Elisabeth Tournier-Lasserve

List of Publications by Year
in descending order

191 papers	17,863 citations	13099 68 h-index	13771 129 g-index

198 all docs	198 docs citations	198 times ranked	12699 citing authors

#	Article	IF	Citations
1	Notch3 mutations in CADASIL, a hereditary adult-onset condition causing stroke and dementia. Nature, 1996, 383, 707-710.	27.8	1,893
2	The Control of Vascular Integrity by Endothelial Cell Junctions: Molecular Basis and Pathological Implications. Developmental Cell, 2009, 16, 209-221.	7.0	692
3	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy maps to chromosome 19q12. Nature Genetics, 1993, 3, 256-259.	21.4	676
4	Strong clustering and stereotyped nature of Notch3 mutations in CADASIL patients. Lancet, The, 1997, 350, 1511-1515.	13.7	651
5	The Clinical Spectrum of Familial Hemiplegic Migraine Associated with Mutations in a Neuronal Calcium Channel. New England Journal of Medicine, 2001, 345, 17-24.	27.0	511
6	The ectodomain of the Notch3 receptor accumulates within the cerebrovasculature of CADASIL patients. Journal of Clinical Investigation, 2000, 105, 597-605.	8.2	503
7	Role of COL4A1 in Small-Vessel Disease and Hemorrhagic Stroke. New England Journal of Medicine, 2006, 354, 1489-1496.	27.0	486
8	Truncating mutations in CCM1, encoding KRIT1, cause hereditary cavernous angiomas. Nature Genetics, 1999, 23, 189-193.	21.4	481
9	Notch3 is required for arterial identity and maturation of vascular smooth muscle cells. Genes and Development, 2004, 18, 2730-2735.	5.9	449
10	EndMT contributes to the onset and progression of cerebral cavernous malformations. Nature, 2013, 498, 492-496.	27.8	403
11	Mutations within the Programmed Cell Death 10 Gene Cause Cerebral Cavernous Malformations. American Journal of Human Genetics, 2005, 76, 42-51.	6.2	400
12	A gene for familial hemiplegic migraine maps to chromosome 19. Nature Genetics, 1993, 5, 40-45.	21.4	369
13	Synopsis of Guidelines for the Clinical Management of Cerebral Cavernous Malformations: Consensus Recommendations Based on Systematic Literature Review by the Angioma Alliance Scientific Advisory Board Clinical Experts Panel. Neurosurgery, 2017, 80, 665-680.	1.1	334
14	Genetics of cavernous angiomas. Lancet Neurology, The, 2007, 6, 237-244.	10.2	314
15	Hereditary cerebral cavernous angiomas: clinical and genetic features in 57 French families. Lancet, The, 1998, 352, 1892-1897.	13.7	257
16	Genotype-phenotype correlations in cerebral cavernous malformations patients. Annals of Neurology, 2006, 60, 550-556.	5.3	222
17	Skin biopsy immunostaining with a Notch3 monoclonal antibody for CADASIL diagnosis. Lancet, The, 2001, 358, 2049-2051.	13.7	221
18	Mutations within the MGC4607 Gene Cause Cerebral Cavernous Malformations. American Journal of Human Genetics, 2004, 74, 326-337.	6.2	219

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19	Transgenic Mice Expressing Mutant Notch3 Develop Vascular Alterations Characteristic of Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy. American Journal of Pathology, 2003, 162, 329-342.	3.8	206
20	Mapping of a second locus for familial hemiplegic migraine to 1q21-q23 and evidence of further heterogeneity. Annals of Neurology, 1997, 42, 885-890.	5.3	196
21	Restricted T-cell receptor V beta gene usage by myelin basic protein-specific T-cell clones in multiple sclerosis: predominant genes vary in individuals.. Proceedings of the National Academy of Sciences of the United States of America, 1991, 88, 2466-2470.	7.1	195
22	Biology of Vascular Malformations of the Brain. Stroke, 2009, 40, e694-702.	2.0	194
23	Notch signalling pathway and human diseases. Seminars in Cell and Developmental Biology, 1998, 9, 619-625.	5.0	180
24	De novo mutations in ATP1A2 and CACNA1A are frequent in early-onset sporadic hemiplegic migraine. Neurology, 2010, 75, 967-972.	1.1	179
25	Cerebral cavernous malformations: from CCM genes to endothelial cell homeostasis. Trends in Molecular Medicine, 2013, 19, 302-308.	6.7	168
26	De novo mutation in theNotch3 gene causing CADASIL. Annals of Neurology, 2000, 47, 388-391.	5.3	167
27	Recent insights into cerebral cavernous malformations: the molecular genetics of CCM. FEBS Journal, 2010, 277, 1070-1075.	4.7	159
28	Notch3 Mutations in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL), a Mendelian Condition Causing Stroke and Vascular Dementia. Annals of the New York Academy of Sciences, 1997, 826, 213-217.	3.8	157
29	Recurrence of the T666M Calcium Channel CACNA1A Gene Mutation in Familial Hemiplegic Migraine with Progressive Cerebellar Ataxia. American Journal of Human Genetics, 1999, 64, 89-98.	6.2	150
30	HeterozygousHTRA1mutations are associated with autosomal dominant cerebral small vessel disease. Brain, 2015, 138, 2347-2358.	7.6	147
31	The Hox-1.3 homeo box protein is a sequence-specific DNA-binding phosphoprotein.. Genes and Development, 1989, 3, 158-172.	5.9	141
32	A gene for hereditary paroxysmal cerebellar ataxia maps to chromosome 19p. Annals of Neurology, 1995, 37, 289-293.	5.3	135
33	Pathogenic Mutations Associated with Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy Differently Affect Jagged1 Binding and Notch3 Activity via the RBP/JK Signaling Pathway. American Journal of Human Genetics, 2004, 74, 338-347.	6.2	135
34	Moyamoya disease and syndromes: from genetics to clinical management. The Application of Clinical Genetics, 2015, 8, 49.	3.0	130
35	Impaired Cerebral Vasoreactivity in a Transgenic Mouse Model of Cerebral Autosomal Dominant Arteriopathy With Subcortical Infarcts and Leukoencephalopathy Arteriopathy. Stroke, 2005, 36, 1053-1058.	2.0	129
36	Regulation of Î²1 Integrin-Klf2-Mediated Angiogenesis by CCM Proteins. Developmental Cell, 2015, 32, 181-190.	7.0	127

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37	Developmental timing of CCM2 loss influences cerebral cavernous malformations in mice. Journal of Experimental Medicine, 2011, 208, 1835-1847.	8.5	118
38	PRRT2 mutations cause hemiplegic migraine. Neurology, 2012, 79, 2122-2124.	1.1	118
39	Migraine With Aura and Brain Magnetic Resonance Imaging Abnormalities in Patients With CADASIL. Archives of Neurology, 2004, 61, 1237-40.	4.5	115
40	Familial Form of Intracranial Cavernous Angioma: MR Imaging Findings in 51 Families. Radiology, 2000, 214, 209-216.	7.3	109
41	Loss of BRCC3 Deubiquitinating Enzyme Leads to Abnormal Angiogenesis and Is Associated with Syndromic Moyamoya. American Journal of Human Genetics, 2011, 88, 718-728.	6.2	109
42	Cognitive Alterations in Non-Demented CADASIL Patients. Cerebrovascular Diseases, 1998, 8, 97-101.	1.7	108
43	Clinical features of cerebral cavernous malformations patients with KRIT1 mutations. Annals of Neurology, 2004, 55, 213-220.	5.3	107
44	Spectrum and expression analysis of KRIT1 mutations in 121 consecutive and unrelated patients with Cerebral Cavernous Malformations. European Journal of Human Genetics, 2002, 10, 733-740.	2.8	105
45	COL4A1 Mutation in a Patient With Sporadic, Recurrent Intracerebral Hemorrhage. Stroke, 2007, 38, 1461-1464.	2.0	104
46	Distinct phenotypic and functional features of CADASIL mutations in the Notch3 ligand binding domain. Brain, 2009, 132, 1601-1612.	7.6	103
47	Sulindac metabolites decrease cerebrovascular malformations in CCM3 -knockout mice. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 8421-8426.	7.1	102
48	Moyamoya disease: diagnosis and interventions. Lancet Neurology, The, 2022, 21, 747-758.	10.2	102
49	Tissue-specific conditionalCCM2knockout mice establish the essential role of endothelial CCM2 in angiogenesis: implications for human cerebral cavernous malformations. DMM Disease Models and Mechanisms, 2009, 2, 168-177.	2.4	100
50	An association between autosomal dominant cerebral cavernomas and a distinctive hyperkeratotic cutaneous vascular malformation in 4 families. Annals of Neurology, 1999, 45, 250-254.	5.3	99
51	New phenotype of the cerebral autosomal dominant arteriopathy mapped to chromosome 19: migraine as the prominent clinical feature.. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 59, 579-585.	1.9	96
52	Loss of Î±1Î²1 Soluble Guanylate Cyclase, the Major Nitric Oxide Receptor, Leads to Moyamoya and Achalasia. American Journal of Human Genetics, 2014, 94, 385-394.	6.2	95
53	The Genetics of Migraine. Lancet Neurology, The, 2002, 1, 285-293.	10.2	94
54	C9ORF72 Repeat Expansions in the Frontotemporal Dementias Spectrum of Diseases: A Flow-chart for Genetic Testing. Journal of Alzheimer's Disease, 2013, 34, 485-499.	2.6	93

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55	CCM1â€“ICAP-1 complex controls Î²1 integrinâ€“dependent endothelial contractility and fibronectin remodeling. Journal of Cell Biology, 2013, 202, 545-561.	5.2	93
56	The archetypal R90C CADASILâ€“NOTCH3 mutation retains NOTCH3 function in vivo. Human Molecular Genetics, 2007, 16, 982-992.	2.9	92
57	Familial hemiplegic migraine and autosomal dominant arteriopathy with leukoencephalopathy (CADASIL). Annals of Neurology, 1995, 38, 817-824.	5.3	91
58	ATP1A2 mutations in 11 families with familial hemiplegic migraine. Human Mutation, 2005, 26, 281-281.	2.5	87
59	Frequency and phenotypes of cutaneous vascular malformations in a consecutive series of 417 patients with familial cerebral cavernous malformations. Journal of the European Academy of Dermatology and Venereology, 2009, 23, 1066-1072.	2.4	87
60	Familial Lupus Erythematosus. Medicine (United States), 2001, 80, 153-158.	1.0	86
61	Impaired Vascular Mechanotransduction in a Transgenic Mouse Model of CADASIL Arteriopathy. Stroke, 2005, 36, 113-117.	2.0	85
62	Research Progresses in Understanding the Pathophysiology of Moyamoya Disease. Cerebrovascular Diseases, 2016, 41, 105-118.	1.7	82
63	Clinical and brain MRI follow-up study of a family with COL4A1 mutation. Neurology, 2007, 69, 1564-1568.	1.1	79
64	Elicited repetitive daily blindness. Neurology, 2009, 72, 1178-1183.	1.1	79
65	Benign paroxysmal tonic upgaze, benign paroxysmal torticollis, episodic ataxia and CACNA1A mutation in a family. Journal of Neurology, 2008, 255, 1600-1602.	3.6	77
66	Rare RNF213 variants in the C-terminal region encompassing the RING-finger domain are associated with moyamoya angiopathy in Caucasians. European Journal of Human Genetics, 2017, 25, 995-1003.	2.8	77
67	Patterns of expression of the three cerebral cavernous malformation (CCM) genes during embryonic and postnatal brain development. Gene Expression Patterns, 2006, 6, 495-503.	0.8	74
68	<i>CCM3</i> Mutations Are Associated with Early-Onset Cerebral Hemorrhage and Multiple Meningiomas. Molecular Syndromology, 2013, 4, 165-172.	0.8	74
69	Familial occurrence and heritable connective tissue disorders in cervical artery dissection. Neurology, 2014, 83, 2023-2031.	1.1	74
70	T cell response to myelin basic protein epitopes in multiple sclerosis patients and healthy subjects. European Journal of Immunology, 1991, 21, 1391-1395.	2.9	71
71	Autoimmune diseases in families of French patients with multiple sclerosis. Acta Neurologica Scandinavica, 2000, 101, 36-40.	2.1	64
72	Cerebral Cavernous Malformation-1 Protein Controls DLL4-Notch3 Signaling Between the Endothelium and Pericytes. Stroke, 2015, 46, 1337-1343.	2.0	62

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73	Familial Cluster Headache: A Series of 186 Index Patients. Headache, 2002, 42, 974-977.	3.9	61
74	Disruption of a mi<scp>R</scp>â€29 binding site leading to <scp>COL4A1</scp> upregulation causes pontine autosomal dominant microangiopathy with leukoencephalopathy. Annals of Neurology, 2016, 80, 741-753.	5.3	61
75	Missense CACNA1A Mutation Causing Episodic Ataxia Type 2. Archives of Neurology, 2001, 58, 292.	4.5	59
76	Adult-onset genetic leukoencephalopathies: A MRI pattern-based approach in a comprehensive study of 154 patients. Brain, 2015, 138, 284-292.	7.6	58
77	Activating NOTCH3 mutation in a patient with small-vessel-disease of the Brain. Human Mutation, 2008, 29, 452-452.	2.5	53
78	Antithrombotic Therapy and Bleeding Risk in a Prospective Cohort Study of Patients With Cerebral Cavernous Malformations. Stroke, 2012, 43, 3196-3199.	2.0	52
79	Krit1/cerebral cavernous malformation 1 mRNA is preferentially expressed in neurons and epithelial cells in embryo and adult. Mechanisms of Development, 2002, 117, 363-367.	1.7	51
80	Lack of the mesodermal homeodomain protein MEOX1 disrupts sclerotome polarity and leads to a remodeling of the cranio-cervical joints of the axial skeleton. Developmental Biology, 2009, 332, 383-395.	2.0	51
81	Human T-cell response to myelin basic protein in multiple sclerosis patients and healthy subjects. Journal of Neuroscience Research, 1988, 19, 149-156.	2.9	50
82	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: a clinicopathological and genetic study of a Swiss family.. Journal of Neurology, Neurosurgery and Psychiatry, 1995, 59, 138-143.	1.9	50
83	CCM molecular screening in a diagnosis context: novel unclassified variants leading to abnormal splicing and importance of large deletions. Neurogenetics, 2013, 14, 133-141.	1.4	50
84	PDGFB Partial Deletion: a New, Rare Mechanism Causing Brain Calcification with Leukoencephalopathy. Journal of Molecular Neuroscience, 2014, 53, 171-175.	2.3	50
85	Evaluation of DHPLC analysis in mutational scanning ofNotch3, a gene with a high G-C content. Human Mutation, 2000, 16, 518-526.	2.5	49
86	New Players in the Genetics of Stroke. New England Journal of Medicine, 2002, 347, 1711-1712.	27.0	49
87	Frequency of Retinal Cavernomas in 60 Patients With Familial Cerebral Cavernomas. JAMA Ophthalmology, 2006, 124, 885.	2.4	49
88	Nontraumatic Pediatric Intracerebral Hemorrhage. Stroke, 2019, 50, 3654-3661.	2.0	49
89	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. Brain, 2019, 142, 1573-1586.	7.6	49
90	New CACNA1A Gene Mutation in a Case of Familial Hemiplegic Migraine with Status epilepticus. European Neurology, 2004, 52, 58-61.	1.4	48

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91	An italian kindred with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). Annals of Neurology, 1995, 38, 231-236.	5.3	46
92	Identification of CACNA1A large deletions in four patients with episodic ataxia. Neurogenetics, 2010, 11, 101-106.	1.4	46
93	A novel hereditary small vessel disease of the brain. Annals of Neurology, 2006, 59, 353-357.	5.3	39
94	Severe Attacks of Familial Hemiplegic Migraine, Childhood Epilepsy and ATP1A2 Mutation. Cephalalgia, 2008, 28, 774-777.	3.9	39
95	Episodic ataxia type 2: unusual aspects in clinical and genetic presentation. Special emphasis in childhood. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 1289-1292.	1.9	39
96	Genetic heterogeneity and absence of founder effect in a series of 36 French cerebral cavernous angiomas families. European Journal of Human Genetics, 1999, 7, 499-504.	2.8	38
97	Anticardiolipin antibodies in patients with multiple sclerosis do not represent a subgroup of patients according to clinical, familial, and biological characteristics. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 72, 647-649.	1.9	38
98	COL4A1 Mutations Associated with a Characteristic Pattern of Intracranial Calcification. Neuropediatrics, 2011, 42, 227-233.	0.6	38
99	Recurrent Episodes of Coma: An Unusual Phenotype of Familial Hemiplegic Migraine with Linkage to Chromosome 1. Neuropediatrics, 1999, 30, 214-217.	0.6	37
100	Familial cavernous malformations in a large French kindred: mapping of the gene to the CCM1 locus on chromosome 7q. Journal of Neurology, Neurosurgery and Psychiatry, 1997, 63, 40-45.	1.9	36
101	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: from stroke to vessel wall physiology. Journal of Neurology, Neurosurgery and Psychiatry, 2001, 70, 285-287.	1.9	36
102	Cerebral Cavernous Malformation 1/2 complex controls ROCK1 and ROCK2 complementary functions for endothelial integrity. Journal of Cell Science, 2018, 131, .	2.0	36
103	Large CACNA1A Deletion in a Family With Episodic Ataxia Type 2. Archives of Neurology, 2008, 65, 817-20.	4.5	34
104	Fetal intracerebral hemorrhage and cataract: think COL4A1. Journal of Perinatology, 2014, 34, 75-77.	2.0	34
105	Systematic pharmacological screens uncover novel pathways involved in cerebral cavernous malformations. EMBO Molecular Medicine, 2018, 10, .	6.9	34
106	The pleiotropy associated with de novo variants in CHD4, CNOT3, and SETD5 extends to moyamoya angiopathy. Genetics in Medicine, 2020, 22, 427-431.	2.4	34
107	Genome-Wide Genotyping Demonstrates a Polygenic Risk Score Associated With White Matter Hyperintensity Volume in CADASIL. Stroke, 2014, 45, 968-972.	2.0	33
108	De novo mutations in CBL causing early-onset paediatric moyamoya angiopathy. Journal of Medical Genetics, 2017, 54, 550-557.	3.2	33

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109	COL4A2 mutation causing adult onset recurrent intracerebral hemorrhage and leukoencephalopathy. Journal of Neurology, 2014, 261, 500-503.	3.6	32
110	Cognitive impairment in children with <scp>CACNA</scp>1A mutations. Developmental Medicine and Child Neurology, 2020, 62, 330-337.	2.1	31
111	Network-based analysis of omics data: the LEAN method. Bioinformatics, 2017, 33, 701-709.	4.1	29
112	APP Mutations in Cerebral Amyloid Angiopathy with or without Cortical Calcifications: Report of Three Families and aÂLiterature Review. Journal of Alzheimer's Disease, 2017, 56, 37-46.	2.6	29
113	APOE É›2 is associated with white matter hyperintensity volume in CADASIL. Journal of Cerebral Blood Flow and Metabolism, 2016, 36, 199-203.	4.3	28
114	Clinical and Molecular Features of 5 European Multigenerational Families With Moyamoya Angiopathy. Stroke, 2019, 50, 789-796.	2.0	27
115	Sporadic hemiplegic migraine and delayed cerebral oedema after minor head trauma: a novel de novo CACNA1A gene mutation. Developmental Medicine and Child Neurology, 2010, 52, 103-104.	2.1	26
116	Mutation in the 3â€™untranslated region of APP as a genetic determinant of cerebral amyloid angiopathy. European Journal of Human Genetics, 2016, 24, 92-98.	2.8	26
117	Differential diagnosis of a vascular leukoencephalopathy within a CADASIL family: use of skin biopsy electron microscopy study and direct genotypic screening. Journal of Neurology, 1998, 245, 734-740.	3.6	25
118	A novel hereditary extensive vascular leukoencephalopathy mapping to chromosome 20q13. Neurology, 2012, 79, 2283-2287.	1.1	25
119	Novel Chronic Mouse Model of Cerebral Cavernous Malformations. Stroke, 2020, 51, 1272-1278.	2.0	25
120	Cerebral small-vessel disease associated with COL4A1 and COL4A2 gene duplications. Neurology, 2014, 83, 1029-1031.	1.1	24
121	CSF1R-related leukoencephalopathy mimicking primary progressive multiple sclerosis. Journal of Neurology, 2016, 263, 1864-1865.	3.6	24
122	Hemiplegic Migraine Associated With PRRT2 Variations. Neurology, 2022, 98, .	1.1	24
123	Phenotypic Variability of Episodic Ataxia Type 2 Mutations: A Family Study. European Neurology, 2010, 64, 114-116.	1.4	23
124	Benign paroxysmal torticollis, benign paroxysmal vertigo, and benign tonic upward gaze are not benign disorders. Developmental Medicine and Child Neurology, 2018, 60, 1256-1263.	2.1	21
125	Presymptomatic genetic testing in CADASIL. Journal of Neurology, 2012, 259, 2131-2136.	3.6	19
126	Cerebral Cavernous Malformations Arise Independent of the Heart of Glass Receptor. Stroke, 2014, 45, 1505-1509.	2.0	19

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127	The immunogenetics of myasthenia gravis, multiple sclerosis and their animal models. Journal of Neuroimmunology, 1993, 47, 103-114.	2.3	18
128	A Human Homolog of Bacterial Acetolactate Synthase Genes Maps within the CADASIL Critical Region. Genomics, 1996, 38, 192-198.	2.9	18
129	Sporadic late onset paroxysmal cerebellar ataxia in four unrelated patients: a new disease?. Journal of Neurology, 2001, 248, 209-214.	3.6	18
130	Stroke-Related Translational Research. Archives of Neurology, 2011, 68, 1110.	4.5	18
131	Natural history of cerebral dot-like cavernomas. Clinical Radiology, 2013, 68, e453-e459.	1.1	18
132	The R131 low-affinity allele of the Fc gamma RIIA receptor is associated with systemic lupus erythematosus but not with other autoimmune diseases in French Caucasians. American Journal of Medicine, 2000, 108, 580-583.	1.5	17
133	Early-Onset Cerebral Amyloid Angiopathy and Alzheimer Disease Related to an APP Locus Triplication. Neurology: Genetics, 2021, 7, e609.	1.9	17
134	COL4A1 Mutation Revealed by an Isolated Brain Hemorrhage. Cerebrovascular Diseases, 2013, 35, 593-594.	1.7	16
135	Deep intronic <scp>KRIT1</scp> mutation in a family with clinically silent multiple cerebral cavernous malformations. Clinical Genetics, 2014, 86, 585-588.	2.0	16
136	Blocking Signalopathic Events to Treat Cerebral Cavernous Malformations. Trends in Molecular Medicine, 2020, 26, 874-887.	6.7	16
137	Healthy monozygous twins do not recognize identical T cell epitopes on the myelin basic protein autoantigen. European Journal of Immunology, 1994, 24, 2299-2303.	2.9	15
138	Intracerebral hemorrhage and COL4A1 mutations, from preterm infants to adult patients. Annals of Neurology, 2009, 65, 1-2.	5.3	15
139	Vascular Remodeling in Moyamoya Angiopathy: From Peripheral Blood Mononuclear Cells to Endothelial Cells. International Journal of Molecular Sciences, 2020, 21, 5763.	4.1	15
140	Hereditary Cerebral Small Vessel Diseases and Stroke: A Guide for Diagnosis and Management. Stroke, 2021, 52, 3025-3032.	2.0	15
141	Late onset hereditary episodic ataxia. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 566-568.	1.9	14
142	Cerebro-retinal microangiopathy with calcifications and cysts due to recessive mutations in the CTC1 gene. Revue Neurologique, 2015, 171, 445-449.	1.5	14
143	Can whole-exome sequencing data be used for linkage analysis?. European Journal of Human Genetics, 2016, 24, 581-586.	2.8	12
144	Heterozygous HTRA1 nonsense or frameshift mutations are pathogenic. Brain, 2021, 144, 2616-2624.	7.6	12

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145	Anti-Myelin Basic Protein Autoreactive T Lymphocytes in Healthy Subjects and Multiple Sclerosis Patients. Annals of the New York Academy of Sciences, 1986, 475, 404-406.	3.8	11
146	Update on the Genetics of Stroke and Cerebrovascular Disease 2004. Stroke, 2005, 36, 179-181.	2.0	11
147	Pulmonary arterial hypertension in familial hemiplegic migraine with ATP1A2 channelopathy. European Respiratory Journal, 2014, 43, 641-643.	6.7	11
148	Cerebral cavernous malformations associated to meningioma: High penetrance in a novel family mutated in thePDCD10gene. Neuroradiology Journal, 2015, 28, 289-293.	1.2	11
149	Acute-Onset Ataxia and Transient Cerebellar Diffusion Restriction Associated with a PRRT2 Mutation. Journal of Stroke and Cerebrovascular Diseases, 2019, 28, e3-e4.	1.6	11
150	In vivo clonal expansion of T lymphocytes specific for an immunodominant N-terminal myelin basic protein epitope in healthy individuals. Journal of Neuroimmunology, 1995, 59, 165-172.	2.3	10
151	Rare variant association testing for multicategory phenotype. Genetic Epidemiology, 2019, 43, 646-656.	1.3	9
152	A case of late-onset CADASIL with interhemispheric disconnection features. Journal of Neurology, 2003, 250, 1242-1244.	3.6	8
153	Xq28 copy number gain causing moyamoya disease and a novel moyamoya syndrome. Journal of Medical Genetics, 2020, 57, 339-346.	3.2	8
154	Spanish families with cavernous angiomas do not share the Hispano- American CCM1 haplotype. Journal of Neurology, Neurosurgery and Psychiatry, 1999, 67, 551-552.	1.9	7
155	Familial central retinal vein occlusion. Eye, 2008, 22, 308-310.	2.1	7
156	An additional monogenic disorder that masquerades as multiple sclerosis. , 1996, 65, 357-358.		6
157	Familial form of typical childhood absence epilepsy in a consanguineous context. Epilepsia, 2010, 51, 1889-1893.	5.1	6
158	Occurrence of multiple Cerebral Cavernous Malformations in a patient with Neurofibromatosis type 1. Journal of the Neurological Sciences, 2015, 350, 98-100.	0.6	6
159	Long-term improvement of paroxysmal dystonic choreathetosis with acetazolamide. Journal of Neurology, 2006, 253, 1362-1364.	3.6	5
160	Multiple cerebral cavernous malformations and a novel CCM3 germline deletion in a German family. Journal of Neurology, 2010, 257, 2097-2098.	3.6	5
161	Advances in Stroke. Stroke, 2013, 44, 309-310.	2.0	5
162	Hyperkeratotic cutaneous vascular malformation associated with familial cerebral cavernous malformations (FCCM) with KRIT1/CCM1 mutation. European Journal of Dermatology, 2014, 24, 255-257.	0.6	5

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163	TREX1 Mutation in Leukodystrophy with Calcifications and Persistent Gadolinium-Enhancement. European Neurology, 2017, 77, 113-114.	1.4	5
164	De novo mutation in the Notch3 gene causing CADASIL. Annals of Neurology, 2000, 47, 388-391.	5.3	5
165	Endâ€Truncated <scp>LAMB1</scp> Causes a Hippocampal Memory Defect and a Leukoencephalopathy. Annals of Neurology, 2021, 90, 962-975.	5.3	5
166	Molecular Genetic Screening of CCM Patients: An Overview. Methods in Molecular Biology, 2020, 2152, 49-57.	0.9	5
167	A Dinucleotide Repeat Polymorphism at the Poly(ADP-ribose) Polymerase Gene is not Associated with Predisposition to Type 1 Diabetes in French Caucasians. Journal of Autoimmunity, 2001, 17, 137-140.	6.5	4
168	Late Diagnosis of COL4A1 Mutation and Problematic Vascular Risk Factor Management. European Neurology, 2014, 72, 150-152.	1.4	4
169	Extension of SKAT to multi-category phenotypes through a geometrical interpretation. European Journal of Human Genetics, 2021, 29, 736-744.	2.8	4
170	Recalibrating vascular malformations and mechanotransduction by pharmacological intervention. Journal of Clinical Investigation, 2022, 132, .	8.2	4
171	To the Editor. Neurology, 1995, 45, 2299-2300.	1.1	3
172	Gene therapy and beyond. Lancet, The, 1999, 354, SIV22.	13.7	3
173	Retinal Ischemic Syndrome, Digestive Tract Small-Vessel Hyalinosis, and Diffuse Cerebral Calcifications: A Pediatric Observation of a Rare Syndrome. JAMA Ophthalmology, 2005, 123, 1141.	2.4	2
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