

# Henry Houlden

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3768129/publications.pdf>

Version: 2024-02-01

653  
papers

44,887  
citations

2795

94  
h-index

3312

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. <i>Nature</i> , 1998, 393, 702-705.	13.7	3,333
2	Genome-wide association study reveals genetic risk underlying Parkinson's disease. <i>Nature Genetics</i> , 2009, 41, 1308-1312.	9.4	1,745
3	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. <i>Nature Genetics</i> , 2014, 46, 989-993.	9.4	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. <i>Lancet Neurology</i> , The, 2019, 18, 1091-1102.	4.9	1,414
5	A pathogenic mutation for probable Alzheimer's disease in the APP gene at the Nâ€²-terminus of Î²â€²-amyloid. <i>Nature Genetics</i> , 1992, 1, 345-347.	9.4	1,384
6	Early-onset Alzheimer's disease caused by mutations at codon 717 of the Î²-amyloid precursor protein gene. <i>Nature</i> , 1991, 353, 844-846.	13.7	1,202
7	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	4.9	1,039
8	Association of an Extended Haplotype in the Tau Gene with Progressive Supranuclear Palsy. <i>Human Molecular Genetics</i> , 1999, 8, 711-715.	1.4	749
9	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. <i>Brain</i> , 2009, 132, 1783-1794.	3.7	612
10	A common polymorphism in the brainâ€²-derived neurotrophic factor gene ( <i>BDNF</i> ) modulates human cortical plasticity and the response to rTMS. <i>Journal of Physiology</i> , 2008, 586, 5717-5725.	1.3	592
11	The structure of the presenilin 1 (S182) gene and identification of six novel mutations in early onset AD families. <i>Nature Genetics</i> , 1995, 11, 219-222.	9.4	461
12	Parkinson's disease induced pluripotent stem cells with triplication of the Î±-synuclein locus. <i>Nature Communications</i> , 2011, 2, 440.	5.8	406
13	Characterization of PLA2G6 as a locus for dystoniaâ€²parkinsonism. <i>Annals of Neurology</i> , 2009, 65, 19-23.	2.8	399
14	A novel Î±-synuclein missense mutation in Parkinson disease. <i>Neurology</i> , 2013, 80, 1062-1064.	1.5	396
15	Corticobasal degeneration and progressive supranuclear palsy share a common tau haplotype. <i>Neurology</i> , 2001, 56, 1702-1706.	1.5	392
16	Î±-Synucleinopathy associated with G51D SNCA mutation: a link between Parkinsonâ€²s disease and multiple system atrophy?. <i>Acta Neuropathologica</i> , 2013, 125, 753-769.	3.9	369
17	A variant of Alzheimer's disease with spastic paraparesis and unusual plaques due to deletion of exon 9 of presenilin 1. <i>Nature Medicine</i> , 1998, 4, 452-455.	15.2	347
18	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. <i>Nature Genetics</i> , 2019, 51, 649-658.	9.4	338

#	ARTICLE	IF	CITATIONS
19	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	13.7	338
20	Hereditary Sensory Neuropathy Type 1 Is Caused by the Accumulation of Two Neurotoxic Sphingolipids. <i>Journal of Biological Chemistry</i> , 2010, 285, 11178-11187.	1.6	320
21	Clinical implications of genetic advances in Charcot-Marie-Tooth disease. <i>Nature Reviews Neurology</i> , 2013, 9, 562-571.	4.9	299
22	Charcot-Marie-Tooth disease: frequency of genetic subtypes and guidelines for genetic testing. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 706-710.	0.9	297
23	Large C9orf72 Hexanucleotide Repeat Expansions Are Seen in Multiple Neurodegenerative Syndromes and Are More Frequent Than Expected in the UK Population. <i>American Journal of Human Genetics</i> , 2013, 92, 345-353.	2.6	297
24	Early onset familial Alzheimer's disease. <i>Neurology</i> , 2003, 60, 235-239.	1.5	292
25	The Parkinson's disease-linked proteins Fbxo7 and Parkin interact to mediate mitophagy. <i>Nature Neuroscience</i> , 2013, 16, 1257-1265.	7.1	292
26	Early-onset L-dopa-responsive parkinsonism with pyramidal signs due to <i>ATP13A2</i> , <i>PLA2G6</i> , <i>FBXO7</i> and <i>spatacsin</i> mutations. <i>Movement Disorders</i> , 2010, 25, 1791-1800.	2.2	287
27	The genetics and neuropathology of Parkinson's disease. <i>Acta Neuropathologica</i> , 2012, 124, 325-338.	3.9	281
28	Mutations in <i>TRPV4</i> cause Charcot-Marie-Tooth disease type 2C. <i>Nature Genetics</i> , 2010, 42, 170-174.	9.4	278
29	Deletion at <i>ITPR1</i> Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. <i>PLoS Genetics</i> , 2007, 3, e108.	1.5	269
30	A locus for familial early-onset Alzheimer's disease on the long arm of chromosome 14, proximal to the <i>±</i> antichymotrypsin gene. <i>Nature Genetics</i> , 1992, 2, 340-342.	9.4	266
31	5' Splice Site Mutations in tau Associated with the Inherited Dementia FTDP-17 Affect a Stem-Loop Structure That Regulates Alternative Splicing of Exon 10. <i>Journal of Biological Chemistry</i> , 1999, 274, 15134-15143.	1.6	266
32	A mutation in Alzheimer's disease destroying a splice acceptor site in the presenilin-1 gene. <i>NeuroReport</i> , 1995, 7, 297-301.	0.6	262
33	<i>SNCA</i> variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , 2009, 65, 610-614.	2.8	257
34	<i>C9orf72</i> expansions are the most common genetic cause of Huntington disease phenocopies. <i>Neurology</i> , 2014, 82, 292-299.	1.5	252
35	Mutations in the Gene <i>PRRT2</i> Cause Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions. <i>Cell Reports</i> , 2012, 1, 2-12.	2.9	250
36	Neuroimaging Features of Neurodegeneration with Brain Iron Accumulation. <i>American Journal of Neuroradiology</i> , 2012, 33, 407-414.	1.2	249

#	ARTICLE	IF	CITATIONS
37	Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism and dystonia. <i>Nature Communications</i> , 2016, 7, 11601.	5.8	233
38	The neuropathology, pathophysiology and genetics of multiple system atrophy. <i>Neuropathology and Applied Neurobiology</i> , 2012, 38, 4-24.	1.8	218
39	Defective FA2H leads to a novel form of neurodegeneration with brain iron accumulation (NBIA). <i>Annals of Neurology</i> , 2010, 68, 611-618.	2.8	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. <i>Lancet Neurology</i> , The, 2018, 17, 539-547.	4.9	192
41	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. <i>Nature Genetics</i> , 2017, 49, 223-237.	9.4	186
42	Mutations in TTBK2, encoding a kinase implicated in tau phosphorylation, segregate with spinocerebellar ataxia type 11. <i>Nature Genetics</i> , 2007, 39, 1434-1436.	9.4	185
43	Widespread Lewy body and tau accumulation in childhood and adult onset dystonia-parkinsonism cases with PLA2G6 mutations. <i>Neurobiology of Aging</i> , 2012, 33, 814-823.	1.5	184
44	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. <i>Annals of Neurology</i> , 2016, 79, 983-990.	2.8	183
45	Spinocerebellar ataxia: an update. <i>Journal of Neurology</i> , 2019, 266, 533-544.	1.8	180
46	G-quadruplex-binding small molecules ameliorate C9orf72 FTD / ALS pathology in vitro and in vivo. <i>EMBO Molecular Medicine</i> , 2018, 10, 22-31.	3.3	178
47	Germline selection shapes human mitochondrial DNA diversity. <i>Science</i> , 2019, 364, .	6.0	178
48	Exome Sequence Reveals Mutations in CoA Synthase as a Cause of Neurodegeneration with Brain Iron Accumulation. <i>American Journal of Human Genetics</i> , 2014, 94, 11-22.	2.6	176
49	Mutation of FA2H underlies a complicated form of hereditary spastic paraplegia (SPG35). <i>Human Mutation</i> , 2010, 31, E1251-E1260.	1.1	174
50	KIF1A, an Axonal Transporter of Synaptic Vesicles, Is Mutated in Hereditary Sensory and Autonomic Neuropathy Type 2. <i>American Journal of Human Genetics</i> , 2011, 89, 219-230.	2.6	172
51	Hereditary spastic paraplegia: from diagnosis to emerging therapeutic approaches. <i>Lancet Neurology</i> , The, 2019, 18, 1136-1146.	4.9	171
52	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. <i>Brain</i> , 2016, 139, 1904-1918.	3.7	170
53	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , 2014, 137, 2480-2492.	3.7	169
54	Mutations in the HSP27 ( HSPB1 ) gene cause dominant, recessive, and sporadic distal HMN/CMT type 2. <i>Neurology</i> , 2008, 71, 1660-1668.	1.5	168

#	ARTICLE	IF	CITATIONS
55	<i>ATP13A2</i> mutations (PARK9) cause neurodegeneration with brain iron accumulation. <i>Movement Disorders</i> , 2010, 25, 979-984.	2.2	163
56	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. <i>Nature Genetics</i> , 2020, 52, 1303-1313.	9.4	163
57	Brown-Vialetto-Van Laere Syndrome, a Ponto-Bulbar Palsy with Deafness, Is Caused by Mutations in C20orf54. <i>American Journal of Human Genetics</i> , 2010, 86, 485-489.	2.6	161
58	The phenotypic spectrum of DYT24 due to ANO3 mutations. <i>Movement Disorders</i> , 2014, 29, 928-934.	2.2	161
59	<i>PRRT2</i> gene mutations. <i>Neurology</i> , 2012, 79, 2115-2121.	1.5	159
60	Mutations in BICD2 Cause Dominant Congenital Spinal Muscular Atrophy and Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2013, 92, 965-973.	2.6	156
61	Complete analysis of the presenilin 1 gene in early onset Alzheimer's disease. <i>NeuroReport</i> , 1996, 7, 801-805.	0.6	150
62	Frequency of tau mutations in three series of non-Alzheimer's degenerative dementia. <i>Annals of Neurology</i> , 1999, 46, 243-248.	2.8	150
63	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. <i>Nature Communications</i> , 2019, 10, 3094.	5.8	150
64	Alpha-synuclein mRNA expression in oligodendrocytes in MSA. <i>Glia</i> , 2014, 62, 964-970.	2.5	149
65	Mutations in the autoregulatory domain of Î²-tubulin 4a cause hereditary dystonia. <i>Annals of Neurology</i> , 2013, 73, 546-553.	2.8	148
66	Genetic Analysis of Inherited Leukodystrophies. <i>JAMA Neurology</i> , 2013, 70, 875.	4.5	147
67	Glucocerebrosidase mutations confer a greater risk of dementia during Parkinson's disease course. <i>Movement Disorders</i> , 2012, 27, 393-399.	2.2	144
68	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. <i>Brain</i> , 2014, 137, 44-56.	3.7	143
69	Cerebral microbleeds and stroke risk after ischaemic stroke or transient ischaemic attack: a pooled analysis of individual patient data from cohort studies. <i>Lancet Neurology</i> , The, 2019, 18, 653-665.	4.9	143
70	<i>ADCY5</i> mutations are another cause of benign hereditary chorea. <i>Neurology</i> , 2015, 85, 80-88.	1.5	140
71	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome due to RFC1 repeat expansion. <i>Brain</i> , 2020, 143, 480-490.	3.7	140
72	A genome-wide association study in multiple system atrophy. <i>Neurology</i> , 2016, 87, 1591-1598.	1.5	139

#	ARTICLE	IF	CITATIONS
73	Clinical and genetic characterization of families with triple A (Allgrove) syndrome. <i>Brain</i> , 2002, 125, 2681-2690.	3.7	137
74	<i>THAP1</i> mutations (DYT6) are an additional cause of early-onset dystonia. <i>Neurology</i> , 2010, 74, 846-850.	1.5	136
75	Genetic risk factors for intracranial aneurysms. <i>Neurology</i> , 2013, 80, 2154-2165.	1.5	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- $\beta$ concentrations. <i>Annals of Neurology</i> , 2000, 48, 806-808.	2.8	135
77	APOE genotype does not modulate age of onset in families with chromosome 14 encoded Alzheimer's disease. <i>Neuroscience Letters</i> , 1994, 169, 179-180.	1.0	130
78	The clinical and genetic heterogeneity of paroxysmal dyskinesias. <i>Brain</i> , 2015, 138, 3567-3580.	3.7	129
79	Regional Distribution of Amyloid-Bri Deposition and Its Association with Neurofibrillary Degeneration in Familial British Dementia. <i>American Journal of Pathology</i> , 2001, 158, 515-526.	1.9	127
80	Pathogenic VCP Mutations Induce Mitochondrial Uncoupling and Reduced ATP Levels. <i>Neuron</i> , 2013, 78, 57-64.	3.8	127
81	<i>THAP1</i> mutations and dystonia phenotypes: Genotype phenotype correlations. <i>Movement Disorders</i> , 2012, 27, 1290-1294.	2.2	126
82	Functional Studies of Missense TREM2 Mutations in Human Stem Cell-Derived Microglia. <i>Stem Cell Reports</i> , 2018, 10, 1294-1307.	2.3	124
83	Clinical, pathological and genetic characterization of hereditary sensory and autonomic neuropathy type 1 (HSAN I). <i>Brain</i> , 2006, 129, 411-425.	3.7	122
84	Expanding the genetic heterogeneity of intellectual disability. <i>Human Genetics</i> , 2017, 136, 1419-1429.	1.8	122
85	The glucocerebrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. <i>Movement Disorders</i> , 2013, 28, 232-236.	2.2	121
86	Mutations in GBA2 Cause Autosomal-Recessive Cerebellar Ataxia with Spasticity. <i>American Journal of Human Genetics</i> , 2013, 92, 245-251.	2.6	120
87	H $\alpha$ ABC syndrome and DYT4: Variable expressivity or pleiotropy of TUBB4 mutations?. <i>Movement Disorders</i> , 2015, 30, 828-833.	2.2	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. <i>Human Molecular Genetics</i> , 2015, 24, 5260-5269.	1.4	116
89	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. <i>Brain</i> , 2012, 135, 2875-2882.	3.7	114
90	Cerebral small vessel disease-related protease HtrA1 processes latent TGF- $\beta$ binding protein 1 and facilitates TGF- $\beta$ signaling. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 16496-16501.	3.3	114

#	ARTICLE	IF	CITATIONS
91	Review: Insights into molecular mechanisms of disease in neurodegeneration with brain iron accumulation: unifying theories. <i>Neuropathology and Applied Neurobiology</i> , 2016, 42, 220-241.	1.8	114
92	Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 cause an unusual spectrum of neuropathies. <i>Brain</i> , 2010, 133, 1798-1809.	3.7	113
93	A novel RAB7 mutation associated with ulcero-mutilating neuropathy. <i>Annals of Neurology</i> , 2004, 56, 586-590.	2.8	112
94	Exome sequencing identifies a significant variant in methionyl-tRNA synthetase ( <i>MARS</i> ) in a family with late-onset CMT2. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 1247-1249.	0.9	112
95	A novel TRK A (NTRK1) mutation associated with hereditary sensory and autonomic neuropathy type V. <i>Annals of Neurology</i> , 2001, 49, 521-525.	2.8	110
96	Large, expanded repeats in SCA8 are not confined to patients with cerebellar ataxia. <i>Nature Genetics</i> , 2000, 24, 214-215.	9.4	109
97	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , 2017, 57, 247.e9-247.e13.	1.5	108
98	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. <i>Cell Reports</i> , 2013, 3, 1795-1805.	2.9	104
99	Age-related penetrance of the C9orf72 repeat expansion. <i>Scientific Reports</i> , 2017, 7, 2116.	1.6	102
100	Rhabdomyolysis: a genetic perspective. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 51.	1.2	101
101	Familial <i>PRRT2</i> mutation with heterogeneous paroxysmal disorders including paroxysmal torticollis and hemiplegic migraine. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 958-960.	1.1	100
102	Genetic dysfunction of <i>MT-ATP6</i> causes axonal Charcot-Marie-Tooth disease. <i>Neurology</i> , 2012, 79, 1145-1154.	1.5	97
103	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. <i>American Journal of Human Genetics</i> , 2016, 98, 763-771.	2.6	96
104	Chromosome 14-encoded Alzheimer's disease: Genetic and clinicopathological description. <i>Annals of Neurology</i> , 1994, 36, 362-367.	2.8	95
105	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021, 78, 464.	4.5	95
106	Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. <i>Brain</i> , 2015, 138, 845-861.	3.7	94
107	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. <i>Neurology</i> , 2021, 96, e1369-e1382.	1.5	93
108	Extended phenotypic spectrum of <i>KIF5A</i> mutations. <i>Neurology</i> , 2014, 83, 612-619.	1.5	92

#	ARTICLE	IF	CITATIONS
109	Forebrain deletion of the dystonia protein torsinA causes dystonic-like movements and loss of striatal cholinergic neurons. <i>ELife</i> , 2015, 4, e08352.	2.8	92
110	Neuropathy target esterase impairments cause Oliverâ€™McFarlane and Laurenceâ€™Moon syndromes. <i>Journal of Medical Genetics</i> , 2015, 52, 85-94.	1.5	91
111	Distinct clinical and neuropathological features of G51D SNCA mutation cases compared with SNCA duplication and H50Q mutation. <i>Molecular Neurodegeneration</i> , 2015, 10, 41.	4.4	90
112	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 611-621.	2.6	89
113	Mutations in the 5' region of the myotubularin-related protein 2 (MTMR2) gene in autosomal recessive hereditary neuropathy with focally folded myelin. <i>Brain</i> , 2001, 124, 907-915.	3.7	88
114	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. <i>American Journal of Human Genetics</i> , 2019, 104, 721-730.	2.6	88
115	Practical approach to the diagnosis of adult-onset leukodystrophies: an updated guide in the genomic era. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 543-555.	0.9	87
116	The inherited ataxias: Genetic heterogeneity, mutation databases, and future directions in research and clinical diagnostics. <i>Human Mutation</i> , 2012, 33, 1324-1332.	1.1	86
117	Genetic variability at the PARK16 locus. <i>European Journal of Human Genetics</i> , 2010, 18, 1356-1359.	1.4	85
118	Excess Î±-synuclein compromises phagocytosis in iPSC-derived macrophages. <i>Scientific Reports</i> , 2017, 7, 9003.	1.6	85
119	Localization of frontotemporal dementia with parkinsonism in an Australian kindred to chromosome 17q21-22. <i>Annals of Neurology</i> , 1997, 42, 794-798.	2.8	83
120	A novel NGF mutation clarifies the molecular mechanism and extends the phenotypic spectrum of the HSN5 neuropathy. <i>Journal of Medical Genetics</i> , 2011, 48, 131-135.	1.5	83
121	Riboflavin Responsive Mitochondrial Dysfunction in Neurodegenerative Diseases. <i>Journal of Clinical Medicine</i> , 2017, 6, 52.	1.0	83
122	Severe infantile neuropathy with diaphragmatic weakness and its relationship to SMARD1. <i>Brain</i> , 2003, 126, 2682-2692.	3.7	82
123	Frequency of mutations in the genes associated with hereditary sensory and autonomic neuropathy in a UK cohort. <i>Journal of Neurology</i> , 2012, 259, 1673-1685.	1.8	82
124	Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. <i>Brain</i> , 2015, 138, 293-310.	3.7	82
125	Defects in the CAPN1 Gene Result in Alterations in Cerebellar Development and Cerebellar Ataxia in Mice and Humans. <i>Cell Reports</i> , 2016, 16, 79-91.	2.9	82
126	Microdeletion in a FAAH pseudogene identified in a patient with high anandamide concentrations and pain insensitivity. <i>British Journal of Anaesthesia</i> , 2019, 123, e249-e253.	1.5	82



#	ARTICLE	IF	CITATIONS
127	Human ataxias: a genetic dissection of inositol triphosphate receptor (ITPR1)-dependent signaling. <i>Trends in Neurosciences</i> , 2010, 33, 211-219.	4.2	81
128	A practical approach to diagnosing adult onset leukodystrophies. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 770-781.	0.9	80
129	The phenotype of Charcot-Marie-Tooth disease type 4C due to SH3TC2 mutations and possible predisposition to an inflammatory neuropathy. <i>Neuromuscular Disorders</i> , 2009, 19, 264-269.	0.3	78
130	A Pathogenic Presenilin-1 Deletion Causes Aberrant A $\beta$ 42 Production in the Absence of Congoophilic Amyloid Plaques. <i>Journal of Biological Chemistry</i> , 2001, 276, 7233-7239.	1.6	76
131	Use of the Barthel Index and the Functional Independence Measure during early inpatient rehabilitation after single incident brain injury. <i>Clinical Rehabilitation</i> , 2006, 20, 153-159.	1.0	75
132	Truncating and Missense Mutations in IGHMBP2 Cause Charcot-Marie Tooth Disease Type 2. <i>American Journal of Human Genetics</i> , 2014, 95, 590-601.	2.6	75
133	DNA repair in the trinucleotide repeat disorders. <i>Lancet Neurology, The</i> , 2017, 16, 88-96.	4.9	75
134	Towards a complete resolution of the genetic architecture of disease. <i>Trends in Genetics</i> , 2010, 26, 438-442.	2.9	74
135	Recent Advances in the Genetics of Cerebellar Ataxias. <i>Current Neurology and Neuroscience Reports</i> , 2012, 12, 227-236.	2.0	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. <i>Lancet Neurology, The</i> , 2022, 21, 234-245.	4.9	74
137	Systemic Amyloid Deposits in Familial British Dementia. <i>Journal of Biological Chemistry</i> , 2001, 276, 43909-43914.	1.6	73
138	Clinical and genetic characterization of leukoencephalopathies in adults. <i>Brain</i> , 2017, 140, 1204-1211.	3.7	73
139	Recessive axonal Charcot-Marie-Tooth disease due to compound heterozygous mitofusin 2 mutations. <i>Neurology</i> , 2011, 77, 168-173.	1.5	72
140	Analysis of Mutations in <i>AARS2</i> in a Series of <i>CSF1R</i> -Negative Patients With Adult-Onset Leukoencephalopathy With Axonal Spheroids and Pigmented Glia. <i>JAMA Neurology</i> , 2016, 73, 1433.	4.5	71
141	Targeted next-generation sequencing panels in the diagnosis of Charcot-Marie-Tooth disease. <i>Neurology</i> , 2020, 94, e51-e61.	1.5	71
142	Loss-of-Function Variants in <i>HOPS</i> Complex Genes <i>VPS16</i> and <i>VPS41</i> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. <i>Annals of Neurology</i> , 2020, 88, 867-877.	2.8	70
143	<i>SPG11</i> mutations are common in familial cases of complicated hereditary spastic paraplegia. <i>Neurology</i> , 2008, 70, 1384-1389.	1.5	69
144	Genetics of neurodegenerative diseases: an overview. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 145, 309-323.	1.0	69

#	ARTICLE	IF	CITATIONS
145	Localization of the Gene for Distal Hereditary Motor Neuronopathy VII (dHMN-VII) to Chromosome 2q14. <i>American Journal of Human Genetics</i> , 2001, 68, 1270-1276.	2.6	68
146	Comprehensive genetic diagnosis of tandem repeat expansion disorders with programmable targeted nanopore sequencing. <i>Science Advances</i> , 2022, 8, eabm5386.	4.7	68
147	PRRT2 Regulates Synaptic Fusion by Directly Modulating SNARE Complex Assembly. <i>Cell Reports</i> , 2018, 22, 820-831.	2.9	67
148	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 460-468.	2.2	66
149	An update on the genetics, clinical presentation, and pathomechanisms of human riboflavin transporter deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 598-607.	1.7	65
150	Redefining the phenotype of ALSP and <i>AARS2</i> mutation-related leukodystrophy. <i>Neurology: Genetics</i> , 2017, 3, e135.	0.9	64
151	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. <i>Brain</i> , 2017, 140, 2820-2837.	3.7	64
152	Somatic copy number gains of $\alpha$ -synuclein (SNCA) in Parkinson's disease and multiple system atrophy brains. <i>Brain</i> , 2018, 141, 2419-2431.	3.7	63
153	<i>RFC1</i> expansions are a common cause of idiopathic sensory neuropathy. <i>Brain</i> , 2021, 144, 1542-1550.	3.7	63
154	Dominant Mutations in GRM1 Cause Spinocerebellar Ataxia Type 44. <i>American Journal of Human Genetics</i> , 2017, 101, 451-458.	2.6	62
155	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. <i>Brain</i> , 2017, 140, 940-952.	3.7	62
156	Hereditary sensory neuropathies. <i>Current Opinion in Neurology</i> , 2004, 17, 569-577.	1.8	61
157	GLUCOCEREBROSIDASE MUTATIONS IN 108 NEUROPATHOLOGICALLY CONFIRMED CASES OF MULTIPLE SYSTEM ATROPHY. <i>Neurology</i> , 2009, 72, 1185-1186.	1.5	60
158	The MAPT p.A152T variant is a risk factor associated with tauopathies with atypical clinical and neuropathological features. <i>Neurobiology of Aging</i> , 2012, 33, 2231.e7-2231.e14.	1.5	60
159	A 6.4 Mb Duplication of the $\alpha$ -Synuclein Locus Causing Frontotemporal Dementia and Parkinsonism. <i>JAMA Neurology</i> , 2014, 71, 1162.	4.5	60
160	Insights From Cerebellar Transcriptomic Analysis Into the Pathogenesis of Ataxia. <i>JAMA Neurology</i> , 2014, 71, 831.	4.5	60
161	A novel tau mutation in exon 9 (1260V) causes a four-repeat tauopathy. <i>Experimental Neurology</i> , 2003, 184, 131-140.	2.0	59
162	Compound heterozygous <i>PANK2</i> mutations confirm HARP and Hallervorden-Spatz syndromes are allelic. <i>Neurology</i> , 2003, 61, 1423-1426.	1.5	59

#	ARTICLE	IF	CITATIONS
163	Validation of next-generation sequencing technologies in genetic diagnosis of dementia. <i>Neurobiology of Aging</i> , 2014, 35, 261-265.	1.5	59
164	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. <i>American Journal of Human Genetics</i> , 2019, 104, 815-834.	2.6	59
165	Hereditary leukoencephalopathy with axonal spheroids: a spectrum of phenotypes from CNS vasculitis to parkinsonism in an adult onset leukodystrophy series. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 512-519.	0.9	58
166	ApoE genotype is a risk factor in nonpresenilin early-onset alzheimer's disease families. , 1998, 81, 117-121.		57
167	Six novel connexin32 (GJB1) mutations in X-linked Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 73, 304-306.	0.9	56
168	Novel Mutations Mapping to the Fourth Sodium Channel Domain of Nav1.7 Result in Variable Clinical Manifestations of Primary Erythromelalgia. <i>NeuroMolecular Medicine</i> , 2013, 15, 265-278.	1.8	56
169	The genetics of intellectual disability: advancing technology and gene editing. <i>F1000Research</i> , 2020, 9, 22.	0.8	56
170	Dopaminergic Neuronal Imaging in Genetic Parkinson's Disease: Insights into Pathogenesis. <i>PLoS ONE</i> , 2013, 8, e69190.	1.1	55
171	A novel presenilin mutation (M233V) causing very early onset Alzheimer's disease with Lewy bodies. <i>Neuroscience Letters</i> , 2001, 313, 93-95.	1.0	54
172	Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 567-569.	2.6	54
173	<i>PDXK</i> mutations cause polyneuropathy responsive to pyridoxal 5-phosphate supplementation. <i>Annals of Neurology</i> , 2019, 86, 225-240.	2.8	54
174	Early onset cerebral amyloid angiopathy following childhood exposure to cadaveric dura. <i>Annals of Neurology</i> , 2019, 85, 284-290.	2.8	54
175	Polymorphism in AACT gene may lower age of onset of Alzheimer's disease. <i>NeuroReport</i> , 1996, 7, 534-536.	0.6	53
176	Somatic alpha-synuclein mutations in Parkinson's disease: Hypothesis and preliminary data. <i>Movement Disorders</i> , 2013, 28, 705-712.	2.2	53
177	Cryptic Amyloidogenic Elements in the 3' UTRs of Neurofilament Genes Trigger Axonal Neuropathy. <i>American Journal of Human Genetics</i> , 2016, 98, 597-614.	2.6	53
178	A novel RFC1 repeat motif (ACAGG) in two Asia-Pacific CANVAS families. <i>Brain</i> , 2020, 143, 2904-2910.	3.7	53
179	Presenilin 1 Mutation in an African American Family Presenting With Atypical Alzheimer Dementia. <i>Archives of Neurology</i> , 2003, 60, 884.	4.9	52
180	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. <i>Brain</i> , 2018, 141, 651-661.	3.7	52

#	ARTICLE	IF	CITATIONS
181	A homozygous <i>loss-of-function</i> mutation in <i>PDE2A</i> associated to early-onset hereditary chorea. <i>Movement Disorders</i> , 2018, 33, 482-488.	2.2	52
182	Hereditary sensory neuropathy is caused by a mutation in the delta subunit of the cytosolic chaperonin-containing t-complex peptide-1 ( <i>Cct4</i> ) gene. <i>Human Molecular Genetics</i> , 2003, 12, 1917-1925.	1.4	51
183	Connexin 32 promoter P2 mutations: A mechanism of peripheral nerve dysfunction. <i>Annals of Neurology</i> , 2004, 56, 730-734.	2.8	51
184	Molecular genetics of autosomal-dominant demyelinating Charcot-Marie-Tooth disease. <i>NeuroMolecular Medicine</i> , 2006, 8, 43-62.	1.8	51
185	c-Jun expression in human neuropathies: a pilot study. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 295-303.	1.4	51
186	The Clinical Relevance of Microbleeds in Stroke study (CROMIS-2): rationale, design, and methods. <i>International Journal of Stroke</i> , 2015, 10, 155-161.	2.9	51
187	Alzheimer disease PS-1 exon 9 deletion defined. <i>Nature Medicine</i> , 1999, 5, 1090-1090.	15.2	50
188	Analysis of <i>tau</i> haplotypes in Pick's disease. <i>Neurology</i> , 2002, 59, 443-445.	1.5	50
189	An <i>ITPR1</i> gene deletion causes spinocerebellar ataxia 15/16: A genetic, clinical and radiological description. <i>Movement Disorders</i> , 2010, 25, 2176-2182.	2.2	49
190	Variable phenotypes are associated with PMP22 missense mutations. <i>Neuromuscular Disorders</i> , 2011, 21, 106-114.	0.3	48
191	Mutation in <i>FAM134B</i> causing severe hereditary sensory neuropathy: Figure 1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 119-120.	0.9	48
192	Autosomal-recessive cerebellar ataxia caused by a novel <i>ADCK3</i> mutation that elongates the protein: clinical, genetic and biochemical characterisation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 493-498.	0.9	48
193	Screening a UK amyotrophic lateral sclerosis cohort provides evidence of multiple origins of the <i>C9orf72</i> expansion. <i>Neurobiology of Aging</i> , 2015, 36, 546.e1-546.e7.	1.5	48
194	Pathological ribonuclease H1 causes R-loop depletion and aberrant DNA segregation in mitochondria. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E4276-85.	3.3	48
195	Homozygous mutations in <i>VAMP1</i> cause a presynaptic congenital myasthenic syndrome. <i>Annals of Neurology</i> , 2017, 81, 597-603.	2.8	48
196	Coenzyme Q10 Levels Are Decreased in the Cerebellum of Multiple-System Atrophy Patients. <i>PLoS ONE</i> , 2016, 11, e0149557.	1.1	48
197	Age of onset in familial early onset Alzheimer's disease correlates with genetic aetiology. <i>American Journal of Medical Genetics Part A</i> , 1993, 48, 129-130.	2.4	47
198	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , 2019, 34, 1851-1863.	2.2	47

#	ARTICLE	IF	CITATIONS
199	An update on advances in magnetic resonance imaging of multiple system atrophy. <i>Journal of Neurology</i> , 2019, 266, 1036-1045.	1.8	47
200	Early versus late anticoagulation for ischaemic stroke associated with atrial fibrillation: multicentre cohort study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 320-325.	0.9	47
201	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. <i>Nature Communications</i> , 2020, 11, 4625.	5.8	47
202	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. <i>American Journal of Human Genetics</i> , 2020, 106, 412-421.	2.6	47
203	Familial paroxysmal exercise-induced dystonia: atypical presentation of autosomal dominant GTP-cyclohydrolase 1 deficiency. <i>Developmental Medicine and Child Neurology</i> , 2010, 52, 583-586.	1.1	46
204	<i>LRRK2</i> exonic variants and risk of multiple system atrophy. <i>Neurology</i> , 2014, 83, 2256-2261.	1.5	46
205	Myoclonus-dystonia syndrome due to tyrosine hydroxylase deficiency. <i>Neurology</i> , 2012, 79, 435-441.	1.5	45
206	Peripheral neuropathy predicts nuclear gene defect in patients with mitochondrial ophthalmoplegia. <i>Brain</i> , 2014, 137, 3200-3212.	3.7	45
207	Brain iron accumulation affects myelin-related molecular systems implicated in a rare neurogenetic disease family with neuropsychiatric features. <i>Molecular Psychiatry</i> , 2016, 21, 1599-1607.	4.1	45
208	Mutations in noncoding regions of <i>GJB1</i> are a major cause of X-linked CMT. <i>Neurology</i> , 2017, 88, 1445-1453.	1.5	45
209	Adaptor protein complex 4 deficiency: a paradigm of childhood-onset hereditary spastic paraplegia caused by defective protein trafficking. <i>Human Molecular Genetics</i> , 2020, 29, 320-334.	1.4	45
210	A MÅori specific RFC1 pathogenic repeat configuration in CANVAS, likely due to a founder allele. <i>Brain</i> , 2020, 143, 2673-2680.	3.7	45
211	High frequency of the expanded C9ORF72 hexanucleotide repeat in familial and sporadic Greek ALS patients. <i>Neurobiology of Aging</i> , 2012, 33, 1851.e1-1851.e5.	1.5	44
212	Creation of an Open-Access, Mutation-Defined Fibroblast Resource for Neurological Disease Research. <i>PLoS ONE</i> , 2012, 7, e43099.	1.1	44
213	Mutations in gamma adducin are associated with inherited cerebral palsy. <i>Annals of Neurology</i> , 2013, 74, 805-814.	2.8	44
214	A loss-of-function homozygous mutation in <i>DDX59</i> implicates a conserved DEAD-box RNA helicase in nervous system development and function. <i>Human Mutation</i> , 2018, 39, 187-192.	1.1	44
215	C9orf72 and its Relevance in Parkinsonism and Movement Disorders: A Comprehensive Review of the Literature. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 575-585.	0.8	44
216	Loss of NARS1 impairs progenitor proliferation in cortical brain organoids and leads to microcephaly. <i>Nature Communications</i> , 2020, 11, 4038.	5.8	44

#	ARTICLE	IF	CITATIONS
217	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome. <i>Nature Communications</i> , 2021, 12, 7342.	5.8	44
218	Indian subcontinent NBIA: Unusual phenotypes, novel <i>PANK2</i> mutations, and undetermined genetic forms. <i>Movement Disorders</i> , 2010, 25, 1424-1431.	2.2	43
219	Pantothenate kinase-associated neurodegeneration is not a synucleinopathy. <i>Neuropathology and Applied Neurobiology</i> , 2013, 39, 121-131.	1.8	43
220	Hereditary spastic paraplegia in Greece: characterisation of a previously unexplored population using next-generation sequencing. <i>European Journal of Human Genetics</i> , 2016, 24, 857-863.	1.4	43
221	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. <i>Brain</i> , 2019, 142, 2948-2964.	3.7	43
222	POLG1 Mutations Manifesting as Autosomal Recessive Axonal Charcot-Marie-Tooth Disease. <i>Archives of Neurology</i> , 2008, 65, 133-6.	4.9	42
223	A Dominant Mutation in FBXO38 Causes Distal Spinal Muscular Atrophy with Calf Predominance. <i>American Journal of Human Genetics</i> , 2013, 93, 976-983.	2.6	42
224	Genome-wide estimate of the heritability of Multiple System Atrophy. <i>Parkinsonism and Related Disorders</i> , 2016, 22, 35-41.	1.1	42
225	The Diagnostic Value of MRI Pattern Recognition in Distal Myopathies. <i>Frontiers in Neurology</i> , 2018, 9, 456.	1.1	42
226	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. <i>BMJ</i> , The, 2021, 375, e066288.	3.0	42
227	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid-beta concentrations. <i>Annals of Neurology</i> , 2000, 48, 806-8.	2.8	42
228	TDP-43 pathology in a patient carrying G2019S LRRK2 mutation and a novel p.Q124E MAPT. <i>Neurobiology of Aging</i> , 2013, 34, 2889.e5-2889.e9.	1.5	41
229	Hereditary sensory and autonomic neuropathy type 1 (HSANI) caused by a novel mutation in <i>SPTLC2</i> . <i>Neurology</i> , 2013, 80, 2106-2111.	1.5	41
230	LETM1 couples mitochondrial DNA metabolism and nutrient preference. <i>EMBO Molecular Medicine</i> , 2018, 10, .	3.3	41
231	Cerebral mitochondrial electron transport chain dysfunction in multiple system atrophy and Parkinson's disease. <i>Scientific Reports</i> , 2019, 9, 6559.	1.6	41
232	What causes intracerebral bleeding after thrombolysis for acute ischaemic stroke? Recent insights into mechanisms and potential biomarkers. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, 1127-1136.	0.9	40
233	Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 98, 1249-1255.	2.6	40
234	Rare variants in SQSTM1 and VCP genes and risk of sporadic inclusion body myositis. <i>Neurobiology of Aging</i> , 2016, 47, 218.e1-218.e9.	1.5	40

#	ARTICLE	IF	CITATIONS
235	Small Vessel Disease and Ischemic Stroke Risk During Anticoagulation for Atrial Fibrillation After Cerebral Ischemia. <i>Stroke</i> , 2021, 52, 91-99.	1.0	40
236	Confirmation that familial clustering and age of onset in late onset Alzheimer's disease are determined at the apolipoprotein E locus. <i>Neuroscience Letters</i> , 1994, 174, 222-224.	1.0	39
237	Dystonic opisthotonus: A red flag for neurodegeneration with brain iron accumulation syndromes?. <i>Movement Disorders</i> , 2013, 28, 1325-1329.	2.2	39
238	Clinical features of childhood-onset paroxysmal kinesigenic dyskinesia with PRRT gene mutations. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 327-334.	1.1	39
239	The analysis of C9orf72 repeat expansions in a large series of clinically and pathologically diagnosed cases with atypical parkinsonism. <i>Neurobiology of Aging</i> , 2015, 36, 1221.e1-1221.e6.	1.5	39
240	iPSC-derived neuronal models of PANK2-associated neurodegeneration reveal mitochondrial dysfunction contributing to early disease. <i>PLoS ONE</i> , 2017, 12, e0184104.	1.1	39
241	Atypical periodic paralysis and myalgia. <i>Neurology</i> , 2018, 90, e412-e418.	1.5	39
242	Rapidly progressive asymmetrical weakness in Charcot-Marie-Tooth disease type 4J resembles chronic inflammatory demyelinating polyneuropathy. <i>Neuromuscular Disorders</i> , 2013, 23, 399-403.	0.3	38
243	CAV3 mutations causing exercise intolerance, myalgia and rhabdomyolysis: Expanding the phenotypic spectrum of caveolinopathies. <i>Neuromuscular Disorders</i> , 2016, 26, 504-510.	0.3	38
244	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. <i>American Journal of Human Genetics</i> , 2017, 100, 969-977.	2.6	38
245	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. <i>Movement Disorders</i> , 2018, 33, 1961-1965.	2.2	38
246	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. <i>Acta Neuropathologica</i> , 2020, 139, 415-442.	3.9	38
247	Neuronal intranuclear inclusion disease is genetically heterogeneous. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1716-1725.	1.7	38
248	Neuropathology of Beta-propeller protein associated neurodegeneration (BPAN): a new tauopathy. <i>Acta Neuropathologica Communications</i> , 2015, 3, 39.	2.4	37
249	±-Synuclein mutations cluster around a putative protein loop. <i>Neuroscience Letters</i> , 2013, 546, 67-70.	1.0	36
250	Next-generation sequencing in neuromuscular diseases. <i>Current Opinion in Neurology</i> , 2016, 29, 527-536.	1.8	36
251	Characteristics of Unruptured Compared to Ruptured Intracranial Aneurysms: A Multicenter Case-Control Study. <i>Neurosurgery</i> , 2018, 83, 43-52.	0.6	36
252	Review: Genetics and neuropathology of primary pure dystonia. <i>Neuropathology and Applied Neurobiology</i> , 2012, 38, 520-534.	1.8	35

#	ARTICLE	IF	CITATIONS
253	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. <i>Neurology</i> , 2012, 79, 127-131.	1.5	35
254	Novel C12orf65 mutations in patients with axonal neuropathy and optic atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 486-492.	0.9	35
255	Investigation of somatic CNVs in brains of synucleinopathy cases using targeted SNCA analysis and single cell sequencing. <i>Acta Neuropathologica Communications</i> , 2019, 7, 219.	2.4	35
256	Clinical heterogeneity and genotype-phenotype correlations in hereditary spastic paraplegia because of Spatacsin mutations (SPG11). <i>European Journal of Neurology</i> , 2008, 15, 1065-1070.	1.7	34
257	Common pathogenic pathways in melanoma and Parkinson disease. <i>Neurology</i> , 2010, 75, 1653-1655.	1.5	34
258	Predictors for a dementia gene mutation based on gene-panel next-generation sequencing of a large dementia referral series. <i>Molecular Psychiatry</i> , 2020, 25, 3399-3412.	4.1	34
259	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , 2014, 83, 1873-1875.	1.5	33
260	The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. <i>Brain</i> , 2017, 140, e49-e49.	3.7	33
261	MRPS25 mutations impair mitochondrial translation and cause encephalomyopathy. <i>Human Molecular Genetics</i> , 2019, 28, 2711-2719.	1.4	33
262	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. <i>Brain</i> , 2021, 144, 769-780.	3.7	33
263	Clinical features, with video documentation, of the original familial lewy body parkinsonism caused by $\alpha$ -synuclein triplication (Iowa kindred). <i>Movement Disorders</i> , 2011, 26, 2134-2136.	2.2	32
264	Investigation of next-generation sequencing technologies as a diagnostic tool for amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2015, 36, 1600.e5-1600.e8.	1.5	32
265	A novel human pain insensitivity disorder caused by a point mutation in ZFH2. <i>Brain</i> , 2018, 141, 365-376.	3.7	32
266	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. <i>American Journal of Human Genetics</i> , 2020, 107, 311-324.	2.6	32
267	Dissecting the Phenotype and Genotype of <i>PLA2G6</i> -Related Parkinsonism. <i>Movement Disorders</i> , 2022, 37, 148-161.	2.2	32
268	Motor neuron pathology in CANVAS due to <i>RFC1</i> expansions. <i>Brain</i> , 2022, 145, 2121-2132.	3.7	32
269	A novel <i>Frabin</i> (FGD4) nonsense mutation p.R275X associated with phenotypic variability in CMT4H. <i>Neurology</i> , 2009, 72, 617-620.	1.5	31
270	Detection of novel mutations and review of published data suggests that hereditary spastic paraplegia caused by spastin (SPAST) mutations is found more often in males. <i>Journal of the Neurological Sciences</i> , 2011, 306, 62-65.	0.3	31



#	ARTICLE	IF	CITATIONS
271	A 30-unit hexanucleotide repeat expansion in C9orf72 induces pathological lesions with dipeptide-repeat proteins and RNA foci, but not TDP-43 inclusions and clinical disease. <i>Acta Neuropathologica</i> , 2015, 130, 599-601.	3.9	31
272	<i>SIGMAR1</i> mutation associated with autosomal recessive Silver-like syndrome. <i>Neurology</i> , 2016, 87, 1607-1612.	1.5	31
273	Phenotypes, genotypes, and the management of paroxysmal movement disorders. <i>Developmental Medicine and Child Neurology</i> , 2018, 60, 559-565.	1.1	31
274	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. <i>American Journal of Human Genetics</i> , 2020, 106, 467-483.	2.6	31
275	Genome-Wide Association Study Identifies Risk Loci for Cluster Headache. <i>Annals of Neurology</i> , 2021, 90, 193-202.	2.8	31
276	Apolipoprotein E4 and traumatic brain injury. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2006, 77, 1106-1107.	0.9	30
277	Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. <i>Brain</i> , 2015, 138, 2859-2874.	3.7	30
278	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 1048-1056.	2.6	30
279	Truncating mutations in <i>SPAST</i> patients are associated with a high rate of psychiatric comorbidities in hereditary spastic paraplegia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 681-687.	0.9	30
280	Chromosome 13 dementia syndromes as models of neurodegeneration. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2001, 8, 277-284.	1.4	29
281	New mutations, genotype phenotype studies and manifesting carriers in giant axonal neuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2007, 78, 1267-1270.	0.9	29
282	GJB1 gene mutations in suspected inflammatory demyelinating neuropathies not responding to treatment. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2009, 80, 699-700.	0.9	29
283	Mutations in Membrin/ GOSR2 Reveal Stringent Secretory Pathway Demands of Dendritic Growth and Synaptic Integrity. <i>Cell Reports</i> , 2017, 21, 97-109.	2.9	29
284	A novel complex neurological phenotype due to a homozygous mutation in FDX2. <i>Brain</i> , 2018, 141, 2289-2298.	3.7	29
285	Proximity extension assay testing reveals novel diagnostic biomarkers of atypical parkinsonian syndromes. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 768-773.	0.9	29
286	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. <i>Brain</i> , 2020, 143, 2929-2944.	3.7	29
287	Sporadic inclusion body myositis: the genetic contributions to the pathogenesis. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 88.	1.2	28
288	C19orf12 mutation leads to a pallido-pyramidal syndrome. <i>Gene</i> , 2014, 537, 352-356.	1.0	28

#	ARTICLE	IF	CITATIONS
289	A <i>de novo</i> dominant mutation in <i>KIF1A</i> associated with axonal neuropathy, spasticity and autism spectrum disorder. <i>Journal of the Peripheral Nervous System</i> , 2017, 22, 460-463.	1.4	28
290	Early-infantile onset epilepsy and developmental delay caused by bi-allelic <i>GAD1</i> variants. <i>Brain</i> , 2020, 143, 2388-2397.	3.7	28
291	Loss of function mutations in <i>GEMIN5</i> cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	5.8	28
292	Apolipoprotein E genotype does not affect the age of onset of dementia in families with defined tau mutations. <i>Neuroscience Letters</i> , 1999, 260, 193-195.	1.0	27
293	Homozygosity mapping through whole genome analysis identifies a <i>COL18A1</i> mutation in an Indian family presenting with an autosomal recessive neurological disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2009, 150B, 993-997.	1.1	27
294	Young-onset parkinsonism due to homozygous duplication of $\alpha$ -synuclein in a consanguineous family. <i>Movement Disorders</i> , 2012, 27, 1829-1830.	2.2	27
295	Pilot phenotype and natural history study of hereditary neuropathies caused by mutations in the <i>HSPB1</i> gene. <i>Neuromuscular Disorders</i> , 2017, 27, 50-56.	0.3	27
296	Mutations in valosin-containing protein (VCP) decrease ADP/ATP translocation across the mitochondrial membrane and impair energy metabolism in human neurons. <i>Journal of Biological Chemistry</i> , 2017, 292, 8907-8917.	1.6	27
297	Nonsyndromic Parkinson disease in a family with autosomal dominant optic atrophy due to <i>OPA1</i> mutations. <i>Neurology: Genetics</i> , 2017, 3, e188.	0.9	27
298	DNA isolation protocol effects on nuclear DNA analysis by microarrays, droplet digital PCR, and whole genome sequencing, and on mitochondrial DNA copy number estimation. <i>PLoS ONE</i> , 2017, 12, e0180467.	1.1	27
299	<i>KDM5A</i> mutations identified in autism spectrum disorder using forward genetics. <i>ELife</i> , 2020, 9, .	2.8	27
300	The instability of the BTB-KELCH protein Gigaxonin causes Giant Axonal Neuropathy and constitutes a new penetrant and specific diagnostic test. <i>Acta Neuropathologica Communications</i> , 2014, 2, 47.	2.4	26
301	Novel <i>HSAN1</i> Mutation in Serine Palmitoyltransferase Resides at a Putative Phosphorylation Site That Is Involved in Regulating Substrate Specificity. <i>NeuroMolecular Medicine</i> , 2015, 17, 47-57.	1.8	26
302	Pure Cerebellar Ataxia with Homozygous Mutations in the <i>PNPLA6</i> Gene. <i>Cerebellum</i> , 2017, 16, 262-267.	1.4	26
303	Genotype-phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. <i>Movement Disorders</i> , 2018, 33, 1119-1129.	2.2	26
304	<i>RFC1</i> Intronic Repeat Expansions Absent in Pathologically Confirmed Multiple Systems Atrophy. <i>Movement Disorders</i> , 2020, 35, 1277-1279.	2.2	26
305	Genetic testing in dementia – utility and clinical strategies. <i>Nature Reviews Neurology</i> , 2021, 17, 23-36.	4.9	26
306	Iatrogenic cerebral amyloid angiopathy: an emerging clinical phenomenon. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 693-700.	0.9	26

#	ARTICLE	IF	CITATIONS
307	The pallidopyramidal syndromes. <i>Current Opinion in Neurology</i> , 2013, 26, 381-394.	1.8	25
308	Multiple system atrophy is not caused by C9orf72 hexanucleotide repeat expansions. <i>Neurobiology of Aging</i> , 2015, 36, 1223.e1-1223.e2.	1.5	25
309	Exome sequencing uncovers hidden pathways in familial and sporadic ALS. <i>Nature Neuroscience</i> , 2015, 18, 611-613.	7.1	25
310	RFC1 expansions can mimic hereditary sensory neuropathy with cough and Sjögren syndrome. <i>Brain</i> , 2020, 143, e82-e82.	3.7	25
311	An update on MSA: premotor and non-motor features open a window of opportunities for early diagnosis and intervention. <i>Journal of Neurology</i> , 2020, 267, 2754-2770.	1.8	25
312	Genetic and Clinical Heterogeneity in Thirteen New Cases with Aceruloplasminemia. Atypical Anemia as a Clue for an Early Diagnosis. <i>International Journal of Molecular Sciences</i> , 2020, 21, 2374.	1.8	25
313	<i>RFC1</i> -related ataxia is a mimic of early multiple system atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 444-446.	0.9	25
314	A novel mutation in the nerve-specific 5'UTR of the <i>GJB1</i> gene causes X-linked Charcot-Marie-Tooth disease. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 65-70.	1.4	24
315	Kohlschütter-Tänzer Syndrome: Mutations in <i>ROGDI</i> and Evidence of Genetic Heterogeneity. <i>Human Mutation</i> , 2013, 34, 296-300.	1.1	24
316	Heterogeneity in clinical features and disease severity in ataxia-associated SYNE1 mutations. <i>Journal of Neurology</i> , 2016, 263, 1503-1510.	1.8	24
317	Gene co-expression networks shed light into diseases of brain iron accumulation. <i>Neurobiology of Disease</i> , 2016, 87, 59-68.	2.1	24
318	Association of enlarged perivascular spaces and anticoagulant-related intracranial hemorrhage. <i>Neurology</i> , 2020, 95, e2192-e2199.	1.5	24
319	Clinical and genetic analysis of spinocerebellar ataxia type 11. <i>Cerebellum</i> , 2008, 7, 159-164.	1.4	23
320	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. <i>Movement Disorders</i> , 2021, 36, 251-255.	2.2	23
321	A novel TRK A (NTRK1) mutation associated with hereditary sensory and autonomic neuropathy type V. <i>Annals of Neurology</i> , 2001, 49, 521-5.	2.8	23
322	X inactivation in females with X-linked Charcot-Marie-Tooth disease. <i>Neuromuscular Disorders</i> , 2012, 22, 617-621.	0.3	22
323	Analysis of Parkinson's disease brain-derived DNA for alpha-synuclein coding somatic mutations. <i>Movement Disorders</i> , 2014, 29, 1060-1064.	2.2	22
324	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. <i>American Journal of Human Genetics</i> , 2020, 106, 484-495.	2.6	22

#	ARTICLE	IF	CITATIONS
325	Biallelic and monoallelic variants in <i>PLXNA1</i> are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. <i>Genetics in Medicine</i> , 2021, 23, 1715-1725.	1.1	22
326	Biallelic variants in <i>HPDL</i> cause pure and complicated hereditary spastic paraplegia. <i>Brain</i> , 2021, 144, 1422-1434.	3.7	22
327	Neuropathological features of genetically confirmed <i>DYT1</i> dystonia: investigating disease-specific inclusions. <i>Acta Neuropathologica Communications</i> , 2014, 2, 159.	2.4	21
328	Concomitant progressive supranuclear palsy and chronic traumatic encephalopathy in a boxer. <i>Acta Neuropathologica Communications</i> , 2014, 2, 24.	2.4	21
329	Complexity of the Genetics and Clinical Presentation of Spinocerebellar Ataxia 17. <i>Frontiers in Cellular Neuroscience</i> , 2018, 12, 429.	1.8	21
330	Assessment of APOE in atypical parkinsonism syndromes. <i>Neurobiology of Disease</i> , 2019, 127, 142-146.	2.1	21
331	Biallelic <i>MADD</i> variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. <i>Brain</i> , 2020, 143, 2437-2453.	3.7	21
332	Biallelic <i>MFSD2A</i> variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. <i>European Journal of Human Genetics</i> , 2020, 28, 1509-1519.	1.4	21
333	Expanding the molecular and phenotypic spectrum of truncating <i>MT-ATP6</i> mutations. <i>Neurology: Genetics</i> , 2020, 6, e381.	0.9	21
334	<i>PANK2</i> gene analysis confirms genetic heterogeneity in neurodegeneration with brain iron accumulation (NBIA) but mutations are rare in other types of adult neurodegenerative disease. <i>Neuroscience Letters</i> , 2006, 407, 162-165.	1.0	20
335	Genetic screening of Greek patients with Huntington's disease phenocopies identifies an <i>SCA8</i> expansion. <i>Journal of Neurology</i> , 2012, 259, 1874-1878.	1.8	20
336	Multiple system atrophy: the application of genetics in understanding etiology. <i>Clinical Autonomic Research</i> , 2015, 25, 19-36.	1.4	20
337	Prevalence of familial cluster headache: a systematic review and meta-analysis. <i>Journal of Headache and Pain</i> , 2020, 21, 37.	2.5	20
338	An ancestral 10-bp repeat expansion in <i>VWA1</i> causes recessive hereditary motor neuropathy. <i>Brain</i> , 2021, 144, 584-600.	3.7	20
339	Expanding the Spectrum of Movement Disorders Associated With <i>C9orf72</i> Hexanucleotide Expansions. <i>Neurology: Genetics</i> , 2021, 7, e575.	0.9	20
340	Recapitulation of endogenous 4R tau expression and formation of insoluble tau in directly reprogrammed human neurons. <i>Cell Stem Cell</i> , 2022, 29, 918-932.e8.	5.2	20
341	Identical twins with Leucine rich repeat kinase type 2 mutations discordant for Parkinson's disease. <i>Movement Disorders</i> , 2012, 27, 1323-1323.	2.2	19
342	A novel p.Glu175X premature stop mutation in the C-terminal end of HSP27 is a cause of CMT2. <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 201-205.	1.4	19

#	ARTICLE	IF	CITATIONS
343	Clinicopathologic and molecular spectrum of <i>RNASEH1</i> -related mitochondrial disease. <i>Neurology: Genetics</i> , 2017, 3, e149.	0.9	19
344	A Review of Copy Number Variants in Inherited Neuropathies. <i>Current Genomics</i> , 2018, 19, 412-419.	0.7	19
345	Neurodegeneration with brain iron accumulation. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 145, 157-166.	1.0	19
346	CANVAS: a late onset ataxia due to biallelic intronic AAGGG expansions. <i>Journal of Neurology</i> , 2021, 268, 1119-1126.	1.8	19
347	Bi-allelic premature truncating variants in <i>LTBP1</i> cause cutis laxa syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 1095-1114.	2.6	19
348	Newly described form of X-linked arthrogyrosis maps to the long arm of the human X chromosome. , 1998, 78, 450-454.		18
349	Exome sequencing expands the mutational spectrum of <i>SPG8</i> in a family with spasticity responsive to L-DOPA treatment. <i>Journal of Neurology</i> , 2013, 260, 2414-2416.	1.8	18
350	Benefit of carbamazepine in a patient with hemiplegic migraine associated with <i>PRRT2</i> mutation. <i>Developmental Medicine and Child Neurology</i> , 2014, 56, 910-910.	1.1	18
351	Ongoing Developments in Sporadic Inclusion Body Myositis. <i>Current Rheumatology Reports</i> , 2014, 16, 477.	2.1	18
352	Assessment of Parkinson's disease risk loci in Greece. <i>Neurobiology of Aging</i> , 2014, 35, 442.e9-442.e16.	1.5	18
353	<i>DYT6</i> Dystonia: A Neuropathological Study. <i>Neurodegenerative Diseases</i> , 2016, 16, 273-278.	0.8	18
354	Homozygous mutation in <i>HSPB1</i> causing distal vacuolar myopathy and motor neuropathy. <i>Neurology: Genetics</i> , 2017, 3, e168.	0.9	18
355	Mutations in <i>XRCC1</i> cause cerebellar ataxia and peripheral neuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1230-1232.	0.9	18
356	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> -associated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	1.1	18
357	The diagnosis of adrenal insufficiency in a patient with Allgrove syndrome and a novel mutation in the <i>ALADIN</i> gene. <i>Metabolism: Clinical and Experimental</i> , 2005, 54, 200-205.	1.5	17
358	Advances in the Genetics of Parkinson's Disease: A Guide for the Clinician. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 3-13.	0.8	17
359	A novel <i>HTRA1</i> exon 2 mutation causes loss of protease activity in a Pakistani <i>CARASIL</i> patient. <i>Journal of Neurology</i> , 2015, 262, 1369-1372.	1.8	17
360	Genotype-phenotype correlations and expansion of the molecular spectrum of <i>AP4M1</i> -related hereditary spastic paraplegia. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 172.	1.2	17

#	ARTICLE	IF	CITATIONS
361	Autonomic dysfunction in genetic forms of synucleinopathies. <i>Movement Disorders</i> , 2018, 33, 359-371.	2.2	17
362	ARSA variants in $\alpha$ -synucleinopathies. <i>Brain</i> , 2019, 142, e70-e70.	3.7	17
363	Development of MRC Centre MRI calf muscle fat fraction protocol as a sensitive outcome measure in Hereditary Sensory Neuropathy Type 1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 895-906.	0.9	17
364	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	3.7	17
365	Small vessel disease burden and intracerebral haemorrhage in patients taking oral anticoagulants. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 805-814.	0.9	17
366	Alzheimer's disease neuropathological change three decades after iatrogenic amyloid- $\beta$ transmission. <i>Acta Neuropathologica</i> , 2021, 142, 211-215.	3.9	17
367	Metabolically induced intracellular pH changes activate mitophagy, autophagy, and cell protection in familial forms of Parkinson's disease. <i>FEBS Journal</i> , 2022, 289, 699-711.	2.2	17
368	TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. <i>Brain</i> , 2022, 145, 3095-3107.	3.7	17
369	Mutational analysis of parkin and PINK1 in multiple system atrophy. <i>Neurobiology of Aging</i> , 2011, 32, 548.e5-548.e7.	1.5	16
370	Comprehensive analysis of the <i>TRPV4</i> gene in a large series of inherited neuropathies and controls. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 1204-1209.	0.9	16
371	The effects of an intronic polymorphism in TOMM40 and APOE genotypes in sporadic inclusion body myositis. <i>Neurobiology of Aging</i> , 2015, 36, 1766.e1-1766.e3.	1.5	16
372	Late-onset Lafora disease with prominent parkinsonism due to a rare mutation in <i>EPM2A</i> . <i>Neurology: Genetics</i> , 2016, 2, e101.	0.9	16
373	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. <i>Movement Disorders</i> , 2021, 36, 449-459.	2.2	16
374	Coding and noncoding variants in EBF3 are involved in HADDs and simplex autism. <i>Human Genomics</i> , 2021, 15, 44.	1.4	16
375	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. <i>Genetics in Medicine</i> , 2021, 23, 2122-2137.	1.1	16
376	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. <i>Human Genetics</i> , 2021, 140, 915-931.	1.8	16
377	Automated Brainstem Segmentation Detects Differential Involvement in Atypical Parkinsonian Syndromes. <i>Journal of Movement Disorders</i> , 2020, 13, 39-46.	0.7	16
378	Familial Idiopathic Brain Calcification – A New and Familial $\alpha$ -Synucleinopathy?. <i>European Neurology</i> , 2003, 49, 223-226.	0.6	15

#	ARTICLE	IF	CITATIONS
379	Neurology and orthopaedics. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 78, 224-232.	0.9	15
380	Triple A syndrome: A novel compound heterozygous mutation in the AAAS gene in an Italian patient without adrenal insufficiency. Journal of the Neurological Sciences, 2010, 290, 150-152.	0.3	15
381	Analysis of ATP13A2 in large neurodegeneration with brain iron accumulation (NBIA) and dystonia-parkinsonism cohorts. Neuroscience Letters, 2012, 523, 35-38.	1.0	15
382	Recent advances in bulbar syndromes. Current Opinion in Neurology, 2014, 27, 506-514.	1.8	15
383	<i>MFN2</i> deletion of exons 7 and 8: founder mutation in the UK population. Journal of the Peripheral Nervous System, 2015, 20, 67-71.	1.4	15
384	The 4H syndrome due to RNF216 mutation. Parkinsonism and Related Disorders, 2015, 21, 1122-1123.	1.1	15
385	3' UTR SNP rs2229611 in G6PC1 affects mRNA stability, expression and Glycogen Storage Disease type-Ia risk. Clinica Chimica Acta, 2017, 471, 46-54.	0.5	15
386	Familial childhood-onset progressive cerebellar syndrome associated with the <i>ATP1A3</i> mutation. Neurology: Genetics, 2017, 3, e145.	0.9	15
387	Association of functional MMP-2 gene variant with intracranial aneurysms: case-control genetic association study and meta-analysis. British Journal of Neurosurgery, 2018, 32, 255-259.	0.4	15
388	Adult-onset neuronal intranuclear inclusion disease mimicking Fragile X-associated tremor-ataxia syndrome in ethnic Chinese patients. Parkinsonism and Related Disorders, 2020, 74, 25-27.	1.1	15
389	Sequencing analysis of the ITPR1 gene in a pure autosomal dominant spinocerebellar ataxia series. Movement Disorders, 2010, 25, 771-773.	2.2	14
390	THAP1 mutations in a Greek primary blepharospasm series. Parkinsonism and Related Disorders, 2013, 19, 404-405.	1.1	14
391	HLA-DRB*1501 associations with magnetic resonance imaging measures of grey matter pathology in multiple sclerosis. Multiple Sclerosis and Related Disorders, 2016, 7, 47-52.	0.9	14
392	Multiple system atrophy: genetic risks and alpha-synuclein mutations. F1000Research, 2017, 6, 2072.	0.8	14
393	Biallelic variants in <i>ADARB1</i> , encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Journal of Medical Genetics, 2021, 58, 495-504.	1.5	14
394	GGC Repeat Expansion in <i>NOTCH2NLC</i> Is Rare in European Leukoencephalopathy. Annals of Neurology, 2020, 88, 641-642.	2.8	14
395	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	1.1	14
396	A geroscience approach for Parkinson's disease: Conceptual framework and design of PROPAG-AGEING project. Mechanisms of Ageing and Development, 2021, 194, 111426.	2.2	14

#	ARTICLE	IF	CITATIONS
397	Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS): genetic and clinical aspects. <i>Practical Neurology</i> , 2022, 22, 14-18.	0.5	14
398	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. <i>Brain</i> , 2022, 145, 1507-1518.	3.7	14
399	Pupil abnormalities in 131 cases of genetically defined inherited peripheral neuropathy. <i>Eye</i> , 2009, 23, 966-974.	1.1	13
400	Friedreich's ataxia and other hereditary ataxias in Greece: An 18-year perspective. <i>Journal of the Neurological Sciences</i> , 2014, 336, 87-92.	0.3	13
401	De novo <i>KCNA2</i> mutations cause hereditary spastic paraplegia. <i>Annals of Neurology</i> , 2017, 81, 326-328.	2.8	13
402	IGHMBP2 mutation associated with organ-specific autonomic dysfunction. <i>Neuromuscular Disorders</i> , 2018, 28, 1012-1015.	0.3	13
403	Novel fluid biomarkers to differentiate frontotemporal dementia and dementia with Lewy bodies from Alzheimer's disease: A systematic review. <i>Journal of the Neurological Sciences</i> , 2020, 415, 116886.	0.3	13
404	<i>MYORG</i> -related disease is associated with central pontine calcifications and atypical parkinsonism. <i>Neurology: Genetics</i> , 2020, 6, e399.	0.9	13
405	GCC repeat expansion in NOTCH2NLC is rare in European patients with essential tremor. <i>Brain</i> , 2020, 143, e57-e57.	3.7	13
406	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. <i>European Journal of Human Genetics</i> , 2021, 29, 411-421.	1.4	13
407	Molecular Genetics of Autosomal-Dominant Demyelinating Charcot-Marie-Tooth Disease. <i>NeuroMolecular Medicine</i> , 2006, 8, 43-62.	1.8	13
408	WARS2 mutations cause dopa-responsive early-onset parkinsonism and progressive myoclonus ataxia. <i>Parkinsonism and Related Disorders</i> , 2022, 94, 54-61.	1.1	13
409	Apolipoprotein E alleles but neither apolipoprotein B nor apolipoprotein AI/CIII alleles are associated with late onset, familial Alzheimer's disease. <i>Neuroscience Letters</i> , 1995, 188, 202-204.	1.0	12
410	The frequency of spinocerebellar ataxia type 23 in a UK population. <i>Journal of Neurology</i> , 2013, 260, 856-859.	1.8	12
411	Madras motor neuron disease (MMND) is distinct from the riboflavin transporter genetic defects that cause Brown-Vialetto-Van Laere syndrome. <i>Journal of the Neurological Sciences</i> , 2013, 334, 119-122.	0.3	12
412	A novel <i>SLC1A4</i> homozygous mutation causing congenital microcephaly, epileptic encephalopathy and spastic tetraparesis: a video-EEG and tractography case study. <i>Journal of Neurogenetics</i> , 2018, 32, 316-321.	0.6	12
413	Kleine-Levin syndrome is associated with LMOD3 variants. <i>Journal of Sleep Research</i> , 2019, 28, e12718.	1.7	12
414	<i>PUS3</i> mutations are associated with intellectual disability, leukoencephalopathy, and nephropathy. <i>Neurology: Genetics</i> , 2019, 5, e306.	0.9	12



#	ARTICLE	IF	CITATIONS
415	Autosomal dominant optic atrophy and cataract "plus" phenotype including axonal neuropathy. <i>Neurology: Genetics</i> , 2019, 5, e322.	0.9	12
416	Longer term stroke risk in intracerebral haemorrhage survivors. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 840-845.	0.9	12
417	Rare novel CYP2U1 and ZFYVE26 variants identified in two Pakistani families with spastic paraplegia. <i>Journal of the Neurological Sciences</i> , 2020, 411, 116669.	0.3	12
418	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. <i>Brain</i> , 2021, 144, e30-e30.	3.7	12
419	Novel <i>NDUFA12</i> variants are associated with isolated complex I defect and variable clinical manifestation. <i>Human Mutation</i> , 2021, 42, 699-710.	1.1	12
420	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. <i>American Journal of Human Genetics</i> , 2021, 108, 2368-2384.	2.6	12
421	A Huntington's disease phenocopy characterized by pallido-nigro-lusian degeneration with brain iron accumulation and p62-positive glial inclusions. <i>Neuropathology and Applied Neurobiology</i> , 2010, 36, 551-557.	1.8	11
422	Paroxysmal Kinesigenic Dyskinesia May Be Misdiagnosed in Co-occurring Gilles de la Tourette Syndrome. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 84-86.	0.8	11
423	Severe axonal neuropathy is a late manifestation of SPG11. <i>Journal of Neurology</i> , 2016, 263, 2278-2286.	1.8	11
424	MSA or SCA 17? A clinicopathological case update. <i>Movement Disorders</i> , 2016, 31, 1582-1584.	2.2	11
425	Calpainopathy with macrophage-rich, regional inflammatory infiltrates. <i>Neuromuscular Disorders</i> , 2017, 27, 738-741.	0.3	11
426	A 30-year history of MPAN case from Russia. <i>Clinical Neurology and Neurosurgery</i> , 2017, 159, 111-113.	0.6	11
427	Brainstem phenotype of cathepsin A related arteriopathy with strokes and leukoencephalopathy. <i>Neurology: Genetics</i> , 2017, 3, e165.	0.9	11
428	Adaptive regulation of riboflavin transport in heart: effect of dietary riboflavin deficiency in cardiovascular pathogenesis. <i>Molecular and Cellular Biochemistry</i> , 2018, 440, 147-156.	1.4	11
429	LRP10 in $\pm$ -synucleinopathies. <i>Lancet Neurology</i> , The, 2018, 17, 1033-1034.	4.9	11
430	Lysosomal storage disorder gene variants in multiple system atrophy. <i>Brain</i> , 2018, 141, e53-e53.	3.7	11
431	Mutation in <i>RNF170</i> causes sensory ataxic neuropathy with vestibular areflexia: a CANVAS mimic. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 1237-1238.	0.9	11
432	Haptoglobin genotype and outcome after aneurysmal subarachnoid haemorrhage. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 305-313.	0.9	11

#	ARTICLE	IF	CITATIONS
433	Expanding the phenotype of <i>PIGS</i> -associated early onset epileptic developmental encephalopathy. <i>Epilepsia</i> , 2021, 62, e35-e41.	2.6	11
434	Biallelic variants in <i>KARS1</i> are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. <i>Genetics in Medicine</i> , 2021, 23, 1933-1943.	1.1	11
435	Loss of <i>C2orf69</i> defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 1301-1317.	2.6	11
436	Biallelic variants in <i>PCDHGC4</i> cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. <i>Genetics in Medicine</i> , 2021, 23, 2138-2149.	1.1	11
437	Bi-allelic variants in <i>SPATA5L1</i> lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. <i>American Journal of Human Genetics</i> , 2021, 108, 2006-2016.	2.6	11
438	Expanding the phenotypic spectrum of <i>BCS1L</i> -related mitochondrial disease. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2155-2165.	1.7	11
439	MRI and CT imaging biomarkers of cerebral amyloid angiopathy in lobar intracerebral hemorrhage. <i>International Journal of Stroke</i> , 2023, 18, 85-94.	2.9	11
440	Genetic characterization of a familial non-specific dementia originating in Jutland, Denmark. <i>Journal of the Neurological Sciences</i> , 1993, 114, 138-143.	0.3	10
441	What are the experiences of adults returning to work following recovery from Guillain-Barré syndrome? An interpretative phenomenological analysis. <i>Disability and Rehabilitation</i> , 2009, 31, 1817-1827.	0.9	10
442	Defective N-linked protein glycosylation pathway in congenital myasthenic syndromes. <i>Brain</i> , 2013, 136, 692-695.	3.7	10
443	Using human induced pluripotent stem cells to model cerebellar disease: Hope and hype. <i>Journal of Neurogenetics</i> , 2015, 29, 95-102.	0.6	10
444	Tubular Aggregates and Cylindrical Spirals Have Distinct Immunohistochemical Signatures. <i>Journal of Neuropathology and Experimental Neurology</i> , 2016, 75, 1171-1178.	0.9	10
445	Paroxysmal Movement Disorder and Epilepsy Caused by a De Novo Truncating Mutation in <i>KAT6A</i> . <i>Journal of Pediatric Genetics</i> , 2018, 07, 114-116.	0.3	10
446	Homozygous missense <i>WIPI2</i> variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. <i>Brain Communications</i> , 2021, 3, fcab183.	1.5	10
447	Cerebral Small Vessel Disease and Functional Outcome Prediction After Intracerebral Hemorrhage. <i>Neurology</i> , 2021, 96, e1954-e1965.	1.5	10
448	A human importin- $\beta 2$ -related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in <i>IPO8</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 1115-1125.	2.6	10
449	Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. <i>Brain Sciences</i> , 2021, 11, 1150.	1.1	10
450	Clinical features of early onset, familial Alzheimer's disease linked to chromosome 14. <i>American Journal of Medical Genetics Part A</i> , 1995, 60, 44-52.	2.4	9

#	ARTICLE	IF	CITATIONS
---	---------	----	-----------

451	Thinning of the Corpus Callosum and Cerebellar Atrophy is Correlated with Phenotypic Severity in a		
-----	--	--	--

#	ARTICLE	IF	CITATIONS
469	Genetic and phenotypic characterisation of inherited myopathies in a tertiary neuromuscular centre. <i>Neuromuscular Disorders</i> , 2019, 29, 747-757.	0.3	8
470	Impaired Bioenergetics in Mutant Mitochondrial DNA Determines Cell Fate During Seizure-Like Activity. <i>Molecular Neurobiology</i> , 2019, 56, 321-334.	1.9	8
471	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. <i>European Journal of Human Genetics</i> , 2021, 29, 271-279.	1.4	8
472	Integrating protein networks and machine learning for disease stratification in the Hereditary Spastic Paraplegias. <i>IScience</i> , 2021, 24, 102484.	1.9	8
473	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome: genetic and clinical insights. <i>Current Opinion in Neurology</i> , 2021, 34, 556-564.	1.8	8
474	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. <i>European Journal of Human Genetics</i> , 2021, 29, 1226-1234.	1.4	8
475	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 1069-1082.	2.6	8
476	Investigating RFC1 expansions in sporadic amyotrophic lateral sclerosis. <i>Journal of the Neurological Sciences</i> , 2021, 430, 118061.	0.3	8
477	Biallelic <i>ADAM22</i> pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. <i>Brain</i> , 2022, 145, 2301-2312.	3.7	8
478	Construction of a Detailed Physical and Transcript Map of the FTDP-17 Candidate Region on Chromosome 17q21. <i>Genomics</i> , 1999, 60, 129-136.	1.3	7
479	Analysis of spinocerebellar ataxias due to expanded triplet repeats in Greek patients with cerebellar ataxia. <i>Journal of the Neurological Sciences</i> , 2012, 318, 178-180.	0.3	7
480	Absence of HINT1 mutations in a UK and Spanish cohort of patients with inherited neuropathies. <i>Journal of Neurology</i> , 2015, 262, 1984-1986.	1.8	7
481	Charcot-Marie-Tooth disease type 2C and scapuloperoneal muscular atrophy overlap syndrome in a patient with the R232C TRPV4 mutation. <i>Journal of Neurology</i> , 2015, 262, 1972-1975.	1.8	7
482	Pathological relationships involving iron and myelin may constitute a shared mechanism linking various rare and common brain diseases. <i>Rare Diseases (Austin, Tex)</i> , 2016, 4, e1198458.	1.8	7
483	Sequencing analysis of the SCA6 CAG expansion excludes an influence of repeat interruptions on disease onset. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1226-1227.	0.9	7
484	Electroclinical history of a five-year-old girl with GRIN1-related early-onset epileptic encephalopathy: a video-case study. <i>Epileptic Disorders</i> , 2018, 20, 423-427.	0.7	7
485	Genotype-Phenotype Correlations in Charcot-Marie-Tooth Disease Due to MTMR2 Mutations and Implications in Membrane Trafficking. <i>Frontiers in Neuroscience</i> , 2019, 13, 974.	1.4	7
486	Continuum of phenotypes in hereditary motor and sensory neuropathy with proximal predominance and Charcot-Marie-Tooth patients with <i>TFG</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1507-1515.	0.7	7

#	ARTICLE	IF	CITATIONS
487	Biotin-Thiamine Responsive Encephalopathy: Report of an Egyptian Family with a Novel SLC19A3 Mutation and Review of the Literature. <i>Journal of Pediatric Genetics</i> , 2019, 08, 100-108.	0.3	7
488	Hereditary polyneuropathy with optic atrophy due to PDXK variant leading to impaired Vitamin B6 metabolism. <i>Neuromuscular Disorders</i> , 2020, 30, 583-589.	0.3	7
489	A glimpse of the genetics of young-onset Parkinson's disease in Central Asia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1671.	0.6	7
490	Two novel biallelic <i>KDEL2</i> missense variants cause osteogenesis imperfecta with neurodevelopmental features. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2241-2249.	0.7	7
491	A Recurrent <i>VPS16</i> p.Arg187* Nonsense Variant in Early-Onset Generalized Dystonia. <i>Movement Disorders</i> , 2021, 36, 1984-1985.	2.2	7
492	<i>SORL1</i> mutation in a Greek family with Parkinson's disease and dementia. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1961-1969.	1.7	7
493	Spastic paraplegia preceding <i>PSEN1</i> -related familial Alzheimer's disease. <i>Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring</i> , 2021, 13, e12186.	1.2	7
494	Heterozygous <i>EIF2AK2</i> Variant Causes Adolescence-Onset Generalized Dystonia Partially Responsive to <i>DBS</i> . <i>Movement Disorders Clinical Practice</i> , 2022, 9, 268-271.	0.8	7
495	<i>El-Hattab-Alkuraya</i> syndrome caused by biallelic <i>WDR45B</i> pathogenic variants: Further delineation of the phenotype and genotype. <i>Clinical Genetics</i> , 2022, 101, 530-540.	1.0	7
496	Genetic defects are common in myopathies with tubular aggregates. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 4-15.	1.7	7
497	Genotypic and phenotypic heterogeneity in familial microcoria. <i>British Journal of Ophthalmology</i> , 2004, 88, 469-473.	2.1	6
498	Screening for mutations in the phosphatidylinositol 4-kinase 2-alpha gene in autosomal recessive hereditary spastic paraplegia. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2011, 12, 148-149.	2.3	6
499	Kjellin syndrome: hereditary spastic paraplegia with pathognomonic macular appearance. <i>Practical Neurology</i> , 2014, 14, 278-279.	0.5	6
500	Diagnostic clues and manifesting carriers in fukutin-related protein (FKRP) limb-girdle muscular dystrophy. <i>Journal of the Neurological Sciences</i> , 2015, 348, 266-268.	0.3	6
501	<i>VPS13C</i> -Another Hint at Mitochondrial Dysfunction in Familial Parkinson's Disease. <i>Movement Disorders</i> , 2016, 31, 1340-1340.	2.2	6
502	Analysis of the prion protein gene in multiple system atrophy. <i>Neurobiology of Aging</i> , 2017, 49, 216.e15-216.e18.	1.5	6
503	<i>CYG1</i> causing progressive limb girdle myopathy with onset during teenage years (polyglucosan body) Tj ETQq1 1 0.784314 rgBT /Ov	0.3	6
504	Quick Flicks: Association of Paroxysmal Kinesigenic Dyskinesia and Tics. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 317-320.	0.8	6

#	ARTICLE	IF	CITATIONS
505	Assessment of the Subarachnoid Hemorrhage International Trialists (SAHIT) Models for Dichotomized Long-Term Functional Outcome Prediction After Aneurysmal Subarachnoid Hemorrhage in a United Kingdom Multicenter Cohort Study. <i>Neurosurgery</i> , 2020, 87, 1269-1276.	0.6	6
506	DRPLA: understanding the natural history and developing biomarkers to accelerate therapeutic trials in a globally rare repeat expansion disorder. <i>Journal of Neurology</i> , 2021, 268, 3031-3041.	1.8	6
507	<i>ANGPTL6</i> Genetic Variants Are an Underlying Cause of Familial Intracranial Aneurysms. <i>Neurology</i> , 2021, 96, e947-e955.	1.5	6
508	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. <i>Brain</i> , 2020, 143, e31-e31.	3.7	6
509	Negative screening for 12 rare LRRK2 pathogenic variants in a cohort of Nigerians with Parkinson's disease. <i>Neurobiology of Aging</i> , 2021, 99, 101.e15-101.e19.	1.5	6
510	Bi-allelic loss-of-function OBSCN variants predispose individuals to severe recurrent rhabdomyolysis. <i>Brain</i> , 2022, 145, 3985-3998.	3.7	6
511	Screening for the APP codon mutations in Alzheimer's disease. <i>Neuroscience Letters</i> , 1993, 154, 161-162.	1.0	5
512	The small, spastic, and furrowed tongue of Allgrove syndrome. <i>Neurology</i> , 2009, 72, 1366-1366.	1.5	5
513	Call for participation in the neurogenetics consortium within the Human Variome Project. <i>Neurogenetics</i> , 2011, 12, 169-173.	0.7	5
514	Extending the clinical spectrum of pain channelopathies. <i>Brain</i> , 2012, 135, 313-316.	3.7	5
515	Ataxia with oculomotor apraxia type 2: an evolving axonal neuropathy. <i>Practical Neurology</i> , 2018, 18, 52-56.	0.5	5
516	Parkinson's Disease in Central Asian and Transcaucasian Countries: A Review of Epidemiology, Genetics, Clinical Characteristics, and Access to Care. <i>Parkinson's Disease</i> , 2019, 2019, 1-7.	0.6	5
517	A de novo truncating mutation in ASXL1 associated with segmental overgrowth. <i>Journal of Genetics</i> , 2019, 98, 1.	0.4	5
518	Sensitivity and specificity of blood-fluid levels for oral anticoagulant-associated intracerebral haemorrhage. <i>Scientific Reports</i> , 2020, 10, 15529.	1.6	5
519	A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. <i>Brain</i> , 2020, 143, e49-e49.	3.7	5
520	Baseline factors associated with early and late death in intracerebral haemorrhage survivors. <i>European Journal of Neurology</i> , 2020, 27, 1257-1263.	1.7	5
521	Brown-Vialetto-Van Laere and Fazio-Londe syndromes: <i>SLC52A3</i> mutations with puzzling phenotypes and inheritance. <i>European Journal of Neurology</i> , 2021, 28, 945-954.	1.7	5
522	R1352Q & CACNA1A Variant in a Patient with Sporadic Hemiplegic Migraine, Ataxia, Seizures and Cerebral Oedema: A Case Report. <i>Case Reports in Neurology</i> , 2021, 13, 123-130.	0.3	5

#	ARTICLE	IF	CITATIONS
523	Allelic and phenotypic heterogeneity in Junctophilin-3 related neurodevelopmental and movement disorders. <i>European Journal of Human Genetics</i> , 2021, 29, 1027-1031.	1.4	5
524	A clinical, molecular genetics and pathological study of a FTDP-17 family with a heterozygous splicing variant c.823-10G>T at the intron 9/exon 10 of the MAPT gene. <i>Neurobiology of Aging</i> , 2021, 106, 343.e1-343.e8.	1.5	5
525	Early-onset phenotype of bi-allelic <i>GRN</i> mutations. <i>Brain</i> , 2021, 144, e22-e22.	3.7	5
526	Genome-Wide Association Study of Clinical Outcome After Aneurysmal Subarachnoid Haemorrhage: Protocol. <i>Translational Stroke Research</i> , 2022, 13, 565-576.	2.3	5
527	Biallelic Loss of Function NDUF12 Variants Cause a Wide Phenotypic Spectrum from Leigh/Leigh-Like Syndrome to Isolated Optic Atrophy. <i>Movement Disorders Clinical Practice</i> , 2022, 9, 218-228.	0.8	5
528	Reply to: Juvenile <i>PLA2G6</i> parkinsonism due to Indian "Asian" p.R741Q mutation, and response to STN DBS. <i>Movement Disorders</i> , 2022, 37, 658-662.	2.2	5
529	How to approach a neurogenetics diagnosis in different European countries: The European Academy of Neurology Neurogenetics Panel survey. <i>European Journal of Neurology</i> , 2022, 29, 1885-1891.	1.7	5
530	Apolipoprotein E and Cerebral Small Vessel Disease Markers in Patients With Intracerebral Haemorrhage. <i>Neurology</i> , 0, , 10.1212/WNL.0000000000200851.	1.5	5
531	Exclusion Mapping in Familial Non-Specific Dementia. <i>Dementia and Geriatric Cognitive Disorders</i> , 1993, 4, 163-166.	0.7	4
532	Spinocerebellar ataxia type 11. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2012, 103, 521-534.	1.0	4
533	Sniffing out the cerebellum: Figure 1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 952-953.	0.9	4
534	K11 C9orf72 Expansions Are The Most Common Genetic Cause Of Huntington's Disease Phenocopy Presentations In A Uk Cohort. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, A82-A82.	0.9	4
535	Novel single base-pair deletion in exon 1 of XK gene leading to McLeod syndrome with chorea, muscle wasting, peripheral neuropathy, acanthocytosis and haemolysis. <i>Journal of the Neurological Sciences</i> , 2014, 339, 220-222.	0.3	4
536	Syndromic associations and RNF216 mutations. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 1389-1390.	1.1	4
537	Triple trouble: a striking new phenotype or competing genes in a family with hereditary spastic paraplegia. <i>Journal of Neurology</i> , 2016, 263, 1232-1233.	1.8	4
538	Are some C19orf12 variants monoallelic for neurological disorders?. <i>Parkinsonism and Related Disorders</i> , 2019, 65, 267-269.	1.1	4
539	The need for biochemical testing in beta-enolase deficiency in the genomic era. <i>JIMD Reports</i> , 2019, 50, 40-43.	0.7	4
540	A new family with GLRB-related hyperekplexia showing chorea in homo- and heterozygous variant carriers. <i>Parkinsonism and Related Disorders</i> , 2020, 79, 97-99.	1.1	4

#	ARTICLE	IF	CITATIONS
541	Haptoglobin genotype and outcome after spontaneous intracerebral haemorrhage. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 298-304.	0.9	4
542	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. <i>Human Molecular Genetics</i> , 2020, 29, 1426-1439.	1.4	4
543	Novel variants broaden the phenotypic spectrum of PLEKHG5 associated neuropathies. <i>European Journal of Neurology</i> , 2021, 28, 1344-1355.	1.7	4
544	Tay-Sachs Disease: Two Novel Rare HEXA Mutations from Pakistan and Morocco. <i>Klinische Padiatrie</i> , 2021, 233, 226-230.	0.2	4
545	Charcot-Marie-Tooth type 4B2 demyelinating neuropathy in miniature Schnauzer dogs caused by a novel splicing SBF2 (MTMR13) genetic variant: a new spontaneous clinical model. <i>PeerJ</i> , 2019, 7, e7983.	0.9	4
546	DYT6 mutated THAP1 is a cell type dependent regulator of the SP1 family. <i>Brain</i> , 2022, 145, 3968-3984.	3.7	4
547	Diagnosing Premotor Multiple System Atrophy. <i>Neurology</i> , 2022, 99, .	1.5	4
548	A Novel DNA Sequence Variation in the First Genetically Confirmed Allgrove Syndrome in Iran. <i>Journal of Clinical Neuromuscular Disease</i> , 2006, 7, 123-127.	0.3	3
549	PAW32 ITPR1 gene deletion causes spinocerebellar ataxia 15/16: a genetic, clinical and radiological description of a novel kindred. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, e32-e32.	0.9	3
550	Clinical variability and l-Dopa responsive Parkinsonism in hereditary spastic paraplegia 11. <i>Journal of Neurology</i> , 2012, 259, 2726-2728.	1.8	3
551	Defects of RNA metabolism in the pathogenesis of spinal muscular atrophy. <i>Neurology</i> , 2014, 82, 1298-1299.	1.5	3
552	G.P.22. <i>Neuromuscular Disorders</i> , 2014, 24, 801.	0.3	3
553	A novel TUBB4A mutation suggests that genotype-phenotype correlation of H-ABC syndrome needs to be revisited: Figure 1. <i>Brain</i> , 2015, 138, e370-e370.	3.7	3
554	PKD or Not PKD: That is the question. <i>Annals of Neurology</i> , 2016, 80, 167-168.	2.8	3
555	Verification of Inter-laboratorial Genotyping Consistency in the Molecular Diagnosis of Polyglutamine Spinocerebellar Ataxias. <i>Journal of Molecular Neuroscience</i> , 2016, 58, 83-87.	1.1	3
556	Further supporting evidence for REEP1 phenotypic and allelic heterogeneity. <i>Neurology: Genetics</i> , 2019, 5, e379.	0.9	3
557	A rare PANK2 deletion in the first north African patient affected with pantothenate kinase associated neurodegeneration. <i>Journal of the Neurological Sciences</i> , 2020, 410, 116639.	0.3	3
558	Identification of common genetic markers of paroxysmal neurological disorders using a network analysis approach. <i>Neurological Sciences</i> , 2020, 41, 851-857.	0.9	3



#	ARTICLE	IF	CITATIONS
559	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCa) syndrome. <i>Journal of Medical Genetics</i> , 2021, 58, 815-831.	1.5	3
560	How to diagnose difficult white matter disorders. <i>Practical Neurology</i> , 2020, 20, 280-286.	0.5	3
561	LRRK2 Mutations and Asian Disease-Associated Variants in the First Parkinson's Disease Cohort from Kazakhstan. <i>Parkinson's Disease</i> , 2020, 2020, 1-10.	0.6	3
562	The repeat variant in MSH3 is not a genetic modifier for spinocerebellar ataxia type 3 and Friedreich's ataxia. <i>Brain</i> , 2020, 143, e25-e25.	3.7	3
563	Novel variants underlying autosomal recessive intellectual disability in Pakistani consanguineous families. <i>BMC Medical Genetics</i> , 2020, 21, 59.	2.1	3
564	Prevalence of <i>C9orf72</i> hexanucleotide repeat expansion in Greek patients with sporadic ALS. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 470-472.	1.1	3
565	Novel likely disease-causing CLN5 variants identified in Pakistani patients with neuronal ceroid lipofuscinosis. <i>Journal of the Neurological Sciences</i> , 2020, 414, 116826.	0.3	3
566	De novo mutation in <i>SLC25A22</i> gene: expansion of the clinical and electroencephalographic phenotype. <i>Journal of Neurogenetics</i> , 2021, 35, 67-73.	0.6	3
567	Editorial: Multifaceted Genes in Amyotrophic Lateral Sclerosis-Frontotemporal Dementia. <i>Frontiers in Neuroscience</i> , 2021, 15, 680185.	1.4	3
568	Whole-genome sequencing. <i>Practical Neurology</i> , 2021, 21, 322-327.	0.5	3
569	A novel variant in the DSE gene leads to Ehlers-Danlos musculocontractural type 2 in a Pakistani family. <i>Congenital Anomalies (discontinued)</i> , 2021, 61, 177-182.	0.3	3
570	A Novel Homozygous <i>ADCY5</i> Variant is Associated with a Neurodevelopmental Disorder and Movement Abnormalities. <i>Movement Disorders Clinical Practice</i> , 2021, 8, 1140-1143.	0.8	3
571	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS1 mutations that lead to exceptionally high amyloid $\beta$ concentrations. <i>Annals of Neurology</i> , 2000, 48, 806-808.	2.8	3
572	A novel frameshift deletion in autosomal recessive SBF1-related syndromic neuropathy with necklace fibres. <i>Journal of Neurology</i> , 2020, 267, 2705-2712.	1.8	3
573	Novel Machado-Joseph disease-modifying genes and pathways identified by whole-exome sequencing. <i>Neurobiology of Disease</i> , 2022, 162, 105578.	2.1	3
574	Prevalence of Fabry Disease among Patients with Parkinson's Disease. <i>Parkinson's Disease</i> , 2022, 2022, 1-8.	0.6	3
575	Bi-allelic variants in <i>CHKA</i> cause a neurodevelopmental disorder with epilepsy and microcephaly. <i>Brain</i> , 2022, 145, 1916-1923.	3.7	3
576	Mutations in <i>TAF8</i> cause a neurodegenerative disorder. <i>Brain</i> , 2022, 145, 3022-3034.	3.7	3

#	ARTICLE	IF	CITATIONS
577	1042â€¦Hereditary sensory and autonomic neuropathy type 1: correlation of severity and plasma atypical deoxy-sphingoid bases. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, e1.68-e1.	0.9	2
578	Mutational analysis of <i><sc>PMP22</sc></i>, <i><sc>EGR2</sc></i>, <i><sc>LITAF</sc></i> and <i><sc>NEFL</sc></i> in Greek Charcotâ€œMarieâ€œTooth type 1 patients. <i>Clinical Genetics</i> , 2013, 83, 388-391.	1.0	2
579	Hypersomnia with dilated pupils in adenosine monophosphate deaminase (<sc>AMPD</sc>) deficiency. <i>Journal of Sleep Research</i> , 2014, 23, 118-120.	1.7	2
580	Response to the commentary of Yates RL and DeLuca GC on the study: HLA-DRB1*1501 associations with magnetic resonance imaging measures of grey matter pathology in multiple sclerosis. <i>Multiple Sclerosis and Related Disorders</i> , 2018, 19, 168-170.	0.9	2
581	Reply to: â€œDopaâ€Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansionsâ€ Movement Disorders, 2020, 35, 1890-1891.	2.2	2
582	Sequence variants in three genes underlying leukodystrophy in Pakistani families. <i>International Journal of Developmental Neuroscience</i> , 2020, 80, 380-388.	0.7	2
583	Questions on <i>NOTCH2NLC</i> Repeat Expansions in Parkinson Disease. <i>JAMA Neurology</i> , 2021, 78, 762.	4.5	2
584	Expanding the mutational landscape and clinical phenotype of the <i>YIF1B</i> related brain disorder. <i>Brain</i> , 2021, 144, e85-e85.	3.7	2
585	GM1 â€Gangliosidosis Type III Associated Parkinsonism. <i>Movement Disorders Clinical Practice</i> , 2021, 8, S21-S23.	0.8	2
586	Genetics of NBIA Disorders. , 2015, , 263-291.		2
587	Age-dependent epileptic encephalopathy associated with an unusual co-occurrence of ZEB2 and SCN1A variants. , 2020, 22, 111-115.		2
588	Genetic Variability and Alzheimerâ€™s Disease. , 1994, , 190-198.		2
589	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. <i>Neurogenetics</i> , 2021, , 1.	0.7	2
590	The clinical characteristics of familial cluster headache. <i>Cephalalgia</i> , 2022, , 033310242210764.	1.8	2
591	A complex epileptic and dysmorphic phenotype associated with a novel frameshift KDM5B variant and deletion of SCN gene cluster. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2022, 97, 20-22.	0.9	2
592	Biallelic Variants in the Ectonucleotidase <sc><i>ENTPD1</i></sc> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. <i>Annals of Neurology</i> , 2022, 92, 304-321.	2.8	2
593	A Commentary on â€Four novel C20ORF54 mutations identified in Brownâ€Violettoâ€Van Laere syndrome patients.â€™. <i>Journal of Human Genetics</i> , 2012, 57, 555-555.	1.1	1
594	Genetic linkage analysis of a large family with photoparoxysmal response. <i>Epilepsy Research</i> , 2012, 99, 38-45.	0.8	1

#	ARTICLE	IF	CITATIONS
595	Severe Dejerineâ€”Sottas disease with respiratory failure and dysmorphic features in association with a PMP22 point mutation and a 3q23 microdeletion. <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 223-225.	1.4	1
596	ADULT LEUKODYSTROPHY: MULTI-DISCIPLINARY STRUCTURED APPROACH. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, e4.196-e4.	0.9	1
597	Neuropathological findings from a human post mortem case of distal hereditary motor neuropathy (dHMN) due to p.Ser135Phe HSPB1 mutation and transgenic mice with mutant or wild-type HSP27 overexpression. <i>Neuromuscular Disorders</i> , 2015, 25, S283.	0.3	1
598	ECG ABNORMALITIES IN ALTERNATING HEMIPLEGIA: A BROADENED PHENOTYPE. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2015, 86, e4.191-e4.	0.9	1
599	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2015, 96, 1008-1009.	2.6	1
600	Reply: The p.Ser107Leu inBICD2is a mutation â€”hot spotâ€” causing distal spinal muscular atrophy. <i>Brain</i> , 2015, 138, e392-e392.	3.7	1
601	Glycogen storage disease type XV: A case report. <i>Neuromuscular Disorders</i> , 2015, 25, S221.	0.3	1
602	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. <i>Brain</i> , 2018, 141, e50-e50.	3.7	1
603	C9orf72 and intracerebral hemorrhage. <i>Neurobiology of Aging</i> , 2019, 84, 237.e1-237.e3.	1.5	1
604	Stem cell transplant arrests decline in case of CSF1R leukoencephalopathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, jnnp-2019-321021.	0.9	1
605	Association between critical care admission and 6-month functional outcome after spontaneous intracerebral haemorrhage. <i>Journal of the Neurological Sciences</i> , 2020, 418, 117141.	0.3	1
606	Genetic epilepsies and the Kv super-family. <i>European Journal of Paediatric Neurology</i> , 2020, 24, 5-6.	0.7	1
607	Parkinsonâ€”s Disease in Kazakhstan: Clinico-Demographic Description of a Large Cohort. <i>Journal of Parkinson's Disease</i> , 2020, 10, 707-709.	1.5	1
608	LRRK2 mutations in Parkinson's disease patients from Central Europe: A case control study. <i>Parkinsonism and Related Disorders</i> , 2021, 83, 110-112.	1.1	1
609	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. <i>Molecular Biology Reports</i> , 2021, 48, 2093-2104.	1.0	1
610	Î±â€”Synuclein (<sc><i>SNCA</i></sc>) <sc>A30G</sc> Mutation as a Cause of a Complex Phenotype Without Parkinsonism. <i>Movement Disorders</i> , 2021, 36, 2209-2212.	2.2	1
611	Magnetic resonance imaging-based scores of small vessel diseases: Associations with intracerebral haemorrhage location. <i>Journal of the Neurological Sciences</i> , 2022, 434, 120165.	0.3	1
612	Biallelic <sc><i>KITLG</i></sc> variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2022, ,	1.3	1

#	ARTICLE	IF	CITATIONS
613	Biallelic loss of <i>EMC10</i> leads to mild to severe intellectual disability. <i>Annals of Clinical and Translational Neurology</i> , 0, , .	1.7	1
614	Primary progressive aphasia: ReADing the clinical GRANularity. <i>Practical Neurology</i> , 2022, 22, 509-514.	0.5	1
615	Microsatellite D21S210 (GT-12) allele frequencies in sporadic Alzheimer's disease. <i>Acta Neurologica Scandinavica</i> , 2009, 91, 145-148.	1.0	0
616	POG11 A novel mutation in the nerve-specific 5'-UTR of the Cx32 gene causing CMTX1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, e50-e50.	0.9	0
617	P61 Diverse phenotypes are associated with missense mutations in the peripheral myelin protein 22 gene. <i>Neuromuscular Disorders</i> , 2010, 20, S21.	0.3	0
618	P62 Characterisation of novel mutations within HSP27 causing Charcot-Marie-Tooth disease 2F and distal hereditary motor neuropathy II. <i>Neuromuscular Disorders</i> , 2010, 20, S21-S22.	0.3	0
619	P63 C-Jun expression in human neuropathies: a pilot study. <i>Neuromuscular Disorders</i> , 2010, 20, S22.	0.3	0
620	P64 Genes for hereditary sensory and autonomic neuropathies: frequency in a UK series and genotype-phenotype correlations. <i>Neuromuscular Disorders</i> , 2010, 20, S22-S22.	0.3	0
621	O08 Characterisation of novel mutations within heat shock protein 27 causing motor axonopathies. <i>Neuromuscular Disorders</i> , 2011, 21, S3.	0.3	0
622	P36 TRPV4 mutations and functional characterisation in a cohort of patients with hereditary neuropathy. <i>Neuromuscular Disorders</i> , 2011, 21, S16.	0.3	0
623	P39 Neurofilament light chain polypeptide gene (NEFL) mutations in autosomal dominant or sporadic Charcot-Marie-Tooth disease. <i>Neuromuscular Disorders</i> , 2011, 21, S17.	0.3	0
624	P41 Genetic mutation frequency in patients with hereditary sensory and autonomic neuropathies (HSAN). <i>Neuromuscular Disorders</i> , 2011, 21, S17-S18.	0.3	0
625	P42 X-inactivation pattern in females with CMTX1. <i>Neuromuscular Disorders</i> , 2011, 21, S18.	0.3	0
626	P43 Genetic modifying factors for the common form of CMT1A due to the chromosome 17 duplication and other causes of CMT1 in non-CMT1A patients. <i>Neuromuscular Disorders</i> , 2011, 21, S18.	0.3	0
627	P45 A clinical study of the hereditary neuropathies due to mutations in the small heat shock proteins. <i>Neuromuscular Disorders</i> , 2011, 21, S18-S19.	0.3	0
628	1030...Charcot-Marie-tooth disease: genetic diagnoses in a specialist clinic. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, e1.57-e1.	0.9	0
629	073...Hereditary spastic paraplegia caused by spastin (SPAST, SPG4) mutations is found more often in males: report of novel mutations from one centre, and review of published literature. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, e1.19-e1.	0.9	0
630	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. <i>Cell Reports</i> , 2013, 4, 402.	2.9	0

#	ARTICLE	IF	CITATIONS
631	CARDIAC FEATURES IN ADULTS WITH ALTERNATING HEMIPLEGIA. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, e4.214-e4.	0.9	0
632	Nueva mutaci3n en SPG11 en una paciente con paraplej3a esp3stica hereditaria complicada: hallazgos cl3nicos-electrofisiol3gicos y moleculares. <i>Neurologia Argentina</i> , 2014, 6, 155-159.	0.1	0
633	Caveolinopathy presenting with muscle pain and rhabdomyolysis. <i>Neuromuscular Disorders</i> , 2015, 25, S297.	0.3	0
634	RYR1-related exertional rhabdomyolysis: Expanding spectrum and diagnostic challenges. <i>Neuromuscular Disorders</i> , 2015, 25, S257-S258.	0.3	0
635	Novel mutation of the ntkr1 gene in a patient with hereditary sensory and autonomic neuropathy type iv phenotype. <i>Journal of the Neurological Sciences</i> , 2015, 357, e348-e349.	0.3	0
636	The utility of immunohistochemistry in the assessment of myopathies with tubular aggregates and cylindrical spirals. <i>Neuromuscular Disorders</i> , 2016, 26, S193-S194.	0.3	0
637	J9...Probing huntington3s disease phenocopy syndromes with next-generation sequencing. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, A78.2-A78.	0.9	0
638	Letter re: Mystery Case: <i>CSF-1R</i> mutation is a cause of intracranial cerebral calcifications, cysts, and leukoencephalopathy. <i>Neurology</i> , 2017, 88, 1978-1978.	1.5	0
639	Next generation sequencing in inherited myopathies. <i>Neuromuscular Disorders</i> , 2017, 27, S42.	0.3	0
640	PO189...Autonomic failure caused by adult onset alexander disease with a novel gfap mutation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, A61.2-A61.	0.9	0
641	Reply: PRUNE1: a disease-causing gene for secondary microcephaly. <i>Brain</i> , 2017, 140, e62-e62.	3.7	0
642	Reply to: No evidence supports genetic heterogeneity of neuronal intranuclear inclusion disease. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 2544-2545.	1.7	0
643	Some pathogenic SETX variants are partially conserved during evolution. <i>Gene</i> , 2021, 771, 145360.	1.0	0
644	Reply: Genetic heterogeneity of neuronal intranuclear inclusion disease. What about the infantile variant?. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1002-1004.	1.7	0
645	Novel ALDH3A2 mutations in structural and functional domains of FALDH causing diverse clinical phenotypes in Sj3gren3-Larsson syndrome patients. <i>Human Mutation</i> , 2021, 42, 1015-1029.	1.1	0
646	Commentary: <scp>GM1</scp>3Gangliosidosis Type <scp>III</scp> Associated Parkinsonism. <i>Movement Disorders Clinical Practice</i> , 2021, 8, S24-S25.	0.8	0
647	Distribution of HLA Alleles and Genotypes in Patients with Chronic Inflammatory Demyelinating Polyneuropathy. <i>Journal of Molecular Neuroscience</i> , 2021, , 1.	1.1	0
648	PO184...Analysis of copy number variants in familial and sporadic parkinson3s disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, A59.4-A60.	0.9	0

#	ARTICLE	IF	CITATIONS
649	SPG11 Presenting with Tremor. Tremor and Other Hyperkinetic Movements, 2012, 2, .	1.1	0
650	Advancing Charcot-Marie-Tooth disease diagnostics, through the UK 100,000 Genomes Project. Medizinische Genetik, 2020, 32, 227-231.	0.1	0
651	Clinical and genetic analysis of spinocerebellar ataxia type 11. Cerebellum, 2008, 7, 1-6.	1.4	0
652	022â€¦ Functional genomics and transcriptomics further characterise and potentially improve diagnostic yield of hereditary ataxias. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A107.3-A108.	0.9	0
653	Genomic features specific to the human lineage are associated with neurological diseases and intelligence. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, A97.1-A97.	0.9	0