

Cinzia Gellera

List of Publications by Year in descending order

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Version: 2024-02-01

206
papers

16,092
citations

18465

62
h-index

18633

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. <i>Genetics in Medicine</i> , 2022, 24, 29-40.	1.1	24
2	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. <i>Npj Genomic Medicine</i> , 2022, 7, 8.	1.7	23
3	Optical coherence tomography in adult adrenoleukodystrophy: a cross-sectional and longitudinal study. <i>Neurological Sciences</i> , 2021, 42, 235-241.	0.9	3
4	Hypomyelinating leukodystrophies in adults: Clinical and genetic features. <i>European Journal of Neurology</i> , 2021, 28, 934-944.	1.7	14
5	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. <i>Neuron</i> , 2021, 109, 448-460.e4.	3.8	56
6	Peripheral nerve enlargement on nerve ultrasound parallels neuropathological changes in adult-onset Krabbe disease. <i>Muscle and Nerve</i> , 2021, 63, E33-E35.	1.0	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. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5673.	1.8	14
8	Progressive Myoclonus Epilepsies. <i>Neurology: Genetics</i> , 2021, 7, e641.	0.9	20
9	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	9.4	223
10	The first case of the <i>TARDBP</i> p.G294V mutation in a homozygous state: is a single pathogenic allele sufficient to cause ALS?. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 273-279.	1.1	10
11	Late-onset Huntington's disease with 40-42 CAG expansion. <i>Neurological Sciences</i> , 2020, 41, 869-876.	0.9	15
12	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in <i>SLC13A5</i> gene. <i>Epilepsia</i> , 2020, 61, 2474-2485.	2.6	31
13	Gabapentin treatment in a patient with KCNQ2 developmental epileptic encephalopathy. <i>Pharmacological Research</i> , 2020, 160, 105200.	3.1	7
14	Huntingtin gene CAG repeat size affects autism risk: Family-based and case-control association study. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2020, 183, 341-351.	1.1	5
15	Teaching NeuroImages: Symmetrical abnormalities of the globi pallidi in succinic semialdehyde dehydrogenase deficiency. <i>Neurology</i> , 2020, 95, e2316-e2317.	1.5	2
16	Frataxin gene editing rescues Friedreich's ataxia pathology in dorsal root ganglia organoid-derived sensory neurons. <i>Nature Communications</i> , 2020, 11, 4178.	5.8	42
17	Missing the pathological expansion in Huntington disease: de novo c. 51C >G variant on the expanded allele causing intrafamilial allele dropout. <i>American Journal of Medical Genetics, Part A</i> , 2020, 185, 397-400.	0.7	1
18	SCN8A splicing mutation causing skipping of the exon 15 associated with intellectual disability and cortical myoclonus. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 82, 56-58.	0.9	0

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19	Conventional and Unconventional Therapeutic Strategies for Sialidosis Type I. <i>Journal of Clinical Medicine</i> , 2020, 9, 695.	1.0	13
20	Molecular Genetics of Niemann-Pick Type C Disease in Italy: An Update on 105 Patients and Description of 18 NPC1 Novel Variants. <i>Journal of Clinical Medicine</i> , 2020, 9, 679.	1.0	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. <i>Journal of Clinical Medicine</i> , 2020, 9, 412.	1.0	24
22	Frequency and distribution of polyQ disease intermediate-length repeat alleles in healthy Italian population. <i>Neurological Sciences</i> , 2020, 41, 1475-1482.	0.9	10
23	Glutamine Synthetase 1 Increases Autophagy Lysosomal Degradation of Mutant Huntingtin Aggregates in Neurons, Ameliorating Motility in a <i>Drosophila</i> Model for Huntington's Disease. <i>Cells</i> , 2020, 9, 196.	1.8	18
24	Circulating MyomiRs as Potential Biomarkers to Monitor Response to Nusinersen in Pediatric SMA Patients. <i>Biomedicines</i> , 2020, 8, 21.	1.4	30
25	Asymptomatic adrenoleukodystrophy in elderly males. <i>Journal of Neurology</i> , 2020, 267, 1849-1851.	1.8	0
26	Early Parkinsonism in a Senegalese girl with Lafora disease. <i>Epileptic Disorders</i> , 2020, 22, 233-236.	0.7	4
27	Progression of Cerebellar Atrophy in Spinocerebellar Ataxia Type 2 Gene Carriers: A Longitudinal MRI Study in Preclinical and Early Disease Stages. <i>Frontiers in Neurology</i> , 2020, 11, 616419.	1.1	16
28	Disorders of lipid metabolism. , 2020, , 731-753.		1
29	Progressive myoclonus epilepsy caused by a gain-of-function KCNA2 mutation. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 65, 106-108.	0.9	16
30	Cognitive Syndromes and C9orf72 Mutation Are Not Related to Cerebellar Degeneration in Amyotrophic Lateral Sclerosis. <i>Frontiers in Neuroscience</i> , 2019, 13, 440.	1.4	10
31	Screening of SLC2A1 in a large cohort of patients suspected for Glut1 deficiency syndrome: identification of novel variants and associated phenotypes. <i>Journal of Neurology</i> , 2019, 266, 1439-1448.	1.8	18
32	HCN ion channels and accessory proteins in epilepsy: genetic analysis of a large cohort of patients and review of the literature. <i>Epilepsy Research</i> , 2019, 153, 49-58.	0.8	32
33	Expanding the central nervous system disease spectrum associated with <i>FLNC</i> mutation. <i>Muscle and Nerve</i> , 2019, 59, E33-E37.	1.0	3
34	Saposin B deficiency as a cause of adult-onset metachromatic leukodystrophy. <i>Neurology</i> , 2019, 93, 310-312.	1.5	6
35	Association of NIPA1 repeat expansions with amyotrophic lateral sclerosis in a large international cohort. <i>Neurobiology of Aging</i> , 2019, 74, 234.e9-234.e15.	1.5	26
36	Familial vitamin E deficiency: Multiorgan complications support the adverse role of oxidative stress. <i>Nutrition</i> , 2019, 63-64, 57-60.	1.1	9

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37	Clinical and genetic characteristics of late-onset Huntington's disease. <i>Parkinsonism and Related Disorders</i> , 2019, 61, 101-105.	1.1	17
38	Spinocerebellar Ataxia Tethering PCR. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 289-297.	1.2	16
39	New <i>FIG4</i> gene mutations causing aggressive <i>ALS</i> . <i>European Journal of Neurology</i> , 2018, 25, e41-e42.	1.7	14
40	Kv7.3 Compound Heterozygous Variants in Early Onset Encephalopathy Reveal Additive Contribution of C-Terminal Residues to PIP2-Dependent K ⁺ Channel Gating. <i>Molecular Neurobiology</i> , 2018, 55, 7009-7024.	1.9	21
41	Multiple system atrophy and CAG repeat length: A genetic screening of polyglutamine disease genes in Italian patients. <i>Neuroscience Letters</i> , 2018, 678, 37-42.	1.0	10
42	Cortical thickness, stance control, and arithmetic skill: An exploratory study in premanifest Huntington disease. <i>Parkinsonism and Related Disorders</i> , 2018, 51, 17-23.	1.1	22
43	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
44	G08...An italian study to estimate the frequency of the intermediate triplet length in the huntingtin gene: 1/20 subject carries an allele with 27-35 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. <i>Italian Journal of Pediatrics</i> , 2018, 44, 128.	1.0	12
46	Concurrent <i>AFG3L2</i> and <i>SPG7</i> mutations associated with syndromic parkinsonism and optic atrophy with aberrant OPA1 processing and mitochondrial network fragmentation. <i>Human Mutation</i> , 2018, 39, 2060-2071.	1.1	32
47	The role of clinical and neuroimaging features in the diagnosis of CADASIL. <i>Journal of Neurology</i> , 2018, 265, 2934-2943.	1.8	25
48	<i>HCN1</i> mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. <i>Brain</i> , 2018, 141, 3160-3178.	3.7	96
49	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. <i>Neurobiology of Aging</i> , 2018, 71, 266.e1-266.e10.	1.5	59
50	A novel de novo HCN1 loss-of-function mutation in genetic generalized epilepsy causing increased neuronal excitability. <i>Neurobiology of Disease</i> , 2018, 118, 55-63.	2.1	47
51	A Loss-of-Function HCN4 Mutation Associated With Familial Benign Myoclonic Epilepsy in Infancy Causes Increased Neuronal Excitability. <i>Frontiers in Molecular Neuroscience</i> , 2018, 11, 269.	1.4	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. <i>Neurotherapeutics</i> , 2018, 15, 1112-1126.	2.1	56
53	Cortical markers of cognitive syndromes in amyotrophic lateral sclerosis. <i>NeuroImage: Clinical</i> , 2018, 19, 675-682.	1.4	24
54	Reduced Cancer Incidence in Huntington's Disease: Analysis in the Registry Study. <i>Journal of Huntington's Disease</i> , 2018, 7, 209-222.	0.9	14

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55	ATXN2 trinucleotide repeat length correlates with risk of ALS. <i>Neurobiology of Aging</i> , 2017, 51, 178.e1-178.e9.	1.5	86
56	Alternating Hemiplegia and Epilepsia Partialis Continua: A new phenotype for a novel compound TBC1D24 mutation. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 47, 71-73.	0.9	16
57	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	129
58	Identification of genetic variants associated with Huntington's disease progression: a genome-wide association study. <i>Lancet Neurology</i> , The, 2017, 16, 701-711.	4.9	248
59	Genetic correlation between amyotrophic lateral sclerosis and schizophrenia. <i>Nature Communications</i> , 2017, 8, 14774.	5.8	114
60	The role of de novo mutations in the development of amyotrophic lateral sclerosis. <i>Human Mutation</i> , 2017, 38, 1534-1541.	1.1	13
61	Stance instability in preclinical SCA1 mutation carriers: A 4-year prospective posturography study. <i>Gait and Posture</i> , 2017, 57, 11-14.	0.6	11
62	Cognitive decline in Huntington's disease expansion gene carriers. <i>Cortex</i> , 2017, 95, 51-62.	1.1	50
63	Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency: delayed hypersensitivity reaction and efficacy of low-dose intermittent supplementation. <i>European Journal of Neurology</i> , 2017, 24, e41-e42.	1.7	6
64	Amyotrophic lateral sclerosis causes small fiber pathology. <i>European Journal of Neurology</i> , 2016, 23, 416-420.	1.7	65
65	No effect of <i>AR</i> polyG polymorphism on spinal and bulbar muscular atrophy phenotype. <i>European Journal of Neurology</i> , 2016, 23, 1134-1136.	1.7	8
66	B35...Glutamine synthetase-1 induces autophagy and neuronal survival in a drosophila model huntington's disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, A21.2-A21.	0.9	1
67	ASAH1 variant causing a mild SMA phenotype with no myoclonic epilepsy: a clinical, biochemical and molecular study. <i>European Journal of Human Genetics</i> , 2016, 24, 1578-1583.	1.4	18
68	Ataxia with vitamin E deficiency caused by a new compound heterozygous mutation. <i>Neurological Sciences</i> , 2016, 37, 1571-1572.	0.9	1
69	In-vivo brain H1-MR-Spectroscopy identification and quantification of 2-hydroxyglutarate in L-2-Hydroxyglutaric aciduria. <i>Brain Research</i> , 2016, 1648, 506-511.	1.1	9
70	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	9.4	494
71	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	9.4	218
72	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, 11253.	5.8	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.	0.9	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.	0.9	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.	0.9	0
76	Clinical Pregenetic Screening for Stroke Monogenic Diseases. Stroke, 2016, 47, 1702-1709.	1.0	34
77	Association of a Locus in the <i>CAMTA1</i> Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	4.5	57
78	Early white matter involvement in an infant carrying a novel mutation in ACOX1. European Journal of Paediatric Neurology, 2016, 20, 431-434.	0.7	4
79	The role of androgen receptor gene variants on SBMA phenotype. Journal of the Neurological Sciences, 2015, 357, e231.	0.3	0
80	Clinical and molecular report of novel GALC mutations in Moroccan patient with Krabbe disease: case report. BMC Pediatrics, 2015, 15, 182.	0.7	5
81	<i>PEX7</i> Mutations 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. <i>Neurobiology of Aging</i> , 2014, 35, 2420.e13-2420.e14.	1.5	16
92	Atypical Friedreich ataxia in patients with FXN p.R165P point mutation or comorbid hemochromatosis. <i>Parkinsonism and Related Disorders</i> , 2014, 20, 919-923.	1.1	10
93	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2014, 23, 2220-2231.	1.4	123
94	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	3.8	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. <i>Pediatric Neurology</i> , 2014, 51, 430-433.	1.0	9
96	E25 Fmri Signal Changes In Frontal Cortex Correlates With Years-to-disease-onset In A Group Of Young Premanifest Huntington Disease Subjects. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, A45-A45.	0.9	0
97	Human Adipose-Derived Mesenchymal Stem Cells as a New Model of Spinal and Bulbar Muscular Atrophy. <i>PLoS ONE</i> , 2014, 9, e112746.	1.1	15
98	SETX mutations are a frequent genetic cause of juvenile and adult onset cerebellar ataxia with neuropathy and elevated serum alpha-fetoprotein. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 123.	1.2	31
99	Granny trips down: is she carrying the big bad wolf?. <i>Neurological Sciences</i> , 2013, 34, 2077-2079.	0.9	2
100	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntington's disease motor onset. <i>Neurogenetics</i> , 2013, 14, 173-179.	0.7	10
101	Suicidal ideation in a European Huntington's disease population. <i>Journal of Affective Disorders</i> , 2013, 151, 248-258.	2.0	74
102	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. <i>Neurobiology of Aging</i> , 2013, 34, 1517.e9-1517.e10.	1.5	35
103	Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2013, 34, 2695.e11-2695.e12.	1.5	30
104	The C9ORF72 expansion mutation is a common cause of ALS+/-FTD in Europe and has a single founder. <i>European Journal of Human Genetics</i> , 2013, 21, 102-108.	1.4	201
105	Ubiqilin 2 mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 183-187.	0.9	74
106	The V471A Polymorphism in Autophagy-Related Gene ATG7 Modifies Age at Onset Specifically in Italian Huntington Disease Patients. <i>PLoS ONE</i> , 2013, 8, e68951.	1.1	49
107	Î2-Defensin Genomic Copy Number Does Not Influence the Age of Onset in Huntington's Disease. <i>Journal of Huntington's Disease</i> , 2013, 2, 107-124.	0.9	1
108	N01...A 6-years experience of genetic counselling in Huntington disease (2006-2011): constant socio-demographic profile of at-risk individuals requesting genetic predictive test. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, A48.4-A49.	0.9	0

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109	Discrepancies in reporting the CAG repeat lengths for Huntington's disease. <i>European Journal of Human Genetics</i> , 2012, 20, 20-26.	1.4	20
110	Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 265-269.	2.3	15
111	CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. <i>Neurology</i> , 2012, 78, 690-695.	1.5	303
112	Mutations in the proflin 1 gene cause familial amyotrophic lateral sclerosis. <i>Nature</i> , 2012, 488, 499-503.	13.7	522
113	Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency with unknown genetic defect. <i>Neurological Sciences</i> , 2012, 33, 1383-1387.	0.9	10
114	Novel and recurrent spastin mutations in a large series of SPG4 Italian families. <i>Neuroscience Letters</i> , 2012, 528, 42-45.	1.0	5
115	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2012, 33, 630.e1-630.e2.	1.5	17
116	ATAXIN2 CAG-repeat length in Italian patients with amyotrophic lateral sclerosis: risk factor or variant phenotype? Implication for genetic testing and counseling. <i>Neurobiology of Aging</i> , 2012, 33, 1847.e15-1847.e21.	1.5	27
117	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. <i>Neurobiology of Aging</i> , 2012, 33, 2528.e7-2528.e14.	1.5	74
118	Cortical myoclonus in childhood and juvenile onset Huntington's disease. <i>Parkinsonism and Related Disorders</i> , 2012, 18, 794-797.	1.1	22
119	TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntington's disease. <i>Biochemical and Biophysical Research Communications</i> , 2012, 424, 404-408.	1.0	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. <i>Neurobiology of Disease</i> , 2012, 46, 41-51.	2.1	159
121	Preferential expression of mutant ABCD1 allele is common in adrenoleukodystrophy female carriers but unrelated to clinical symptoms. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 10.	1.2	29
122	An Optimized Method for Manufacturing a Clinical Scale Dendritic Cell-Based Vaccine for the Treatment of Glioblastoma. <i>PLoS ONE</i> , 2012, 7, e52301.	1.1	30
123	Observing Huntington's disease: the European Huntington's Disease Network's REGISTRY. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2011, 82, 1409-1412.	0.9	82
124	Paroxysmal exercise-induced dyskinesia with self-limiting partial epilepsy: A novel GLUT-1 mutation with benign phenotype. <i>Parkinsonism and Related Disorders</i> , 2011, 17, 479-481.	1.1	16
125	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. <i>Neurobiology of Aging</i> , 2011, 32, 966-967.	1.5	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. <i>Neuromuscular Disorders</i> , 2011, 21, 129-131.	0.3	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. <i>Neurogenetics</i> , 2011, 12, 193-201.	0.7	46
128	Brain diffusion-weighted imaging in Friedreich's ataxia. <i>Movement Disorders</i> , 2011, 26, 705-712.	2.2	52
129	Novel optineurin mutations in patients with familial and sporadic amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2011, 82, 1239-1243.	0.9	86
130	NMDA receptor gene variations as modifiers in Huntington disease: a replication study. <i>PLOS Currents</i> , 2011, 3, RRN1247.	1.4	20
131	Paraoxonase gene mutations in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2010, 68, 102-107.	2.8	67
132	A novel SOD1 mutation in a young amyotrophic lateral sclerosis patient with a very slowly progressive clinical course. <i>Muscle and Nerve</i> , 2010, 42, 596-597.	1.0	11
133	Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. <i>Nature Genetics</i> , 2010, 42, 313-321.	9.4	291
134	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. <i>Journal of Medical Genetics</i> , 2010, 47, 190-194.	1.5	152
135	Identification of novel and recurrent CACNA1A gene mutations in fifteen patients with episodic ataxia type 2. <i>Journal of the Neurological Sciences</i> , 2010, 291, 30-36.	0.3	63
136	Predictive Genetic Tests in Neurodegenerative Disorders: A Methodological Approach Integrating Psychological Counseling for At-Risk Individuals and Referring Clinicians. <i>European Neurology</i> , 2010, 64, 33-41.	0.6	12
137	Age and founder effect of <i>SOD1</i> A4V mutation causing ALS. <i>Neurology</i> , 2009, 72, 1634-1639.	1.5	84
138	Visual system involvement in patients with Friedreich's ataxia. <i>Brain</i> , 2009, 132, 116-123.	3.7	146
139	Analysis of <i>FUS</i> gene mutation in familial amyotrophic lateral sclerosis within an Italian cohort. <i>Neurology</i> , 2009, 73, 1180-1185.	1.5	139
140	High frequency of <i>TARDBP</i> gene mutations in Italian patients with amyotrophic lateral sclerosis. <i>Human Mutation</i> , 2009, 30, 688-694.	1.1	184
141	Rare association of motor neuron disease and spinocerebellar ataxia type 2 (SCA2): a new case and review of the literature. <i>Journal of Neurology</i> , 2009, 256, 1926-1928.	1.8	42
142	The gene coding for PGC-1 β modifies age at onset in Huntington's Disease. <i>Molecular Neurodegeneration</i> , 2009, 4, 3.	4.4	119
143	A Simple Multiplex Real-Time PCR Methodology for the SMN1 Gene Copy Number Quantification. <i>Genetic Testing and Molecular Biomarkers</i> , 2009, 13, 37-42.	0.3	16
144	A majority of Huntington's disease patients may be treatable by individualized allele-specific RNA interference. <i>Experimental Neurology</i> , 2009, 217, 312-319.	2.0	109

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145	Spinocerebellar ataxia type 28: A novel autosomal dominant cerebellar ataxia characterized by slow progression and ophthalmoparesis. <i>Cerebellum</i> , 2008, 7, 184-188.	1.4	57
146	Identification of new ANG gene mutations in a large cohort of Italian patients with amyotrophic lateral sclerosis. <i>Neurogenetics</i> , 2008, 9, 33-40.	0.7	102
147	Spinal and bulbar muscular atrophy: Skeletal muscle pathology in male patients and heterozygous females. <i>Journal of the Neurological Sciences</i> , 2008, 264, 100-105.	0.3	133
148	The rare G93D mutation causes a slowly progressing lower motor neuron disease. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2008, 9, 35-39.	2.3	18
149	Ataxia With Oculomotor Apraxia Type 1 (AOA1): Clinical and Neuropsychological Features in 2 New Patients and Differential Diagnosis. <i>Journal of Child Neurology</i> , 2008, 23, 895-900.	0.7	22
150	Isolated Vitamin E Deficiency Mimicking Distal Hereditary Motor Neuropathy in a 13-Year-Old Boy. <i>Journal of Child Neurology</i> , 2008, 23, 1328-1330.	0.7	10
151	Paroxysmal movement disorders in <i>GLUT1</i> deficiency syndrome. <i>Neurology</i> , 2008, 71, 146-148.	1.5	73
152	Biological abnormalities of peripheral A2A receptors in a large representation of polyglutamine disorders and Huntington's disease stages. <i>Neurobiology of Disease</i> , 2007, 27, 36-43.	2.1	35
153	Spinocerebellar ataxia type 17 (SCA17): Oculomotor phenotype and clinical characterization of 15 Italian patients. <i>Journal of Neurology</i> , 2007, 254, 1538-1546.	1.8	78
154	Frataxin gene point mutations in Italian Friedreich ataxia patients. <i>Neurogenetics</i> , 2007, 8, 289-299.	0.7	71
155	Large Pathogenic Expansions in the SCA2 and SCA7 Genes Can Be Detected by Fluorescent Repeat-Primed Polymerase Chain Reaction Assay. <i>Journal of Molecular Diagnostics</i> , 2006, 8, 128-132.	1.2	31
156	The A467T and W748S POLG substitutions are a rare cause of adult-onset ataxia in Europe. <i>Brain</i> , 2006, 130, E69-E69.	3.7	14
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