Leonardo Salviati

List of Publications by Year
in descending order

154 papers	10,907 citations	38742 50 h-index	34986 98 g-index

160 all docs	160 docs citations	160 times ranked	15388 citing authors

#	Article	IF	Citations
1	Mitochondrial Cristae Shape Determines Respiratory Chain Supercomplexes Assembly and Respiratory Efficiency. Cell, 2013, 155, 160-171.	28.9	955
2	Sarcopenia: Aging-Related Loss of Muscle Mass and Function. Physiological Reviews, 2019, 99, 427-511.	28.8	767
3	Assessment of mitochondrial respiratory chain enzymatic activities on tissues and cultured cells. Nature Protocols, 2012, 7, 1235-1246.	12.0	732
4	Age-Associated Loss of OPA1 in Muscle Impacts Muscle Mass, Metabolic Homeostasis, Systemic Inflammation, and Epithelial Senescence. Cell Metabolism, 2017, 25, 1374-1389.e6.	16.2	388
5	COQ6 mutations in human patients produce nephrotic syndrome with sensorineural deafness. Journal of Clinical Investigation, 2011, 121, 2013-2024.	8.2	343
6	A Mutation in Para-Hydroxybenzoate-Polyprenyl Transferase (COQ2) Causes Primary Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2006, 78, 345-349.	6.2	322
7	Genetic bases and clinical manifestations of coenzyme Q₁₀ (CoQ₁₀) deficiency. Journal of Inherited Metabolic Disease, 2015, 38, 145-156.	3.6	301
8	Mitochondrial dysfunction in inherited renal disease and acute kidney injury. Nature Reviews Nephrology, 2016, 12, 267-280.	9.6	276
9	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. Journal of Clinical Investigation, 2013, 123, 5179-5189.	8.2	275
10	DRP1-mediated mitochondrial shape controls calcium homeostasis and muscle mass. Nature Communications, 2019, 10, 2576.	12.8	274
11	Early Coenzyme Q10 Supplementation in Primary Coenzyme Q10 Deficiency. New England Journal of Medicine, 2008, 358, 2849-2850.	27.0	217
12	Coenzyme Q biosynthesis in health and disease. Biochimica Et Biophysica Acta - Bioenergetics, 2016, 1857, 1079-1085.	1.0	185
13	Coenzyme Q deficiency triggers mitochondria degradation by mitophagy. Autophagy, 2009, 5, 19-32.	9.1	179
14	LETM1, deleted in Wolf Hirschhorn syndrome is required for normal mitochondrial morphology and cellular viability. Human Molecular Genetics, 2007, 17, 201-214.	2.9	163
15	The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. American Journal of Human Genetics, 2013, 93, 744-751.	6.2	154
16	Mitochondrial DNA depletion and dGK gene mutations. Annals of Neurology, 2002, 52, 311-317.	5.3	152
17	Respiratory chain dysfunction and oxidative stress correlate with severity of primary CoQ ₁₀ deficiency. FASEB Journal, 2008, 22, 1874-1885.	0.5	150
18	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.5	142

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19	Missense mutation of the COQ2 gene causes defects of bioenergetics and de novo pyrimidine synthesis. Human Molecular Genetics, 2007, 16, 1091-1097.	2.9	129
20	Newborn screening for lysosomal storage disorders by tandem mass spectrometry in North East Italy. Journal of Inherited Metabolic Disease, 2018, 41, 209-219.	3.6	114
21	Renal involvement in mitochondrial cytopathies. Pediatric Nephrology, 2012, 27, 539-550.	1.7	109
22	Transcriptional programming of lipid and amino acid metabolism by the skeletal muscle circadian clock. PLoS Biology, 2018, 16, e2005886.	5.6	107
23	A functionally dominant mitochondrial DNA mutation. Human Molecular Genetics, 2008, 17, 1814-1820.	2.9	104
24	Genetics of Coenzyme Q10 Deficiency. Molecular Syndromology, 2014, 5, 156-162.	0.8	102
25	Cytochrome c Oxidase Deficiency Due to a Novel SCO2 Mutation Mimics Werdnig-Hoffmann Disease. Archives of Neurology, 2002, 59, 862-5.	4.5	95
26	Haploinsufficiency ofCOQ4causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	3.2	95
27	Treatment of CoQ10 Deficient Fibroblasts with Ubiquinone, CoQ Analogs, and Vitamin C: Time- and Compound-Dependent Effects. PLoS ONE, 2010, 5, e11897.	2.5	92
28	A novel deletion in the GTPase domain of OPA1 causes defects in mitochondrial morphology and distribution, but not in function. Human Molecular Genetics, 2008, 17, 3291-3302.	2.9	91
29	Molecular Analysis, Pathogenic Mechanisms, and Readthrough Therapy on a Large Cohort of<scp>K</scp>abuki Syndrome Patients. Human Mutation, 2014, 35, 841-850.	2.5	87
30	Coenzyme Q10 is frequently reduced in muscle of patients with mitochondrial myopathy. Neuromuscular Disorders, 2010, 20, 44-48.	0.6	84
31	A novel CRYAB mutation resulting in multisystemic disease. Neuromuscular Disorders, 2012, 22, 66-72.	0.6	84
32	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€“Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
33	Comorbidity between headache and epilepsy in a pediatric headache center. Journal of Headache and Pain, 2010, 11, 235-240.	6.0	77
34	Coenzyme Q deficiency in muscle. Current Opinion in Neurology, 2011, 24, 449-456.	3.6	73
35	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
36	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. European Journal of Human Genetics, 2015, 23, 753-760.	2.8	73

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37	Whole exome sequencing in dominant cataract identifies a new causative factor, CRYBA2, and a variety of novel alleles in known genes. Human Genetics, 2013, 132, 761-770.	3.8	72
38	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
39	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	3.4	70
40	Abnormalities of cerebral arteries are frequent in patients with late-onset Pompe disease. Journal of Neurology, 2010, 257, 1730-1733.	3.6	68
41	Copper supplementation restores cytochrome c oxidase activity in cultured cells from patients with SCO2 mutations. Biochemical Journal, 2002, 363, 321-327.	3.7	66
42	Analysis of Coenzyme Q10 in muscle and fibroblasts for the diagnosis of CoQ10 deficiency syndromes. Clinical Biochemistry, 2008, 41, 697-700.	1.9	65
43	Mutations of cytochrome c identified in patients with thrombocytopenia THC4 affect both apoptosis and cellular bioenergetics. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 269-274.	3.8	65
44	Effect of vanillic acid on COQ6 mutants identified in patients with coenzyme Q10 deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1-6.	3.8	64
45	Mutation Screening in Patients With Isolated Cytochrome c Oxidase Deficiency. Pediatric Research, 2003, 53, 224-230.	2.3	62
46	Coenzyme Q10 distribution in blood is altered in patients with Fibromyalgia. Clinical Biochemistry, 2009, 42, 732-735.	1.9	60
47	MCM5: a new actor in the link between DNA replication and Meier-Gorlin syndrome. European Journal of Human Genetics, 2017, 25, 646-650.	2.8	60
48	Dominant Noonan syndrome-causing LZTR1 mutations specifically affect the Kelch domain substrate-recognition surface and enhance RAS-MAPK signaling. Human Molecular Genetics, 2019, 28, 1007-1022.	2.9	58
49	Acute Disseminated Encephalomyelitis Associated With Hepatitis C Virus Infection. Archives of Neurology, 2001, 58, 1679.	4.5	57
50	Copper supplementation restores cytochrome c oxidase activity in cultured cells from patients with SCO2 mutations. Biochemical Journal, 2002, 363, 321.	3.7	55
51	Patients with a phenotype consistent with facioscapulohumeral muscular dystrophy display genetic and epigenetic heterogeneity. Journal of Medical Genetics, 2012, 49, 41-46.	3.2	55
52	Prevalence of AIP mutations in a large series of sporadic Italian acromegalic patients and evaluation of CDKN1B status in acromegalic patients with multiple endocrine neoplasia. European Journal of Endocrinology, 2010, 163, 369-376.	3.7	53
53	TheCOQ2genotype predicts the severity of coenzyme Q₁₀deficiency. Human Molecular Genetics, 2016, 25, 4256-4265.	2.9	53
54	The chaperone co-inducer BGP-15 alleviates ventilation-induced diaphragm dysfunction. Science Translational Medicine, 2016, 8, 350ra103.	12.4	53

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55	Coenzyme Q10 deficiency in mitochondrial DNA depletion syndromes. Mitochondrion, 2013, 13, 337-341.	3.4	51
56	In mammalian skeletal muscle, phosphorylation of TOMM22 by protein kinase CSNK2/CK2 controls mitophagy. Autophagy, 2018, 14, 311-335.	9.1	51
57	Optimization of respiratory chain enzymatic assays in muscle for the diagnosis of mitochondrial disorders. Mitochondrion, 2011, 11, 893-904.	3.4	50
58	FSHD1 and FSHD2 form a disease continuum. Neurology, 2019, 92, e2273-e2285.	1.1	50
59	Functional characterization of human COQ4, a gene required for Coenzyme Q10 biosynthesis. Biochemical and Biophysical Research Communications, 2008, 372, 35-39.	2.1	49
60	Recessive MYL2 mutations cause infantile type I muscle fibre disease and cardiomyopathy. Brain, 2013, 136, 282-293.	7.6	48
61	Molecular characterization of the human COQ5 C-methyltransferase in coenzyme Q10 biosynthesis. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2014, 1841, 1628-1638.	2.4	48
62	Facioscapulohumeral muscular dystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 607-614.	3.8	47
63	Argininosuccinate lyase deficiency: mutational spectrum in Italian patients and identification of a novelASLpseudogene. Human Mutation, 2007, 28, 694-702.	2.5	46
64	Implementation of Second-Tier Tests in Newborn Screening for Lysosomal Disorders in North Eastern Italy. International Journal of Neonatal Screening, 2019, 5, 24.	3.2	45
65	Mutation Screening in Patients With Isolated Cytochrome c Oxidase Deficiency. Pediatric Research, 2003, 53, 224-230.	2.3	44
66	Mutations in COQ8B (ADCK4) found in patients with steroid-resistant nephrotic syndrome alter COQ8B function. Human Mutation, 2018, 39, 406-414.	2.5	43
67	High frequency of mosaic CREBBP deletions in Rubinsteinâ€“Taybi syndrome patients and mapping of somatic and germ-line breakpoints. Genomics, 2007, 90, 567-573.	2.9	42
68	A novel deletion in the GJA12 gene causes Pelizaeusâ€“Merzbacher-like disease. Neurogenetics, 2007, 8, 57-60.	1.4	42
69	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. European Journal of Human Genetics, 2015, 23, 1254-1258.	2.8	42
70	ALDH18A1gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. Brain, 2016, 139, e3-e3.	7.6	42
71	Deletion of PTEN and BMPR1A on Chromosome 10q23 Is Not Always Associated with Juvenile Polyposis of Infancy. American Journal of Human Genetics, 2006, 79, 593-596.	6.2	40
72	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

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73	The pyruvate kinase activator mitapivat reduces hemolysis and improves anemia in a Î²-thalassemia mouse model. Journal of Clinical Investigation, 2021, 131, .	8.2	39
74	Characterization of 14 novel deletions underlying Rubinsteinâ€“Taybi syndrome: an update of the CREBBP deletion repertoire. Human Genetics, 2015, 134, 613-626.	3.8	38
75	Increased mitophagy in the skeletal muscle of spinal and bulbar muscular atrophy patients. Human Molecular Genetics, 2017, 26, ddx019.	2.9	37
76	Vanillic Acid Restores Coenzyme Q Biosynthesis and ATP Production in Human Cells LackingCOQ6. Oxidative Medicine and Cellular Longevity, 2019, 2019, 1-11.	4.0	35
77	The Conserved Mitochondrial Twin Cx9C Protein Cmc2 Is a Cmc1 Homologue Essential for Cytochrome c Oxidase Biogenesis. Journal of Biological Chemistry, 2010, 285, 15088-15099.	3.4	34
78	Molecular diagnosis of coenzyme Q₁₀ deficiency: an update. Expert Review of Molecular Diagnostics, 2018, 18, 491-498.	3.1	33
79	Complex Neurologic Syndrome Associated With the G1606A Mutation of Mitochondrial DNA. Archives of Neurology, 2002, 59, 1013.	4.5	32
80	Mitochondrial cytopathies and the kidney. Nephrologie Et Therapeutique, 2017, 13, S23-S28.	0.5	31
81	The idebenone metabolite QS10 restores electron transfer in complex I and coenzyme Q defects. Biochimica Et Biophysica Acta - Bioenergetics, 2018, 1859, 901-908.	1.0	31
82	Functional Complementation in Yeast Allows Molecular Characterization of Missense Argininosuccinate Lyase Mutations. Journal of Biological Chemistry, 2009, 284, 28926-28934.	3.4	30
83	Mitochondrial myopathy and ophthalmoplegia in a sporadic patient with the G12315A mutation in mitochondrial DNA. Neuromuscular Disorders, 2002, 12, 865-868.	0.6	29
84	NovelSURF1 mutation in a child with subacute encephalopathy and without the radiological features of Leigh Syndrome. American Journal of Medical Genetics Part A, 2004, 128A, 195-198.	2.4	29
85	Copper and bezafibrate cooperate to rescue cytochrome c oxidase deficiency in cells of patients with sco2 mutations. Orphanet Journal of Rare Diseases, 2012, 7, 21.	2.7	29
86	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
87	hCOX18 and hCOX19: Two human genes involved in cytochrome c oxidase assembly. Biochemical and Biophysical Research Communications, 2005, 337, 832-839.	2.1	28
88	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. Pediatric Nephrology, 2011, 26, 717-724.	1.7	27
89	Renal Mitochondrial Cytopathies. International Journal of Nephrology, 2011, 2011, 1-10.	1.3	27
90	Genetic susceptibility to teratogens: State of the art. Reproductive Toxicology, 2012, 34, 186-191.	2.9	27

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91	ADCK2 Haploinsufficiency Reduces Mitochondrial Lipid Oxidation and Causes Myopathy Associated with CoQ Deficiency. Journal of Clinical Medicine, 2019, 8, 1374.	2.4	27
92	The Arg1038Gly missense variant in the NF1 gene causes a mild phenotype without neurofibromas. Molecular Genetics & Genomic Medicine, 2019, 7, e616.	1.2	26
93	Hepatocerebral Mitochondrial DNA Depletion Syndrome: Clinical and Morphologic Features of a Nuclear Gene Mutation. Journal of Pediatric Gastroenterology and Nutrition, 2004, 38, 216-220.	1.8	25
94	Renal hypoplasia without optic coloboma associated with PAX2 gene deletion. Nephrology Dialysis Transplantation, 2007, 22, 2076-2078.	0.7	25
95	COX16 is required for assembly of cytochrome c oxidase in human cells and is involved in copper delivery to COX2. Biochimica Et Biophysica Acta - Bioenergetics, 2018, 1859, 244-252.	1.0	25
96	The multiple roles of coenzyme Q in cellular homeostasis and their relevance for the pathogenesis of coenzyme Q deficiency. Free Radical Biology and Medicine, 2021, 166, 277-286.	2.9	24
97	Functional Analysis of Missense Mutations ofOAT, Causing Gyrate Atrophy of Choroid and Retina. Human Mutation, 2013, 34, 229-236.	2.5	23
98	Multiple acyl-COA dehydrogenase deficiency in elderly carriers. Journal of Neurology, 2020, 267, 1414-1419.	3.6	23
99	Deficit of human ornithine aminotransferase in gyrate atrophy: Molecular, cellular, and clinical aspects. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2021, 1869, 140555.	2.3	22
100	Coenzyme Q10 Deficiencies in Neuromuscular Diseases. Advances in Experimental Medicine and Biology, 2009, 652, 117-128.	1.6	21
101	A synonymous splicing mutation in the SF3B4 gene segregates in a family with highly variable Nager syndrome. European Journal of Human Genetics, 2017, 25, 371-375.	2.8	20
102	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	12.8	20
103	Survival transcriptome in the coenzyme Q₁₀deficiency syndrome is acquired by epigenetic modifications: a modelling study for human coenzyme Q₁₀deficiencies. BMJ Open, 2013, 3, e002524.	1.9	19
104	Glycogen synthase binds to sarcoplasmic reticulum and is phosphorylated by CaMKII in fast-twitch skeletal muscle. Archives of Biochemistry and Biophysics, 2007, 459, 115-121.	3.0	18
105	Neurofibromatosis type 1 in two siblings due to maternal germline mosaicism. Clinical Genetics, 2014, 85, 386-389.	2.0	18
106	Vitamin K2 cannot substitute Coenzyme Q10 as electron carrier in the mitochondrial respiratory chain of mammalian cells. Scientific Reports, 2019, 9, 6553.	3.3	18
107	Molecular analysis of two uncharacterized sequence variants of the VHL gene. Journal of Human Genetics, 2006, 51, 964-968.	2.3	17
108	Severe encephalopathy associated to pyruvate dehydrogenase mutations and unbalanced coenzyme Q10 content. European Journal of Human Genetics, 2016, 24, 367-372.	2.8	17

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109	New pathogenic variants in COQ4 cause ataxia and neurodevelopmental disorder without detectable CoQ10 deficiency in muscle or skin fibroblasts. Journal of Neurology, 2021, 268, 3381-3389.	3.6	17
110	Increased level of N-acetylaspartylglutamate (NAAG) in the CSF of a patient with Pelizaeus-Merzbacher-like disease due to mutation in the GJA12 gene. European Journal of Paediatric Neurology, 2008, 12, 348-350.	1.6	16
111	Molecular diagnosis of coenzyme Q₁₀deficiency. Expert Review of Molecular Diagnostics, 2015, 15, 1049-1059.	3.1	16
112	Atrio-ventricular block requiring pacemaker in patients with late onset Pompe disease. Neuromuscular Disorders, 2014, 24, 648-650.	0.6	15
113	FBXO28 is a critical gene of the 1q41q42 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1418-1420.	1.2	15
114	Prenatal detection of trisomy 8 mosaicism: Pregnancy outcome and follow up of a series of 17 consecutive cases. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 221, 23-27.	1.1	15
115	Dandy-Walker Malformation Masking the Molar Tooth Sign: An Illustrative Case With Magnetic Resonance Imaging Follow-up. Journal of Child Neurology, 2010, 25, 1419-1422.	1.4	14
116	Is CFTR 621+3 A>G a cystic fibrosis causing mutation?. Journal of Human Genetics, 2010, 55, 23-26.	2.3	14
117	Acute Quadriplegic Myopathy in a 17-Month-Old Boy. Journal of Child Neurology, 2000, 15, 63-66.	1.4	13
118	Impaired copper and iron metabolism in blood cells and muscles of patients affected by copper deficiency myeloneuropathy. Neuropathology and Applied Neurobiology, 2014, 40, 888-898.	3.2	12
119	Validation of CFTR intronic variants identified during cystic fibrosis population screening by a minigene splicing assay. Clinical Chemistry and Laboratory Medicine, 2015, 53, 1719-23.	2.3	12
120	Molecular and cellular basis of ornithine Î´-aminotransferase deficiency caused by the V332M mutation associated with gyrate atrophy of the choroid and retina. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 3629-3638.	3.8	12
121	Biallelic mutations in the TOGARAM1 gene cause a novel primary ciliopathy. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-106833.	3.2	12
122	Motor axonal neuropathy associated with <scp>GNE</scp> mutations. Muscle and Nerve, 2021, 63, 396-401.	2.2	12
123	The combined use of enzyme activity and metabolite assays as a strategy for newborn screening of mucopolysaccharidosis type I. Clinical Chemistry and Laboratory Medicine, 2020, 58, 2063-2072.	2.3	12
124	Two splice variants of CaMKII-anchoring protein are present in the sarcoplasmic reticulum of rabbit fast-twitch muscle. Biochemical and Biophysical Research Communications, 2003, 302, 73-83.	2.1	11
125	Challenges in diagnosis and treatment of late-onset Pompe disease. Current Opinion in Neurology, 2011, 24, 443-448.	3.6	11
126	Exome sequencing analysis in a pair of monozygotic twins re-evaluates the genetics behind their intellectual disability and reveals a CHD2 mutation. Brain and Development, 2016, 38, 590-596.	1.1	11

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127	Drug Repurposing for Duchenne Muscular Dystrophy: The Monoamine Oxidase B Inhibitor Safinamide Ameliorates the Pathological Phenotype in mdx Mice and in Myogenic Cultures From DMD Patients. Frontiers in Physiology, 2018, 9, 1087.	2.8	11
128	A new Italian family with HTRA1 mutation associated with autosomal-dominant variant of CARASIL: Are we pointing towards a disease spectrum?. Journal of the Neurological Sciences, 2019, 396, 108-111.	0.6	11
129	Crosstalk between Long-Term Sublethal Oxidative Stress and Detrimental Inflammation as Potential Drivers for Age-Related Retinal Degeneration. Antioxidants, 2021, 10, 25.	5.1	11
130	Xâ€linked brachytelephalangic chondrodysplasia punctata: A simple trait that is not so simple. American Journal of Medical Genetics, Part A, 2009, 149A, 2464-2468.	1.2	10
131	Novel mutations in the L1CAM gene support the complexity of L1 syndrome. Journal of the Neurological Sciences, 2010, 294, 124-126.	0.6	10
132	The displacement of frataxin from the mitochondrial cristae correlates with abnormal respiratory supercomplexes formation and bioenergetic defects in cells of Friedreich ataxia patients. FASEB Journal, 2021, 35, e21362.	0.5	9
133	Complexity of the 5â€²UTR region of the CLCN5gene: eleven 5â€²UTR ends are differentially expressed in the human kidney. BMC Medical Genomics, 2014, 7, 41.	1.5	8
134	Mutation analysis of COX18 in 29 patients with isolated cytochrome c oxidase deficiency. Journal of Human Genetics, 2009, 54, 419-421.	2.3	7
135	Is there a link between COQ6 and schwannomatosis?. Genetics in Medicine, 2015, 17, 312-313.	2.4	7
136	Hybrid Minigene Assay: An Efficient Tool to Characterize mRNA Splicing Profiles of NF1 Variants. Cancers, 2021, 13, 999.	3.7	7
137	6q27 subtelomeric deletions: Is there a specific phenotype?. American Journal of Medical Genetics, Part A, 2011, 155, 1213-1214.	1.2	6
138	Photosensitive epilepsy and long QT: expanding Timothy syndrome phenotype. Clinical Neurophysiology, 2019, 130, 2134-2136.	1.5	6
139	Polymorphisms of the SCN1A gene in children and adolescents with primary headache and idiopathic or cryptogenic epilepsy: is there a linkage?. Journal of Headache and Pain, 2011, 12, 435-441.	6.0	5
140	Genotypeâ€“phenotype correlation in Gordonâ€™s syndrome: report of two cases carrying novel heterozygous mutations. Journal of Nephrology, 2022, 35, 859-862.	2.0	5
141	Yeast complementation is sufficiently sensitive to detect the residual activity of ASL alleles associated with mild forms of argininosuccinic aciduria. Journal of Inherited Metabolic Disease, 2012, 35, 557-558.	3.6	4
142	Ichthyosis and Kallmann syndrome: Not always a contiguous gene syndrome. Journal of Dermatological Science, 2015, 78, 158-160.	1.9	4
143	Heterologous Expression in Yeast of Human Ornithine Carriers ORNT1 and ORNT2 and of ORNT1 Alleles Implicated in HHH Syndrome in Humans. JIMD Reports, 2015, 28, 119-126.	1.5	4
144	Correlation between Î±1-Antitrypsin Deficiency and SARS-CoV-2 Infection: Epidemiological Data and Pathogenetic Hypotheses. Journal of Clinical Medicine, 2021, 10, 4493.	2.4	4

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145	Toll-like receptor 3 pathway deficiency, herpes simplex encephalitis, and anti-NMDAR encephalitis: more questions than answers. Pediatric Research, 2021, 89, 1043-1043.	2.3	3
146	Craniosynostosis is a feature of <scp>CHD7</scp>â€related <scp>CHARGE</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2160-2163.	1.2	2
147	Molecular and Cellular Studies Reveal Folding Defects of Human Ornithine Aminotransferase Variants Associated With Gyrate Atrophy of the Choroid and Retina. Frontiers in Molecular Biosciences, 2021, 8, 695205.	3.5	2
148	The Splicing of the Mitochondrial Calcium Uniporter Genuine Activator MICU1 Is Driven by RBFOX2 Splicing Factor during Myogenic Differentiation. International Journal of Molecular Sciences, 2022, 23, 2517.	4.1	2
149	A novel RRM2B mutation associated with mitochondrial DNA depletion syndrome. Molecular Genetics and Metabolism Reports, 2022, 32, 100887.	1.1	2
150	Post-natal developmental expression of Î±KAP splice variants in rabbit fast-twitch and slow-twitch skeletal muscle. Journal of Muscle Research and Cell Motility, 2004, 25, 309-314.	2.0	0
151	Two unusual cases of Gitelmanâ€™s syndrome with a complex inheritance: how the phenotype can help interpret the genotype: lesson for the clinical nephrologist. Journal of Nephrology, 2021, 34, 1327-1330.	2.0	0
152	Cytochrome c Defects in Human Disease. , 2021, , 191-200.		0
153	The Kidney in Mitochondrial Diseases. , 2021, , 1-13.		0
154	Distinct Phenotypic and microRNA Expression in X-Linked Charcotâ€“Marieâ€“Tooth Correlated with a Novel Mutation in the GJB1 Gene. , 2022, 1, 66-74.		0