Gabriella Silvestri

List of Publications by Year
in descending order

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159	5,859	81900	82547
papers	citations	39 h-index	72 g-index

171	171	171	6015
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	MELAS: Clinical features, biochemistry, and molecular genetics. Annals of Neurology, 1992, 31, 391-398.	5.3	514
2	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Neurology, 1994, 44, 721-721.	1.1	402
3	Progress in the molecular diagnosis of facioscapulohumeral muscular dystrophy and correlation between the number ofKpnI repeats at the 4q35 locus and clinical phenotype. Annals of Neurology, 1999, 45, 751-757.	5.3	263
4	Atypical clinical presentations associated with the MELAS mutation at position 3243 of human mitochondrial DNA. Neuromuscular Disorders, 1993, 3, 43-50.	0.6	219
5	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. FASEB Journal, 2007, 21, 1210-1226.	0.5	209
6	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
7	A new mtDNA mutation in the tRNA(Lys) gene associated with myoclonic epilepsy and ragged-red fibers (MERRF). American Journal of Human Genetics, 1992, 51, 1213-7.	6.2	179
8	A new mtDNA mutation in the tRNALeu(UUR) gene associated with maternally inherited cardiomyopathy. Human Mutation, 1994, 3, 37-43.	2.5	178
9	Coenzyme Q ₁₀ reverses pathological phenotype and reduces apoptosis in familial CoQ ₁₀ deficiency. Neurology, 2001, 57, 515-518.	1.1	157
10	Characterization of the Pattern of Cognitive Impairment in Myotonic Dystrophy Type 1. Archives of Neurology, 2004, 61, 1943-7.	4.5	147
11	Fatal infantile liver failure associated with mitochondrial DNA depletion. Journal of Pediatrics, 1992, 121, 896-901.	1.8	123
12	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. Neurology, 1991, 41, 1053-1053.	1.1	120
13	Apoptosis in mitochondrial encephalomyopathies with mitochondrial DNA mutations: a potential pathogenic mechanism. Brain, 2000, 123, 93-104.	7.6	117
14	Cardiac features of Emeryâ€“Dreifuss muscular dystrophy caused by lamin A/C gene mutations. European Heart Journal, 2003, 24, 2227-2236.	2.2	103
15	Widespread tissue distribution of a tRNA ^{Leu(UUR)} mutation in the mitochondrial DNA of a patient with MELAS syndrome. Neurology, 1991, 41, 1663-1663.	1.1	100
16	Abnormal brain and muscle energy metabolism shown by 31P-MRS in familial hemiplegic migraine. Journal of the Neurological Sciences, 1995, 129, 214-222.	0.6	82
17	Cognitive impairment in myotonic dystrophy type 1 (DM1). Journal of Neurology, 2008, 255, 1737-1742.	3.6	76
18	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64

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19	Novel mutations in SPG11 cause hereditary spastic paraplegia associated with earlyâ€onset levodopaâ€responsive Parkinsonism. Movement Disorders, 2011, 26, 553-556.	3.9	62
20	Abnormal Functional Brain Connectivity and Personality Traits in Myotonic Dystrophy Type 1. JAMA Neurology, 2014, 71, 603.	9.0	62
21	Prevalence and clinical correlates of sleep disordered breathing in myotonic dystrophy types 1 and 2. Sleep and Breathing, 2014, 18, 579-589.	1.7	58
22	Muscle imaging findings in GNE myopathy. Journal of Neurology, 2012, 259, 1358-1365.	3.6	57
23	A Novel Mitochondrial DNA Point Mutation in the tRNAIleGene Is Associated with Progressive External Ophtalmoplegia. Biochemical and Biophysical Research Communications, 1996, 220, 623-627.	2.1	54
24	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
25	Homozygosity mapping of Marinescoâ€“SjÃ¶gren syndrome to 5q31. European Journal of Human Genetics, 2003, 11, 770-778.	2.8	53
26	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
27	NovelGNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy. Human Mutation, 2004, 23, 632-632.	2.5	52
28	Myotonic dystrophy type 1: role of <scp>CCG</scp>, <scp>CTC</scp> and <scp>CGG</scp> interruptions within <scp>DMPK</scp> alleles in the pathogenesis and molecular diagnosis. Clinical Genetics, 2017, 92, 355-364.	2.0	52
29	GCG genetic expansions in Italian patients with oculopharyngeal muscular dystrophy. Neurology, 2000, 54, 608-608.	1.1	51
30	Maternally inherited cardiomyopathy: A new phenotype associated with the A to G at nt.3243 of mitochondrial DNA (MELAS mutation). , 1997, 20, 221-225.		50
31	Apoptosis and ROS Detoxification Enzymes Correlate with Cytochrome c Oxidase Deficiency in Mitochondrial Encephalomyopathies. Molecular and Cellular Neurosciences, 2001, 17, 696-705.	2.2	50
32	â€œI Know that You Know that I Knowâ€: Neural Substrates Associated with Social Cognition Deficits in DM1 Patients. PLoS ONE, 2016, 11, e0156901.	2.5	50
33	Ekbom's syndrome: Lipomas, ataxia, and neuropathy with MERRF. Muscle and Nerve, 1994, 17, 943-945.	2.2	49
34	Validation of plasma microRNAs as biomarkers for myotonic dystrophy type 1. Scientific Reports, 2016, 6, 38174.	3.3	49
35	A new mtDNA mutation associated with a progressive encephalopathy and cytochrome c oxidase deficiency. Neurology, 2000, 54, 1693-1696.	1.1	48
36	Ophthalmoplegia, demyelinating neuropathy, leukoencephalopathy, myopathy, and gastrointestinal dysfunction with multiple deletions of mitochondrial DNA: A mitochondrial multisystem disorder in search of a name. Muscle and Nerve, 1994, 17, 667-674.	2.2	46

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37	Implicit and explicit aspects of sequence learning in pre-symptomatic Huntington's disease. Parkinsonism and Related Disorders, 2008, 14, 457-464.	2.2	45
38	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45
39	The complex phenotype of spinocerebellar ataxia type 48 in eight unrelated Italian families. European Journal of Neurology, 2020, 27, 498-505.	3.3	44
40	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
41	Prevalence of spinocerebellar ataxia type 2 mutation among Italian Parkinsonian patients. Movement Disorders, 2007, 22, 324-327.	3.9	42
42	New phenotype and pathology features in MYH7-related distal myopathy. Neuromuscular Disorders, 2012, 22, 640-647.	0.6	41
43	Alternative splicing of human insulin receptor gene (INSR) in type I and type II skeletal muscle fibers of patients with myotonic dystrophy type 1 and type 2. Molecular and Cellular Biochemistry, 2013, 380, 259-265.	3.1	41
44	Molecular, clinical, and muscle studies in myotonic dystrophy type 1 (DM1) associated with novel variant CCG expansions. Journal of Neurology, 2013, 260, 1245-1257.	3.6	41
45	A late-onset mitochondrial myopathy is associated with a novel mitochondrial DNA (mtDNA) point mutation in the tRNATrp gene. Neuromuscular Disorders, 1998, 8, 291-295.	0.6	39
46	Risk of Arrhythmias in MYotonic Dystrophy: trial design of the RAMYD study. Journal of Cardiovascular Medicine, 2009, 10, 51-58.	1.5	37
47	An Age-Standardized Prevalence Estimate and a Sex and Age Distribution of Myotonic Dystrophy Types 1 and 2 in the Rome Province, Italy. Neuroepidemiology, 2016, 46, 191-197.	2.3	37
48	Dysregulation of Circular RNAs in Myotonic Dystrophy Type 1. International Journal of Molecular Sciences, 2019, 20, 1938.	4.1	37
49	Sporadic late-onset nemaline myopathy: clinical, pathology and imaging findings in a single center cohort. Journal of Neurology, 2018, 265, 542-551.	3.6	36
50	Alternative splicing alterations of <scp>Ca</scp>²⁺ handling genes are associated with <scp>Ca</scp>²⁺ signal dysregulation in myotonic dystrophy type 1 (<scp>DM</scp>1) and type 2 (<scp>DM</scp>2) myotubes. Neuropathology and Applied Neurobiology, 2014, 40, 464-476.	3.2	35
51	<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
52	Increased risk of tumor in DM1 is not related to exposure to common lifestyle risk factors. Journal of Neurology, 2016, 263, 492-498.	3.6	32
53	Muscle <scp>MRI</scp> in female carriers of dystrophinopathy. European Journal of Neurology, 2012, 19, 1256-1260.	3.3	31
54	Expansion size and presence of CCG/CTC/CGG sequence interruptions in the expanded CTG array are independently associated to hypermethylation at the DMPK locus in myotonic dystrophy type 1 (DM1). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 2645-2652.	3.8	31

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55	Prevalence and predictor factors of respiratory impairment in a large cohort of patients with Myotonic Dystrophy type 1 (DM1): A retrospective, cross sectional study. Journal of the Neurological Sciences, 2019, 399, 118-124.	0.6	31
56	Substrate reduction therapy with miglustat in chronic GM2 gangliosidosis type Sandhoff: results of a 3â€year followâ€up. Journal of Inherited Metabolic Disease, 2010, 33, 355-361.	3.6	30
57	An Italian Neurology Outpatient Clinic Facing SARS-CoV-2 Pandemic: Data From 2,167 Patients. Frontiers in Neurology, 2020, 11, 564.	2.4	30
58	A novel KIF5A/SPG10 mutation in spastic paraplegia associated with axonal neuropathy. Journal of Neurology, 2008, 255, 1090-1092.	3.6	29
59	Brain Connectomicsâ€™ Modification to Clarify Motor and Nonmotor Features of Myotonic Dystrophy Type 1. Neural Plasticity, 2016, 2016, 1-10.	2.2	28
60	MELAS point mutation with unusual clinical presentation. Neuromuscular Disorders, 1993, 3, 191-193.	0.6	27
61	An Italian family with autosomal recessive inclusion-body myopathy and mutations in the GNE gene. Neurology, 2002, 59, 1808-1809.	1.1	27
62	Chronic autoimmune autonomic neuropathy responsive to immunosuppressive therapy. Neurology, 2007, 68, 161-162.	1.1	27
63	Successful Treatment of Acute Autoimmune Limbic Encephalitis With Negative VGKC and NMDAR Antibodies. Cognitive and Behavioral Neurology, 2009, 22, 63-66.	0.9	25
64	Dysplastic nevi, cutaneous melanoma, and other skin neoplasms in patients with myotonic dystrophy type 1: A cross-sectional study. Journal of the American Academy of Dermatology, 2015, 72, 85-91.	1.2	25
65	Manifesting heterozygotes in McArdle's disease: clinical, morphological and biochemical studies in a family. Journal of the Neurological Sciences, 1993, 115, 91-94.	0.6	24
66	Hereditary spastic paraplegia: Novel mutations and expansion of the phenotype variability in SPG56. European Journal of Paediatric Neurology, 2016, 20, 444-448.	1.6	22
67	Single-fiber PCR in MELAS3243 patients: Correlations between intratissue distribution and phenotypic expression of the mtDNAA3243G genotype. American Journal of Medical Genetics Part A, 2000, 94, 201-206.	2.4	21
68	Low-Rate Repetitive Nerve Stimulation Protocol in an Italian Cohort of Patients Affected by Recessive Myotonia Congenita. Journal of Clinical Neurophysiology, 2011, 28, 39-44.	1.7	21
69	Abnormal Cortical Thickness Is Associated With Deficits in Social Cognition in Patients With Myotonic Dystrophy Type 1. Frontiers in Neurology, 2020, 11, 113.	2.4	21
70	Prefrontal cortex controls human balance during overground ataxic gait. Restorative Neurology and Neuroscience, 2012, 30, 397-405.	0.7	18
71	Prefrontal cortex as a compensatory network in ataxic gait: A correlation study between cortical activity and gait parameters. Restorative Neurology and Neuroscience, 2015, 33, 177-187.	0.7	18
72	Spastic paraplegia with thinning of the corpus callosum and white matter abnormalities: Further mutations and relative frequency in ZFYVE26/SPG15 in the Italian population. Journal of the Neurological Sciences, 2009, 277, 22-25.	0.6	17

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73	Severe 5,10â€methylenetetrahydrofolate reductase deficiency: a rare, treatable cause of complicated hereditary spastic paraplegia. European Journal of Neurology, 2018, 25, 602-605.	3.3	17
74	Analysis of MTMR1 expression and correlation with muscle pathological features in juvenile/adult onset myotonic dystrophy type 1 (DM1) and in myotonic dystrophy type 2 (DM2). Experimental and Molecular Pathology, 2010, 89, 158-168.	2.1	16
75	Myotonic dystrophy type 1 and de novo FSHD mutation double trouble: A clinical and muscle MRI study. Neuromuscular Disorders, 2013, 23, 427-431.	0.6	16
76	Dystrophin is not essential for the integrity of the cytoskeleton. Acta Neuropathologica, 1994, 87, 377-384.	7.7	15
77	Pathogenic role of mtDNA duplications in mitochondrial diseases associated with mtDNA deletions. American Journal of Medical Genetics Part A, 2003, 118A, 247-254.	2.4	15
78	Somatic mosaicism in TPM2-related myopathy with nemaline rods and cap structures. Acta Neuropathologica, 2013, 125, 169-171.	7.7	15
79	A channelopathy mutation in the voltage-sensor discloses contributions of a conserved phenylalanine to gating properties of Kv1.1 channels and ataxia. Scientific Reports, 2017, 7, 4583.	3.3	15
80	Chronic GM2 gangliosidosis type Sandhoff associated with a novel missense HEXB gene mutation causing a double pathogenic effect. Molecular Genetics and Metabolism, 2007, 91, 111-114.	1.1	14
81	A case of CMT 1B due to Val 102/fs null mutation of the MPZ gene presenting as hyperCKemia. Clinical Neurology and Neurosurgery, 2010, 112, 794-797.	1.4	13
82	Frontotemporal dementia, Parkinsonism and lower motor neuron involvement in a patient with C9ORF72 expansion. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 66-69.	1.7	13
83	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.	2.4	13
84	Mitochondrial DNA deletions in oculopharyngeal muscular dystrophy. FEBS Letters, 1997, 418, 167-170.	2.8	12
85	Homozygosity for c 6325T>G transition in the ATM gene causes an atypical, late-onset variant form of ataxia-telangiectasia. Journal of Neurology, 2010, 257, 1738-1740.	3.6	12
86	Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 917-924.	3.8	12
87	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.	3.3	12
88	High Prevalence and Gender-Related Differences of Gastrointestinal Manifestations in a Cohort of DM1 Patients: A Perspective, Cross-Sectional Study. Frontiers in Neurology, 2020, 11, 394.	2.4	12
89	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12
90	Immunocytochemical localization of vinculin in muscle and nerve. Muscle and Nerve, 1995, 18, 1277-1284.	2.2	11

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91	SIADH in a patient with sensory ataxic neuropathy with anti-disialosyl antibodies (CANOMAD). Journal of Neurology, 2009, 256, 1177-1179.	3.6	11
92	fNIRS evaluation during a phonemic verbal task reveals prefrontal hypometabolism in patients affected by myotonic dystrophy type 1. Clinical Neurophysiology, 2013, 124, 2269-2276.	1.5	11
93	A novel nonsense EIF1AX mutation identified in a thyroid nodule histologically diagnosed as oncocytic carcinoma. Endocrine, 2018, 62, 492-495.	2.3	11
94	Clinical features and outcome of patients with autoimmune cerebellar ataxia evaluated with the Scale for the Assessment and Rating of Ataxia. European Journal of Neurology, 2022, 29, 564-572.	3.3	11
95	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.	2.3	10
96	Translational control of polyamine metabolism by CNBP is required for Drosophila locomotor function. ELife, 2021, 10, .	6.0	10
97	Muscle magnetic resonance imaging in myotonic dystrophy type 1 (DM1): Refining muscle involvement and implications for clinical trials. European Journal of Neurology, 2022, 29, 843-854.	3.3	10
98	Functional involvement of central nervous system in mitochondrial disorders. Electroencephalography and Clinical Neurophysiology - Electromyography and Motor Control, 1997, 105, 171-180.	1.4	9
99	A novel mutation in the SACS gene associated with a complicated form of spastic ataxia. Journal of Neurology, 2008, 255, 1429-1431.	3.6	9
100	New Wearable System for Step-Counting Telemonitoring and Telerehabilitation Based on the Codivilla Spring. Telemedicine Journal and E-Health, 2008, 14, 1096-1100.	2.8	9
101	Positive outcome in a patient with Wilson's disease treated with reduced zinc dosage in pregnancy. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2011, 159, 237-238.	1.1	9
102	Spectral domain optical coherence tomography findings in myotonic dystrophy. Neuromuscular Disorders, 2020, 30, 144-150.	0.6	9
103	Lesion distribution and substrate of white matter damage in myotonic dystrophy type 1: Comparison with multiple sclerosis. NeuroImage: Clinical, 2021, 29, 102562.	2.7	9
104	Evidence of white matter involvement in SCA 7. Journal of Neurology, 2007, 254, 536-538.	3.6	8
105	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.	1.9	8
106	Novel <scp>SACS</scp> mutations in two unrelated Italian patients with spastic ataxia: clinicoâ€diagnostic characterization and results of serial brain <scp>MRI</scp> studies. European Journal of Neurology, 2012, 19, e77-8.	3.3	7
107	MRI Neurography Findings in Patients with Idiopathic Brachial Plexopathy: Correlations with Clinical-neurophysiological Data in Eight Consecutive Cases. Internal Medicine, 2013, 52, 2031-2039.	0.7	7
108	Restless legs syndrome and daytime sleepiness are prominent in myotonic dystrophy type 2. Neurology, 2014, 82, 283-284.	1.1	7

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109	Ventral tegmental area dysfunction affects decision-making in patients with myotonic dystrophy type-1. Cortex, 2020, 128, 192-202.	2.4	7
110	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.	3.6	7
111	Phase angle and impedance ratio: Two specular ways to analyze body composition. Annals of Clinical Nutrition, 2018, 1, .	0.2	7
112	Toward the Integration of Novel Wearable Step-Counters in Gait Telerehabilitation After Stroke. Telemedicine Journal and E-Health, 2009, 15, 105-111.	2.8	6
113	Clinical characteristics of metabolic associated fatty liver disease (MAFLD) in subjects with myotonic dystrophy type 1 (DM1). Digestive and Liver Disease, 2021, 53, 1451-1457.	0.9	6
114	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.	3.3	6
115	Nuclear Factor Erythroid 2-Related Factor 2 Activation Might Mitigate Clinical Symptoms in Friedreichâ€™s Ataxia: Clues of an â€œOut-Brain Originâ€•of the Disease From a Family Study. Frontiers in Neuroscience, 2021, 15, 638810.	2.8	5
116	Resveratrol corrects aberrant splicing of RYR1 pre-mRNA and Ca²⁺ signal in myotonic dystrophy type 1 myotubes. Neural Regeneration Research, 2020, 15, 1757.	3.0	5
117	Restless legs syndrome and daytime sleepiness are prominent in myotonic dystrophy type 2. Neurology, 2014, 83, 572-573.	1.1	4
118	Response to â€œAutosomal recessive axonal neuropathy caused by HINT1 mutation: New association of a psychiatric disorder to the neurological phenotypeâ€• Neuromuscular Disorders, 2020, 30, 265-266.	0.6	4
119	Caregiversâ€™ and Physiciansâ€™ Perspectives on Alpha-Mannosidosis: A Report from Italy. Advances in Therapy, 2021, 38, 1-10.	2.9	4
120	P2.38 Lower limb muscle MRI in a large cohort of FSHD patients. Neuromuscular Disorders, 2011, 21, 671.	0.6	3
121	Lack of Any Cardiac Involvement in a Patient with Andersen-Tawil Syndrome Associated with the c.574Aâ†'G Mutation in <i>KCNJ2</i>. Cardiology, 2011, 120, 200-203.	1.4	3
122	Teaching Neuro Images : Autosomal dominant leukodystrophy in a sporadic case. Neurology, 2014, 83, e121.	1.1	3
123	NGS-based detection of a novel mutation in PRKCG (SCA14) in sporadic adult-onset ataxia plus dystonic tremor. Neurological Sciences, 2020, 41, 2989-2991.	1.9	3
124	Elevated serum Neurofilament Light chain (NfL) as a potential biomarker of neurological involvement in Myotonic Dystrophy type 1 (DM1). Journal of Neurology, 2022, , .	3.6	3
125	Letter to the editor. Muscle and Nerve, 1995, 18, 478-478.	2.2	2
126	Glycogen Storage Disease Type II Diagnosed in a 74-Year-Old Woman. Journal of the American Geriatrics Society, 2004, 52, 1034-1035.	2.6	2

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127	Do not jump to easy conclusions! Lessons from pitfall in the molecular diagnosis of <scp>ARSACS</scp>. Clinical Genetics, 2014, 86, 396-397.	2.0	2
128	Concentric muscle involvement in POLG -related distal myopathy. Neuromuscular Disorders, 2017, 27, 500-501.	0.6	2
129	Reader response: High frequency of gastrointestinal manifestations in myotonic dystrophy type 1 and type 2. Neurology, 2018, 90, 814.1-814.	1.1	2
130	Secondary hypokalemic periodic paralysis as a rare clinical presentation of Conn syndrome. Clinical Neurophysiology, 2018, 129, 2505-2506.	1.5	2
131	Clinical use of bioelectrical impedance analysis in patients affected by myotonic dystrophy type 1: A cross-sectional study. Nutrition, 2019, 67-68, 110546.	2.4	2
132	Compound heterozygosity for an expanded (GAA) and a (GAAGGA) repeat at FXN locus: from a diagnostic pitfall to potential clues to the pathogenesis of Friedreich ataxia. Neurogenetics, 2020, 21, 279-287.	1.4	2
133	Apoptosis and Oxidative Stress in Mitochondrial Disorders. , 2002, , 37-45.		2
134	Dystrophin is not essential for the integrity of the cytoskeleton. Acta Neuropathologica, 1994, 87, 377-384.	7.7	2
135	Immunohistochemical studies of muscle and nerve in merosin-deficient congenital muscular dystrophy. Neuromuscular Disorders, 1996, 6, S18.	0.6	1
136	Transient MRI Abnormalities in a Case of Occipital Lobe Epilepsy with Favorable Outcome. Clinical EEG and Neuroscience, 2006, 37, 219-222.	1.7	1
137	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay in the Time of Next-Generation Sequencing. Archives of Neurology, 2012, 69, 1661.	4.5	1
138	Serial neuroimaging findings in a novel case of sporadic progressive ataxia and palatal tremor (PAPT). Journal of the Neurological Sciences, 2017, 379, 16-17.	0.6	1
139	Dysautonomia as Onset Symptom of Myotonic Dystrophy Type 2. European Neurology, 2018, 79, 166-170.	1.4	1
140	A unique case of multiphasic ADEM or what else?. Multiple Sclerosis and Related Disorders, 2019, 35, 73-75.	2.0	1
141	A man with sarcoidosis and slurred speech. European Journal of Neurology, 2020, 27, e7-e8.	3.3	1
142	Maternally inherited cardiomyopathy: A new phenotype associated with the A to G at nt.3243 of mitochondrial DNA (MELAS mutation). , 1997, 20, 221.		1
143	The impact of the molecular data on clinical practice in facio-scapulohumeral muscular dystrophy (FSHD). Neuromuscular Disorders, 1997, 7, 443-444.	0.6	0
144	G.P.17.05 Predictive role of NCAM in the identification of patients with HIBM due to GNE mutations with atypical clinical phenotype. Neuromuscular Disorders, 2007, 17, 885.	0.6	0

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145	Subdural hematoma in a young woman with an â€œoldâ€•brain. Acta Neurologica Belgica, 2012, 112, 385-387.	1.1	0
146	P593: fNIRS evaluation during a phonemic verbal task reveals prefrontal hypometabolism in patients affected by myotonic dystrophy type 1. Clinical Neurophysiology, 2014, 125, S209-S210.	1.5	0
147	Clarification on Uveal Melanoma Associated With Myotonic Dystrophy. JAMA Ophthalmology, 2018, 136, 1426.	2.5	0
148	Malnutrition estimate among patients affected by myotonic dystrophy type 1. Phase angle and impedance ratio correlate with disease staging. Clinical Nutrition, 2018, 37, S119.	5.0	0
149	Imaging Features of Varicella Zoster Virus Cranial Multiple Mononeuropathies. European Neurology, 2018, 79, 315-316.	1.4	0
150	Reply to the letter entitled â€œPredictors of respiratory impairment in patients with myotonic dystrophy type 1â€• Journal of the Neurological Sciences, 2019, 403, 166-167.	0.6	0
151	Letter of response to â€œMyotonic dystrophy type 1, individualised respiratory care rather than standart prognosticationâ€• Journal of the Neurological Sciences, 2019, 401, 66.	0.6	0
152	Editorial: Myotonic Dystrophies: Developments in Research From Bench to Bedside. Frontiers in Neurology, 2020, 11, 594836.	2.4	0
153	Myotonic dystrophy type 1 cosegregating with autosomal dominant polycystic kidney disease type 2. Neurological Sciences, 2020, 41, 3761-3763.	1.9	0
154	Unusual case of long survival patient with leptomeningeal carcinomatosis from breast cancer. British Journal of Neurosurgery, 2021, , 1-4.	0.8	0
155	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.	0.6	0
156	Myotonic dystrophy: A new perspective on the treatment of a multisystemic disease. Drugs of the Future, 2010, 35, 237.	0.1	0
157	Remitting-Relapsing Carbamazepine Overdosage Mimicking Vertebrobasilar Transient Ischemic Attacks. International Journal of Case Reports in Medicine, 0, , 1-4.	0.0	0
158	Morphological observations in mitochondrial diseases. Progress in Cell Research, 1995, , 217-221.	0.3	0
159	A clinical and epidemiological prevalence study on Friedreichâ€™s Ataxia in Latium, Italy.. Neuroepidemiology, 0, , .	2.3	0