Filippo M Santorelli

List of Publications by Year
in descending order

581	18,404	16791 66	37326 100
papers	citations	h-index	g-index

594	594	594	20236
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Adult-onset mitochondrial movement disorders: a national picture from the Italian Network. Journal of Neurology, 2022, 269, 1413-1421.	1.8	10
2	POLR3A variants in hereditary spastic paraparesis and ataxia: clinical, genetic, and neuroradiological findings in a cohort of Italian patients. Neurological Sciences, 2022, 43, 1071-1077.	0.9	8
3	Episodic ataxia and severe infantile phenotype in spinocerebellar ataxia type 14: expansion of the phenotype and novel mutations. Journal of Neurology, 2022, 269, 1476-1484.	1.8	7
4	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	1.8	12
5	Biâ€allelic variants in <scp>MDH2</scp>: Expanding the clinical phenotype. Clinical Genetics, 2022, 101, 260-264.	1.0	4
6	Correlation among maternal risk factors, gene methylation and disease severity in females with autism spectrum disorder. Epigenomics, 2022, 14, 175-185.	1.0	5
7	Clinical-Genetic Features Influencing Disability in Spastic Paraplegia Type 4. Neurology: Genetics, 2022, 8, e664.	0.9	9
8	Automatic Recognition of Ragged Red Fibers in Muscle Biopsy from Patients with Mitochondrial Disorders. Healthcare (Switzerland), 2022, 10, 574.	1.0	2
9	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study. Acta Neuropathologica Communications, 2022, 10, 54.	2.4	3
10	Iron-sensitive MR imaging of the primary motor cortex to differentiate hereditary spastic paraplegia from other motor neuron diseases. European Radiology, 2022, 32, 8058-8064.	2.3	6
11	In vitro study of polydopamine nanoparticles as protective antioxidant agents in fibroblasts derived from ARSACS patients. Biomaterials Science, 2022, 10, 3770-3792.	2.6	10
12	Screening for RFC-1 pathological expansion in late-onset ataxias: a contribution to the differential diagnosis. Journal of Neurology, 2022, 269, 5431-5435.	1.8	4
13	Lysosomal Proteomics Links Disturbances in Lipid Homeostasis and Sphingolipid Metabolism to CLN5 Disease. Cells, 2022, 11, 1840.	1.8	8
14	Multiple sclerosis in patients with hereditary spastic paraplegia: a case report and systematic review. Neurological Sciences, 2022, 43, 5501-5511.	0.9	2
15	Trehalose Treatment in Zebrafish Model of Lafora Disease. International Journal of Molecular Sciences, 2022, 23, 6874.	1.8	9
16	Enzymatic diagnosis of neuronal lipofuscinoses in dried blood spots using substrates for concomitant tandem mass spectrometry and fluorimetry. Journal of Mass Spectrometry, 2021, 56, e4675.	0.7	5
17	Spinocerebellar ataxia type 48: last but not least. Neurological Sciences, 2021, 42, 1577-1577.	0.9	1
18	Leopard-like retinopathy and severe early-onset portal hypertension expand the phenotype of KARS1-related syndrome: a case report. BMC Medical Genomics, 2021, 14, 25.	0.7	2

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19	Protein aggregates and autophagy involvement in a family with a mutation in Z-band alternatively spliced PDZ-motif protein. Neuromuscular Disorders, 2021, 31, 44-51.	0.3	3
20	Phenotypic Definition and Genotype-Phenotype Correlates in PMPCA-Related Disease. Applied Sciences (Switzerland), 2021, 11, 748.	1.3	1
21	Application of a Clinical Workflow May Lead to Increased Diagnostic Precision in Hereditary Spastic Paraplegias and Cerebellar Ataxias: A Single Center Experience. Brain Sciences, 2021, 11, 246.	1.1	10
22	Neuroacanthocytosis Syndromes in an Italian Cohort: Clinical Spectrum, High Genetic Variability and Muscle Involvement. Genes, 2021, 12, 344.	1.0	6
23	Nutraceutical Screening in a Zebrafish Model of Muscular Dystrophy: Gingerol as a Possible Food Aid. Nutrients, 2021, 13, 998.	1.7	12
24	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.	1.8	17
25	Partial Lipodystrophy and LMNA p.R545H Variant. Journal of Clinical Medicine, 2021, 10, 1142.	1.0	1
26	A next generation sequencingâ€based analysis of a large cohort of ataxic patients refines the clinical spectrum associated with spinocerebellar ataxia 21. European Journal of Neurology, 2021, 28, 2784-2788.	1.7	6
27	Proximal weakness involvement in the first Italian case of Charcotâ€Marieâ€Tooth 2CC harboring a novel frameshift variant in NEFH. Journal of the Peripheral Nervous System, 2021, 26, 231-234.	1.4	7
28	Focal status and acute encephalopathy in a 13-year-old boy with de novo DNM1L mutation: Video-polygraphic pattern and clues for differential diagnosis. Brain and Development, 2021, 43, 644-651.	0.6	5
29	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
30	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063.	1.0	8
31	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. Brain, 2021, 144, 3020-3035.	3.7	11
32	Bi-allelic variants in MTMR5/SBF1 cause Charcot-Marie-Tooth type 4B3 featuring mitochondrial dysfunction. BMC Medical Genomics, 2021, 14, 157.	0.7	2
33	The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. Frontiers in Neurology, 2021, 12, 677551.	1.1	15
34	Learning from massive testing of mitochondrial disorders: UPD explaining unorthodox transmission. Journal of Medical Genetics, 2021, 58, 543-546.	1.5	4
35	Neuroimaging patterns in paediatric onset hereditary spastic paraplegias. Journal of the Neurological Sciences, 2021, 425, 117441.	0.3	4
36	The Diagnostic Approach to Mitochondrial Disorders in Children in the Era of Next-Generation Sequencing: A 4-Year Cohort Study. Journal of Clinical Medicine, 2021, 10, 3222.	1.0	4

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37	Gordon Holmes syndrome caused by two novel mutations in the PNPLA6 gene. Clinical Neurology and Neurosurgery, 2021, 207, 106763.	0.6	7
38	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	1.8	12
39	Efficient Neuroprotective Rescue of Sacsin-Related Disease Phenotypes in Zebrafish. International Journal of Molecular Sciences, 2021, 22, 8401.	1.8	7
40	Tackling Dysfunction of Mitochondrial Bioenergetics in the Brain. International Journal of Molecular Sciences, 2021, 22, 8325.	1.8	5
41	Expanding the clinical and genetic spectrum of pathogenic variants in <scp>STIM1</scp>. Muscle and Nerve, 2021, 64, 567-575.	1.0	7
42	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	4.2	13
43	High-throughput imaging of ATG9A distribution as a diagnostic functional assay for adaptor protein complex 4-associated hereditary spastic paraplegia. Brain Communications, 2021, 3, fcab221.	1.5	11
44	Kir4.1 Dysfunction in the Pathophysiology of Depression: A Systematic Review. Cells, 2021, 10, 2628.	1.8	4
45	Evaluation of the therapeutic potential of resveratrol-loaded nanostructured lipid carriers on autosomal recessive spastic ataxia of Charlevoix-Saguenay patient-derived fibroblasts. Materials and Design, 2021, 209, 110012.	3.3	6
46	Reconsidering NMIHBA Core Features: Macrocephaly Is Not a So Unusual Sign in PRUNE1-Related Encephalopathy. Journal of Pediatric Neurology, 2021, 19, 116-123.	0.0	1
47	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.	1.2	16
48	Adult onset cerebellar ataxia due to novel mutations in BRAT1. Journal of the Neurological Sciences, 2021, 429, 118261.	0.3	0
49	The role of the neurologist in the diagnostic route of HSP and cerebellar ataxias in the next generation sequencing era: A single center experience. Journal of the Neurological Sciences, 2021, 429, 118282.	0.3	0
50	Assessment of Sacsin Turnover in Patients With ARSACS. Neurology, 2021, 97, e2315-e2327.	1.5	11
51	Proximal weakness involvement in the first Italian case of Charcot-Marie-Tooth 2CC harboring a novel frameshift variant in NEFH. Journal of the Neurological Sciences, 2021, 429, 118392.	0.3	0
52	Clinical and genetic features of a large cohort of Italian SPG4 patients from the D.A.I.S.Y. collaborative network. Journal of the Neurological Sciences, 2021, 429, 118251.	0.3	0
53	Mitochondrial neuro-gastro-intestinal encephalomyopathy: A case report. Journal of the Neurological Sciences, 2021, 429, 119371.	0.3	0
54	Oxytocin Receptor Gene Polymorphism in Lactating Dogs. Animals, 2021, 11, 3099.	1.0	5

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55	Spinal Nerve Roots Abnormalities on MRI in a Child with SURF1 Mitochondrial Disease. Neuropediatrics, 2021, , .	0.3	0
56	Identification of a Thyroid Hormone Derivative as a Pleiotropic Agent for the Treatment of Alzheimerâ€™s Disease. Pharmaceuticals, 2021, 14, 1330.	1.7	6
57	Intrafamilial â€œDOAâ€plusâ€•phenotype variability related to different OMI/HTRA2 expression. American Journal of Medical Genetics, Part A, 2020, 182, 176-182.	0.7	2
58	The complex phenotype of spinocerebellar ataxia type 48 in eight unrelated Italian families. European Journal of Neurology, 2020, 27, 498-505.	1.7	44
59	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.	1.4	45
60	SPG8 mutations in Italian families: clinical data and literature review. Neurological Sciences, 2020, 41, 699-703.	0.9	9
61	Customized multigene panels in epilepsy: the best things come in small packages. Neurogenetics, 2020, 21, 1-18.	0.7	9
62	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96.	0.7	14
63	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29
64	Multimodal evaluation of an Italian family with a hereditary spastic paraplegia and POLR3A mutations. Annals of Clinical and Translational Neurology, 2020, 7, 2326-2331.	1.7	4
65	Impaired flickering of the permeability transition pore causes SPG7 spastic paraplegia. EBioMedicine, 2020, 61, 103050.	2.7	28
66	STUB1 â€Related Ataxias: A Challenging Diagnosis. Movement Disorders Clinical Practice, 2020, 7, 733-734.	0.8	4
67	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	1.1	35
68	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genetics in Medicine, 2020, 22, 1851-1862.	1.1	30
69	Tumor Suppressor Role of hsa-miR-193a-3p and -5p in Cutaneous Melanoma. International Journal of Molecular Sciences, 2020, 21, 6183.	1.8	16
70	Evaluation of Chromosome Microarray Analysis in a Large Cohort of Females with Autism Spectrum Disorders: A Single Center Italian Study. Journal of Personalized Medicine, 2020, 10, 160.	1.1	9
71	Protein Delivery by Peptide-Based Stealth Liposomes: A Biomolecular Insight into Enzyme Replacement Therapy. Molecular Pharmaceutics, 2020, 17, 4510-4521.	2.3	10
72	Docosahexaenoic acid in ARSACS: observations in two patients. BMC Neurology, 2020, 20, 215.	0.8	8

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73	New AARS2 Mutations in Two Siblings With Tremor, Downbeat Nystagmus, and Primary Amenorrhea: A Benign Phenotype Without Leukoencephalopathy. Movement Disorders Clinical Practice, 2020, 7, 684-687.	0.8	8
74	Ataxia-myoclonus syndrome due to a novel homozygous ATP13A2 mutation. Parkinsonism and Related Disorders, 2020, 76, 42-43.	1.1	7
75	Development of Nanostructured Lipid Carriers for the Delivery of Idebenone in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. ACS Omega, 2020, 5, 12451-12466.	1.6	16
76	Rare phenotype of ALS4 associated with heterozygous missense mutation c.5842Aâ€‰>â€‰G/p.M1948V in helicase domain of SETX gene. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 312-313.	1.1	1
77	Expansion of the genetic landscape of ERLIN2â€related disorders. Annals of Clinical and Translational Neurology, 2020, 7, 573-578.	1.7	12
78	Cerebello-Cortical Alterations Linked to Cognitive and Social Problems in Patients With Spastic Paraplegia Type 7: A Preliminary Study. Frontiers in Neurology, 2020, 11, 82.	1.1	13
79	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	1.1	49
80	Proteomic and functional analyses in disease models reveal CLN5 protein involvement in mitochondrial dysfunction. Cell Death Discovery, 2020, 6, 18.	2.0	23
81	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	1.1	18
82	Of cognition and cerebellum in SCA48. Neurogenetics, 2020, 21, 145-146.	0.7	4
83	Optic Atrophy and Generalized Chorea in a Patient Harboring an OPA10/RTN4IP1 Pathogenic Variant. Neuropediatrics, 2020, 51, 425-429.	0.3	4
84	MYH2 myopathy, a new case expands the clinical and pathological spectrum of the recessive form. Molecular Genetics & Genomic Medicine, 2020, 8, e1320.	0.6	10
85	A new paraplegin mutation in a patient with primary progressive multiple sclerosis. Multiple Sclerosis and Related Disorders, 2020, 44, 102302.	0.9	3
86	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	0.9	2
87	Distal motor neuropathy associated with novel EMILIN1 mutation. Neurobiology of Disease, 2020, 137, 104757.	2.1	6
88	Expanding the clinical and genetic heterogeneity of SPAX5. Annals of Clinical and Translational Neurology, 2020, 7, 595-601.	1.7	11
89	Evolution of Epileptiform Activity in Zebrafish by Statistical-Based Integration of Electrophysiology and 2-Photon Ca2+ Imaging. Cells, 2020, 9, 769.	1.8	12
90	Biâ€allelic mutations in HARS1 severely impair histidylâ€tRNA synthetase expression and enzymatic activity causing a novel multisystem ataxic syndrome. Human Mutation, 2020, 41, 1232-1237.	1.1	15

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91	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.	2.8	52
92	Spinocerebellar ataxia type 48: last but not least. Neurological Sciences, 2020, 41, 2423-2432.	0.9	31
93	A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. Neurobiology of Disease, 2020, 141, 104880.	2.1	29
94	Loss of ap4s1 in zebrafish leads to neurodevelopmental defects resembling spastic paraplegia 52. Annals of Clinical and Translational Neurology, 2020, 7, 584-589.	1.7	15
95	A novel mutation of Twinkle in Perrault syndrome: A not rare diagnosis?. Annals of Human Genetics, 2020, 84, 417-422.	0.3	7
96	Genetics Influences Drug Consumption in Medication Overuse Headache, Not in Migraine: Evidence From Wolframin His611Arg Polymorphism Analysis. Frontiers in Neurology, 2020, 11, 599517.	1.1	1
97	Functional Network Profiles in ARSACS Disclosed by Aptamer-Based Proteomic Technology. Frontiers in Neurology, 2020, 11, 603774.	1.1	9
98	Social Preference Tests in Zebrafish: A Systematic Review. Frontiers in Veterinary Science, 2020, 7, 590057.	0.9	46
99	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
100	Electrophysiological Profile Remodeling via Selective Suppression of Voltage-Gated Currents by CLN1/PPT1 Overexpression in Human Neuronal-Like Cells. Frontiers in Cellular Neuroscience, 2020, 14, 569598.	1.8	5
101	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. Acta Myologica, 2020, 39, 57-66.	1.5	24
102	Alpha-sarcoglycanopathy presenting as myalgia and hyperCKemia in two adults with a long-term follow-up. Case reports. Acta Myologica, 2020, 39, 218-221.	1.5	1
103	Prevalence and phenotype of the c.1529C>T <scp>SPG</scp>7 variant in adultâ€onset cerebellar ataxia in Italy. European Journal of Neurology, 2019, 26, 80-86.	1.7	12
104	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.	4.3	41
105	Functional Transcriptome Analysis in ARSACS KO Cell Model Reveals a Role of Sacsin in Autophagy. Scientific Reports, 2019, 9, 11878.	1.6	26
106	Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. Journal of Neurology, 2019, 266, 2657-2664.	1.8	19
107	Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. Neurogenetics, 2019, 20, 165-172.	0.7	8
108	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism and Related Disorders, 2019, 68, 8-16.	1.1	25

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109	P.110Clinical, morphological and genetic data in Italian patients with fiber-type-disproportion. Neuromuscular Disorders, 2019, 29, S80-S81.	0.3	0
110	Overt Hypogonadism May Not Be a Sentinel Sign of RING Finger Protein 216: Two Novel Mutations Associated with Ataxia, Chorea, and Fertility. Movement Disorders Clinical Practice, 2019, 6, 724-726.	0.8	12
111	Management of Hereditary Spastic Paraplegia: A Systematic Review of the Literature. Frontiers in Neurology, 2019, 10, 3.	1.1	47
112	Clinical and molecular studies in two new cases of ARSACS. Neurogenetics, 2019, 20, 45-49.	0.7	15
113	Muscle pain in mitochondrial diseases: a picture from the Italian network. Journal of Neurology, 2019, 266, 953-959.	1.8	9
114	VARS2-linked mitochondrial encephalopathy: two case reports enlarging the clinical phenotype. BMC Medical Genetics, 2019, 20, 77.	2.1	8
115	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.	1.1	43
116	A Novel CAPN1 Mutation Causes a Pure Hereditary Spastic Paraplegia in an Italian Family. Frontiers in Neurology, 2019, 10, 580.	1.1	14
117	Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	1.0	29
118	Fishing in the Cell Powerhouse: Zebrafish as A Tool for Exploration of Mitochondrial Defects Affecting the Nervous System. International Journal of Molecular Sciences, 2019, 20, 2409.	1.8	16
119	The features of the m.10197G>A mtDNA mutation. Journal of the Neurological Sciences, 2019, 400, 184-185.	0.3	0
120	Degenerative and acquired sporadic adult onset ataxia. Neurological Sciences, 2019, 40, 1335-1342.	0.9	26
121	Complex multisystem phenotype associated with the mitochondrial DNA m.5522G>A mutation. Neurological Sciences, 2019, 40, 1705-1708.	0.9	10
122	The genetic etiology in cerebral palsy mimics: The results from a Greek tertiary care center. European Journal of Paediatric Neurology, 2019, 23, 427-437.	0.7	26
123	Clinical and neuroimaging features of the m.10197G>A mtDNA mutation: New case reports and expansion of the phenotype variability. Journal of the Neurological Sciences, 2019, 399, 69-75.	0.3	8
124	MRIndex: A tool for evaluating muscle involvement in neuromuscular diseases from MRI images. , 2019, , .		3
125	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	0.9	31
126	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.	1.7	63

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127	NeuroExam: a tool for neurological examination in neuromuscular diseases. , 2019, , .		4
128	Swimming in Deep Water: Zebrafish Modeling of Complicated Forms of Hereditary Spastic Paraplegia and Spastic Ataxia. Frontiers in Neuroscience, 2019, 13, 1311.	1.4	14
129	Kufs disease due to mutation ofCLN6: clinical, pathological and molecular genetic features. Brain, 2019, 142, 59-69.	3.7	28
130	Italian recommendations for diagnosis and management of congenital myasthenic syndromes. Neurological Sciences, 2019, 40, 457-468.	0.9	24
131	Autophagic vacuolar myopathy caused by a CLN3 mutation. A case report. Neuromuscular Disorders, 2019, 29, 67-69.	0.3	1
132	Progressive myoclonus epilepsy and ceroidolipofuscinosis 14: The multifaceted phenotypic spectrum of KCTD7-related disorders. European Journal of Medical Genetics, 2019, 62, 103591.	0.7	15
133	SCN11A variant as possible pain generator in sensory axonal neuropathy. Neurological Sciences, 2019, 40, 1295-1297.	0.9	6
134	Teaching NeuroImages: Leigh-like features expand the picture of PMPCA-related disorders. Neurology, 2019, 92, e168-e169.	1.5	6
135	Late adult-onset adrenomyeloneuropathy evolving with atypical severe frontal lobe syndrome: Importance of neuroimaging. Radiology Case Reports, 2019, 14, 309-314.	0.2	2
136	PhysioTest: A Dedicated Module to Collect Data from Physiotherapy Assessments in Neuromuscular Diseases. Biosystems and Biorobotics, 2019, , 805-809.	0.2	3
137	A Novel Approach to Gene Analysis: Gene Panels and Cluster Definition to Assist Genotyping Patients with Congenital Myopathies. , 2019, , .		2
138	Myoclonus epilepsy, retinitis pigmentosa, leukoencephalopathy and cerebral calcifications associated with a novel m.5513G>A mutation in the MT-TW gene. Biochemical and Biophysical Research Communications, 2018, 500, 158-162.	1.0	5
139	A V1143F mutation in the neuronal-enriched isoform 2 of the PMCA pump is linked with ataxia. Neurobiology of Disease, 2018, 115, 157-166.	2.1	15
140	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.3	24
141	Clinical and neuroimaging features of autosomal recessive spastic paraplegia 35 (SPG35): case reports, new mutations, and brief literature review. Neurogenetics, 2018, 19, 123-130.	0.7	29
142	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	4.5	69
143	A novel homozygous MFN2 mutation associated with severe and atypical CMT2 phenotype. European Journal of Paediatric Neurology, 2018, 22, 563-567.	0.7	21
144	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. Human Molecular Genetics, 2018, 27, 1892-1904.	1.4	29

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145	CYP2U1 activity is altered by missense mutations in hereditary spastic paraplegia 56. Human Mutation, 2018, 39, 140-151.	1.1	19
146	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.	0.7	54
147	DUCHENNE MUSCULAR DYSTROPHY - GENETICS. Neuromuscular Disorders, 2018, 28, S97.	0.3	0
148	InGene: a multimodal approach to the genotype-phenotype association in neuromuscular diseases. , 2018, , .		1
149	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	1.1	64
150	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 170.	1.2	26
151	CLN8 is an endoplasmic reticulum cargo receptor that regulates lysosome biogenesis. Nature Cell Biology, 2018, 20, 1370-1377.	4.6	80
152	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	1.0	7
153	Reversible Valproate-Induced Subacute Encephalopathy Associated With a MT-ATP8 Variant in the Mitochondrial Genome. Frontiers in Neurology, 2018, 9, 728.	1.1	13
154	Predictors of survival in spinocerebellar ataxia type 2 population from Southern Italy. Neurological Sciences, 2018, 39, 1857-1860.	0.9	6
155	Understanding Spreading Depression from Headache to Sudden Unexpected Death. Frontiers in Neurology, 2018, 9, 19.	1.1	51
156	Relapsing-Remitting Course of Cystic Leukoencephalopathy. Pediatric Neurology, 2018, 89, 63-65.	1.0	4
157	Cross-Linked Enzyme Aggregates as Versatile Tool for Enzyme Delivery: Application to Polymeric Nanoparticles. Bioconjugate Chemistry, 2018, 29, 2225-2231.	1.8	34
158	C09â€…SCAS genes as disease modifiers in huntingtonâ€™s disease. , 2018, , .		0
159	Fast Progression of Cerebellar Atrophy in PLA2G6-Associated Infantile Neuronal Axonal Dystrophy. Cerebellum, 2017, 16, 742-745.	1.4	6
160	Loss of spatacsin function alters lysosomal lipid clearance leading to upper and lower motor neuron degeneration. Neurobiology of Disease, 2017, 102, 21-37.	2.1	85
161	Concentric muscle involvement in POLG -related distal myopathy. Neuromuscular Disorders, 2017, 27, 500-501.	0.3	2
162	SPG2 mimicking multiple sclerosis in a family identified using next generation sequencing. Journal of the Neurological Sciences, 2017, 375, 198-202.	0.3	18

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163	Sporadic chronic progressive external ophthalmoplegia with single large mitochondrial DNA deletion and neurogenic findings. Journal of Neurology, 2017, 264, 597-599.	1.8	2
164	Widening the Heterogeneity of Leigh Syndrome: Clinical, Biochemical, and Neuroradiologic Features in a Patient Harboring a NDUFA10 Mutation. JIMD Reports, 2017, 37, 37-43.	0.7	13
165	Late onset of neutral lipid storage disease due to novel PNPLA2 mutations causing total loss of lipase activity in a patient with myopathy and slight cardiac involvement. Neuromuscular Disorders, 2017, 27, 481-486.	0.3	21
166	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1. European Journal of Paediatric Neurology, 2017, 21, 671-677.	0.7	25
167	Mutations in GMPPB Presenting with Pseudometabolic Myopathy. JIMD Reports, 2017, 38, 23-31.	0.7	8
168	Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. Neurology, 2017, 88, 2132-2140.	1.5	41
169	Diagnostic methods and emerging treatments for adult neuronal ceroid lipofuscinoses (Kufs disease). Expert Opinion on Orphan Drugs, 2017, 5, 487-501.	0.5	2
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