Henry Houlden

List of Publications by Year
in descending order

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

#	Article	IF	Citations
1	MRI and CT imaging biomarkers of cerebral amyloid angiopathy in lobar intracerebral hemorrhage. International Journal of Stroke, 2023, 18, 85-94.	5.9	11
2	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
3	Cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS): genetic and clinical aspects. Practical Neurology, 2022, 22, 14-18.	1.1	14
4	Metabolically induced intracellular pH changes activate mitophagy, autophagy, and cell protection in familial forms of Parkinson's disease. FEBS Journal, 2022, 289, 699-711.	4.7	17
5	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	7.6	14
6	Dissecting the Phenotype and Genotype of <scp>PLA2G6</scp>â€Related Parkinsonism. Movement Disorders, 2022, 37, 148-161.	3.9	32
7	Heterozygous <scp>EIF2AK2</scp> Variant Causes Adolescenceâ€Onset Generalized Dystonia Partially Responsive to <scp>DBS</scp>. Movement Disorders Clinical Practice, 2022, 9, 268-271.	1.5	7
8	WARS2 mutations cause dopa-responsive early-onset parkinsonism and progressive myoclonus ataxia. Parkinsonism and Related Disorders, 2022, 94, 54-61.	2.2	13
9	Novel Machado-Joseph disease-modifying genes and pathways identified by whole-exome sequencing. Neurobiology of Disease, 2022, 162, 105578.	4.4	3
10	Genome-Wide Association Study of Clinical Outcome After Aneurysmal Subarachnoid Haemorrhage: Protocol. Translational Stroke Research, 2022, 13, 565-576.	4.2	5
11	DYT6 mutated THAP1 is a cell type dependent regulator of the SP1 family. Brain, 2022, 145, 3968-3984.	7.6	4
12	Prevalence of Fabry Disease among Patients with Parkinsonâ€™s Disease. Parkinson's Disease, 2022, 2022, 1-8.	1.1	3
13	Biallelic Lossâ€ofâ€Function NDUFA12 Variants Cause a Wide Phenotypic Spectrum from Leigh/Leighâ€Like Syndrome to Isolated Optic Atrophy. Movement Disorders Clinical Practice, 2022, 9, 218-228.	1.5	5
14	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
15	Magnetic resonance imaging-based scores of small vessel diseases: Associations with intracerebral haemorrhage location. Journal of the Neurological Sciences, 2022, 434, 120165.	0.6	1
16	Reply to: Juvenile PLA2G6â€parkinsonism due to Indian â€˜Asianâ€™ p.R741Q mutation, and response to STN DBS. Movement Disorders, 2022, 37, 658-662.	3.9	5
17	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3
18	The clinical characteristics of familial cluster headache. Cephalalgia, 2022, , 033310242210764.	3.9	2

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19	How to approach a neurogenetics diagnosis in different European countries: The European Academy of Neurology Neurogenetics Panel survey. European Journal of Neurology, 2022, 29, 1885-1891.	3.3	5
20	<scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540.	2.0	7
21	Comprehensive genetic diagnosis of tandem repeat expansion disorders with programmable targeted nanopore sequencing. Science Advances, 2022, 8, eabm5386.	10.3	68
22	Whole genome sequencing for the diagnosis of neurological repeat expansion disorders in the UK: a retrospective diagnostic accuracy and prospective clinical validation study. Lancet Neurology, The, 2022, 21, 234-245.	10.2	74
23	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	7.6	8
24	A complex epileptic and dysmorphic phenotype associated with a novel frameshift KDM5B variant and deletion of SCN gene cluster. Seizure: the Journal of the British Epilepsy Association, 2022, 97, 20-22.	2.0	2
25	Bi-allelic loss-of-function OBSCN variants predispose individuals to severe recurrent rhabdomyolysis. Brain, 2022, 145, 3985-3998.	7.6	6
26	Motor neuron pathology in CANVAS due to RFC1 expansions. Brain, 2022, 145, 2121-2132.	7.6	32
27	Genetic defects are common in myopathies with tubular aggregates. Annals of Clinical and Translational Neurology, 2022, 9, 4-15.	3.7	7
28	TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. Brain, 2022, 145, 3095-3107.	7.6	17
29	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	5.3	2
30	Mutations inTAF8cause a neurodegenerative disorder. Brain, 2022, 145, 3022-3034.	7.6	3
31	Biallelic <scp> KITLG </scp> variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. Journal of the European Academy of Dermatology and Venereology, 2022, , .	2.4	1
32	Combining biomarkers for prognostic modelling of Parkinsonâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 707-715.	1.9	9
33	Iatrogenic cerebral amyloid angiopathy: an emerging clinical phenomenon. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 693-700.	1.9	26
34	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.	1.9	0
35	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.	1.9	0
36	Recapitulation of endogenous 4R tau expression and formation of insoluble tau in directly reprogrammed human neurons. Cell Stem Cell, 2022, 29, 918-932.e8.	11.1	20

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37	Primary progressive aphasia: ReADing the clinical GRANularity. Practical Neurology, 2022, 22, 509-514.	1.1	1
38	Diagnosing Premotor Multiple System Atrophy. Neurology, 2022, 99, .	1.1	4
39	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. Journal of Medical Genetics, 2021, 58, 815-831.	3.2	3
40	Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Journal of Medical Genetics, 2021, 58, 495-504.	3.2	14
41	DRPLA: understanding the natural history and developing biomarkers to accelerate therapeutic trials in a globally rare repeat expansion disorder. Journal of Neurology, 2021, 268, 3031-3041.	3.6	6
42	ANGPTL6 Genetic Variants Are an Underlying Cause of Familial Intracranial Aneurysms. Neurology, 2021, 96, e947-e955.	1.1	6
43	Genetic testing in dementia â€" utility and clinical strategies. Nature Reviews Neurology, 2021, 17, 23-36.	10.1	26
44	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. Movement Disorders, 2021, 36, 251-255.	3.9	23
45	Negative screening for 12 rare LRRK2 pathogenic variants in a cohort of Nigerians with Parkinson's disease. Neurobiology of Aging, 2021, 99, 101.e15-101.e19.	3.1	6
46	Novel variants broaden the phenotypic spectrum of PLEKHG5 â€ɐssociated neuropathies. European Journal of Neurology, 2021, 28, 1344-1355.	3.3	4
47	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
48	Small Vessel Disease and Ischemic Stroke Risk During Anticoagulation for Atrial Fibrillation After Cerebral Ischemia. Stroke, 2021, 52, 91-99.	2.0	40
49	Some pathogenic SETX variants are partially conserved during evolution. Gene, 2021, 771, 145360.	2.2	0
50	Brownâ^'Vialettoâ^'Van Laere and Fazioâ^'Londe syndromes: SLC52A3 mutations with puzzling phenotypes and inheritance. European Journal of Neurology, 2021, 28, 945-954.	3.3	5
51	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. Movement Disorders, 2021, 36, 449-459.	3.9	16
52	CANVAS: a late onset ataxia due to biallelic intronic AAGGG expansions. Journal of Neurology, 2021, 268, 1119-1126.	3.6	19
53	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	2.8	8
54	Homozygous missense WIPI2 variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Brain Communications, 2021, 3, fcab183.	3.3	10

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55	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	7.6	12
56	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
57	Natural History, Phenotypic Spectrum, and Discriminative Features of Multisystemic RFC1 Disease. Neurology, 2021, 96, e1369-e1382.	1.1	93
58	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
59	Cerebral Small Vessel Disease and Functional Outcome Prediction After Intracerebral Hemorrhage. Neurology, 2021, 96, e1954-e1965.	1.1	10
60	R1352Q <i>CACNA1A</i> Variant in a Patient with Sporadic Hemiplegic Migraine, Ataxia, Seizures and Cerebral Oedema: A Case Report. Case Reports in Neurology, 2021, 13, 123-130.	0.7	5
61	LRRK2 mutations in Parkinson's disease patients from Central Europe: A case control study. Parkinsonism and Related Disorders, 2021, 83, 110-112.	2.2	1
62	Expanding the Spectrum of <scp>AP5Z1â€</scp>Related Hereditary Spastic Paraplegia (<scp>HSPâ€SPG48</scp>): A Multicenter Study on a Rare Disease. Movement Disorders, 2021, 36, 1034-1038.	3.9	9
63	RFC1-related ataxia is a mimic of early multiple system atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 444-446.	1.9	25
64	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
65	A geroscience approach for Parkinsonâ€™s disease: Conceptual framework and design of PROPAG-AGEING project. Mechanisms of Ageing and Development, 2021, 194, 111426.	4.6	14
66	Small vessel disease burden and intracerebral haemorrhage in patients taking oral anticoagulants. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 805-814.	1.9	17
67	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	2.5	12
68	Uniparental isodisomy of chromosome 2 causing MRPL44-related multisystem mitochondrial disease. Molecular Biology Reports, 2021, 48, 2093-2104.	2.3	1
69	Expanding the Spectrum of Movement Disorders Associated With C9orf72 Hexanucleotide Expansions. Neurology: Genetics, 2021, 7, e575.	1.9	20
70	Reply: Genetic heterogeneity of neuronal intranuclear inclusion disease. What about the infantile variant?. Annals of Clinical and Translational Neurology, 2021, 8, 1002-1004.	3.7	0
71	Human-lineage-specific genomic elements are associated with neurodegenerative disease and APOE transcript usage. Nature Communications, 2021, 12, 2076.	12.8	9
72	Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. European Journal of Human Genetics, 2021, 29, 1027-1031.	2.8	5

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73	Tay-Sachs Disease: Two Novel Rare HEXA Mutations from Pakistan and Morocco. Klinische Padiatrie, 2021, 233, 226-230.	0.6	4
74	Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltageâ€gated sodium channel function. Epilepsia, 2021, 62, e82-e87.	5.1	9
75	De novo mutation in SLC25A22 gene: expansion of the clinical and electroencephalographic phenotype. Journal of Neurogenetics, 2021, 35, 67-73.	1.4	3
76	Editorial: Multifaceted Genes in Amyotrophic Lateral Sclerosis-Frontotemporal Dementia. Frontiers in Neuroscience, 2021, 15, 680185.	2.8	3
77	A glimpse of the genetics of youngâ€onset Parkinsonâ€™s disease in Central Asia. Molecular Genetics & Genomic Medicine, 2021, 9, e1671.	1.2	7
78	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	9.0	95
79	Two novel biâ€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249.	1.2	7
80	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18
81	Integrating protein networks and machine learning for disease stratification in the Hereditary Spastic Paraplegias. IScience, 2021, 24, 102484.	4.1	8
82	Alzheimerâ€™s disease neuropathological change three decades after iatrogenic amyloid-Î² transmission. Acta Neuropathologica, 2021, 142, 211-215.	7.7	17
83	A Recurrent <scp>VPS16</scp> p.Arg187* Nonsense Variant in Earlyâ€Onset Generalized Dystonia. Movement Disorders, 2021, 36, 1984-1985.	3.9	7
84	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
85	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
86	Whole-genome sequencing. Practical Neurology, 2021, 21, 322-327.	1.1	3
87	RFC1 expansions are a common cause of idiopathic sensory neuropathy. Brain, 2021, 144, 1542-1550.	7.6	63
88	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
89	Cerebellar ataxia, neuropathy, vestibular areflexia syndrome: genetic and clinical insights. Current Opinion in Neurology, 2021, 34, 556-564.	3.6	8
90	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	6.2	10

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91	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. European Journal of Human Genetics, 2021, 29, 1226-1234.	2.8	8
92	Questions on NOTCH2NLC Repeat Expansions in Parkinson Disease. JAMA Neurology, 2021, 78, 762.	9.0	2
93	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
94	Novel ALDH3A2 mutations in structural and functional domains of FALDH causing diverse clinical phenotypes in SjÃ¶grenâ€“Larsson syndrome patients. Human Mutation, 2021, 42, 1015-1029.	2.5	0
95	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
96	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	2.4	11
97	A novel variant in the DSE gene leads to Ehlersâ€“Danlos musculocontractural type 2 in a Pakistani family. Congenital Anomalies (discontinued), 2021, 61, 177-182.	0.6	3
98	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	6.2	11
99	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Genetics in Medicine, 2021, 23, 2138-2149.	2.4	11
100	A Novel Homozygous <scp>ADCY5</scp> Variant is Associated with a Neurodevelopmental Disorder and Movement Abnormalities. Movement Disorders Clinical Practice, 2021, 8, 1140-1143.	1.5	3
101	Coding and noncoding variants in EBF3 are involved in HADDS and simplex autism. Human Genomics, 2021, 15, 44.	2.9	16
102	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	5.3	31
103	Expanding the mutational landscape and clinical phenotype of the YIF1B related brain disorder. Brain, 2021, 144, e85-e85.	7.6	2
104	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
105	Commentary: <scp>GM1</scp>â€Gangliosidosis Type <scp>III</scp> Associated Parkinsonism. Movement Disorders Clinical Practice, 2021, 8, S24-S25.	1.5	0
106	Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. Brain Sciences, 2021, 11, 1150.	2.3	10
107	GM1 â€Gangliosidosis Type III Associated Parkinsonism. Movement Disorders Clinical Practice, 2021, 8, S21-S23.	1.5	2
108	Î±â€Synuclein (<scp>SNCA</scp>) <scp>A30G</scp> Mutation as a Cause of a Complex Phenotype Without Parkinsonism. Movement Disorders, 2021, 36, 2209-2212.	3.9	1

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109	SORL1 mutation in a Greek family with Parkinson's disease and dementia. Annals of Clinical and Translational Neurology, 2021, 8, 1961-1969.	3.7	7
110	Distribution of HLA Alleles and Genotypes in Patients with Chronic Inflammatory Demyelinating Polyneuropathy. Journal of Molecular Neuroscience, 2021, , 1.	2.3	0
111	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. Neurobiology of Aging, 2021, 106, 343.e1-343.e8.	3.1	5
112	Investigating RFC1 expansions in sporadic amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2021, 430, 118061.	0.6	8
113	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	3.8	16
114	Spastic paraplegia preceding PSEN1 â€related familial Alzheimer's disease. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12186.	2.4	7
115	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
116	Expanding the phenotypic spectrum of BCS1L â€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
117	Early-onset phenotype of bi-allelic GRN mutations. Brain, 2021, 144, e22-e22.	7.6	5
118	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. Neurogenetics, 2021, , 1.	1.4	2
119	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	6.0	42
120	Epileptic Phenotypes Associated With SNAREs and Related Synaptic Vesicle Exocytosis Machinery. Frontiers in Neurology, 2021, 12, 806506.	2.4	9
121	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	12.8	44
122	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
123	Predictors for a dementia gene mutation based on gene-panel next-generation sequencing of a large dementia referral series. Molecular Psychiatry, 2020, 25, 3399-3412.	7.9	34
124	Adaptor protein complex 4 deficiency: a paradigm of childhood-onset hereditary spastic paraplegia caused by defective protein trafficking. Human Molecular Genetics, 2020, 29, 320-334.	2.9	45
125	A rare PANK2 deletion in the first north African patient affected with pantothenate kinase associated neurodegeneration. Journal of the Neurological Sciences, 2020, 410, 116639.	0.6	3
126	Identification of common genetic markers of paroxysmal neurological disorders using a network analysis approach. Neurological Sciences, 2020, 41, 851-857.	1.9	3

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127	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. Acta Neuropathologica, 2020, 139, 415-442.	7.7	38
128	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29
129	Association between critical care admission and 6-month functional outcome after spontaneous intracerebral haemorrhage. Journal of the Neurological Sciences, 2020, 418, 117141.	0.6	1
130	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. Nature Genetics, 2020, 52, 1303-1313.	21.4	163
131	A new family with GLRB-related hyperekplexia showing chorea in homo- and heterozygous variant carriers. Parkinsonism and Related Disorders, 2020, 79, 97-99.	2.2	4
132	Neuronal intranuclear inclusion disease is genetically heterogeneous. Annals of Clinical and Translational Neurology, 2020, 7, 1716-1725.	3.7	38
133	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
134	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
135	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. Neurosurgery, 2020, 87, 1269-1276.	1.1	6
136	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
137	Loss of NARS1 impairs progenitor proliferation in cortical brain organoids and leads to microcephaly. Nature Communications, 2020, 11, 4038.	12.8	44
138	A novel RFC1 repeat motif (ACAGG) in two Asia-Pacific CANVAS families. Brain, 2020, 143, 2904-2910.	7.6	53
139	Reply to: â€œDopaâ€Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansionsâ€• Movement Disorders, 2020, 35, 1890-1891.	3.9	2
140	Reply to: No evidence supports genetic heterogeneity of neuronal intranuclear inclusion disease. Annals of Clinical and Translational Neurology, 2020, 7, 2544-2545.	3.7	0
141	Sensitivity and specificity of blood-fluid levels for oral anticoagulant-associated intracerebral haemorrhage. Scientific Reports, 2020, 10, 15529.	3.3	5
142	RFC1 expansions can mimic hereditary sensory neuropathy with cough and SjÃ¶gren syndrome. Brain, 2020, 143, e82-e82.	7.6	25
143	Mutation in RNF170 causes sensory ataxic neuropathy with vestibular areflexia: a CANVAS mimic. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1237-1238.	1.9	11
144	A MÄori specific RFC1 pathogenic repeat configuration in CANVAS, likely due to a founder allele. Brain, 2020, 143, 2673-2680.	7.6	45

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145	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	5.3	70
146	Association of enlarged perivascular spaces and anticoagulant-related intracranial hemorrhage. Neurology, 2020, 95, e2192-e2199.	1.1	24
147	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. European Journal of Human Genetics, 2020, 28, 1763-1768.	2.8	9
148	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	12.8	47
149	A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. Brain, 2020, 143, e49-e49.	7.6	5
150	How to diagnose difficult white matter disorders. Practical Neurology, 2020, 20, 280-286.	1.1	3
151	An update on MSA: premotor and non-motor features open a window of opportunities for early diagnosis and intervention. Journal of Neurology, 2020, 267, 2754-2770.	3.6	25
152	Genetic and Clinical Heterogeneity in Thirteen New Cases with Aceruloplasminemia. Atypical Anemia as a Clue for an Early Diagnosis. International Journal of Molecular Sciences, 2020, 21, 2374.	4.1	25
153	Sequence variants in three genes underlying leukodystrophy in Pakistani families. International Journal of Developmental Neuroscience, 2020, 80, 380-388.	1.6	2
154	Novel fluid biomarkers to differentiate frontotemporal dementia and dementia with Lewy bodies from Alzheimer's disease: A systematic review. Journal of the Neurological Sciences, 2020, 415, 116886.	0.6	13
155	Hereditary polyneuropathy with optic atrophy due to PDXK variant leading to impaired Vitamin B6 metabolism. Neuromuscular Disorders, 2020, 30, 583-589.	0.6	7
156	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	1.9	13
157	Haptoglobin genotype and outcome after spontaneous intracerebral haemorrhage. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 298-304.	1.9	4
158	<scp>GGC</scp> Repeat Expansion in <scp>NOTCH2NLC</scp> Is Rare in European Leukoencephalopathy. Annals of Neurology, 2020, 88, 641-642.	5.3	14
159	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
160	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
161	Longer term stroke risk in intracerebral haemorrhage survivors. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 840-845.	1.9	12
162	GGC repeat expansion in NOTCH2NLC is rare in European patients with essential tremor. Brain, 2020, 143, e57-e57.	7.6	13

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163	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	6.2	47
164	LRRK2 Mutations and Asian Disease-Associated Variants in the First Parkinsonâ€™s Disease Cohort from Kazakhstan. Parkinson's Disease, 2020, 2020, 1-10.	1.1	3
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