Filippo M Santorelli

List of Publications by Year
in descending order

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582 papers	18,404 citations	14655 66 h-index	32842 100 g-index
594 all docs	594 docs citations	594 times ranked	18826 citing authors

#	Article	IF	Citations
1	Mitochondrial encephalomyopathy with coenzyme Q ₁₀ deficiency. Neurology, 1997, 48, 1238-1243.	1.1	313
2	Mutations in SPG11, encoding spatacsin, are a major cause of spastic paraplegia with thin corpus callosum. Nature Genetics, 2007, 39, 366-372.	21.4	303
3	COQ2 Nephropathy. Journal of the American Society of Nephrology: JASN, 2007, 18, 2773-2780.	6.1	297
4	Hereditary spastic paraplegia: Clinical-genetic characteristics and evolving molecular mechanisms. Experimental Neurology, 2014, 261, 518-539.	4.1	281
5	The mutation at nt 8993 of mitochondrial DNA is a common cause of Leigh's syndrome. Annals of Neurology, 1993, 34, 827-834.	5.3	252
6	Methylmalonic and propionic aciduria. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2006, 142C, 104-112.	1.6	214
7	Mutations in SPG11 are frequent in autosomal recessive spastic paraplegia with thin corpus callosum, cognitive decline and lower motor neuron degeneration. Brain, 2008, 131, 772-784.	7.6	206
8	Clinical features associated with the A â† ' G transition at nucleotide 8344 of mtDNA (â€œMERRF mutationâ€). Neurology, 1993, 43, 1200-1200.	1.1	205
9	Infantile-Onset Ascending Hereditary Spastic Paralysis Is Associated with Mutations in the Alsin Gene. American Journal of Human Genetics, 2002, 71, 518-527.	6.2	203
10	Identification of the SPG15 Gene, Encoding Spastizin, as a Frequent Cause of Complicated Autosomal-Recessive Spastic Paraplegia, Including Kjellin Syndrome. American Journal of Human Genetics, 2008, 82, 992-1002.	6.2	192
11	Supercomplexes and subcomplexes of mitochondrial oxidative phosphorylation. Biochimica Et Biophysica Acta - Bioenergetics, 2006, 1757, 1066-1072.	1.0	189
12	Identification of a Novel Mutation in the mtDNA ND5 Gene Associated with MELAS. Biochemical and Biophysical Research Communications, 1997, 238, 326-328.	2.1	187
13	SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. Brain, 2007, 130, 862-874.	7.6	180
14	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	6.2	179
15	A new mtDNA mutation in the tRNALeu(UUR) gene associated with maternally inherited cardiomyopathy. Human Mutation, 1994, 3, 37-43.	2.5	178
16	Congenital muscular dystrophies with defective glycosylation of dystroglycan. Neurology, 2009, 72, 1802-1809.	1.1	166
17	A homoplasmic mitochondrial transfer Ribonucleic Acid mutation as a cause of maternally inherited hypertrophic cardiomyopathy. Journal of the American College of Cardiology, 2003, 41, 1786-1796.	2.8	161
18	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157

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19	Glutathione in blood of patients with Friedreich's ataxia. European Journal of Clinical Investigation, 2001, 31, 1007-1011.	3.4	154
20	Multiple mitochondria1 DNA deletions associated with autosomal recessive ophthalmoplegia and severe cardiomyopathy. Neurology, 1996, 46, 1329-1329.	1.1	153
21	Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase. Human Mutation, 2008, 29, 330-331.	2.5	144
22	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.5	142
23	Infantile-onset ascending hereditary spastic paralysis: A case report and brief literature review. European Journal of Paediatric Neurology, 2014, 18, 235-239.	1.6	139
24	Maternally inherited cardiomyopathy and hearing loss associated with a novel mutation in the mitochondrial tRNA(Lys) gene (G8363A). American Journal of Human Genetics, 1996, 58, 933-9.	6.2	132
25	Leigh syndrome and hypertrophic cardiomyopathy in an infant with a mitochondrial DNA point mutation (T8993G). American Journal of Medical Genetics Part A, 1994, 50, 265-271.	2.4	124
26	DPM2â€CDG: A muscular dystrophyâ€“dystroglycanopathy syndrome with severe epilepsy. Annals of Neurology, 2012, 72, 550-558.	5.3	121
27	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
28	The Mitochondrial Ornithine Transporter. Journal of Biological Chemistry, 2003, 278, 32778-32783.	3.4	117
29	Ataxia with oculomotor apraxia type 2: A clinical, pathologic, and genetic study. Neurology, 2006, 66, 1207-1210.	1.1	114
30	Autism with Seizures and Intellectual Disability: Possible Causative Role of Gain-of-function of the Inwardly-Rectifying K+ Channel Kir4.1. Neurobiology of Disease, 2011, 43, 239-247.	4.4	108
31	Multiple mitochondrial DNA deletions in sporadic inclusion body myositis: A study of 56 patients. Annals of Neurology, 1996, 39, 789-795.	5.3	107
32	Maternally Inherited Cardiomyopathy: An Atypical Presentation of the mtDNA 12S rRNA Gene A1555G Mutation. American Journal of Human Genetics, 1999, 64, 295-300.	6.2	107
33	A Novel mtDNA Point Mutation in Maternally Inherited Cardiomyopathy. Biochemical and Biophysical Research Communications, 1995, 213, 588-593.	2.1	104
34	Somatic and germline mosaicisms in Severe Myoclonic Epilepsy of Infancy. Biochemical and Biophysical Research Communications, 2006, 341, 489-493.	2.1	102
35	Spectrum of Brain Changes in Patients With Congenital Muscular Dystrophy and FKRP Gene Mutations. Archives of Neurology, 2006, 63, 251.	4.5	97
36	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

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37	Autosomal dominant external ophthalmoplegia and bipolar affective disorder associated with a mutation in the ANT1 gene. Neuromuscular Disorders, 2003, 13, 162-165.	0.6	95
38	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
39	POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes. Biochemical and Biophysical Research Communications, 2007, 363, 1033-1037.	2.1	91
40	Maternally inherited Leigh syndrome. Journal of Pediatrics, 1993, 122, 419-422.	1.8	87
41	Epilepsy, Behavioral Abnormalities, and Physiological Comorbidities in Syntaxin-Binding Protein 1 (STXBP1) Mutant Zebrafish. PLoS ONE, 2016, 11, e0151148.	2.5	87
42	Familial basilar migraine associated with a new mutation in the ATP1A2 gene. Neurology, 2005, 65, 1826-1828.	1.1	86
43	Loss of spatacsin function alters lysosomal lipid clearance leading to upper and lower motor neuron degeneration. Neurobiology of Disease, 2017, 102, 21-37.	4.4	85
44	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. Neuromuscular Disorders, 2006, 16, 548-552.	0.6	83
45	Clinical and genetic studies in hereditary spastic paraplegia with thin corpus callosum. Neurology, 2004, 62, 262-268.	1.1	82
46	A Novel Mitochondrial DNA Point Mutation Associated with Mitochondrial Encephalocardiomyopathy. Biochemical and Biophysical Research Communications, 1995, 216, 835-840.	2.1	81
47	Multiple mtDNA deletions features in autosomal dominant and recessive diseases suggest distinct pathogeneses. Neurology, 1998, 50, 99-106.	1.1	81
48	Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. Molecular Genetics and Metabolism, 2008, 93, 475-480.	1.1	80
49	Congenital myopathies: clinical phenotypes and new diagnostic tools. Italian Journal of Pediatrics, 2017, 43, 101.	2.6	80
50	CLN8 is an endoplasmic reticulum cargo receptor that regulates lysosome biogenesis. Nature Cell Biology, 2018, 20, 1370-1377.	10.3	80
51	A T â†' C mutation at nt 8993 of mitochondrial DNA in a child with Leigh syndrome. Neurology, 1994, 44, 972-972.	1.1	80
52	The T9176G mtDNA mutation severely affects ATP production and results in Leigh syndrome. Neurology, 2001, 56, 687-690.	1.1	79
53	Long period fiber grating nano-optrode for cancer biomarker detection. Biosensors and Bioelectronics, 2016, 80, 590-600.	10.1	79
54	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

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55	Mitochondrial DNA metabolism in early development of zebrafish (Danio rerio). Biochimica Et Biophysica Acta - Bioenergetics, 2012, 1817, 1002-1011.	1.0	78
56	Revelation of a New Mitochondrial DNA Mutation (G12147A) in a MELAS/MERFF Phenotype. Archives of Neurology, 2004, 61, 269.	4.5	77
57	Mitochondrial DNA haplogroups influence the therapeutic response to riboflavin in migraineurs. Neurology, 2009, 72, 1588-1594.	1.1	77
58	Novel DyneinDYNC1H1Neck and Motor Domain Mutations Link Distal Spinal Muscular Atrophy and Abnormal Cortical Development. Human Mutation, 2014, 35, 298-302.	2.5	77
59	MotorPlex provides accurate variant detection across large muscle genes both in single myopathic patients and in pools of DNA samples. Acta Neuropathologica Communications, 2014, 2, 100.	5.2	76
60	Malignant Pancreatic Endocrine Tumor in a Child With Tuberous Sclerosis. American Journal of Surgical Pathology, 2003, 27, 1386-1389.	3.7	75
61	Subcomplexes of human ATP synthase mark mitochondrial biosynthesis disorders. Annals of Neurology, 2006, 59, 265-275.	5.3	75
62	Friedreich's ataxia: Oxidative stress and cytoskeletal abnormalities. Journal of the Neurological Sciences, 2009, 287, 111-118.	0.6	75
63	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.1	75
64	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	7.6	74
65	Maternally inherited encephalopathy associated with a single-base insertion in the mitochondrial tRNATrp gene. Annals of Neurology, 1997, 42, 256-260.	5.3	72
66	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
67	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	2.7	70
68	Mutation analysis in 16 patients with mtDNA depletion. Human Mutation, 2003, 21, 453-454.	2.5	69
69	A novel mutation in SACS gene in a family from southern Italy. Neurology, 2004, 62, 100-102.	1.1	69
70	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	9.0	69
71	Maternally inherited deafness associated with a T1095C mutation in the mDNA. European Journal of Human Genetics, 2001, 9, 147-149.	2.8	68
72	Genetic heterogeneity of megalencephalic leukoencephalopathy and subcortical cysts. Neurology, 2003, 61, 534-537.	1.1	68

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73	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
74	Novel SACS mutations in autosomal recessive spastic ataxia of Charlevoix-Saguenay type. Neurology, 2004, 62, 103-106.	1.1	66
75	Genetically induced dysfunctions of Kir2.1 channels: implications for short QT3 syndrome and autismâ€“epilepsy phenotype. Human Molecular Genetics, 2014, 23, 4875-4886.	2.9	65
76	Fatal infantile leukodystrophy. Neurology, 2001, 57, 265-270.	1.1	64
77	Infantile ascending hereditary spastic paralysis (IAHSP). Neurology, 2003, 60, 674-682.	1.1	64
78	Mutation inCPT1CAssociated With Pure Autosomal Dominant Spastic Paraplegia. JAMA Neurology, 2015, 72, 561.	9.0	64
79	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
80	The Extra-Virgin Olive Oil Polyphenols Oleocanthal and Oleacein Counteract Inflammation-Related Gene and miRNA Expression in Adipocytes by Attenuating NF-ÎºB Activation. Nutrients, 2019, 11, 2855.	4.1	63
81	Clinical and molecular findings in patients with giant axonal neuropathy (GAN). Neurology, 2004, 62, 13-16.	1.1	62
82	Novel mutations in SPG11 cause hereditary spastic paraplegia associated with earlyâ€onset levodopaâ€responsive Parkinsonism. Movement Disorders, 2011, 26, 553-556.	3.9	62
83	Mutations in MFSD8/CLN7 are a frequent cause of variant-late infantile neuronal ceroid lipofuscinosis. Human Mutation, 2009, 30, E530-E540.	2.5	59
84	Familyâ€based association study of 5â€HTTLPR, TPH, MAOâ€A, and DRD4 polymorphisms in mood disorders. American Journal of Medical Genetics Part A, 2002, 114, 361-369.	2.4	57
85	Identification of novel mutations in theSLC25A15gene in hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome: A clinical, molecular, and functional study. Human Mutation, 2009, 30, 741-748.	2.5	57
86	Drug consumption in medication overuse headache is influenced by brain-derived neurotrophic factor Val66Met polymorphism. Journal of Headache and Pain, 2009, 10, 349-355.	6.0	57
87	Supratentorial and pontine <scp>MRI</scp> abnormalities characterize recessive spastic ataxia of <scp>C</scp>harlevoixâ€<scp>S</scp>aguenay. A comprehensive study of an <scp>I</scp>talian series. European Journal of Neurology, 2013, 20, 138-146.	3.3	57
88	Clinical and molecular findings in hyperornithinemia-hyperammonemia-homocitrullinuria syndrome. Neurology, 2001, 57, 911-914.	1.1	56
89	Gain-of-function defects of astrocytic Kir4.1 channels in children with autism spectrum disorders and epilepsy. Scientific Reports, 2016, 6, 34325.	3.3	56
90	Clinical Heterogeneity Associated with the Mitochondrial DNA T8993C Point Mutation. Pediatric Research, 1996, 39, 914-917.	2.3	56

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91	A Combined Nucleic Acid and Protein Analysis in Friedreich Ataxia: Implications for Diagnosis, Pathogenesis and Clinical Trial Design. PLoS ONE, 2011, 6, e17627.	2.5	55
92	Autism-epilepsy phenotype with macrocephaly suggests PTEN, but not GLIALCAM, genetic screening. BMC Medical Genetics, 2014, 15, 26.	2.1	55
93	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
94	Clinical application of next generation sequencing in hereditary spinocerebellar ataxia: increasing the diagnostic yield and broadening the ataxia-spasticity spectrum. A retrospective analysis. Neurogenetics, 2018, 19, 1-8.	1.4	54
95	Proficiency testing of clinical microbiology laboratories using modified decontamination procedures for detection of nontuberculous mycobacteria in sputum samples from cystic fibrosis patients. The Nontuberculous Mycobacteria in Cystic Fibrosis Study Group. Journal of Clinical Microbiology, 1997, 35, 2706-2708.	3.9	54
96	The mitochondrial A3243G mutation presenting as severe cardiomyopathy.. Journal of Medical Genetics, 1997, 34, 607-609.	3.2	53
97	Screening of ARHSP-TCC patients expands the spectrum ofSPG11mutations and includes a large scale gene deletion. Human Mutation, 2009, 30, E500-E519.	2.5	53
98	Expanding the clinical spectrum of POMT1 phenotype. Neurology, 2006, 66, 1564-1567.	1.1	52
99	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	5.3	52
100	Novel mutation in the mitochondrial DNA tRNA glycine gene associated with sudden unexpected death. Pediatric Neurology, 1996, 15, 145-149.	2.1	51
101	Identification of novel WFS1 mutations in Italian children with Wolfram syndrome. Human Mutation, 2001, 17, 348-349.	2.5	51
102	Pontocerebellar hypoplasia. Neurology, 2010, 75, 1459-1464.	1.1	51
103	Centronuclear myopathies: genotypeâ€“phenotype correlation and frequency of defined genetic forms in an Italian cohort. Journal of Neurology, 2015, 262, 1728-1740.	3.6	51
104	Understanding Spreading Depression from Headache to Sudden Unexpected Death. Frontiers in Neurology, 2018, 9, 19.	2.4	51
105	Comparative biochemical studies in fibroblasts from patients with different forms of Leigh syndrome. Journal of Inherited Metabolic Disease, 1996, 19, 43-50.	3.6	50
106	Protein glutathionylation in human central nervous system: Potential role in redox regulation of neuronal defense against free radicals. Journal of Neuroscience Research, 2006, 83, 256-263.	2.9	50
107	Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease?. Cerebellum, 2014, 13, 79-88.	2.5	50
108	Heterogeneous clinical presentation of the mtDNA NARP/T8993G mutation. Neurology, 1997, 49, 270-273.	1.1	49

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109	Cerium oxide nanoparticles: the regenerative redox machine in bioenergetic imbalance. Nanomedicine, 2017, 12, 403-416.	3.3	49
110	Neutral Lipid Storage Diseases: clinical/genetic features and natural history in a large cohort of Italian patients. Orphanet Journal of Rare Diseases, 2017, 12, 90.	2.7	49
111	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	2.3	49
112	White matter lesions in spastic paraplegia with mutations in SPG5/CYP7B1. Neuromuscular Disorders, 2009, 19, 62-65.	0.6	48
113	Motor Chip: A Comparative Genomic Hybridization Microarray for Copy-Number Mutations in 245 Neuromuscular Disorders. Clinical Chemistry, 2011, 57, 1584-1596.	3.2	48
114	Leigh-type neuropathology in Pearson syndrome associated with impaired ATP production and a novel mtDNA deletion. Neurology, 1996, 47, 1320-1323.	1.1	47
115	Novel L2HGDH mutations in 21 patients with L-2-hydroxyglutaric aciduria of Portuguese origin. Human Mutation, 2005, 26, 395-396.	2.5	47
116	Management of Hereditary Spastic Paraplegia: A Systematic Review of the Literature. Frontiers in Neurology, 2019, 10, 3.	2.4	47
117	Social Preference Tests in Zebrafish: A Systematic Review. Frontiers in Veterinary Science, 2020, 7, 590057.	2.2	46
118	SPG3A. Neurology, 2002, 59, 2002-2005.	1.1	45
119	Autosomal dominant hereditary spastic paraplegia: DHPLC-based mutation analysis ofSPG4 reveals eleven novel mutations. Human Mutation, 2005, 25, 506-506.	2.5	45
120	â€œBartter-likeâ€•phenotype in Kearnsâ€“Sayre syndrome. Pediatric Nephrology, 2006, 21, 355-360.	1.7	45
121	Variant late infantile ceroid lipofuscinoses associated with novel mutations in CLN6. Biochemical and Biophysical Research Communications, 2009, 379, 892-897.	2.1	45
122	Syndromes associated with mitochondrial DNA depletion. Italian Journal of Pediatrics, 2014, 40, 34.	2.6	45
123	Adaptor protein complex 4 deficiency: a paradigm of childhood-onset hereditary spastic paraplegia caused by defective protein trafficking. Human Molecular Genetics, 2020, 29, 320-334.	2.9	45
124	Intrafamilial variability in hereditary spastic paraplegia associated with an SPG4 gene mutation. Neurology, 2000, 55, 702-705.	1.1	44
125	The complex phenotype of spinocerebellar ataxia type 48 in eight unrelated Italian families. European Journal of Neurology, 2020, 27, 498-505.	3.3	44
126	Progressive exercise intolerance associated with a new muscle-restricted nonsense mutation (G142X) in the mitochondrial cytochromeb gene. Muscle and Nerve, 2003, 28, 508-511.	2.2	43

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127	Novel mutations in CLN8 in Italian variant late infantile neuronal ceroid lipofuscinosis: another genetic hit in the Mediterranean. Neurogenetics, 2006, 7, 111-117.	1.4	43
128	Spastic paraplegia with thin corpus callosum: description of 20 new families, refinement of the SPG11 locus, candidate gene analysis and evidence of genetic heterogeneity. Neurogenetics, 2006, 7, 149-156.	1.4	43
129	Progressive cavitating leukoencephalopathy associated with respiratory chain complex I deficiency and a novel mutation in NDUFS1. Neurogenetics, 2011, 12, 9-17.	1.4	43
130	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Porettiâ€“Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267.	2.8	43
131	Motor neuron degeneration in spastic paraplegia 11 mimics amyotrophic lateral sclerosis lesions. Brain, 2016, 139, aww061.	7.6	43
132	Spinocerebellar ataxia 48 presenting with ataxia associated with cognitive, psychiatric, and extrapyramidal features: A report of two Italian families. Parkinsonism and Related Disorders, 2019, 65, 91-96.	2.2	43
133	Uncoupling Protein-1 mRNA Expression in Lipomas from Patients Bearing Pathogenic Mitochondrial DNA Mutations. Biochemical and Biophysical Research Communications, 2000, 278, 800-802.	2.1	42
134	POMT2 mutation in a patient with â€˜MEB-likeâ€™ phenotype. Neuromuscular Disorders, 2006, 16, 446-448.	0.6	42
135	Cortical response to somatosensory stimulation in medication overuse headache patients is influenced by angiotensin converting enzyme (ACE) I/D genetic polymorphism. Cephalalgia, 2012, 32, 1189-1197.	3.9	42
136	Molecular epidemiology of childhood neuronal ceroid-lipofuscinosis in Italy. Orphanet Journal of Rare Diseases, 2013, 8, 19.	2.7	42
137	Novel TTC19 mutation in a family with severe psychiatric manifestations and complex III deficiency. Neurogenetics, 2013, 14, 153-160.	1.4	42
138	Powerhouse failure and oxidative damage in autosomal recessive spastic ataxia of Charlevoix-Saguenay. Journal of Neurology, 2015, 262, 2755-2763.	3.6	42
139	A double mutation (A8296G and G8363A) in the mitochondrial DNA tRNA ^{Lys} gene associated with myoclonus epilepsy with ragged-red fibers. Neurology, 1999, 52, 377-377.	1.1	42
140	Mitochondrial myopathy, parkinsonism, and multiple mtDNA deletions in a Sephardic Jewish family. Neurology, 2001, 56, 802-805.	1.1	41
141	Revelation of a Novel CLN5 Mutation in Early Juvenile Neuronal Ceroid Lipofuscinosis. Neuropediatrics, 2007, 38, 46-49.	0.6	41
142	Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. Neurology, 2017, 88, 2132-2140.	1.1	41
143	ZFYVE26/SPASTIZIN and SPG11/SPATACSIN mutations in hereditary spastic paraplegia types AR-SPG15 and AR-SPG11 have different effects on autophagy and endocytosis. Autophagy, 2019, 15, 34-57.	9.1	41
144	A Novel ATP1A2 Mutation in a Family with FHM Type II. Cephalalgia, 2006, 26, 324-328.	3.9	39

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145	The wolframin His611Arg polymorphism influences medication overuse headache. Neuroscience Letters, 2007, 424, 179-184.	2.1	39
146	New findings in the ataxia of Charlevoixâ€“Saguenay. Journal of Neurology, 2012, 259, 869-878.	3.6	39
147	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
148	Comparative biochemical studies of ATPases in cells from patients with the T8993G or T8993C mitochondrial DNA mutations. Journal of Inherited Metabolic Disease, 1998, 21, 829-836.	3.6	38
149	X-linked congenital ataxia: A clinical and genetic study. , 2000, 92, 53-56.		38
150	POMGnT1 Mutations in Congenital Muscular Dystrophy. Archives of Neurology, 2006, 63, 1491.	4.5	38
151	POMT1 and POMT2 mutations in CMD patients: A multicentric Italian study. Neuromuscular Disorders, 2008, 18, 565-571.	0.6	38
152	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
153	Incomplete penetrance in an SPG3A -linked family with a new mutation in the atlastin gene. Neurology, 2004, 62, 2138-2139.	1.1	37
154	HHH syndrome (hyperornithinaemia, hyperammonaemia, homocitrullinuria), with fulminant hepatitis-like presentation. Journal of Inherited Metabolic Disease, 2006, 29, 186-189.	3.6	37
155	The mitochondrial DNA A8344G mutation in leigh syndrome revealed by analysis in paraffin-embedded sections: Revisiting the past. Annals of Neurology, 1998, 44, 962-964.	5.3	35
156	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	2.4	35
157	Late-onset MNGIE without peripheral neuropathy due to incomplete loss of thymidine phosphorylase activity. Neuromuscular Disorders, 2009, 19, 837-840.	0.6	34
158	Cross-Linked Enzyme Aggregates as Versatile Tool for Enzyme Delivery: Application to Polymeric Nanoparticles. Bioconjugate Chemistry, 2018, 29, 2225-2231.	3.6	34
159	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
160	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33
161	The emerging concept of mitochondrial cardiomyopathies. American Heart Journal, 2001, 141, 5A-12A.	2.7	32
162	Phenotypic Heterogeneity in Two Unrelated Danon Patients Associated with the SameLAMPâ€2Gene Mutation. Neuropediatrics, 2005, 36, 309-313.	0.6	32

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163	Migraine headache: a review of the molecular genetics of a common disorder. Journal of Headache and Pain, 2012, 13, 571-580.	6.0	32
164	Identification of mutations in AP4S1/SPG52 through next generation sequencing in three families. European Journal of Neurology, 2016, 23, 1580-1587.	3.3	32
165	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
166	The R495W mutation in SPG3A causes spastic paraplegia associated with axonal neuropathy. Journal of Neurology, 2005, 252, 901-903.	3.6	31
167	Peroxisomal acylâ€CoAâ€oxidase deficiency: Two new cases. American Journal of Medical Genetics, Part A, 2008, 146A, 1676-1681.	1.2	31
168	Collapsing glomerulopathy associated with inherited mitochondrial injury. Kidney International, 2008, 74, 237-243.	5.2	31
169	Rippling muscle disease and cardiomyopathy associated with a mutation in the CAV3 gene. Neuromuscular Disorders, 2009, 19, 779-783.	0.6	31
170	Myelinated retinal fibers in autosomal recessive spastic ataxia of Charlevoix-Saguenay. European Journal of Neurology, 2011, 18, 1187-1190.	3.3	31
171	TRPV4 mutations in children with congenital distal spinal muscular atrophy. Neurogenetics, 2012, 13, 195-203.	1.4	31
172	Respiratory and cardiac function in congenital muscular dystrophies with alpha dystroglycan deficiency. Neuromuscular Disorders, 2012, 22, 685-689.	0.6	31
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