

Cinzia Gellera

List of Publications by Year in descending order

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206
papers

16,092
citations

18465

62
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209
docs citations

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times ranked

16026
citing authors

#	ARTICLE	IF	CITATIONS
1	Friedreich's Ataxia: Autosomal Recessive Disease Caused by an Intronic GAA Triplet Repeat Expansion. <i>Science</i> , 1996, 271, 1423-1427.	6.0	2,642
2	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. <i>Science</i> , 2015, 347, 1436-1441.	6.0	823
3	An autosomal dominant disorder with multiple deletions of mitochondrial DNA starting at the D-loop region. <i>Nature</i> , 1989, 339, 309-311.	13.7	640
4	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. <i>Nature</i> , 2012, 488, 499-503.	13.7	522
5	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
6	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
7	Maternally inherited myopathy and cardiomyopathy: association with mutation in mitochondrial DNA tRNA ^{Leu} (UUR). <i>Lancet, The</i> , 1991, 338, 143-147.	6.3	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. <i>Human Molecular Genetics</i> , 1999, 8, 425-430.	1.4	349
9	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	3.8	308
10	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
11	Mutations in the mitochondrial protease gene AFG3L2 cause dominant hereditary ataxia SCA28. <i>Nature Genetics</i> , 2010, 42, 313-321.	9.4	291
12	Identification of genetic variants associated with Huntington's disease progression: a genome-wide association study. <i>Lancet Neurology, The</i> , 2017, 16, 701-711.	4.9	248
13	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
14	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	9.4	218
15	The C9ORF72 expansion mutation is a common cause of ALS+ α^{FTD} in Europe and has a single founder. <i>European Journal of Human Genetics</i> , 2013, 21, 102-108.	1.4	201
16	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
17	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, 11253.	5.8	174
18	Homozygosity for CAG mutation in Huntington disease is associated with a more severe clinical course. <i>Brain</i> , 2003, 126, 946-955.	3.7	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. <i>Neurobiology of Disease</i> , 2012, 46, 41-51.	2.1	159
20	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. <i>Journal of Medical Genetics</i> , 2010, 47, 190-194.	1.5	152
21	Molecular characterization of inherited carnitine palmitoyltransferase II deficiency.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1992, 89, 8429-8433.	3.3	151
22	Visual system involvement in patients with Friedreich's ataxia. <i>Brain</i> , 2009, 132, 116-123.	3.7	146
23	Human frataxin maintains mitochondrial iron homeostasis in <i>Saccharomyces cerevisiae</i> . <i>Human Molecular Genetics</i> , 2000, 9, 2523-2530.	1.4	140
24	Analysis of FUS gene mutation in familial amyotrophic lateral sclerosis within an Italian cohort. <i>Neurology</i> , 2009, 73, 1180-1185.	1.5	139
25	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
26	Overlapping phenotypes in complex spastic paraplegias SPG11, SPG15, SPG35 and SPG48. <i>Brain</i> , 2014, 137, 1907-1920.	3.7	133
27	Ataxia with isolated vitamin E deficiency: neurological phenotype, clinical follow-up and novel mutations in TTPA gene in Italian families. <i>Neurological Sciences</i> , 2004, 25, 130-137.	0.9	131
28	Molecular Genetics of Hereditary Spinocerebellar Ataxia. <i>Archives of Neurology</i> , 2004, 61, 727.	4.9	130
29	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017, 9, .	5.8	129
30	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
31	SCA28, a novel form of autosomal dominant cerebellar ataxia on chromosome 18p11.22-q11.2. <i>Brain</i> , 2006, 129, 235-242.	3.7	122
32	Dominantly inherited mitochondrial myopathy with multiple deletions of mitochondrial DNA. <i>Neurology</i> , 1991, 41, 1053-1053.	1.5	120
33	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
34	Genetic correlation between amyotrophic lateral sclerosis and schizophrenia. <i>Nature Communications</i> , 2017, 8, 14774.	5.8	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. <i>Neuromuscular Disorders</i> , 2000, 10, 391-397.	0.3	112
36	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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37	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
38	Tissue distribution and transmission of mitochondrial DNA deletions in mitochondrial myopathies. <i>Annals of Neurology</i> , 1990, 28, 94-97.	2.8	98
39	Psychiatric Symptoms Do Not Correlate With Cognitive Decline, Motor Symptoms, or CAG Repeat Length in Huntington's Disease. <i>Archives of Neurology</i> , 1996, 53, 493-497.	4.9	97
40	<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
41	<i>FMR1</i> gene premutation is a frequent genetic cause of late-onset sporadic cerebellar ataxia. <i>Neurology</i> , 2005, 64, 145-147.	1.5	90
42	Carnitine palmitoyltransferase II deficiency: structure of the gene and characterization of two novel disease-causing mutations. <i>Human Molecular Genetics</i> , 1995, 4, 19-29.	1.4	89
43	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
44	ATXN2 trinucleotide repeat length correlates with risk of ALS. <i>Neurobiology of Aging</i> , 2017, 51, 178.e1-178.e9.	1.5	86
45	Frataxin expression rescues mitochondrial dysfunctions in FRDA cells. <i>Human Molecular Genetics</i> , 2001, 10, 2099-2107.	1.4	84
46	Age and founder effect of <i>SOD1</i> A4V mutation causing ALS. <i>Neurology</i> , 2009, 72, 1634-1639.	1.5	84
47	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
48	Loss of AP-5 results in accumulation of aberrant endolysosomes: defining a new type of lysosomal storage disease. <i>Human Molecular Genetics</i> , 2015, 24, 4984-4996.	1.4	80
49	Fumarase deficiency is an autosomal recessive encephalopathy affecting both the mitochondrial and the cytosolic enzymes. <i>Neurology</i> , 1990, 40, 495-495.	1.5	79
50	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
51	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
52	Suicidal ideation in a European Huntington's disease population. <i>Journal of Affective Disorders</i> , 2013, 151, 248-258.	2.0	74
53	<i>Ubiquilin 2</i> 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
54	Paroxysmal movement disorders in <i>GLUT1</i> deficiency syndrome. <i>Neurology</i> , 2008, 71, 146-148.	1.5	73

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55	Atypical movement disorders in the early stages of Huntington's disease: clinical and genetic analysis. <i>Clinical Genetics</i> , 2001, 58, 50-56.	1.0	72
56	The complex clinical and genetic classification of inherited ataxias. II. Autosomal recessive ataxias. <i>Neurological Sciences</i> , 2001, 22, 219-228.	0.9	71
57	Clinical and Genetic Findings in 26 Italian Patients with Lafora Disease. <i>Epilepsia</i> , 2006, 47, 640-643.	2.6	71
58	Frataxin gene point mutations in Italian Friedreich ataxia patients. <i>Neurogenetics</i> , 2007, 8, 289-299.	0.7	71
59	Paraoxonase gene mutations in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2010, 68, 102-107.	2.8	67
60	Assignment of the Human Carnitine Palmitoyltransferase II Gene (CPT1) to Chromosome 1p32. <i>Genomics</i> , 1994, 24, 195-197.	1.3	65
61	Amyotrophic lateral sclerosis causes small fiber pathology. <i>European Journal of Neurology</i> , 2016, 23, 416-420.	1.7	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. <i>Journal of Neurology</i> , 1999, 246, 389-393.	1.8	63
63	Relative Frequencies of CAG Expansions in Spinocerebellar Ataxia and Dentatorubropallidolusian Atrophy in 116 Italian Families. <i>European Neurology</i> , 2000, 44, 31-36.	0.6	63
64	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
65	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
66	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
67	Association of a Locus in the <i>CAMTA1</i> Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2016, 73, 812.	4.5	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. <i>Neurotherapeutics</i> , 2018, 15, 1112-1126.	2.1	56
69	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. <i>Neuron</i> , 2021, 109, 448-460.e4.	3.8	56
70	Brain diffusion-weighted imaging in Friedreich's ataxia. <i>Movement Disorders</i> , 2011, 26, 705-712.	2.2	52
71	Cognitive decline in Huntington's disease expansion gene carriers. <i>Cortex</i> , 2017, 95, 51-62.	1.1	50
72	Normalization of short-chain acylcoenzyme a dehydrogenase after riboflavin treatment in a girl with multiple acylcoenzyme a dehydrogenase-deficient myopathy. <i>Annals of Neurology</i> , 1989, 25, 479-484.	2.8	49

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73	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
74	Detection of Large Pathogenic Expansions in FRDA1, SCA10, and SCA12 Genes Using a Simple Fluorescent Repeat-Primed PCR Assay. <i>Journal of Molecular Diagnostics</i> , 2004, 6, 96-100.	1.2	48
75	Superoxide dismutase gene mutations in Italian patients with familial and sporadic amyotrophic lateral sclerosis: identification of three novel missense mutations. <i>Neuromuscular Disorders</i> , 2001, 11, 404-410.	0.3	47
76	The S18Y polymorphism in the UCHL1 gene is a genetic modifier in Huntingtonâ€™s disease. <i>Neurogenetics</i> , 2006, 7, 27-30.	0.7	47
77	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
78	Ataxia with oculomotor apraxia type 1 (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
79	cDNA cloning and mitochondrial import of the beta-subunit of the human electron-transfer flavoprotein. <i>FEBS Journal</i> , 1993, 213, 1003-1008.	0.2	44
80	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. <i>Journal of Neurology</i> , 2015, 262, 1376-1378.	1.8	44
81	Prevalence of Inherited Ataxias in the Province of Padua, Italy. <i>Neuroepidemiology</i> , 2004, 23, 275-280.	1.1	43
82	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
83	Frataxin gene editing rescues Friedreichâ€™s ataxia pathology in dorsal root ganglia organoid-derived sensory neurons. <i>Nature Communications</i> , 2020, 11, 4178.	5.8	42
84	Multiple founder effects in spinal and bulbar muscular atrophy (SBMA, Kennedy disease) around the world. <i>European Journal of Human Genetics</i> , 2001, 9, 431-436.	1.4	41
85	The gender effect in juvenile Huntington disease patients of Italian origin. <i>American Journal of Medical Genetics Part A</i> , 2004, 125B, 92-98.	2.4	41
86	Family and molecular data for a fine analysis of age at onset in Huntington disease. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 366-373.	2.4	40
87	Genetic analysis of candidate genes modifying the age-at-onset in Huntingtonâ€™s disease. <i>Human Genetics</i> , 2006, 120, 285-292.	1.8	39
88	Pathogenic effect of an intermediate-size SCA-6 allele (CAG) ₁₉ in a homozygous patient. <i>Neurology</i> , 2001, 57, 1502-1504.	1.5	37
89	Expanding sialidosis spectrum by genome-wide screening. <i>Neurology</i> , 2014, 82, 2003-2006.	1.5	37
90	Myoclonic dystonia as unique presentation of isolated vitamin E deficiency in a young patient. <i>Movement Disorders</i> , 2002, 17, 612-614.	2.2	36

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91	Very late onset Friedreich's ataxia without cardiomyopathy is associated with limited GAA expansion in the <i>X25</i> gene. <i>Neurology</i> , 1997, 49, 1153-1155.	1.5	35
92	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
93	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
94	Clinical Pre-genetic Screening for Stroke Monogenic Diseases. <i>Stroke</i> , 2016, 47, 1702-1709.	1.0	34
95	Heterozygous D90A-SOD1 mutation in a patient with facial onset sensory motor neuronopathy (FOSMN) syndrome: a bridge to amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 1009-1011.	0.9	32
96	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
97	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
98	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
99	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
100	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
101	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
102	Circulating MyomiRs as Potential Biomarkers to Monitor Response to Nusinersen in Pediatric SMA Patients. <i>Biomedicines</i> , 2020, 8, 21.	1.4	30
103	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
104	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
105	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. <i>Neurobiology of Aging</i> , 2011, 32, 966-967.	1.5	28
106	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
107	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
108	The role of clinical and neuroimaging features in the diagnosis of CADASIL. <i>Journal of Neurology</i> , 2018, 265, 2934-2943.	1.8	25

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109	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
110	Cortical markers of cognitive syndromes in amyotrophic lateral sclerosis. <i>NeuroImage: Clinical</i> , 2018, 19, 675-682.	1.4	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. <i>Journal of Clinical Medicine</i> , 2020, 9, 412.	1.0	24
112	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
113	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. <i>Npj Genomic Medicine</i> , 2022, 7, 8.	1.7	23
114	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
115	Cortical myoclonus in childhood and juvenile onset Huntington's disease. <i>Parkinsonism and Related Disorders</i> , 2012, 18, 794-797.	1.1	22
116	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
117	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
118	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
119	Evidence for two distinct mitochondrial malic enzymes in human skeletal muscle: purification and properties of the NAD(P) ⁺ -dependent enzyme. <i>BBA - Proteins and Proteomics</i> , 1987, 916, 446-454.	2.1	20
120	Efficacious vitamin E treatment in a child with ataxia with isolated vitamin E deficiency. <i>European Journal of Pediatrics</i> , 2006, 165, 494-495.	1.3	20
121	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
122	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
123	NMDA receptor gene variations as modifiers in Huntington disease: a replication study. <i>PLOS Currents</i> , 2011, 3, RRN1247.	1.4	20
124	Progressive Myoclonus Epilepsies. <i>Neurology: Genetics</i> , 2021, 7, e641.	0.9	20
125	Cognitive and psychiatric characterization of patients with Huntington's disease and their at-risk relatives. <i>Neurological Sciences</i> , 2002, 23, s105-s106.	0.9	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. <i>Neuromuscular Disorders</i> , 2011, 21, 129-131.	0.3	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.	1.4	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.	2.3	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.	1.4	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.	1.8	18
131	Glutamine Synthetase 1 Increases Autophagy Lysosomal Degradation of Mutant Huntingtin Aggregates in Neurons, Ameliorating Motility in a Drosophila Model for Huntingtin's Disease. Cells, 2020, 9, 196.	1.8	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.	1.1	17
133	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e1-630.e2.	1.5	17
134	Clinical and genetic characteristics of late-onset Huntington's disease. Parkinsonism and Related Disorders, 2019, 61, 101-105.	1.1	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.	0.3	16
136	Paroxysmal exercise-induced dyskinesia with self-limiting partial epilepsy: A novel GLUT-1 mutation with benign phenotype. Parkinsonism and Related Disorders, 2011, 17, 479-481.	1.1	16
137	Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. Neurobiology of Aging, 2014, 35, 2420.e13-2420.e14.	1.5	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.	0.9	16
139	Spinocerebellar Ataxia Tethering PCR. Journal of Molecular Diagnostics, 2018, 20, 289-297.	1.2	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.	0.9	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.	1.1	16
142	Clinical and molecular findings in the first identified Italian family with dentatorubral-pallidoluysian atrophy. Acta Neurologica Scandinavica, 1998, 98, 324-327.	1.0	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.	2.3	15
144	Late-onset Huntington's disease with 40-42 CAG expansion. Neurological Sciences, 2020, 41, 869-876.	0.9	15

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145	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
146	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
147	New <i>FIG4</i> gene mutations causing aggressive <i>ALS</i> . <i>European Journal of Neurology</i> , 2018, 25, e41-e42.	1.7	14
148	Hypomyelinating leukodystrophies in adults: Clinical and genetic features. <i>European Journal of Neurology</i> , 2021, 28, 934-944.	1.7	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. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5673.	1.8	14
150	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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