Douglas A Marchuk

List of Publications by Year
in descending order

95 papers	10,788 citations	41339 49 h-index	51602 86 g-index
106 all docs	106 docs citations	106 times ranked	6602 citing authors

#	Article	IF	Citations
1	Developmental venous anomalies are a genetic primer for cerebral cavernous malformations. , 2022, 1, 246-252.		21
2	Propranolol as therapy for cerebral cavernous malformations: a cautionary note. Journal of Translational Medicine, 2022, 20, 160.	4.4	5
3	Rapamycin in Cerebral Cavernous Malformations: What Doses to Test in Mice and Humans. ACS Pharmacology and Translational Science, 2022, 5, 266-277.	4.9	5
4	Genetic genealogy uncovers a founder deletion mutation in the cerebral cavernous malformations 2 gene. Human Genetics, 2022, , 1.	3.8	0
5	A novel somatic mutation in GNAQ in a capillary malformation provides insight into molecular pathogenesis. Angiogenesis, 2022, 25, 493-502.	7.2	8
6	Propranolol inhibits cavernous vascular malformations by Î²1 adrenergic receptor antagonism in animal models. Journal of Clinical Investigation, 2021, 131, .	8.2	28
7	Abstract MP11: Circulating Plasma Biomarkers Associated With Brain Arteriovenous Malformations. Stroke, 2021, 52, .	2.0	0
8	PIK3CA and CCM mutations fuel cavernomas through a cancer-like mechanism. Nature, 2021, 594, 271-276.	27.8	103
9	Cerebral Cavernous Malformation: From Mechanism to Therapy. Circulation Research, 2021, 129, 195-215.	4.5	82
10	A Neuroprotective Locus Modulates Ischemic Stroke Infarction Independent of Collateral Vessel Anatomy. Frontiers in Neuroscience, 2021, 15, 705160.	2.8	4
11	Pilot investigation of circulating angiogenic and inflammatory biomarkers associated with vascular malformations. Orphanet Journal of Rare Diseases, 2021, 16, 372.	2.7	8
12	Predictors of mortality in patients with hereditary hemorrhagic telangiectasia. Orphanet Journal of Rare Diseases, 2021, 16, 12.	2.7	12
13	Hereditary Hemorrhagic Telangiectasia (Oslerâ€“Weberâ€“Rendu Syndrome). , 2020, , 115-140.		1
14	A Brain-Targeted Orally Available ROCK2 Inhibitor Benefits Mild and Aggressive Cavernous Angioma Disease. Translational Stroke Research, 2020, 11, 365-376.	4.2	22
15	Novel Murine Models of Cerebral Cavernous Malformations. Angiogenesis, 2020, 23, 651-666.	7.2	36
16	Cerebral cavernous malformations are driven by ADAMTS5 proteolysis of versican. Journal of Experimental Medicine, 2020, 217, .	8.5	30
17	Phenotypic characterization of murine models of cerebral cavernous malformations. Laboratory Investigation, 2019, 99, 319-330.	3.7	24
18	Somatic Mutations in Vascular Malformations of Hereditary Hemorrhagic Telangiectasia Result in Bi-allelic Loss of ENG or ACVRL1. American Journal of Human Genetics, 2019, 105, 894-906.	6.2	77

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19	Transcriptome clarifies mechanisms of lesion genesis versus progression in models of Ccm3 cerebral cavernous malformations. Acta Neuropathologica Communications, 2019, 7, 132.	5.2	27
20	Rho Kinase Inhibition Blunts Lesion Development and Hemorrhage in Murine Models of Aggressive Pdcd10/Ccm3 Disease. Stroke, 2019, 50, 738-744.	2.0	40
21	Physical and Family History Variables Associated With Neurological and Cognitive Development in Sturge-Weber Syndrome. Pediatric Neurology, 2019, 96, 30-36.	2.1	32
22	Distinct cellular roles for PDCD10 define a gut-brain axis in cerebral cavernous malformation. Science Translational Medicine, 2019, 11, .	12.4	51
23	Comprehensive transcriptome analysis of cerebral cavernous malformation across multiple species and genotypes. JCI Insight, 2019, 4, .	5.0	40
24	Biomarkers of cavernous angioma with symptomatic hemorrhage. JCI Insight, 2019, 4, .	5.0	25
25	Novel Neuroprotective Loci Modulating Ischemic Stroke Volume in Wild-Derived Inbred Mouse Strains. Genetics, 2019, 213, 1079-1092.	2.9	11
26	Abstract 64: Novel and Known Genes Elucidated in Cerebral Cavernous Malformation Through Comparative Transcriptomic Analysis of Multiple Model Species and Human Microdissected Lesional Endothelial Cells. Stroke, 2019, 50, .	2.0	0
27	Cerebral Cavernous Malformations Develop Through Clonal Expansion of Mutant Endothelial Cells. Circulation Research, 2018, 123, 1143-1151.	4.5	83
28	Neuronal <scp>IL</scp>â€4RÎ± modulates neuronal apoptosis and cell viability during the acute phases of cerebral ischemia. FEBS Journal, 2018, 285, 2785-2798.	4.7	18
29	BAG3 (Bcl-2â€“Associated Athanogene-3) Coding Variant in Mice Determines Susceptibility to Ischemic Limb Muscle Myopathy by Directing Autophagy. Circulation, 2017, 136, 281-296.	1.6	51
30	The pathobiology of vascular malformations: insights from human and model organism genetics. Journal of Pathology, 2017, 241, 281-293.	4.5	64
31	RhoA Kinase Inhibition With Fasudil Versus Simvastatin in Murine Models of Cerebral Cavernous Malformations. Stroke, 2017, 48, 187-194.	2.0	86
32	Natural allelic variation of the IL-21 receptor modulates ischemic stroke infarct volume. Journal of Clinical Investigation, 2016, 126, 2827-2838.	8.2	25
33	B-Cell Depletion Reduces the Maturation of Cerebral Cavernous Malformations in Murine Models. Journal of NeuroImmune Pharmacology, 2016, 11, 369-377.	4.1	39
34	Micro-computed tomography in murine models of cerebral cavernous malformations as a paradigm for brain disease. Journal of Neuroscience Methods, 2016, 271, 14-24.	2.5	25
35	Sturgeâ€“Weber Syndrome. , 2015, , 945-953.		0
36	ADAM12: a genetic modifier of preclinical peripheral arterial disease. American Journal of Physiology - Heart and Circulatory Physiology, 2015, 309, H790-H803.	3.2	34

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37	Exceptional aggressiveness of cerebral cavernous malformation disease associated with PDCD10 mutations. Genetics in Medicine, 2015, 17, 188-196.	2.4	116
38	Abstract T P231: B- Cell Depletion Decreases Lesion Burden in a Murine Model of Cerebral Cavernous Malformation Disease. Stroke, 2015, 46, .	2.0	0
39	Lesions from patients with sporadic cerebral cavernous malformations harbor somatic mutations in the CCM genes: evidence for a common biochemical pathway for CCM pathogenesis. Human Molecular Genetics, 2014, 23, 4357-4370.	2.9	117
40	Hereditary Hemorrhagic Telangiectasia (Oslerâ€“Weberâ€“Rendu Syndrome). , 2013, , 1-18.		2
41	Sturgeâ€“Weber Syndrome and Port-Wine Stains Caused by Somatic Mutation inGNAQ. New England Journal of Medicine, 2013, 368, 1971-1979.	27.0	865
42	A novel genetic locus modulates infarct volume independently of the extent of collateral circulation. Physiological Genomics, 2013, 45, 751-763.	2.3	27
43	Natural Genetic Variation of Integrin Alpha L (Itgal) Modulates Ischemic Brain Injury in Stroke. PLoS Genetics, 2013, 9, e1003807.	3.5	26
44	Brain Vascular Malformation Consortium: Overview, Progress and Future Directions. The Journal of Rare Disorders, 2013, 1, 5.	1.5	21
45	Fasudil Decreases Lesion Burden in a Murine Model of Cerebral Cavernous Malformation Disease. Stroke, 2012, 43, 571-574.	2.0	130
46	Skeletal Muscleâ€“Specific Genetic Determinants Contribute to the Differential Strain-Dependent Effects of Hindlimb Ischemia in Mice. American Journal of Pathology, 2012, 180, 2156-2169.	3.8	66
47	A novel mouse model of cerebral cavernous malformations based on the two-hit mutation hypothesis recapitulates the human disease. Human Molecular Genetics, 2011, 20, 211-222.	2.9	120
48	A founder mutation in the Ashkenazi Jewish population affecting messenger RNA splicing of the CCM2 gene causes cerebral cavernous malformations. Genetics in Medicine, 2011, 13, 662-666.	2.4	12
49	Overlapping spectra of SMAD4 mutations in juvenile polyposis (JP) and JPâ€“HHT syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 333-339.	1.2	128
50	Biallelic somatic and germline mutations in cerebral cavernous malformations (CCMs): evidence for a two-hit mechanism of CCM pathogenesis. Human Molecular Genetics, 2009, 18, 919-930.	2.9	244
51	A Locus Mapping to Mouse Chromosome 7 Determines Infarct Volume in a Mouse Model of Ischemic Stroke. Circulation: Cardiovascular Genetics, 2009, 2, 591-598.	5.1	54
52	The cerebral cavernous malformation signaling pathway promotes vascular integrity via Rho GTPases. Nature Medicine, 2009, 15, 177-184.	30.7	340
53	A Quantitative Trait Locus (LSq-1) on Mouse Chromosome 7 Is Linked to the Absence of Tissue Loss After Surgical Hindlimb Ischemia. Circulation, 2008, 117, 1207-1215.	1.6	105
54	Deletions in CCM2 Are a Common Cause of Cerebral Cavernous Malformations. American Journal of Human Genetics, 2007, 80, 69-75.	6.2	80

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55	Genetic analysis of a family with hereditary glomuvenous malformations. Australasian Journal of Dermatology, 2007, 48, 170-173.	0.7	8
56	Redefining Heart Failure. Journal of the American College of Cardiology, 2006, 48, 1289-1298.	2.8	50
57	SMAD4 mutations found in unselected HHT patients. Journal of Medical Genetics, 2006, 43, 793-797.	3.2	212
58	Neuronal expression of the Ccm2 gene in a new mouse model of cerebral cavernous malformations. Mammalian Genome, 2006, 17, 119-128.	2.2	68
59	Genetics of cerebral cavernous malformations. Current Neurology and Neuroscience Reports, 2005, 5, 391-396.	4.2	62
60	CCM1 and CCM2 protein interactions in cell signaling: implications for cerebral cavernous malformations pathogenesis. Human Molecular Genetics, 2005, 14, 2521-2531.	2.9	238
61	Human Retroviral gag- and gag-pol-like Proteins Interact with the Transforming Growth Factor-Î² Receptor Activin Receptor-like Kinase 1. Journal of Biological Chemistry, 2005, 280, 8482-8493.	3.4	54
62	Molecular Classification of Patients With Unexplained Hamartomatous and Hyperplastic Polyposis. JAMA - Journal of the American Medical Association, 2005, 294, 2465.	7.4	218
63	The Molecular Basis of Cerebrovascular Malformations. , 2005, , 437-450.		0
64	Ccm1is required for arterial morphogenesis: implications for the etiology of human cavernous malformations. Development (Cambridge), 2004, 131, 1437-1448.	2.5	180
65	Loss of p53 Sensitizes Mice with a Mutation in Ccm1 (KRIT1) to Development of Cerebral Vascular Malformations. American Journal of Pathology, 2004, 165, 1509-1518.	3.8	110
66	A combined syndrome of juvenile polyposis and hereditary haemorrhagic telangiectasia associated with mutations in MADH4 (SMAD4). Lancet, The, 2004, 363, 852-859.	13.7	667
67	Polymorphisms in Genes Involved in Inflammatory and Angiogenic Pathways and the Risk of Hemorrhagic Presentation of Brain Arteriovenous Malformations. Stroke, 2004, 35, 2294-2300.	2.0	134
68	Mutations in a Gene Encoding a Novel Protein Containing a Phosphotyrosine-Binding Domain Cause Type 2 Cerebral Cavernous Malformations. American Journal of Human Genetics, 2003, 73, 1459-1464.	6.2	319
69	Vascular morphogenesis: tales of two syndromes. Human Molecular Genetics, 2003, 12, 97R-112.	2.9	134
70	A mouse model for hereditary hemorrhagic telangiectasia (HHT) type 2. Human Molecular Genetics, 2003, 12, 473-482.	2.9	172
71	KRIT1 association with the integrin-binding protein ICAP-1: a new direction in the elucidation of cerebral cavernous malformations (CCM1) pathogenesis. Human Molecular Genetics, 2002, 11, 389-396.	2.9	176
72	Fine mapping and genetic heterogeneity in the pure form of autosomal dominant familial spastic paraplegia. Neurogenetics, 2001, 3, 91-97.	1.4	13

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73	Clinical manifestations in a large hereditary hemorrhagic telangiectasia (HHT) type 2 kindred. American Journal of Medical Genetics Part A, 2000, 93, 320-327.	2.4	95
74	Two common endoglin mutations in families with hereditary hemorrhagic telangiectasia in the Netherlands Antilles: evidence for a founder effect. Human Genetics, 2000, 107, 40-44.	3.8	6
75	Expression analysis of endoglin missense and truncation mutations: insights into protein structure and disease mechanisms. Human Molecular Genetics, 2000, 9, 745-755.	2.9	59
76	Endoglin, an Ancillary TGFÎ² Receptor, Is Required for Extraembryonic Angiogenesis and Plays a Key Role in Heart Development. Developmental Biology, 2000, 217, 42-53.	2.0	418
77	Two common endoglin mutations in families with hereditary hemorrhagic telangiectasia in the Netherlands Antilles: evidence for a founder effect. Human Genetics, 2000, 107, 40-44.	3.8	38
78	Expression Analysis of Four Endoglin Missense Mutations Suggests That Haploinsufficiency Is the Predominant Mechanism for Hereditary Hemorrhagic Telangiectasia Type 1. Human Molecular Genetics, 1999, 8, 2171-2181.	2.9	113
79	Mutations in the Gene Encoding KRIT1, a Krev-1/rap1a Binding Protein, Cause Cerebral Cavernous Malformations (CCM1). Human Molecular Genetics, 1999, 8, 2325-2333.	2.9	373
80	Assignment of Transforming Growth Factor Î²1 and Î²3 and a Third New Ligand to the Type I Receptor ALK-1. Journal of Biological Chemistry, 1999, 274, 9984-9992.	3.4	132
81	Genetic mapping of a novel familial form of infantile hemangioma. American Journal of Medical Genetics Part A, 1999, 82, 77-83.	2.4	141
82	Report on the workshop on hereditary hemorrhagic telangiectasis, July 10-11, 1997. , 1998, 76, 269-273.		41
83	Mutation and expression analysis of the endoglin gene in hereditary hemorrhagic telangiectasia reveals null alleles. Human Mutation, 1998, 11, 286-294.	2.5	104
84	Novel missense and frameshift mutations in the activin receptor-like kinase-1 gene in hereditary hemorrhagic telangiectasia. Human Mutation, 1998, 12, 137-137.	2.5	25
85	Cloning of the Promoter Region of Human Endoglin, the Target Gene for Hereditary Hemorrhagic Telangiectasia Type 1. Blood, 1998, 92, 4677-4690.	1.4	98
86	Cloning of the Promoter Region of Human Endoglin, the Target Gene for Hereditary Hemorrhagic Telangiectasia Type 1. Blood, 1998, 92, 4677-4690.	1.4	5
87	The Activin Receptor-Like Kinase 1 Gene: Genomic Structure and Mutations in Hereditary Hemorrhagic Telangiectasia Type 2. American Journal of Human Genetics, 1997, 61, 60-67.	6.2	225
88	Endoglin gene polymorphism as a risk factor for sporadic intracerebral hemorrhage. Annals of Neurology, 1997, 41, 683-686.	5.3	52
89	Mutations in the activin receptorâ€“like kinase 1 gene in hereditary haemorrhagic telangiectasia type 2. Nature Genetics, 1996, 13, 189-195.	21.4	1,031
90	Clinical heterogeneity in hereditary haemorrhagic telangiectasia: are pulmonary arteriovenous malformations more common in families linked to endoglin?. Journal of Medical Genetics, 1996, 33, 256-257.	3.2	106

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91	Six novel mutations in the endoglin gene in hereditary hemorrhagic telangiectasia type 1 suggest a dominant-negative effect of receptor function. Human Molecular Genetics, 1995, 4, 1983-1985.	2.9	110
92	Hereditary Hemorrhagic Telangiectasia. New England Journal of Medicine, 1995, 333, 918-924.	27.0	986
93	A second locus for hereditary hemorrhagic telangiectasia maps to chromosome 12.. Genome Research, 1995, 5, 21-28.	5.5	143
94	Genetic heterogeneity in hereditary haemorrhagic telangiectasia: possible correlation with clinical phenotype.. Journal of Medical Genetics, 1994, 31, 927-932.	3.2	80
95	Circulating Plasma miRNA Homologs in Mice and Humans Reflect Familial Cerebral Cavernous Malformation Disease. Translational Stroke Research, 0, , .	4.2	0