Douglas A Marchuk

List of Publications by Year in Descending Order

92 papers	8,752 citations	45 h-index	93 g-index
106 ext. papers	9,891 ext. citations	10.1 avg, IF	5.39 L-index

#	Paper	IF	Citations
92	Developmental venous anomalies are a genetic primer for cerebral cavernous malformations. **2022**, 1, 246-252		2
91	Propranolol as therapy for cerebral cavernous malformations: a cautionary note.. *Journal of Translational Medicine*, **2022**, 20, 160	8.5	1
90	Rapamycin in Cerebral Cavernous Malformations: What Doses to Test in Mice and Humans.. *ACS Pharmacology and Translational Science*, **2022**, 5, 266-277	5.9	0
89	Genetic genealogy uncovers a founder deletion mutation in the cerebral cavernous malformations 2 gene.. *Human Genetics*, **2022**, 1	6.3	
88	PIK3CA and CCM mutations fuel cavernomas through a cancer-like mechanism. *Nature*, **2021**, 594, 271-276	50.4	22
87	Cerebral Cavernous Malformation: From Mechanism to Therapy. *Circulation Research*, **2021**, 129, 195-215	15.7	12
86	Propranolol inhibits cavernous vascular malformations by 1 adrenergic receptor antagonism in animal models. *Journal of Clinical Investigation*, **2021**, 131,	15.9	11
85	A Neuroprotective Locus Modulates Ischemic Stroke Infarction Independent of Collateral Vessel Anatomy. *Frontiers in Neuroscience*, **2021**, 15, 705160	5.1	0
84	Pilot investigation of circulating angiogenic and inflammatory biomarkers associated with vascular malformations. *Orphanet Journal of Rare Diseases*, **2021**, 16, 372	4.2	2
83	Predictors of mortality in patients with hereditary hemorrhagic telangiectasia. *Orphanet Journal of Rare Diseases*, **2021**, 16, 12	4.2	4
82	Novel Murine Models of Cerebral Cavernous Malformations. *Angiogenesis*, **2020**, 23, 651-666	10.6	18
81	Cerebral cavernous malformations are driven by ADAMTS5 proteolysis of versican. *Journal of Experimental Medicine*, **2020**, 217,	16.6	13
80	Hereditary Hemorrhagic Telangiectasia (Osler Weber Rendu Syndrome) **2020**, 115-140		1
79	A Brain-Targeted Orally Available ROCK2 Inhibitor Benefits Mild and Aggressive Cavernous Angioma Disease. *Translational Stroke Research*, **2020**, 11, 365-376	7.8	9
78	Transcriptome clarifies mechanisms of lesion genesis versus progression in models of Ccm3 cerebral cavernous malformations. *Acta Neuropathologica Communications*, **2019**, 7, 132	7.3	11
77	Rho Kinase Inhibition Blunts Lesion Development and Hemorrhage in Murine Models of Aggressive Pdcd10/Ccm3 Disease. *Stroke*, **2019**, 50, 738-744	6.7	23
76	Phenotypic characterization of murine models of cerebral cavernous malformations. *Laboratory Investigation*, **2019**, 99, 319-330	5.9	18

75	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	11	39
74	Comprehensive transcriptome analysis of cerebral cavernous malformation across multiple species and genotypes. *JCI Insight*, **2019**, 4,	9.9	25
73	Biomarkers of cavernous angioma with symptomatic hemorrhage. *JCI Insight*, **2019**, 4,	9.9	15
72	Novel Neuroprotective Loci Modulating Ischemic Stroke Volume in Wild-Derived Inbred Mouse Strains. *Genetics*, **2019**, 213, 1079-1092	4	2
71	Physical and Family History Variables Associated With Neurological and Cognitive Development in Sturge-Weber Syndrome. *Pediatric Neurology*, **2019**, 96, 30-36	2.9	18
70	Distinct cellular roles for PDCD10 define a gut-brain axis in cerebral cavernous malformation. *Science Translational Medicine*, **2019**, 11,	17.5	25
69	Cerebral Cavernous Malformations Develop Through Clonal Expansion of Mutant Endothelial Cells. *Circulation Research*, **2018**, 123, 1143-1151	15.7	49
68	Neuronal IL-4R modulates neuronal apoptosis and cell viability during the acute phases of cerebral ischemia. *FEBS Journal*, **2018**, 285, 2785-2798	5.7	10
67	BAG3 (Bcl-2-Associated Athanogene-3) Coding Variant in Mice Determines Susceptibility to Ischemic Limb Muscle Myopathy by Directing Autophagy. *Circulation*, **2017**, 136, 281-296	16.7	31
66	The pathobiology of vascular malformations: insights from human and model organism genetics. *Journal of Pathology*, **2017**, 241, 281-293	9.4	46
65	RhoA Kinase Inhibition With Fasudil Versus Simvastatin in Murine Models of Cerebral Cavernous Malformations. *Stroke*, **2017**, 48, 187-194	6.7	63
64	Micro-computed tomography in murine models of cerebral cavernous malformations as a paradigm for brain disease. *Journal of Neuroscience Methods*, **2016**, 271, 14-24	3	21
63	Natural allelic variation of the IL-21 receptor modulates ischemic stroke infarct volume. *Journal of Clinical Investigation*, **2016**, 126, 2827-38	15.9	15
62	B-Cell Depletion Reduces the Maturation of Cerebral Cavernous Malformations in Murine Models. *Journal of NeuroImmune Pharmacology*, **2016**, 11, 369-77	6.9	28
61	ADAM12: a genetic modifier of preclinical peripheral arterial disease. *American Journal of Physiology - Heart and Circulatory Physiology*, **2015**, 309, H790-803	5.2	22
60	Exceptional aggressiveness of cerebral cavernous malformation disease associated with PDCD10 mutations. *Genetics in Medicine*, **2015**, 17, 188-196	8.1	97
59	Sturge Weber Syndrome **2015**, 945-953		
58	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, 4357-70	5.6	93

57	Hereditary Hemorrhagic Telangiectasia (Osler Weber Rendu Syndrome) **2013**, 1-18		1
56	Sturge-Weber syndrome and port-wine stains caused by somatic mutation in GNAQ. *New England Journal of Medicine*, **2013**, 368, 1971-9	59.2	672
55	A novel genetic locus modulates infarct volume independently of the extent of collateral circulation. *Physiological Genomics*, **2013**, 45, 751-63	3.6	21
54	Natural genetic variation of integrin alpha L (Itgal) modulates ischemic brain injury in stroke. *PLoS Genetics*, **2013**, 9, e1003807	6	20
53	Brain Vascular Malformation Consortium: Overview, Progress and Future Directions **2013**, 1, 5		20
52	Skeletal muscle-specific genetic determinants contribute to the differential strain-dependent effects of hindlimb ischemia in mice. *American Journal of Pathology*, **2012**, 180, 2156-69	5.8	46
51	Fasudil decreases lesion burden in a murine model of cerebral cavernous malformation disease. *Stroke*, **2012**, 43, 571-4	6.7	105
50	A novel mouse model of cerebral cavernous malformations based on the two-hit mutation hypothesis recapitulates the human disease. *Human Molecular Genetics*, **2011**, 20, 211-22	5.6	98
49	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, 662-6	8.1	11
48	Overlapping spectra of SMAD4 mutations in juvenile polyposis (JP) and JP-HHT syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 333-9	2.5	99
47	Biallelic somatic and germline mutations in cerebral cavernous malformations (CCMs): evidence for a two-hit mechanism of CCM pathogenesis. *Human Molecular Genetics*, **2009**, 18, 919-30	5.6	203
46	A locus mapping to mouse chromosome 7 determines infarct volume in a mouse model of ischemic stroke. *Circulation: Cardiovascular Genetics*, **2009**, 2, 591-8		46
45	The cerebral cavernous malformation signaling pathway promotes vascular integrity via Rho GTPases. *Nature Medicine*, **2009**, 15, 177-84	50.5	280
44	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, 1207-15	16.7	85
43	Genetic analysis of a family with hereditary glomuvenous malformations. *Australasian Journal of Dermatology*, **2007**, 48, 170-3	1.3	7
42	Deletions in CCM2 are a common cause of cerebral cavernous malformations. *American Journal of Human Genetics*, **2007**, 80, 69-75	11	64
41	Redefining heart failure: the utility of genomics. *Journal of the American College of Cardiology*, **2006**, 48, 1289-98	15.1	46
40	SMAD4 mutations found in unselected HHT patients. *Journal of Medical Genetics*, **2006**, 43, 793-7	5.8	164

39	Neuronal expression of the Ccm2 gene in a new mouse model of cerebral cavernous malformations. *Mammalian Genome*, **2006**, 17, 119-28	3.2	59
38	Molecular classification of patients with unexplained hamartomatous and hyperplastic polyposis. *JAMA - Journal of the American Medical Association*, **2005**, 294, 2465-73	27.4	189
37	Genetics of cerebral cavernous malformations. *Current Neurology and Neuroscience Reports*, **2005**, 5, 391-6	6.6	53
36	CCM1 and CCM2 protein interactions in cell signaling: implications for cerebral cavernous malformations pathogenesis. *Human Molecular Genetics*, **2005**, 14, 2521-31	5.6	202
35	Human retroviral gag- and gag-pol-like proteins interact with the transforming growth factor-beta receptor activin receptor-like kinase 1. *Journal of Biological Chemistry*, **2005**, 280, 8482-93	5.4	45
34	The Molecular Basis of Cerebrovascular Malformations **2005**, 437-450		
33	Ccm1 is required for arterial morphogenesis: implications for the etiology of human cavernous malformations. *Development (Cambridge)*, **2004**, 131, 1437-48	6.6	166
32	Loss of p53 sensitizes mice with a mutation in Ccm1 (KRIT1) to development of cerebral vascular malformations. *American Journal of Pathology*, **2004**, 165, 1509-18	5.8	92
31	A combined syndrome of juvenile polyposis and hereditary haemorrhagic telangiectasia associated with mutations in MADH4 (SMAD4). *Lancet, The*, **2004**, 363, 852-9	40	569
30	Polymorphisms in genes involved in inflammatory and angiogenic pathways and the risk of hemorrhagic presentation of brain arteriovenous malformations. *Stroke*, **2004**, 35, 2294-300	6.7	110
29	A mouse model for hereditary hemorrhagic telangiectasia (HHT) type 2. *Human Molecular Genetics*, **2003**, 12, 473-82	5.6	149
28	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, 1459-64	11	279
27	Vascular morphogenesis: tales of two syndromes. *Human Molecular Genetics*, **2003**, 12 Spec No 1, R97-112	5.6	119
26	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, 389-96	5.6	161
25	Fine mapping and genetic heterogeneity in the pure form of autosomal dominant familial spastic paraplegia. *Neurogenetics*, **2001**, 3, 91-7	3	11
24	Clinical manifestations in a large hereditary hemorrhagic telangiectasia (HHT) type 2 kindred. *American Journal of Medical Genetics Part A*, **2000**, 93, 320-7		79
23	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	6.3	5
22	Expression analysis of endoglin missense and truncation mutations: insights into protein structure and disease mechanisms. *Human Molecular Genetics*, **2000**, 9, 745-55	5.6	48

21	Endoglin, an ancillary TGFbeta receptor, is required for extraembryonic angiogenesis and plays a key role in heart development. *Developmental Biology*, **2000**, 217, 42-53	3.1	382
20	Two common endoglin mutations in families with hereditary hemorrhagic telangiectasia in the Netherlands Antilles: evidence for a founder effect. *Human Genetics*, **2000**, 107, 40-4	6.3	34
19	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, 2171-81	5.6	102
18	Mutations in the gene encoding KRIT1, a Krev-1/rap1a binding protein, cause cerebral cavernous malformations (CCM1). *Human Molecular Genetics*, **1999**, 8, 2325-33	5.6	327
17	Assignment of transforming growth factor beta1 and beta3 and a third new ligand to the type I receptor ALK-1. *Journal of Biological Chemistry*, **1999**, 274, 9984-92	5.4	109
16	Genetic mapping of a novel familial form of infantile hemangioma. *American Journal of Medical Genetics Part A*, **1999**, 82, 77-83		118
15	Report on the workshop on hereditary hemorrhagic telangiectasis, July 10-11, 1997 **1998**, 76, 269-273		39
14	Mutation and expression analysis of the endoglin gene in hereditary hemorrhagic telangiectasia reveals null alleles. *Human Mutation*, **1998**, 11, 286-94	4.7	87
13	Novel missense and frameshift mutations in the activin receptor-like kinase-1 gene in hereditary hemorrhagic telangiectasia. *Human Mutation*, **1998**, 12, 137-137	4.7	18
12	Cloning of the Promoter Region of Human Endoglin, the Target Gene for Hereditary Hemorrhagic Telangiectasia Type 1. *Blood*, **1998**, 92, 4677-4690	2.2	90
11	Cloning of the Promoter Region of Human Endoglin, the Target Gene for Hereditary Hemorrhagic Telangiectasia Type 1. *Blood*, **1998**, 92, 4677-4690	2.2	5
10	The activin receptor-like kinase 1 gene: genomic structure and mutations in hereditary hemorrhagic telangiectasia type 2. *American Journal of Human Genetics*, **1997**, 61, 60-7	11	199
9	Endoglin gene polymorphism as a risk factor for sporadic intracerebral hemorrhage. *Annals of Neurology*, **1997**, 41, 683-6	9.4	45
8	Mutations in the activin receptor-like kinase 1 gene in hereditary haemorrhagic telangiectasia type 2. *Nature Genetics*, **1996**, 13, 189-95	36.3	904
7	Clinical heterogeneity in hereditary haemorrhagic telangiectasia: are pulmonary arteriovenous malformations more common in families linked to endoglin?. *Journal of Medical Genetics*, **1996**, 33, 256-7	5.8	92
6	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, 1983-5	5.6	93
5	Hereditary hemorrhagic telangiectasia. *New England Journal of Medicine*, **1995**, 333, 918-24	59.2	841
4	A second locus for hereditary hemorrhagic telangiectasia maps to chromosome 12. *Genome Research*, **1995**, 5, 21-8	9.7	114

3	Genetic heterogeneity in hereditary haemorrhagic telangiectasia: possible correlation with clinical phenotype. *Journal of Medical Genetics*, **1994**, 31, 927-32	5.8	65
2	Novel hemorrhage models of cerebral cavernous malformations		2
1	Somatic Mutations in Vascular Malformations of Hereditary Hemorrhagic Telangiectasia Result in Biallelic Loss of ENG or ACVRL1		1