Cinzia Gellera

List of Publications by Year
in descending order

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206 papers	16,092 citations	18482 62 h-index	18647 119 g-index
209 all docs	209 docs citations	209 times ranked	16026 citing authors

#	Article	IF	Citations
1	Digenic inheritance of STUB1 variants and TBP polyglutamine expansions explains the incomplete penetrance of SCA17 and SCA48. Genetics in Medicine, 2022, 24, 29-40.	2.4	24
2	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8.	3.8	23
3	Optical coherence tomography in adult adrenoleukodystrophy: a cross-sectional and longitudinal study. Neurological Sciences, 2021, 42, 235-241.	1.9	3
4	Hypomyelinating leukodystrophies in adults: Clinical and genetic features. European Journal of Neurology, 2021, 28, 934-944.	3.3	14
5	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
6	Peripheral nerve enlargement on nerve ultrasound parallels neuropathological changes in adultâ€onset Krabbe disease. Muscle and Nerve, 2021, 63, E33-E35.	2.2	2
7	Dysregulation of Muscle-Specific MicroRNAs as Common Pathogenic Feature Associated with Muscle Atrophy in ALS, SMA and SBMA: Evidence from Animal Models and Human Patients. International Journal of Molecular Sciences, 2021, 22, 5673.	4.1	14
8	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	1.9	20
9	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	21.4	223
10	The first case of the TARDBP p.G294V mutation in a homozygous state: is a single pathogenic allele sufficient to cause ALS?. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 273-279.	1.7	10
11	Late-onset Huntingtonâ€™s disease with 40â€“42 CAG expansion. Neurological Sciences, 2020, 41, 869-876.	1.9	15
12	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia, 2020, 61, 2474-2485.	5.1	31
13	Gabapentin treatment in a patient with KCNQ2 developmental epileptic encephalopathy. Pharmacological Research, 2020, 160, 105200.	7.1	7
14	Huntingtin gene CAG repeat size affects autism risk: Familyâ€based and caseâ€“control association study. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 341-351.	1.7	5
15	Teaching NeuroImages: Symmetrical abnormalities of the globi pallidi in succinic semialdehyde dehydrogenase deficiency. Neurology, 2020, 95, e2316-e2317.	1.1	2
16	Frataxin gene editing rescues Friedreichâ€™s ataxia pathology in dorsal root ganglia organoid-derived sensory neurons. Nature Communications, 2020, 11, 4178.	12.8	42
17	Missing the pathological expansion in Huntington disease: de novo c. 51C >G variant on the expanded allele causing intrafamilial allele dropout. American Journal of Medical Genetics, Part A, 2020, 185, 397-400.	1.2	1
18	SCN8A splicing mutation causing skipping of the exon 15 associated with intellectual disability and cortical myoclonus. Seizure: the Journal of the British Epilepsy Association, 2020, 82, 56-58.	2.0	0

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19	Conventional and Unconventional Therapeutic Strategies for Sialidosis Type I. Journal of Clinical Medicine, 2020, 9, 695.	2.4	13
20	Molecular Genetics of Niemannâ€“Pick Type C Disease in Italy: An Update on 105 Patients and Description of 18 NPC1 Novel Variants. Journal of Clinical Medicine, 2020, 9, 679.	2.4	21
21	Sorting Rare ALS Genetic Variants by Targeted Re-Sequencing Panel in Italian Patients: OPTN, VCP, and SQSTM1 Variants Account for 3% of Rare Genetic Forms. Journal of Clinical Medicine, 2020, 9, 412.	2.4	24
22	Frequency and distribution of polyQ disease intermediate-length repeat alleles in healthy Italian population. Neurological Sciences, 2020, 41, 1475-1482.	1.9	10
23	Glutamine Synthetase 1 Increases Autophagy Lysosomal Degradation of Mutant Huntingtin Aggregates in Neurons, Ameliorating Motility in a Drosophila Model for Huntingtonâ€™s Disease. Cells, 2020, 9, 196.	4.1	18
24	Circulating MyomiRs as Potential Biomarkers to Monitor Response to Nusinersen in Pediatric SMA Patients. Biomedicines, 2020, 8, 21.	3.2	30
25	Asymptomatic adrenoleukodystrophy in elderly males. Journal of Neurology, 2020, 267, 1849-1851.	3.6	0
26	Early Parkinsonism in a Senegalese girl with Lafora disease. Epileptic Disorders, 2020, 22, 233-236.	1.3	4
27	Progression of Cerebellar Atrophy in Spinocerebellar Ataxia Type 2 Gene Carriers: A Longitudinal MRI Study in Preclinical and Early Disease Stages. Frontiers in Neurology, 2020, 11, 616419.	2.4	16
28	Disorders of lipid metabolism. , 2020, , 731-753.		1
29	Progressive myoclonus epilepsy caused by a gain-of-function KCNA2 mutation. Seizure: the Journal of the British Epilepsy Association, 2019, 65, 106-108.	2.0	16
30	Cognitive Syndromes and C9orf72 Mutation Are Not Related to Cerebellar Degeneration in Amyotrophic Lateral Sclerosis. Frontiers in Neuroscience, 2019, 13, 440.	2.8	10
31	Screening of SLC2A1 in a large cohort of patients suspected for Glut1 deficiency syndrome: identification of novel variants and associated phenotypes. Journal of Neurology, 2019, 266, 1439-1448.	3.6	18
32	HCN ion channels and accessory proteins in epilepsy: genetic analysis of a large cohort of patients and review of the literature. Epilepsy Research, 2019, 153, 49-58.	1.6	32
33	Expanding the central nervous system disease spectrum associated with FLNC mutation. Muscle and Nerve, 2019, 59, E33-E37.	2.2	3
34	Saposin B deficiency as a cause of adult-onset metachromatic leukodystrophy. Neurology, 2019, 93, 310-312.	1.1	6
35	Association of NIPA1 repeat expansions with amyotrophic lateral sclerosis in a large international cohort. Neurobiology of Aging, 2019, 74, 234.e9-234.e15.	3.1	26
36	Familial vitamin E deficiency: Multiorgan complications support the adverse role of oxidative stress. Nutrition, 2019, 63-64, 57-60.	2.4	9

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37	Clinical and genetic characteristics of late-onset Huntington's disease. Parkinsonism and Related Disorders, 2019, 61, 101-105.	2.2	17
38	Spinocerebellar Ataxia Tethering PCR. Journal of Molecular Diagnostics, 2018, 20, 289-297.	2.8	16
39	New <scp>FIG</scp>4 gene mutations causing aggressive <scp>ALS</scp>. European Journal of Neurology, 2018, 25, e41-e42.	3.3	14
40	Kv7.3 Compound Heterozygous Variants in Early Onset Encephalopathy Reveal Additive Contribution of C-Terminal Residues to PIP2-Dependent K+ Channel Gating. Molecular Neurobiology, 2018, 55, 7009-7024.	4.0	21
41	Multiple system atrophy and CAG repeat length: A genetic screening of polyglutamine disease genes in Italian patients. Neuroscience Letters, 2018, 678, 37-42.	2.1	10
42	Cortical thickness, stance control, and arithmetic skill: An exploratory study in premanifest Huntington disease. Parkinsonism and Related Disorders, 2018, 51, 17-23.	2.2	22
43	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
44	G08â€...An italian study to estimate the frequency of the intermediate triplet length in the huntingtin gene: 1/20 subject carries an allele with 27â€“35 cag. , 2018, , .		0
45	A new case report of severe mucopolysaccharidosis type VII: diagnosis, treatment with haematopoietic cell transplantation and prenatal diagnosis in a second pregnancy. Italian Journal of Pediatrics, 2018, 44, 128.	2.6	12
46	Concurrent AFG3L2 and SPG7 mutations associated with syndromic parkinsonism and optic atrophy with aberrant OPA1 processing and mitochondrial network fragmentation. Human Mutation, 2018, 39, 2060-2071.	2.5	32
47	The role of clinical and neuroimaging features in the diagnosis of CADASIL. Journal of Neurology, 2018, 265, 2934-2943.	3.6	25
48	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
49	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. Neurobiology of Aging, 2018, 71, 266.e1-266.e10.	3.1	59
50	A novel de novo HCN1 loss-of-function mutation in genetic generalized epilepsy causing increased neuronal excitability. Neurobiology of Disease, 2018, 118, 55-63.	4.4	47
51	A Loss-of-Function HCN4 Mutation Associated With Familial Benign Myoclonic Epilepsy in Infancy Causes Increased Neuronal Excitability. Frontiers in Molecular Neuroscience, 2018, 11, 269.	2.9	25
52	Early Treatment with Quinidine in 2 Patients with Epilepsy of Infancy with Migrating Focal Seizures (EIMFS) Due to Gain-of-Function KCNT1 Mutations: Functional Studies, Clinical Responses, and Critical Issues for Personalized Therapy. Neurotherapeutics, 2018, 15, 1112-1126.	4.4	56
53	Cortical markers of cognitive syndromes in amyotrophic lateral sclerosis. NeuroImage: Clinical, 2018, 19, 675-682.	2.7	24
54	Reduced Cancer Incidence in Huntington's Disease: Analysis in the Registry Study. Journal of Huntington's Disease, 2018, 7, 209-222.	1.9	14

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55	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
56	Alternating Hemiplegia and Epilepsia Partialis Continua: A new phenotype for a novel compound TBC1D24 mutation. Seizure: the Journal of the British Epilepsy Association, 2017, 47, 71-73.	2.0	16
57	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
58	Identification of genetic variants associated with Huntington's disease progression: a genome-wide association study. Lancet Neurology, The, 2017, 16, 701-711.	10.2	248
59	Genetic correlation between amyotrophic lateral sclerosis and schizophrenia. Nature Communications, 2017, 8, 14774.	12.8	114
60	The role of de novo mutations in the development of amyotrophic lateral sclerosis. Human Mutation, 2017, 38, 1534-1541.	2.5	13
61	Stance instability in preclinical SCA1 mutation carriers: A 4-year prospective posturography study. Gait and Posture, 2017, 57, 11-14.	1.4	11
62	Cognitive decline in Huntington's disease expansion gene carriers. Cortex, 2017, 95, 51-62.	2.4	50
63	Riboflavinâ€responsive multiple acylâ€CoA dehydrogenase deficiency: delayed hypersensitivity reaction and efficacy of lowâ€dose intermittent supplementation. European Journal of Neurology, 2017, 24, e41-e42.	3.3	6
64	Amyotrophic lateral sclerosis causes small fiber pathology. European Journal of Neurology, 2016, 23, 416-420.	3.3	65
65	No effect of <scp>AR</scp> polyG polymorphism on spinal and bulbar muscular atrophy phenotype. European Journal of Neurology, 2016, 23, 1134-1136.	3.3	8
66	B35â€...Glutamine synthetase-1 induces autophagy and neuronal survival in a drosophila model huntingtonâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A21.2-A21.	1.9	1
67	ASAH1 variant causing a mild SMA phenotype with no myoclonic epilepsy: a clinical, biochemical and molecular study. European Journal of Human Genetics, 2016, 24, 1578-1583.	2.8	18
68	Ataxia with vitamin E deficiency caused by a new compound heterozygous mutation. Neurological Sciences, 2016, 37, 1571-1572.	1.9	1
69	In-vivo brain H1-MR-Spectroscopy identification and quantification of 2-hydroxyglutarate in L-2-Hydroxyglutaric aciduria. Brain Research, 2016, 1648, 506-511.	2.2	9
70	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	21.4	494
71	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
72	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174

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73	I16â€…Very slow disease progression in two hd patients carrying 40 and 45 cag repeats: a 10-year follow-up observational report. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A64.2-A64.	1.9	0
74	F6â€…Does premanifest HD perform worse in arithmetic? Symbol digit and calculation tests confirm early cognitive impairment in preHD and correlate with brain MRI abnormalities. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A50.2-A50.	1.9	0
75	E7â€…Posturography and mri study in premanifest huntingtonâ€™s disease subjects to investigate stance abnormalities and early markers of neurodegeneration. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A47.3-A48.	1.9	0
76	Clinical Pregenetic Screening for Stroke Monogenic Diseases. Stroke, 2016, 47, 1702-1709.	2.0	34
77	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
78	Early white matter involvement in an infant carrying a novel mutation in ACOX1. European Journal of Paediatric Neurology, 2016, 20, 431-434.	1.6	4
79	The role of androgen receptor gene variants on SBMA phenotype. Journal of the Neurological Sciences, 2015, 357, e231.	0.6	0
80	Clinical and molecular report of novel GALC mutations in Moroccan patient with Krabbe disease: case report. BMC Pediatrics, 2015, 15, 182.	1.7	5
81	PEX7Mutations Cause Congenital Cataract Retinopathy and Late-Onset Ataxia and Cognitive		

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91	Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. Neurobiology of Aging, 2014, 35, 2420.e13-2420.e14.	3.1	16
92	Atypical Friedreich ataxia in patients with FXN p.R165P point mutation or comorbid hemochromatosis. Parkinsonism and Related Disorders, 2014, 20, 919-923.	2.2	10
93	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	2.9	123
94	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
95	Hyperargininemia: 7-Month Follow-Up Under Sodium Benzoate Therapy in an Italian Child Presenting Progressive Spastic Paraparesis, Cognitive Decline, and Novel Mutation in ARG1 Gene. Pediatric Neurology, 2014, 51, 430-433.	2.1	9
96	E25 Fmri Signal Changes In Frontal Cortex Correlates With Years-to-disease-onset In A Group Of Young Premanifest Huntington Disease Subjects. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, A45-A45.	1.9	0
97	Human Adipose-Derived Mesenchymal Stem Cells as a New Model of Spinal and Bulbar Muscular Atrophy. PLoS ONE, 2014, 9, e112746.	2.5	15
98	SETX mutations are a frequent genetic cause of juvenile and adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. Orphanet Journal of Rare Diseases, 2013, 8, 123.	2.7	31
99	Granny trips down: is she carrying the big bad wolf?. Neurological Sciences, 2013, 34, 2077-2079.	1.9	2
100	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntingtonâ€™s disease motor onset. Neurogenetics, 2013, 14, 173-179.	1.4	10
101	Suicidal ideation in a European Huntington's disease population. Journal of Affective Disorders, 2013, 151, 248-258.	4.1	74
102	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. Neurobiology of Aging, 2013, 34, 1517.e9-1517.e10.	3.1	35
103	Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2695.e11-2695.e12.	3.1	30
104	The C9ORF72 expansion mutation is a common cause of ALS+/â^'FTD in Europe and has a single founder. European Journal of Human Genetics, 2013, 21, 102-108.	2.8	201
105	Ubiquilin 2mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 183-187.	1.9	74
106	The V471A Polymorphism in Autophagy-Related Gene ATG7 Modifies Age at Onset Specifically in Italian Huntington Disease Patients. PLoS ONE, 2013, 8, e68951.	2.5	49
107	Î²-Defensin Genomic Copy Number Does Not Influence the Age of Onset in Huntington's Disease. Journal of Huntington's Disease, 2013, 2, 107-124.	1.9	1
108	N01â€…A 6-years experience of genetic counselling in Huntington disease (2006â€“2011): constant socio-demographic profile of at-risk individuals requesting genetic predictive test. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, A48.4-A49.	1.9	0

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109	Discrepancies in reporting the CAG repeat lengths for Huntington's disease. European Journal of Human Genetics, 2012, 20, 20-26.	2.8	20
110	Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 265-269.	2.1	15
111	CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. Neurology, 2012, 78, 690-695.	1.1	303
112	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. Nature, 2012, 488, 499-503.	27.8	522
113	Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency with unknown genetic defect. Neurological Sciences, 2012, 33, 1383-1387.	1.9	10
114	Novel and recurrent spastin mutations in a large series of SPG4 Italian families. Neuroscience Letters, 2012, 528, 42-45.	2.1	5
115	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e1-630.e2.	3.1	17
116	ATAXIN2 CAG-repeat length in Italian patients with amyotrophic lateral sclerosis: risk factor or variant phenotype? Implication for genetic testing and counseling. Neurobiology of Aging, 2012, 33, 1847.e15-1847.e21.	3.1	27
117	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. Neurobiology of Aging, 2012, 33, 2528.e7-2528.e14.	3.1	74
118	Cortical myoclonus in childhood and juvenile onset Huntington's disease. Parkinsonism and Related Disorders, 2012, 18, 794-797.	2.2	22
119	TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntingtonâ€™s disease. Biochemical and Biophysical Research Communications, 2012, 424, 404-408.	2.1	20
120	The first reported generation of several induced pluripotent stem cell lines from homozygous and heterozygous Huntington's disease patients demonstrates mutation related enhanced lysosomal activity. Neurobiology of Disease, 2012, 46, 41-51.	4.4	159
121	Preferential expression of mutant ABCD1 allele is common in adrenoleukodystrophy female carriers but unrelated to clinical symptoms. Orphanet Journal of Rare Diseases, 2012, 7, 10.	2.7	29
122	An Optimized Method for Manufacturing a Clinical Scale Dendritic Cell-Based Vaccine for the Treatment of Glioblastoma. PLoS ONE, 2012, 7, e52301.	2.5	30
123	Observing Huntington's disease: the European Huntington's Disease Network's REGISTRY. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1409-1412.	1.9	82
124	Paroxysmal exercise-induced dyskinesia with self-limiting partial epilepsy: A novel GLUT-1Âmutation with benign phenotype. Parkinsonism and Related Disorders, 2011, 17, 479-481.	2.2	16
125	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. Neurobiology of Aging, 2011, 32, 966-967.	3.1	28
126	Co-occurrence of amyotrophic lateral sclerosis and Charcot-Marie-Tooth disease type 2A in a patient with a novel mutation in the mitofusin-2 gene. Neuromuscular Disorders, 2011, 21, 129-131.	0.6	19

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127	Ataxia with oculomotor apraxia type1 (AOA1): novel and recurrent aprataxin mutations, coenzyme Q10 analyses, and clinical findings in Italian patients. Neurogenetics, 2011, 12, 193-201.	1.4	46
128	Brain diffusionâ€weighted imaging in Friedreich's ataxia. Movement Disorders, 2011, 26, 705-712.	3.9	52
129	Novel optineurin mutations in patients with familial and sporadic amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1239-1243.	1.9	86
130	NMDA receptor gene variations as modifiers in Huntington disease: a replication study. PLOS Currents, 2011, 3, RRN1247.	1.4	20
131	Paraoxonase gene mutations in amyotrophic lateral sclerosis. Annals of Neurology, 2010, 68, 102-107.	5.3	67
132	A novel SOD1 mutation in a young amyotrophic lateral sclerosis patient with a very slowly progressive clinical course. Muscle and Nerve, 2010, 42, 596-597.	2.2	11
133	Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. Nature Genetics, 2010, 42, 313-321.	21.4	291
134	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2010, 47, 190-194.	3.2	152
135	Identification of novel and recurrent CACNA1A gene mutations in fifteen patients with episodic ataxia type 2. Journal of the Neurological Sciences, 2010, 291, 30-36.	0.6	63
136	Predictive Genetic Tests in Neurodegenerative Disorders: A Methodological Approach Integrating Psychological Counseling for At-Risk Individuals and Referring Clinicians. European Neurology, 2010, 64, 33-41.	1.4	12
137	Age and founder effect of SOD1 A4V mutation causing ALS. Neurology, 2009, 72, 1634-1639.	1.1	84
138	Visual system involvement in patients with Friedreich's ataxia. Brain, 2009, 132, 116-123.	7.6	146
139	Analysis of FUS gene mutation in familial amyotrophic lateral sclerosis within an Italian cohort. Neurology, 2009, 73, 1180-1185.	1.1	139
140	High frequency ofTARDBPgene mutations in Italian patients with amyotrophic lateral sclerosis. Human Mutation, 2009, 30, 688-694.	2.5	184
141	Rare association of motor neuron disease and spinocerebellar ataxia type 2 (SCA2): a new case and review of the literature. Journal of Neurology, 2009, 256, 1926-1928.	3.6	42
142	The gene coding for PGC-1Î± modifies age at onset in Huntington's Disease. Molecular Neurodegeneration, 2009, 4, 3.	10.8	119
143	A Simple Multiplex Real-Time PCR Methodology for the SMN1 Gene Copy Number Quantification. Genetic Testing and Molecular Biomarkers, 2009, 13, 37-42.	0.7	16
144	A majority of Huntington's disease patients may be treatable by individualized allele-specific RNA interference. Experimental Neurology, 2009, 217, 312-319.	4.1	109

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145	Spinocerebellar ataxia type 28: A novel autosomal dominant cerebellar ataxia characterized by slow progression and ophthalmoparesis. Cerebellum, 2008, 7, 184-188.	2.5	57
146	Identification of new ANG gene mutations in a large cohort of Italian patients with amyotrophic lateral sclerosis. Neurogenetics, 2008, 9, 33-40.	1.4	102
147	Spinal and bulbar muscular atrophy: Skeletal muscle pathology in male patients and heterozygous females. Journal of the Neurological Sciences, 2008, 264, 100-105.	0.6	133
148	The rare G93D mutation causes a slowly progressing lower motor neuron disease. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 35-39.	2.1	18
149	Ataxia With Oculomotor Apraxia Type 1 (AOA1): Clinical and Neuropsychological Features in 2 New Patients and Differential Diagnosis. Journal of Child Neurology, 2008, 23, 895-900.	1.4	22
150	Isolated Vitamin E Deficiency Mimicking Distal Hereditary Motor Neuropathy in a 13-Year-Old Boy. Journal of Child Neurology, 2008, 23, 1328-1330.	1.4	10
151	Paroxysmal movement disorders in GLUT1 deficiency syndrome. Neurology, 2008, 71, 146-148.	1.1	73
152	Biological abnormalities of peripheral A2A receptors in a large representation of polyglutamine disorders and Huntington's disease stages. Neurobiology of Disease, 2007, 27, 36-43.	4.4	35
153	Spinocerebellar ataxia type 17 (SCA17): Oculomotor phenotype and clinical characterization of 15 Italian patients. Journal of Neurology, 2007, 254, 1538-1546.	3.6	78
154	Frataxin gene point mutations in Italian Friedreich ataxia patients. Neurogenetics, 2007, 8, 289-299.	1.4	71
155	Large Pathogenic Expansions in the SCA2 and SCA7 Genes Can Be Detected by Fluorescent Repeat-Primed Polymerase Chain Reaction Assay. Journal of Molecular Diagnostics, 2006, 8, 128-132.	2.8	31
156	The A467T and W748S POLG substitutions are a rare cause of adult-onset ataxia in Europe. Brain, 2006, 130, E69-E69.	7.6	14
157	Clinical and Genetic Findings in 26 Italian Patients with Lafora Disease. Epilepsia, 2006, 47, 640-643.	5.1	71
158	The S18Y polymorphism in the UCHL1 gene is a genetic modifier in Huntingtonâ€™s disease. Neurogenetics, 2006, 7, 27-30.	1.4	47
159	Efficacious vitamin E treatment in a child with ataxia with isolated vitamin E deficiency. European Journal of Pediatrics, 2006, 165, 494-495.	2.7	20
160	Genetic analysis of candidate genes modifying the age-at-onset in Huntingtonâ€™s disease. Human Genetics, 2006, 120, 285-292.	3.8	39
161	SCA28, a novel form of autosomal dominant cerebellar ataxia on chromosome 18p11.22â€“q11.2. Brain, 2006, 129, 235-242.	7.6	122
162	FMR1 gene premutation is a frequent genetic cause of late-onset sporadic cerebellar ataxia. Neurology, 2005, 64, 145-147.	1.1	90

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163	Molecular Genetics of Hereditary Spinocerebellar Ataxia. Archives of Neurology, 2004, 61, 727.	4.5	130
164	Prevalence of Inherited Ataxias in the Province of Padua, Italy. Neuroepidemiology, 2004, 23, 275-280.	2.3	43
165	Ataxia with isolated vitamin E deficiency: neurological phenotype, clinical follow-up and novel mutations in TTPAgene in Italian families. Neurological Sciences, 2004, 25, 130-137.	1.9	131
166	The gender effect in juvenile Huntington disease patients of Italian origin. American Journal of Medical Genetics Part A, 2004, 125B, 92-98.	2.4	41
167	Detection of Large Pathogenic Expansions in FRDA1, SCA10, and SCA12 Genes Using a Simple Fluorescent Repeat-Primed PCR Assay. Journal of Molecular Diagnostics, 2004, 6, 96-100.	2.8	48
168	A whole genome screen for linkage disequilibrium in multiple sclerosis performed in a continental Italian population. Journal of Neuroimmunology, 2003, 143, 97-100.	2.3	17
169	Homozygosity for CAG mutation in Huntington disease is associated with a more severe clinical course. Brain, 2003, 126, 946-955.	7.6	173
170	Myoclonic dystonia as unique presentation of isolated vitamin E deficiency in a young patient. Movement Disorders, 2002, 17, 612-614.	3.9	36
171	Cognitive and psychiatric characterization of patients with Huntington's disease and their at-risk relatives. Neurological Sciences, 2002, 23, s105-s106.	1.9	19
172	Superoxide dismutase gene mutations in Italian patients with familial and sporadic amyotrophic lateral sclerosis: identification of three novel missense mutations. Neuromuscular Disorders, 2001, 11, 404-410.	0.6	47
173	The complex clinical and genetic classification of inherited ataxias. II. Autosomal recessive ataxias. Neurological Sciences, 2001, 22, 219-228.	1.9	71
174	Atypical movement disorders in the early stages of Huntington's disease: clinical and genetic analysis. Clinical Genetics, 2001, 58, 50-56.	2.0	72
175	Multiple founder effects in spinal and bulbar muscular atrophy (SBMA, Kennedy disease) around the world. European Journal of Human Genetics, 2001, 9, 431-436.	2.8	41
176	Genetics of ALS in Italian families. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2001, 2, s43-s46.	1.2	18
177	Pathogenic effect of an intermediate-size SCA-6 allele (CAG) ₁₉ in a homozygous patient. Neurology, 2001, 57, 1502-1504.	1.1	37
178	Frataxin expression rescues mitochondrial dysfunctions in FRDA cells. Human Molecular Genetics, 2001, 10, 2099-2107.	2.9	84
179	Family and molecular data for a fine analysis of age at onset in Huntington disease. American Journal of Medical Genetics Part A, 2000, 95, 366-373.	2.4	40
180	Sensitivity of FRDA Lymphoblasts to Salts of Transition Metal Ions. Antioxidants and Redox Signaling, 2000, 2, 461-465.	5.4	13

#	Article	IF	Citations
181	Human frataxin maintains mitochondrial iron homeostasis in Saccharomyces cerevisiae. Human Molecular Genetics, 2000, 9, 2523-2530.	2.9	140
182	Relative Frequencies of CAG Expansions in Spinocerebellar Ataxia and Dentatorubropallidoluysian Atrophy in 116 Italian Families. European Neurology, 2000, 44, 31-36.	1.4	63
183	Phenotypic manifestations associated with CAG-repeat expansion in the androgen receptor gene in male patients and heterozygous females: a clinical and molecular study of 30 families. Neuromuscular Disorders, 2000, 10, 391-397.	0.6	112
184	The Friedreich's ataxia mutation confers cellular sensitivity to oxidant stress which is rescued by chelators of iron and calcium and inhibitors of apoptosis. Human Molecular Genetics, 1999, 8, 425-430.	2.9	349
185	Clinical and molecular studies of 73 Italian families with autosomal dominant cerebellar ataxia type I: SCA1 and SCA2 are the most common genotypes. Journal of Neurology, 1999, 246, 389-393.	3.6	63
186	Clinical and molecular findings in the first identified Italian family with dentatorubral-pallidoluysian atrophy. Acta Neurologica Scandinavica, 1998, 98, 324-327.	2.1	15
187	Very late onset Friedreich's ataxia without cardiomyopathy is associated with limited GAA expansion in the X25 gene. Neurology, 1997, 49, 1153-1155.	1.1	35
188	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. Science, 1996, 271, 1423-1427.	12.6	2,642
189	Psychiatric Symptoms Do Not Correlate With Cognitive Decline, Motor Symptoms, or CAG Repeat Length in Huntington's Disease. Archives of Neurology, 1996, 53, 493-497.	4.5	97
190	Kennedy's disease: clinical and molecular study of two Italian families. Italian Journal of Neurological Sciences, 1995, 16, 467-471.	0.1	1
191	Carnitine palmitoyltransferase II deficiency: structure of the gene and characterization of two novel disease-causing mutations. Human Molecular Genetics, 1995, 4, 19-29.	2.9	89
192	Molecular Pathogenesis of Mitochondrial Diseases. Progress in Cell Research, 1995, 5, 223-224.	0.3	2
193	Assignment of the Human Carnitine Palmitoyltransferase II Gene (CPT1) to Chromosome 1p32. Genomics, 1994, 24, 195-197.	2.9	65
194	cDNA cloning and mitochondrial import of the beta-subunit of the human electron-transfer flavoprotein. FEBS Journal, 1993, 213, 1003-1008.	0.2	44
195	Molecular characterization of inherited carnitine palmitoyltransferase II deficiency.. Proceedings of the National Academy of Sciences of the United States of America, 1992, 89, 8429-8433.	7.1	151
196	Maternally inherited myopathy and cardiomyopathy: association with mutation in mitochondrial DNA tRNALeu(UUR). Lancet, The, 1991, 338, 143-147.	13.7	395
197	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. Neurology, 1991, 41, 1053-1053.	1.1	120
198	Tissue distribution and transmission of mitochondrial DNA deletions in mitochondrial myopathies. Annals of Neurology, 1990, 28, 94-97.	5.3	98

#	Article	IF	Citations
199	Fumarase deficiency is an autosomal recessive encephalopathy affecting both the mitochondrial and the cytosolic enzymes. Neurology, 1990, 40, 495-495.	1.1	79
200	Normalization of short-chain acylcoenzyme a dehydrogenase after riboflavin treatment in a girl with multiple acylcoenzyme a dehydrogenase?deficient myopathy. Annals of Neurology, 1989, 25, 479-484.	5.3	49
201	An autosomal dominant disorder with multiple deletions of mitochondrial DNA starting at the D-loop region. Nature, 1989, 339, 309-311.	27.8	640
202	Evidence for two distinct mitochondrial malic enzymes in human skeletal muscle: purification and properties of the NAD(P)+-dependent enzyme. BBA - Proteins and Proteomics, 1987, 916, 446-454.	2.1	20
203	Myoadenylate deaminase deficiency in twins with recessive olivopontocerebellar atrophy. Italian Journal of Neurological Sciences, 1986, 7, 107-112.	0.1	1
204	Production of polyclonal antibodies against protein antigens purified by electroelution from SDS-polyacrylamide gel. Protocol Exchange, 0, , .	0.3	2
205	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
206	Functional Characterization of Two Variants at the Intron 6â€”Exon 7 Boundary of the KCNQ2 Potassium Channel Gene Causing Distinct Epileptic Phenotypes. Frontiers in Pharmacology, 0, 13, .	3.5	0