Gabriella Silvestri

List of Publications by Year
in descending order

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159	5,859	81889	82542
papers	citations	39 h-index	72 g-index

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

#	Article	IF	Citations
1	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
2	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
3	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
4	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
5	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
6	Caregiversâ€™ and Physiciansâ€™ Perspectives on Alpha-Mannosidosis: A Report from Italy. Advances in Therapy, 2021, 38, 1-10.	2.9	4
7	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
8	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
9	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
10	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
11	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
12	Unusual case of long survival patient with leptomeningeal carcinomatosis from breast cancer. British Journal of Neurosurgery, 2021, , 1-4.	0.8	0
13	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12
14	Translational control of polyamine metabolism by CNBP is required for Drosophila locomotor function. ELife, 2021, 10, .	6.0	10
15	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
16	A man with sarcoidosis and slurred speech. European Journal of Neurology, 2020, 27, e7-e8.	3.3	1
17	The complex phenotype of spinocerebellar ataxia type 48 in eight unrelated Italian families. European Journal of Neurology, 2020, 27, 498-505.	3.3	44
18	Spectral domain optical coherence tomography findings in myotonic dystrophy. Neuromuscular Disorders, 2020, 30, 144-150.	0.6	9

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19	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
20	Editorial: Myotonic Dystrophies: Developments in Research From Bench to Bedside. Frontiers in Neurology, 2020, 11, 594836.	2.4	0
21	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
22	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
23	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
24	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
25	An Italian Neurology Outpatient Clinic Facing SARS-CoV-2 Pandemic: Data From 2,167 Patients. Frontiers in Neurology, 2020, 11, 564.	2.4	30
26	Myotonic dystrophy type 1 cosegregating with autosomal dominant polycystic kidney disease type 2. Neurological Sciences, 2020, 41, 3761-3763.	1.9	0
27	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
28	Ventral tegmental area dysfunction affects decision-making in patients with myotonic dystrophy type-1. Cortex, 2020, 128, 192-202.	2.4	7
29	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
30	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
31	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
32	A unique case of multiphasic ADEM or what else?. Multiple Sclerosis and Related Disorders, 2019, 35, 73-75.	2.0	1
33	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
34	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
35	Dysregulation of Circular RNAs in Myotonic Dystrophy Type 1. International Journal of Molecular Sciences, 2019, 20, 1938.	4.1	37
36	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

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37	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
38	Reader response: High frequency of gastrointestinal manifestations in myotonic dystrophy type 1 and type 2. Neurology, 2018, 90, 814.1-814.	1.1	2
39	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
40	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
41	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
42	A novel nonsense EIF1AX mutation identified in a thyroid nodule histologically diagnosed as oncocytic carcinoma. Endocrine, 2018, 62, 492-495.	2.3	11
43	Dysautonomia as Onset Symptom of Myotonic Dystrophy Type 2. European Neurology, 2018, 79, 166-170.	1.4	1
44	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
45	Clarification on Uveal Melanoma Associated With Myotonic Dystrophy. JAMA Ophthalmology, 2018, 136, 1426.	2.5	0
46	Secondary hypokalemic periodic paralysis as a rare clinical presentation of Conn syndrome. Clinical Neurophysiology, 2018, 129, 2505-2506.	1.5	2
47	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
48	Imaging Features of Varicella Zoster Virus Cranial Multiple Mononeuropathies. European Neurology, 2018, 79, 315-316.	1.4	0
49	Phase angle and impedance ratio: Two specular ways to analyze body composition. Annals of Clinical Nutrition, 2018, 1, .	0.2	7
50	Concentric muscle involvement in POLG -related distal myopathy. Neuromuscular Disorders, 2017, 27, 500-501.	0.6	2
51	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
52	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
53	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
54	Clinical and genetic characteristics of sporadic adult-onset degenerative ataxia. Neurology, 2017, 89, 1043-1049.	1.1	45

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55	<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
56	Brain Connectomicsâ€™ Modification to Clarify Motor and Nonmotor Features of Myotonic Dystrophy Type 1. Neural Plasticity, 2016, 2016, 1-10.	2.2	28
57	Validation of plasma microRNAs as biomarkers for myotonic dystrophy type 1. Scientific Reports, 2016, 6, 38174.	3.3	49
58	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
59	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
60	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
61	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
62	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
63	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
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	Abnormal Functional Brain Connectivity and Personality Traits in Myotonic Dystrophy Type 1. JAMA Neurology, 2014, 71, 603.	9.0	62
66	Restless legs syndrome and daytime sleepiness are prominent in myotonic dystrophy type 2. Neurology, 2014, 83, 572-573.	1.1	4
67	Restless legs syndrome and daytime sleepiness are prominent in myotonic dystrophy type 2. Neurology, 2014, 82, 283-284.	1.1	7
68	Teaching Neuro Images : Autosomal dominant leukodystrophy in a sporadic case. Neurology, 2014, 83, e121.	1.1	3
69	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
70	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
71	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
72	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

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73	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
74	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
75	Somatic mosaicism in TPM2-related myopathy with nemaline rods and cap structures. Acta Neuropathologica, 2013, 125, 169-171.	7.7	15
76	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
77	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
78	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
79	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
80	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay in the Time of Next-Generation Sequencing. Archives of Neurology, 2012, 69, 1661.	4.5	1
81	New phenotype and pathology features in MYH7-related distal myopathy. Neuromuscular Disorders, 2012, 22, 640-647.	0.6	41
82	Subdural hematoma in a young woman with an â€œoldâ€•brain. Acta Neurologica Belgica, 2012, 112, 385-387.	1.1	0
83	Prefrontal cortex controls human balance during overground ataxic gait. Restorative Neurology and Neuroscience, 2012, 30, 397-405.	0.7	18
84	Muscle imaging findings in GNE myopathy. Journal of Neurology, 2012, 259, 1358-1365.	3.6	57
85	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
86	Muscle <scp>MRI</scp> in female carriers of dystrophinopathy. European Journal of Neurology, 2012, 19, 1256-1260.	3.3	31
87	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
88	P2.38 Lower limb muscle MRI in a large cohort of FSHD patients. Neuromuscular Disorders, 2011, 21, 671.	0.6	3
89	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
90	Novel mutations in SPG11 cause hereditary spastic paraplegia associated with earlyâ€onset levodopaâ€responsive Parkinsonism. Movement Disorders, 2011, 26, 553-556.	3.9	62

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91	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
92	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
93	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
94	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
95	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
96	Myotonic dystrophy: A new perspective on the treatment of a multisystemic disease. Drugs of the Future, 2010, 35, 237.	0.1	0
97	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
98	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
99	SIADH in a patient with sensory ataxic neuropathy with anti-disialosyl antibodies (CANOMAD). Journal of Neurology, 2009, 256, 1177-1179.	3.6	11
100	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
101	Successful Treatment of Acute Autoimmune Limbic Encephalitis With Negative VGKC and NMDAR Antibodies. Cognitive and Behavioral Neurology, 2009, 22, 63-66.	0.9	25
102	Risk of Arrhythmias in MYotonic Dystrophy: trial design of the RAMYD study. Journal of Cardiovascular Medicine, 2009, 10, 51-58.	1.5	37
103	Cognitive impairment in myotonic dystrophy type 1 (DM1). Journal of Neurology, 2008, 255, 1737-1742.	3.6	76
104	A novel KIF5A/SPG10 mutation in spastic paraplegia associated with axonal neuropathy. Journal of Neurology, 2008, 255, 1090-1092.	3.6	29
105	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
106	Implicit and explicit aspects of sequence learning in pre-symptomatic Huntington's disease. Parkinsonism and Related Disorders, 2008, 14, 457-464.	2.2	45
107	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
108	Chronic autoimmune autonomic neuropathy responsive to immunosuppressive therapy. Neurology, 2007, 68, 161-162.	1.1	27

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109	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
110	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
111	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
112	Prevalence of spinocerebellar ataxia type 2 mutation among Italian Parkinsonian patients. Movement Disorders, 2007, 22, 324-327.	3.9	42
113	Evidence of white matter involvement in SCA 7. Journal of Neurology, 2007, 254, 536-538.	3.6	8
114	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
115	Transient MRI Abnormalities in a Case of Occipital Lobe Epilepsy with Favorable Outcome. Clinical EEG and Neuroscience, 2006, 37, 219-222.	1.7	1
116	Characterization of the Pattern of Cognitive Impairment in Myotonic Dystrophy Type 1. Archives of Neurology, 2004, 61, 1943-7.	4.5	147
117	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
118	NovelGNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy. Human Mutation, 2004, 23, 632-632.	2.5	52
119	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
120	Homozygosity mapping of Marinescoâ€“SjÃ¶gren syndrome to 5q31. European Journal of Human Genetics, 2003, 11, 770-778.	2.8	53
121	Cardiac features of Emeryâ€“Dreifuss muscular dystrophy caused by lamin A/C gene mutations. European Heart Journal, 2003, 24, 2227-2236.	2.2	103
122	An Italian family with autosomal recessive inclusion-body myopathy and mutations in the GNE gene. Neurology, 2002, 59, 1808-1809.	1.1	27
123	Apoptosis and Oxidative Stress in Mitochondrial Disorders. , 2002, , 37-45.		2
124	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
125	Coenzyme Q ₁₀ reverses pathological phenotype and reduces apoptosis in familial CoQ ₁₀ deficiency. Neurology, 2001, 57, 515-518.	1.1	157
126	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

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127	A new mtDNA mutation associated with a progressive encephalopathy and cytochrome c oxidase deficiency. Neurology, 2000, 54, 1693-1696.	1.1	48
128	Apoptosis in mitochondrial encephalomyopathies with mitochondrial DNA mutations: a potential pathogenic mechanism. Brain, 2000, 123, 93-104.	7.6	117
129	GCG genetic expansions in Italian patients with oculopharyngeal muscular dystrophy. Neurology, 2000, 54, 608-608.	1.1	51
130	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
131	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
132	The impact of the molecular data on clinical practice in facio-scapulohumeral muscular dystrophy (FSHD). Neuromuscular Disorders, 1997, 7, 443-444.	0.6	0
133	Functional involvement of central nervous system in mitochondrial disorders. Electroencephalography and Clinical Neurophysiology - Electromyography and Motor Control, 1997, 105, 171-180.	1.4	9
134	Mitochondrial DNA deletions in oculopharyngeal muscular dystrophy. FEBS Letters, 1997, 418, 167-170.	2.8	12
135	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
136	Maternally inherited cardiomyopathy: A new phenotype associated with the A to G at nt.3243 of mitochondrial DNA (MELAS mutation). , 1997, 20, 221.		1
137	Immunohistochemical studies of muscle and nerve in merosin-deficient congenital muscular dystrophy. Neuromuscular Disorders, 1996, 6, S18.	0.6	1
138	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
139	Letter to the editor. Muscle and Nerve, 1995, 18, 478-478.	2.2	2
140	Immunocytochemical localization of vinculin in muscle and nerve. Muscle and Nerve, 1995, 18, 1277-1284.	2.2	11
141	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
142	Morphological observations in mitochondrial diseases. Progress in Cell Research, 1995, , 217-221.	0.3	0
143	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE). Neurology, 1994, 44, 721-721.	1.1	402
144	Dystrophin is not essential for the integrity of the cytoskeleton. Acta Neuropathologica, 1994, 87, 377-384.	7.7	15

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145	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
146	Ekbom's syndrome: Lipomas, ataxia, and neuropathy with MERRF. Muscle and Nerve, 1994, 17, 943-945.	2.2	49
147	A new mtDNA mutation in the tRNALeu(UUR) gene associated with maternally inherited cardiomyopathy. Human Mutation, 1994, 3, 37-43.	2.5	178
148	Dystrophin is not essential for the integrity of the cytoskeleton. Acta Neuropathologica, 1994, 87, 377-384.	7.7	2
149	Atypical clinical presentations associated with the MELAS mutation at position 3243 of human mitochondrial DNA. Neuromuscular Disorders, 1993, 3, 43-50.	0.6	219
150	MELAS point mutation with unusual clinical presentation. Neuromuscular Disorders, 1993, 3, 191-193.	0.6	27
151	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
152	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
153	Fatal infantile liver failure associated with mitochondrial DNA depletion. Journal of Pediatrics, 1992, 121, 896-901.	1.8	123
154	MELAS: Clinical features, biochemistry, and molecular genetics. Annals of Neurology, 1992, 31, 391-398.	5.3	514
155	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
156	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
157	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. Neurology, 1991, 41, 1053-1053.	1.1	120
158	Remitting-Relapsing Carbamazepine Overdosage Mimicking Vertebrobasilar Transient Ischemic Attacks. International Journal of Case Reports in Medicine, 0, , 1-4.	0.0	0
159	A clinical and epidemiological prevalence study on Friedreichâ€™s Ataxia in Latium, Italy.. Neuroepidemiology, 0, , .	2.3	0