Henry Houlden

List of Publications by Year
in descending order

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653 papers	44,887 citations	2802 94 h-index	3323 184 g-index
723 all docs	723 docs citations	723 times ranked	39744 citing authors

#	Article	IF	Citations
1	Association of missense and 5â€²-splice-site mutations in tau with the inherited dementia FTDP-17. Nature, 1998, 393, 702-705.	27.8	3,333
2	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	21.4	1,745
3	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nature Genetics, 2014, 46, 989-993.	21.4	1,685
4	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet Neurology, The, 2019, 18, 1091-1102.	10.2	1,414
5	A pathogenic mutation for probable Alzheimer's disease in the APP gene at the Nâ€“terminus of Î²â€“amyloid. Nature Genetics, 1992, 1, 345-347.	21.4	1,384
6	Early-onset Alzheimer's disease caused by mutations at codon 717 of the Î²-amyloid precursor protein gene. Nature, 1991, 353, 844-846.	27.8	1,202
7	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	10.2	1,039
8	Association of an Extended Haplotype in the Tau Gene with Progressive Supranuclear Palsy. Human Molecular Genetics, 1999, 8, 711-715.	2.9	749
9	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. Brain, 2009, 132, 1783-1794.	7.6	612
10	A common polymorphism in the brainâ€derived neurotrophic factor gene (BDNF) modulates human cortical plasticity and the response to rTMS. Journal of Physiology, 2008, 586, 5717-5725.	2.9	592
11	The structure of the presenilin 1 (S182) gene and identification of six novel mutations in early onset AD families. Nature Genetics, 1995, 11, 219-222.	21.4	461
12	Parkinson's disease induced pluripotent stem cells with triplication of the Î±-synuclein locus. Nature Communications, 2011, 2, 440.	12.8	406
13	Characterization of PLA2G6 as a locus for dystoniaâ€parkinsonism. Annals of Neurology, 2009, 65, 19-23.	5.3	399
14	A novel Î±-synuclein missense mutation in Parkinson disease. Neurology, 2013, 80, 1062-1064.	1.1	396
15	Corticobasal degeneration and progressive supranuclear palsy share a common tau haplotype. Neurology, 2001, 56, 1702-1706.	1.1	392
16	Î±-Synucleinopathy associated with G51D SNCA mutation: a link between Parkinsonâ€™s disease and multiple system atrophy?. Acta Neuropathologica, 2013, 125, 753-769.	7.7	369
17	A variant of Alzheimer's disease with spastic paraparesis and unusual plaques due to deletion of exon 9 of presenilin 1. Nature Medicine, 1998, 4, 452-455.	30.7	347
18	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. Nature Genetics, 2019, 51, 649-658.	21.4	338

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19	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
20	Hereditary Sensory Neuropathy Type 1 Is Caused by the Accumulation of Two Neurotoxic Sphingolipids. Journal of Biological Chemistry, 2010, 285, 11178-11187.	3.4	320
21	Clinical implications of genetic advances in Charcotâ€“Marieâ€“Tooth disease. Nature Reviews Neurology, 2013, 9, 562-571.	10.1	299
22	Charcotâ€“Marieâ€“Tooth disease: frequency of genetic subtypes and guidelines for genetic testing. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 706-710.	1.9	297
23	Large C9orf72 Hexanucleotide Repeat Expansions Are Seen in Multiple Neurodegenerative Syndromes and Are More Frequent Than Expected in the UK Population. American Journal of Human Genetics, 2013, 92, 345-353.	6.2	297
24	Early onset familial Alzheimerâ€™s disease. Neurology, 2003, 60, 235-239.	1.1	292
25	The Parkinson's diseaseâ€“linked proteins Fbxo7 and Parkin interact to mediate mitophagy. Nature Neuroscience, 2013, 16, 1257-1265.	14.8	292
26	Earlyâ€onset Lâ€dopaâ€responsive parkinsonism with pyramidal signs due to ATP13A2, PLA2G6, FBXO7 and spatacsin mutations. Movement Disorders, 2010, 25, 1791-1800.	3.9	287
27	The genetics and neuropathology of Parkinsonâ€™s disease. Acta Neuropathologica, 2012, 124, 325-338.	7.7	281
28	Mutations in TRPV4 cause Charcot-Marie-Tooth disease type 2C. Nature Genetics, 2010, 42, 170-174.	21.4	278
29	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. PLoS Genetics, 2007, 3, e108.	3.5	269
30	A locus for familial earlyâ€“onset Alzhelmer's disease on the long arm of chromosome 14, proximal to the Î±1â€“antichymotrypsin gene. Nature Genetics, 1992, 2, 340-342.	21.4	266
31	5â€² Splice Site Mutations in tau Associated with the Inherited Dementia FTDP-17 Affect a Stem-Loop Structure That Regulates Alternative Splicing of Exon 10. Journal of Biological Chemistry, 1999, 274, 15134-15143.	3.4	266
32	A mutation in AlzheimerÊ¼s disease destroying a splice acceptor site in the presenilin-1 gene. NeuroReport, 1995, 7, 297-301.	1.2	262
33	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	5.3	257
34	C9orf72 expansions are the most common genetic cause of Huntington disease phenocopies. Neurology, 2014, 82, 292-299.	1.1	252
35	Mutations in the Gene PRRT2 Cause Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions. Cell Reports, 2012, 1, 2-12.	6.4	250
36	Neuroimaging Features of Neurodegeneration with Brain Iron Accumulation. American Journal of Neuroradiology, 2012, 33, 407-414.	2.4	249

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37	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonismâ€“dystonia. Nature Communications, 2016, 7, 11601.	12.8	233
38	The neuropathology, pathophysiology and genetics of multiple system atrophy. Neuropathology and Applied Neurobiology, 2012, 38, 4-24.	3.2	218
39	Defective FA2H leads to a novel form of neurodegeneration with brain iron accumulation (NBIA). Annals of Neurology, 2010, 68, 611-618.	5.3	202
40	Cerebral microbleeds and intracranial haemorrhage risk in patients anticoagulated for atrial fibrillation after acute ischaemic stroke or transient ischaemic attack (CROMIS-2): a multicentre observational cohort study. Lancet Neurology, The, 2018, 17, 539-547.	10.2	192
41	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
42	Mutations in TTBK2, encoding a kinase implicated in tau phosphorylation, segregate with spinocerebellar ataxia type 11. Nature Genetics, 2007, 39, 1434-1436.	21.4	185
43	Widespread Lewy body and tau accumulation in childhood and adult onset dystonia-parkinsonism cases with PLA2G6 mutations. Neurobiology of Aging, 2012, 33, 814-823.	3.1	184
44	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. Annals of Neurology, 2016, 79, 983-990.	5.3	183
45	Spinocerebellar ataxia: an update. Journal of Neurology, 2019, 266, 533-544.	3.6	180
46	Gâ€quadruplexâ€binding small molecules ameliorate C9orf72 <scp>FTD</scp> / <scp>ALS</scp> pathology inÂvitro and inÂvivo. EMBO Molecular Medicine, 2018, 10, 22-31.	6.9	178
47	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
48	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2014, 94, 11-22.	6.2	176
49	Mutation of FA2H underlies a complicated form of hereditary spastic paraplegia (SPG35). Human Mutation, 2010, 31, E1251-E1260.	2.5	174
50	KIF1A, an Axonal Transporter of Synaptic Vesicles, Is Mutated in Hereditary Sensory and Autonomic Neuropathy Type 2. American Journal of Human Genetics, 2011, 89, 219-230.	6.2	172
51	Hereditary spastic paraplegia: from diagnosis to emerging therapeutic approaches. Lancet Neurology, The, 2019, 18, 1136-1146.	10.2	171
52	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. Brain, 2016, 139, 1904-1918.	7.6	170
53	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	7.6	169
54	Mutations in the HSP27 (HSPB1) gene cause dominant, recessive, and sporadic distal HMN/CMT type 2. Neurology, 2008, 71, 1660-1668.	1.1	168

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55	ATP13A2 mutations (PARK9) cause neurodegeneration with brain iron accumulation. Movement Disorders, 2010, 25, 979-984.	3.9	163
56	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. Nature Genetics, 2020, 52, 1303-1313.	21.4	163
57	Brown-Vialetto-Van Laere Syndrome, a Ponto-Bulbar Palsy with Deafness, Is Caused by Mutations in C20orf54. American Journal of Human Genetics, 2010, 86, 485-489.	6.2	161
58	The phenotypic spectrum of DYT24 due to ANO3 mutations. Movement Disorders, 2014, 29, 928-934.	3.9	161
59	PRRT2 gene mutations. Neurology, 2012, 79, 2115-2121.	1.1	159
60	Mutations in BICD2 Cause Dominant Congenital Spinal Muscular Atrophy and Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2013, 92, 965-973.	6.2	156
61	Complete analysis of the presenilin 1 gene in early onset AlzheimerÊ¼s disease. NeuroReport, 1996, 7, 801-805.	1.2	150
62	Frequency oftau mutations in three series of non-Alzheimer's degenerative dementia. Annals of Neurology, 1999, 46, 243-248.	5.3	150
63	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
64	Alphaâ€synuclein mRNA expression in oligodendrocytes in MSA. Glia, 2014, 62, 964-970.	4.9	149
65	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	5.3	148
66	Genetic Analysis of Inherited Leukodystrophies. JAMA Neurology, 2013, 70, 875.	9.0	147
67	Glucocerebrosidase mutations confer a greater risk of dementia during Parkinson's disease course. Movement Disorders, 2012, 27, 393-399.	3.9	144
68	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	7.6	143
69	Cerebral microbleeds and stroke risk after ischaemic stroke or transient ischaemic attack: a pooled analysis of individual patient data from cohort studies. Lancet Neurology, The, 2019, 18, 653-665.	10.2	143
70	ADCY5 mutations are another cause of benign hereditary chorea. Neurology, 2015, 85, 80-88.	1.1	140
71	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. Brain, 2020, 143, 480-490.	7.6	140
72	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.1	139

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73	Clinical and genetic characterization of families with triple A (Allgrove) syndrome. Brain, 2002, 125, 2681-2690.	7.6	137
74	THAP1 mutations (DYT6) are an additional cause of early-onset dystonia. Neurology, 2010, 74, 846-850.	1.1	136
75	Genetic risk factors for intracranial aneurysms. Neurology, 2013, 80, 2154-2165.	1.1	136
76	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid-? concentrations. Annals of Neurology, 2000, 48, 806-808.	5.3	135
77	APOE genotype does not modulate age of onset in families with chromosome 14 encoded Alzheimer's disease. Neuroscience Letters, 1994, 169, 179-180.	2.1	130
78	The clinical and genetic heterogeneity of paroxysmal dyskinesias. Brain, 2015, 138, 3567-3580.	7.6	129
79	Regional Distribution of Amyloid-Bri Deposition and Its Association with Neurofibrillary Degeneration in Familial British Dementia. American Journal of Pathology, 2001, 158, 515-526.	3.8	127
80	Pathogenic VCP Mutations Induce Mitochondrial Uncoupling and Reduced ATP Levels. Neuron, 2013, 78, 57-64.	8.1	127
81	THAP1 mutations and dystonia phenotypes: Genotype phenotype correlations. Movement Disorders, 2012, 27, 1290-1294.	3.9	126
82	Functional Studies of Missense TREM2 Mutations in Human Stem Cell-Derived Microglia. Stem Cell Reports, 2018, 10, 1294-1307.	4.8	124
83	Clinical, pathological and genetic characterization of hereditary sensory and autonomic neuropathy type 1 (HSAN I). Brain, 2006, 129, 411-425.	7.6	122
84	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	122
85	The glucocerobrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. Movement Disorders, 2013, 28, 232-236.	3.9	121
86	Mutations in GBA2 Cause Autosomal-Recessive Cerebellar Ataxia with Spasticity. American Journal of Human Genetics, 2013, 92, 245-251.	6.2	120
87	Hâ€ABC syndrome and DYT4: Variable expressivity or pleiotropy of TUBB4 mutations?. Movement Disorders, 2015, 30, 828-833.	3.9	117
88	Developmental regulation of tau splicing is disrupted in stem cell-derived neurons from frontotemporal dementia patients with the 10 + 16 splice-site mutation in MAPT. Human Molecular Genetics, 2015, 24, 5260-5269.	2.9	116
89	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain, 2012, 135, 2875-2882.	7.6	114
90	Cerebral small vessel disease-related protease HtrA1 processes latent TGF-Î² binding protein 1 and facilitates TGF-Î² signaling. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 16496-16501.	7.1	114

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91	Review: Insights into molecular mechanisms of disease in neurodegeneration with brain iron accumulation: unifying theories. Neuropathology and Applied Neurobiology, 2016, 42, 220-241.	3.2	114
92	Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies. Brain, 2010, 133, 1798-1809.	7.6	113
93	A novel RAB7 mutation associated with ulceroâ€mutilating neuropathy. Annals of Neurology, 2004, 56, 586-590.	5.3	112
94	Exome sequencing identifies a significant variant in methionyl-tRNA synthetase (MARS) in a family with late-onset CMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 1247-1249.	1.9	112
95	A novel TRK A (NTRK1) mutation associated with hereditary sensory and autonomic neuropathy type V. Annals of Neurology, 2001, 49, 521-525.	5.3	110
96	Large, expanded repeats in SCA8 are not confined to patients with cerebellar ataxia. Nature Genetics, 2000, 24, 214-215.	21.4	109
97	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13.	3.1	108
98	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. Cell Reports, 2013, 3, 1795-1805.	6.4	104
99	Age-related penetrance of the C9orf72 repeat expansion. Scientific Reports, 2017, 7, 2116.	3.3	102
100	Rhabdomyolysis: a genetic perspective. Orphanet Journal of Rare Diseases, 2015, 10, 51.	2.7	101
101	Familial PRRT2 mutation with heterogeneous paroxysmal disorders including paroxysmal torticollis and hemiplegic migraine. Developmental Medicine and Child Neurology, 2012, 54, 958-960.	2.1	100
102	Genetic dysfunction of MT-ATP6 causes axonal Charcot-Marie-Tooth disease. Neurology, 2012, 79, 1145-1154.	1.1	97
103	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
104	Chromosome 14-encoded Alzheimer's disease: Genetic and clinicopathological description. Annals of Neurology, 1994, 36, 362-367.	5.3	95
105	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	9.0	95
106	Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. Brain, 2015, 138, 845-861.	7.6	94
107	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
108	Extended phenotypic spectrum of KIF5A mutations. Neurology, 2014, 83, 612-619.	1.1	92

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109	Forebrain deletion of the dystonia protein torsinA causes dystonic-like movements and loss of striatal cholinergic neurons. ELife, 2015, 4, e08352.	6.0	92
110	Neuropathy target esterase impairments cause Oliverâ€“McFarlane and Laurenceâ€“Moon syndromes. Journal of Medical Genetics, 2015, 52, 85-94.	3.2	91
111	Distinct clinical and neuropathological features of G51D SNCA mutation cases compared with SNCA duplication and H50Q mutation. Molecular Neurodegeneration, 2015, 10, 41.	10.8	90
112	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. American Journal of Human Genetics, 2014, 95, 611-621.	6.2	89
113	Mutations in the 5' region of the myotubularin-related protein 2 (MTMR2) gene in autosomal recessive hereditary neuropathy with focally folded myelin. Brain, 2001, 124, 907-915.	7.6	88
114	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
115	Practical approach to the diagnosis of adult-onset leukodystrophies: an updated guide in the genomic era. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 543-555.	1.9	87
116	The inherited ataxias: Genetic heterogeneity, mutation databases, and future directions in research and clinical diagnostics. Human Mutation, 2012, 33, 1324-1332.	2.5	86
117	Genetic variability at the PARK16 locus. European Journal of Human Genetics, 2010, 18, 1356-1359.	2.8	85
118	Excess Î±-synuclein compromises phagocytosis in iPSC-derived macrophages. Scientific Reports, 2017, 7, 9003.	3.3	85
119	Localization of frontotemporal dementia with parkinsonism in an Australian kindred to chromosome 17q21-22. Annals of Neurology, 1997, 42, 794-798.	5.3	83
120	A novel NGF mutation clarifies the molecular mechanism and extends the phenotypic spectrum of the HSAN5 neuropathy. Journal of Medical Genetics, 2011, 48, 131-135.	3.2	83
121	Riboflavin Responsive Mitochondrial Dysfunction in Neurodegenerative Diseases. Journal of Clinical Medicine, 2017, 6, 52.	2.4	83
122	Severe infantile neuropathy with diaphragmatic weakness and its relationship to SMARD1. Brain, 2003, 126, 2682-2692.	7.6	82
123	Frequency of mutations in the genes associated with hereditary sensory and autonomic neuropathy in a UK cohort. Journal of Neurology, 2012, 259, 1673-1685.	3.6	82
124	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. Brain, 2015, 138, 293-310.	7.6	82
125	Defects in the CAPN1 Gene Result in Alterations in Cerebellar Development and Cerebellar Ataxia in Mice and Humans. Cell Reports, 2016, 16, 79-91.	6.4	82
126	Microdeletion in a FAAH pseudogene identified in a patient with high anandamide concentrations and pain insensitivity. British Journal of Anaesthesia, 2019, 123, e249-e253.	3.4	82

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127	Human ataxias: a genetic dissection of inositol triphosphate receptor (ITPR1)-dependent signaling. Trends in Neurosciences, 2010, 33, 211-219.	8.6	81
128	A practical approach to diagnosing adult onset leukodystrophies. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 770-781.	1.9	80
129	The phenotype of Charcotâ€“Marieâ€“Tooth disease type 4C due to SH3TC2 mutations and possible predisposition to an inflammatory neuropathy. Neuromuscular Disorders, 2009, 19, 264-269.	0.6	78
130	A Pathogenic Presenilin-1 Deletion Causes Abberrant Aî²42 Production in the Absence of Congophilic Amyloid Plaques. Journal of Biological Chemistry, 2001, 276, 7233-7239.	3.4	76
131	Use of the Barthel Index and the Functional Independence Measure during early inpatient rehabilitation after single incident brain injury. Clinical Rehabilitation, 2006, 20, 153-159.	2.2	75
132	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. American Journal of Human Genetics, 2014, 95, 590-601.	6.2	75
133	DNA repair in the trinucleotide repeat disorders. Lancet Neurology, The, 2017, 16, 88-96.	10.2	75
134	Towards a complete resolution of the genetic architecture of disease. Trends in Genetics, 2010, 26, 438-442.	6.7	74
135	Recent Advances in the Genetics of Cerebellar Ataxias. Current Neurology and Neuroscience Reports, 2012, 12, 227-236.	4.2	74
136	Whole genome sequencing for the diagnosis of neurological repeat expansion disorders in the UK: a retrospective diagnostic accuracy and prospective clinical validation study. Lancet Neurology, The, 2022, 21, 234-245.	10.2	74
137	Systemic Amyloid Deposits in Familial British Dementia. Journal of Biological Chemistry, 2001, 276, 43909-43914.	3.4	73
138	Clinical and genetic characterization of leukoencephalopathies in adults. Brain, 2017, 140, 1204-1211.	7.6	73
139	Recessive axonal Charcot-Marie-Tooth disease due to compound heterozygous mitofusin 2 mutations. Neurology, 2011, 77, 168-173.	1.1	72
140	Analysis of Mutations in AARS2 in a Series of CSF1R-Negative Patients With Adult-Onset Leukoencephalopathy With Axonal Spheroids and Pigmented Glia. JAMA Neurology, 2016, 73, 1433.	9.0	71
141	Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. Neurology, 2020, 94, e51-e61.	1.1	71
142	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
143	SPG11 mutations are common in familial cases of complicated hereditary spastic paraplegia. Neurology, 2008, 70, 1384-1389.	1.1	69
144	Genetics of neurodegenerative diseases: an overview. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 145, 309-323.	1.8	69

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145	Localization of the Gene for Distal Hereditary Motor Neuronopathy VII (dHMN-VII) to Chromosome 2q14. American Journal of Human Genetics, 2001, 68, 1270-1276.	6.2	68
146	Comprehensive genetic diagnosis of tandem repeat expansion disorders with programmable targeted nanopore sequencing. Science Advances, 2022, 8, eabm5386.	10.3	68
147	PRRT2 Regulates Synaptic Fusion by Directly Modulating SNARE Complex Assembly. Cell Reports, 2018, 22, 820-831.	6.4	67
148	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	3.9	66
149	An update on the genetics, clinical presentation, and pathomechanisms of human riboflavin transporter deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 598-607.	3.6	65
150	Redefining the phenotype of ALSP and AARS2 mutationâ€“related leukodystrophy. Neurology: Genetics, 2017, 3, e135.	1.9	64
151	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	7.6	64
152	Somatic copy number gains of Î±-synuclein (SNCA) in Parkinsonâ€™s disease and multiple system atrophy brains. Brain, 2018, 141, 2419-2431.	7.6	63
153	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	7.6	63
154	Dominant Mutations in GRM1 Cause Spinocerebellar Ataxia Type 44. American Journal of Human Genetics, 2017, 101, 451-458.	6.2	62
155	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
156	Hereditary sensory neuropathies. Current Opinion in Neurology, 2004, 17, 569-577.	3.6	61
157	GLUCOCEREBROSIDASE MUTATIONS IN 108 NEUROPATHOLOGICALLY CONFIRMED CASES OF MULTIPLE SYSTEM ATROPHY. Neurology, 2009, 72, 1185-1186.	1.1	60
158	The MAPT p.A152T variant is a risk factor associated with tauopathies with atypical clinical and neuropathological features. Neurobiology of Aging, 2012, 33, 2231.e7-2231.e14.	3.1	60
159	A 6.4 Mb Duplication of the Î±-Synuclein Locus Causing Frontotemporal Dementia and Parkinsonism. JAMA Neurology, 2014, 71, 1162.	9.0	60
160	Insights From Cerebellar Transcriptomic Analysis Into the Pathogenesis of Ataxia. JAMA Neurology, 2014, 71, 831.	9.0	60
161	A novel tau mutation in exon 9 (1260V) causes a four-repeat tauopathy. Experimental Neurology, 2003, 184, 131-140.	4.1	59
162	Compound heterozygous PANK2 mutations confirm HARP and Hallervorden-Spatz syndromes are allelic. Neurology, 2003, 61, 1423-1426.	1.1	59

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163	Validation of next-generation sequencing technologies in genetic diagnosis of dementia. Neurobiology of Aging, 2014, 35, 261-265.	3.1	59
164	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
165	Hereditary leukoencephalopathy with axonal spheroids: a spectrum of phenotypes from CNS vasculitis to parkinsonism in an adult onset leukodystrophy series. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 512-519.	1.9	58
166	ApoE genotype is a risk factor in nonpresenilin early-onset alzheimer's disease families. American Journal of Medical Genetics Part A, 1998, 81, 117-121.	2.4	57
167	Six novel connexin32 (GJB1) mutations in X-linked Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 73, 304-306.	1.9	56
168	Novel Mutations Mapping to the Fourth Sodium Channel Domain of Nav1.7 Result in Variable Clinical Manifestations of Primary Erythromelalgia. NeuroMolecular Medicine, 2013, 15, 265-278.	3.4	56
169	The genetics of intellectual disability: advancing technology and gene editing. F1000Research, 2020, 9, 22.	1.6	56
170	Dopaminergic Neuronal Imaging in Genetic Parkinson's Disease: Insights into Pathogenesis. PLoS ONE, 2013, 8, e69190.	2.5	55
171	A novel presenilin mutation (M233V) causing very early onset Alzheimer's disease with Lewy bodies. Neuroscience Letters, 2001, 313, 93-95.	2.1	54
172	Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. American Journal of Human Genetics, 2010, 87, 567-569.	6.2	54
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