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	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. Science, 1996, 271, 1423-1427.	12.6	2,642
2	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
3	An autosomal dominant disorder with multiple deletions of mitochondrial DNA starting at the D-loop region. Nature, 1989, 339, 309-311.	27.8	640
4	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. Nature, 2012, 488, 499-503.	27.8	522
5	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
6	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
7	Maternally inherited myopathy and cardiomyopathy: association with mutation in mitochondrial DNA tRNALeu(UUR). Lancet, The, 1991, 338, 143-147.	13.7	395
8	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
9	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
10	CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. Neurology, 2012, 78, 690-695.	1.1	303
11	Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. Nature Genetics, 2010, 42, 313-321.	21.4	291
12	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
13	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
14	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
15	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
16	High frequency ofTARDBPgene mutations in Italian patients with amyotrophic lateral sclerosis. Human Mutation, 2009, 30, 688-694.	2.5	184
17	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174
18	Homozygosity for CAG mutation in Huntington disease is associated with a more severe clinical course. Brain, 2003, 126, 946-955.	7.6	173

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19	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
20	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2010, 47, 190-194.	3.2	152
21	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
22	Visual system involvement in patients with Friedreich's ataxia. Brain, 2009, 132, 116-123.	7.6	146
23	Human frataxin maintains mitochondrial iron homeostasis in Saccharomyces cerevisiae. Human Molecular Genetics, 2000, 9, 2523-2530.	2.9	140
24	Analysis of FUS gene mutation in familial amyotrophic lateral sclerosis within an Italian cohort. Neurology, 2009, 73, 1180-1185.	1.1	139
25	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
26	Overlapping phenotypes in complex spastic paraplegias SPG11, SPG15, SPG35 and SPG48. Brain, 2014, 137, 1907-1920.	7.6	133
27	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
28	Molecular Genetics of Hereditary Spinocerebellar Ataxia. Archives of Neurology, 2004, 61, 727.	4.5	130
29	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
30	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
31	SCA28, a novel form of autosomal dominant cerebellar ataxia on chromosome 18p11.22â€“q11.2. Brain, 2006, 129, 235-242.	7.6	122
32	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. Neurology, 1991, 41, 1053-1053.	1.1	120
33	The gene coding for PGC-1Î± modifies age at onset in Huntington's Disease. Molecular Neurodegeneration, 2009, 4, 3.	10.8	119
34	Genetic correlation between amyotrophic lateral sclerosis and schizophrenia. Nature Communications, 2017, 8, 14774.	12.8	114
35	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
36	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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37	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
38	Tissue distribution and transmission of mitochondrial DNA deletions in mitochondrial myopathies. Annals of Neurology, 1990, 28, 94-97.	5.3	98
39	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
40	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	7.6	96
41	FMR1 gene premutation is a frequent genetic cause of late-onset sporadic cerebellar ataxia. Neurology, 2005, 64, 145-147.	1.1	90
42	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
43	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
44	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
45	Frataxin expression rescues mitochondrial dysfunctions in FRDA cells. Human Molecular Genetics, 2001, 10, 2099-2107.	2.9	84
46	Age and founder effect of SOD1 A4V mutation causing ALS. Neurology, 2009, 72, 1634-1639.	1.1	84
47	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
48	Loss of AP-5 results in accumulation of aberrant endolysosomes: defining a new type of lysosomal storage disease. Human Molecular Genetics, 2015, 24, 4984-4996.	2.9	80
49	Fumarase deficiency is an autosomal recessive encephalopathy affecting both the mitochondrial and the cytosolic enzymes. Neurology, 1990, 40, 495-495.	1.1	79
50	Spinocerebellar ataxia type 17 (SCA17): Oculomotor phenotype and clinical characterization of 15 Italian patients. Journal of Neurology, 2007, 254, 1538-1546.	3.6	78
51	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
52	Suicidal ideation in a European Huntington's disease population. Journal of Affective Disorders, 2013, 151, 248-258.	4.1	74
53	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
54	Paroxysmal movement disorders in GLUT1 deficiency syndrome. Neurology, 2008, 71, 146-148.	1.1	73

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55	Atypical movement disorders in the early stages of Huntington's disease: clinical and genetic analysis. Clinical Genetics, 2001, 58, 50-56.	2.0	72
56	The complex clinical and genetic classification of inherited ataxias. II. Autosomal recessive ataxias. Neurological Sciences, 2001, 22, 219-228.	1.9	71
57	Clinical and Genetic Findings in 26 Italian Patients with Lafora Disease. Epilepsia, 2006, 47, 640-643.	5.1	71
58	Frataxin gene point mutations in Italian Friedreich ataxia patients. Neurogenetics, 2007, 8, 289-299.	1.4	71
59	Paraoxonase gene mutations in amyotrophic lateral sclerosis. Annals of Neurology, 2010, 68, 102-107.	5.3	67
60	Assignment of the Human Carnitine Palmitoyltransferase II Gene (CPT1) to Chromosome 1p32. Genomics, 1994, 24, 195-197.	2.9	65
61	Amyotrophic lateral sclerosis causes small fiber pathology. European Journal of Neurology, 2016, 23, 416-420.	3.3	65
62	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
63	Relative Frequencies of CAG Expansions in Spinocerebellar Ataxia and Dentatorubropallidoluysian Atrophy in 116 Italian Families. European Neurology, 2000, 44, 31-36.	1.4	63
64	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
65	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
66	Spinocerebellar ataxia type 28: A novel autosomal dominant cerebellar ataxia characterized by slow progression and ophthalmoparesis. Cerebellum, 2008, 7, 184-188.	2.5	57
67	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
68	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
69	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
70	Brain diffusionâ€weighted imaging in Friedreich's ataxia. Movement Disorders, 2011, 26, 705-712.	3.9	52
71	Cognitive decline in Huntington's disease expansion gene carriers. Cortex, 2017, 95, 51-62.	2.4	50
72	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

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73	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
74	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
75	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
76	The S18Y polymorphism in the UCHL1 gene is a genetic modifier in Huntingtonâ€™s disease. Neurogenetics, 2006, 7, 27-30.	1.4	47
77	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
78	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
79	cDNA cloning and mitochondrial import of the beta-subunit of the human electron-transfer flavoprotein. FEBS Journal, 1993, 213, 1003-1008.	0.2	44
80	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	3.6	44
81	Prevalence of Inherited Ataxias in the Province of Padua, Italy. Neuroepidemiology, 2004, 23, 275-280.	2.3	43
82	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
83	Frataxin gene editing rescues Friedreichâ€™s ataxia pathology in dorsal root ganglia organoid-derived sensory neurons. Nature Communications, 2020, 11, 4178.	12.8	42
84	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
85	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
86	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
87	Genetic analysis of candidate genes modifying the age-at-onset in Huntingtonâ€™s disease. Human Genetics, 2006, 120, 285-292.	3.8	39
88	Pathogenic effect of an intermediate-size SCA-6 allele (CAG) ₁₉ in a homozygous patient. Neurology, 2001, 57, 1502-1504.	1.1	37
89	Expanding sialidosis spectrum by genome-wide screening. Neurology, 2014, 82, 2003-2006.	1.1	37
90	Myoclonic dystonia as unique presentation of isolated vitamin E deficiency in a young patient. Movement Disorders, 2002, 17, 612-614.	3.9	36

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91	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
92	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
93	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
94	Clinical Pregenetic Screening for Stroke Monogenic Diseases. Stroke, 2016, 47, 1702-1709.	2.0	34
95	Heterozygous D90A-SOD1 mutation in a patient with facial onset sensory motor neuronopathy (FOSMN) syndrome: a bridge to amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1009-1011.	1.9	32
96	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
97	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
98	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
99	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
100	Neonatal developmental and epileptic encephalopathy due to autosomal recessive variants in SLC13A5 gene. Epilepsia, 2020, 61, 2474-2485.	5.1	31
101	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
102	Circulating MyomiRs as Potential Biomarkers to Monitor Response to Nusinersen in Pediatric SMA Patients. Biomedicines, 2020, 8, 21.	3.2	30
103	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
104	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
105	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. Neurobiology of Aging, 2011, 32, 966-967.	3.1	28
106	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
107	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
108	The role of clinical and neuroimaging features in the diagnosis of CADASIL. Journal of Neurology, 2018, 265, 2934-2943.	3.6	25

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109	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
110	Cortical markers of cognitive syndromes in amyotrophic lateral sclerosis. NeuroImage: Clinical, 2018, 19, 675-682.	2.7	24
111	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
112	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
113	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8.	3.8	23
114	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
115	Cortical myoclonus in childhood and juvenile onset Huntington's disease. Parkinsonism and Related Disorders, 2012, 18, 794-797.	2.2	22
116	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
117	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
118	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
119	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
120	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
121	Discrepancies in reporting the CAG repeat lengths for Huntington's disease. European Journal of Human Genetics, 2012, 20, 20-26.	2.8	20
122	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
123	NMDA receptor gene variations as modifiers in Huntington disease: a replication study. PLOS Currents, 2011, 3, RRN1247.	1.4	20
124	Progressive Myoclonus Epilepsies. Neurology: Genetics, 2021, 7, e641.	1.9	20
125	Cognitive and psychiatric characterization of patients with Huntington's disease and their at-risk relatives. Neurological Sciences, 2002, 23, s105-s106.	1.9	19
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	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
128	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
129	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
130	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
131	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
132	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
133	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e1-630.e2.	3.1	17
134	Clinical and genetic characteristics of late-onset Huntington's disease. Parkinsonism and Related Disorders, 2019, 61, 101-105.	2.2	17
135	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
136	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
137	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
138	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
139	Spinocerebellar Ataxia Tethering PCR. Journal of Molecular Diagnostics, 2018, 20, 289-297.	2.8	16
140	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
141	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
142	Clinical and molecular findings in the first identified Italian family with dentatorubral-pallidoluysian atrophy. Acta Neurologica Scandinavica, 1998, 98, 324-327.	2.1	15
143	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
144	Late-onset Huntingtonâ€™s disease with 40â€“42 CAG expansion. Neurological Sciences, 2020, 41, 869-876.	1.9	15

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145	Human Adipose-Derived Mesenchymal Stem Cells as a New Model of Spinal and Bulbar Muscular Atrophy. PLoS ONE, 2014, 9, e112746.	2.5	15
146	The A467T and W748S POLG substitutions are a rare cause of adult-onset ataxia in Europe. Brain, 2006, 130, E69-E69.	7.6	14
147	New <scp>FIG</scp>4 gene mutations causing aggressive <scp>ALS</scp>. European Journal of Neurology, 2018, 25, e41-e42.	3.3	14
148	Hypomyelinating leukodystrophies in adults: Clinical and genetic features. European Journal of Neurology, 2021, 28, 934-944.	3.3	14
149	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
150	Reduced Cancer Incidence in Huntington's Disease: Analysis in the Registry Study. Journal of Huntington's Disease, 2018, 7, 209-222.	1.9	14
151	Sensitivity of FRDA Lymphoblasts to Salts of Transition Metal Ions. Antioxidants and Redox Signaling, 2000, 2, 461-465.	5.4	13
152	The role of de novo mutations in the development of amyotrophic lateral sclerosis. Human Mutation, 2017, 38, 1534-1541.	2.5	13
153	Conventional and Unconventional Therapeutic Strategies for Sialidosis Type I. Journal of Clinical Medicine, 2020, 9, 695.	2.4	13
154	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
155	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
156	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
157	Stance instability in preclinical SCA1 mutation carriers: A 4-year prospective posturography study. Gait and Posture, 2017, 57, 11-14.	1.4	11
158	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
159	Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency with unknown genetic defect. Neurological Sciences, 2012, 33, 1383-1387.	1.9	10
160	Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntingtonâ€™s disease motor onset. Neurogenetics, 2013, 14, 173-179.	1.4	10
161	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
162	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

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163	Cognitive Syndromes and C9orf72 Mutation Are Not Related to Cerebellar Degeneration in Amyotrophic Lateral Sclerosis. Frontiers in Neuroscience, 2019, 13, 440.	2.8	10
164	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
165	Frequency and distribution of polyQ disease intermediate-length repeat alleles in healthy Italian population. Neurological Sciences, 2020, 41, 1475-1482.	1.9	10
166	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
167	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
168	Familial vitamin E deficiency: Multiorgan complications support the adverse role of oxidative stress. Nutrition, 2019, 63-64, 57-60.	2.4	9
169	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
170	PEX7Mutations Cause Congenital Cataract Retinopathy and Late-Onset Ataxia and Cognitive		

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