinsonism. Parkinsonism and Related Disorders, 2020, 72, 75-79.	2.2	16
102	Respiratory chain defects in hereditary spastic paraplegias. Neuromuscular Disorders, 2001, 11, 565-569.	0.6	15
103	Further pitfalls in the diagnosis of mtDNA mutations: homoplasmic mt-tRNA mutations. Journal of Medical Genetics, 2007, 45, 55-61.	3.2	15
104	Deep sequencing unearths Nuclear mitochondrial Sequences under Leber's hereditary optic neuropathy-associated false heteroplasmic mitochondrial DNA variants. Human Molecular Genetics, 2012, 21, 3753-3764.	2.9	15
105	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
106	Expression of muscle-type phosphorylase in innervated and aneural cultured muscle of patients with myophosphorylase deficiency.. Journal of Clinical Investigation, 1993, 92, 1774-1780.	8.2	15
107	3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clinica Chimica Acta, 2017, 471, 95-100.	1.1	14
108	Novel 7-DHCR mutation in a child with Smith-Lemli-Opitz syndrome. , 2000, 91, 138-140.		13

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109	Mitochondrial Neurogastrointestinal Encephalomyopathy: Novel Pathogenic Mutations in Thymidine Phosphorylase Gene in Two Italian Brothers. Neuropediatrics, 2012, 43, 201-208.	0.6	13
110	Clinical and molecular findings in four new patients harbouring the mtDNA 8993T'C mutation. Journal of Inherited Metabolic Disease, 2001, 24, 883-884.	3.6	12
111	Novel mutation in mitochondrial Elongation Factor EF-Tu associated to dysplastic leukoencephalopathy and defective mitochondrial DNA translation. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 961-967.	3.8	12
112	The urinary organic acids profile in single large-scale mitochondrial DNA deletion disorders. Clinica Chimica Acta, 2018, 481, 156-160.	1.1	12
113	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	2.5	12
114	Biallelic hypomorphic variants in ALDH1A2 cause a novel lethal human multiple congenital anomaly syndrome encompassing diaphragmatic, pulmonary, and cardiovascular defects. Human Mutation, 2021, 42, 506-519.	2.5	12
115	Infantile Mitochondrial Disorders. Bioscience Reports, 2007, 27, 105-112.	2.4	10
116	Infantile-Onset Disorders of Mitochondrial Replication and Protein Synthesis. Journal of Child Neurology, 2011, 26, 866-875.	1.4	10
117	Succinic semialdehyde dehydrogenase deficiency: clinical, biochemical and molecular characterization of a new patient with severe phenotype and a novel mutation. Clinical Genetics, 2006, 69, 294-296.	2.0	9
118	A mitochondrial ATPase 6 mutation is associated with Leigh syndrome in a family and affects proton flow and adenosine triphosphate output when modeled in Escherichia coli. Acta Paediatrica, International Journal of Paediatrics, 2004, 93, 65-67.	1.5	8
119	Protein glutathionylation in cellular compartments: A constitutive redox signal. Redox Report, 2012, 17, 63-71.	4.5	8
120	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8
121	Cellular and molecular studies in muscle and cultures from patients with multiple mitochondrial DNA deletions. Journal of the Neurological Sciences, 1999, 170, 24-31.	0.6	7
122	A novel mtDNA mutation in the ATPase6 gene studied by E. coli modeling. Neurological Sciences, 2000, 21, S983-S984.	1.9	7
123	Novel large-range mitochondrial DNA deletions and fatal multisystemic disorder with prominent hepatopathy. Biochemical and Biophysical Research Communications, 2011, 415, 300-304.	2.1	7
124	ISCA1 mutation in a patient with infantile-onset leukodystrophy causes defects in mitochondrial [4Feâ€“4S] proteins. Human Molecular Genetics, 2018, 27, 3650-3650.	2.9	6
125	Novel dilated cardiomyopathy associated to Calreticulin and Myo7A gene mutation in Usher syndrome. ESC Heart Failure, 2021, 8, 2310-2315.	3.1	6
126	Hyperactive HRAS dysregulates energetic metabolism in fibroblasts from patients with Costello syndrome via enhanced production of reactive oxidizing species. Human Molecular Genetics, 2022, 31, 561-575.	2.9	6

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127	Variability of the expression of muscle mitochondrial damage in ocular mitochondrial myopathy. Neuromuscular Disorders, 1992, 2, 397-404.	0.6	5
128	Cardiolipin content in mitochondria from cultured skin fibroblasts harboring mutations in the mitochondrial ATP6 gene. Journal of Bioenergetics and Biomembranes, 2011, 43, 683-690.	2.3	5
129	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. European Journal of Paediatric Neurology, 2017, 21, 873-883.	1.6	4
130	Delayed appearance of 3â€methylglutaconic aciduria in neonates with early onset metabolic cardiomyopathies: A potential pitfall for the diagnosis. American Journal of Medical Genetics, Part A, 2020, 182, 64-70.	1.2	4
131	Neuromyopathy with congenital cataracts and glaucoma: a distinct syndrome caused by POLG variants. European Journal of Human Genetics, 2018, 26, 367-373.	2.8	3
132	A novel homozygous variant in <scp>COX5A</scp> causes an attenuated phenotype with failure to thrive, lactic acidosis, hypoglycemia, and short stature. Clinical Genetics, 2022, 102, 56-60.	2.0	3
133	<scp>SHP2</scp>'s gainâ€ofâ€function in <scp>Werner</scp> syndrome causes childhood disease onset likely resulting from negative genetic interaction. Clinical Genetics, 2022, 102, 12-21.	2.0	2
134	Biallelic CLPB mutation associated with isolated neutropenia and 3â€MGAâ€uria. Pediatric Allergy and Immunology, 2022, 33, .	2.6	2
135	Clinical and audiological follow up of a family with the 8363G>A mutation in the mitochondrial DNA. Neuromuscular Disorders, 2009, 19, 291-296.	0.6	1
136	Mitochondrial DNA depletion syndromes: an update. Paediatrics and Child Health (United Kingdom), 2009, 19, S32-S37.	0.4	1
137	Progressive axonal polyneuropathy in a mitochondrial disorder: an uncommon association with familial amyloid neuropathy. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2018, 25, 261-262.	3.0	1
138	OXPHOS and mtDNA alterations in a family with spastic paraparesis. Acta Neurologica Scandinavica, 2000, 101, 255-258.	2.1	1
139	A novel 7-DHCR mutation in a lebanese child with Smith-Lemli-Opitz syndrome. Atherosclerosis, 1999, 144, 21-22.	0.8	0
140	P.P.7 01 Confocal imaging and electron microscopy analysis to identify secondary collagen VI defects. Neuromuscular Disorders, 2006, 16, 713-714.	0.6	0
141	M.P.1.04 A novel mtDNA mutation in COIII impairs assembly of cytochrome c oxidase in a MELAS patient. Neuromuscular Disorders, 2007, 17, 768-769.	0.6	0
142	G.P.3.02 Late-onset MNGIE without peripheral neuropathy due to incomplete loss of thymidine phosphorylase activity. Neuromuscular Disorders, 2009, 19, 561-562.	0.6	0
143	EM.P.5.07 Abnormal elastin deposits and altered organization of elastic fibers in collagen VI- related disorders. Neuromuscular Disorders, 2009, 19, 631-632.	0.6	0
144	Response to: Phenotypic heterogeneity of Leigh syndrome due to NDUFA12 variants is multicausal. Human Mutation, 2022, 43, 99-100.	2.5	0