

Douglas A Marchuk

List of Publications by Year in descending order

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95
papers

10,788
citations

47409

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60403

85
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106
all docs

106
docs citations

106
times ranked

7128
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.	1.8	5
3	Rapamycin in Cerebral Cavernous Malformations: What Doses to Test in Mice and Humans. ACS Pharmacology and Translational Science, 2022, 5, 266-277.	2.5	5
4	Genetic genealogy uncovers a founder deletion mutation in the cerebral cavernous malformations 2 gene. Human Genetics, 2022, , 1.	1.8	0
5	A novel somatic mutation in GNAQ in a capillary malformation provides insight into molecular pathogenesis. Angiogenesis, 2022, 25, 493-502.	3.7	8
6	Propranolol inhibits cavernous vascular malformations by β_1 adrenergic receptor antagonism in animal models. Journal of Clinical Investigation, 2021, 131, .	3.9	28
7	Abstract MP11: Circulating Plasma Biomarkers Associated With Brain Arteriovenous Malformations. Stroke, 2021, 52, .	1.0	0
8	PIK3CA and CCM mutations fuel cavernomas through a cancer-like mechanism. Nature, 2021, 594, 271-276.	13.7	103
9	Cerebral Cavernous Malformation: From Mechanism to Therapy. Circulation Research, 2021, 129, 195-215.	2.0	82
10	A Neuroprotective Locus Modulates Ischemic Stroke Infarction Independent of Collateral Vessel Anatomy. Frontiers in Neuroscience, 2021, 15, 705160.	1.4	4
11	Pilot investigation of circulating angiogenic and inflammatory biomarkers associated with vascular malformations. Orphanet Journal of Rare Diseases, 2021, 16, 372.	1.2	8
12	Predictors of mortality in patients with hereditary hemorrhagic telangiectasia. Orphanet Journal of Rare Diseases, 2021, 16, 12.	1.2	12
13	Hereditary Hemorrhagic Telangiectasia (Osler-Weber-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.	2.3	22
15	Novel Murine Models of Cerebral Cavernous Malformations. Angiogenesis, 2020, 23, 651-666.	3.7	36
16	Cerebral cavernous malformations are driven by ADAMTS5 proteolysis of versican. Journal of Experimental Medicine, 2020, 217, .	4.2	30
17	Phenotypic characterization of murine models of cerebral cavernous malformations. Laboratory Investigation, 2019, 99, 319-330.	1.7	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.	2.6	77

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19	Transcriptome clarifies mechanisms of lesion genesis versus progression in models of Ccm3 cerebral cavernous malformations. <i>Acta Neuropathologica Communications</i> , 2019, 7, 132.	2.4	27
20	Rho Kinase Inhibition Blunts Lesion Development and Hemorrhage in Murine Models of Aggressive <i><i>Pcd10/Ccm3</i></i> Disease. <i>Stroke</i> , 2019, 50, 738-744.	1.0	40
21	Physical and Family History Variables Associated With Neurological and Cognitive Development in Sturge-Weber Syndrome. <i>Pediatric Neurology</i> , 2019, 96, 30-36.	1.0	32
22	Distinct cellular roles for PDCD10 define a gut-brain axis in cerebral cavernous malformation. <i>Science Translational Medicine</i> , 2019, 11, .	5.8	51
23	Comprehensive transcriptome analysis of cerebral cavernous malformation across multiple species and genotypes. <i>JCI Insight</i> , 2019, 4, .	2.3	40
24	Biomarkers of cavernous angioma with symptomatic hemorrhage. <i>JCI Insight</i> , 2019, 4, .	2.3	25
25	Novel Neuroprotective Loci Modulating Ischemic Stroke Volume in Wild-Derived Inbred Mouse Strains. <i>Genetics</i> , 2019, 213, 1079-1092.	1.2	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. <i>Stroke</i> , 2019, 50, .	1.0	0
27	Cerebral Cavernous Malformations Develop Through Clonal Expansion of Mutant Endothelial Cells. <i>Circulation Research</i> , 2018, 123, 1143-1151.	2.0	83
28	Neuronal <i><sc>IL</sc></i> modulates neuronal apoptosis and cell viability during the acute phases of cerebral ischemia. <i>FEBS Journal</i> , 2018, 285, 2785-2798.	2.2	18
29	BAG3 (Bcl-2 Associated Athanogene-3) Coding Variant in Mice Determines Susceptibility to Ischemic Limb Muscle Myopathy by Directing Autophagy. <i>Circulation</i> , 2017, 136, 281-296.	1.6	51
30	The pathobiology of vascular malformations: insights from human and model organism genetics. <i>Journal of Pathology</i> , 2017, 241, 281-293.	2.1	64
31	RhoA Kinase Inhibition With Fasudil Versus Simvastatin in Murine Models of Cerebral Cavernous Malformations. <i>Stroke</i> , 2017, 48, 187-194.	1.0	86
32	Natural allelic variation of the IL-21 receptor modulates ischemic stroke infarct volume. <i>Journal of Clinical Investigation</i> , 2016, 126, 2827-2838.	3.9	25
33	B-Cell Depletion Reduces the Maturation of Cerebral Cavernous Malformations in Murine Models. <i>Journal of NeuroImmune Pharmacology</i> , 2016, 11, 369-377.	2.1	39
34	Micro-computed tomography in murine models of cerebral cavernous malformations as a paradigm for brain disease. <i>Journal of Neuroscience Methods</i> , 2016, 271, 14-24.	1.3	25
35	Sturge-Weber Syndrome. , 2015, , 945-953.		0
36	ADAM12: a genetic modifier of preclinical peripheral arterial disease. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , 2015, 309, H790-H803.	1.5	34

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37	Exceptional aggressiveness of cerebral cavernous malformation disease associated with PDCD10 mutations. <i>Genetics in Medicine</i> , 2015, 17, 188-196.	1.1	116
38	Abstract T P231: B- Cell Depletion Decreases Lesion Burden in a Murine Model of Cerebral Cavernous Malformation Disease. <i>Stroke</i> , 2015, 46, .	1.0	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. <i>Human Molecular Genetics</i> , 2014, 23, 4357-4370.	1.4	117
40	Hereditary Hemorrhagic Telangiectasia (Osler-Weber-Rendu Syndrome). , 2013, , 1-18.		2
41	Sturge-Weber Syndrome and Port-Wine Stains Caused by Somatic Mutation in <i>GNAQ</i> . <i>New England Journal of Medicine</i> , 2013, 368, 1971-1979.	13.9	865
42	A novel genetic locus modulates infarct volume independently of the extent of collateral circulation. <i>Physiological Genomics</i> , 2013, 45, 751-763.	1.0	27
43	Natural Genetic Variation of Integrin Alpha L (<i>Itgal</i>) Modulates Ischemic Brain Injury in Stroke. <i>PLoS Genetics</i> , 2013, 9, e1003807.	1.5	26
44	Brain Vascular Malformation Consortium: Overview, Progress and Future Directions. <i>The Journal of Rare Disorders</i> , 2013, 1, 5.	1.5	21
45	Fasudil Decreases Lesion Burden in a Murine Model of Cerebral Cavernous Malformation Disease. <i>Stroke</i> , 2012, 43, 571-574.	1.0	130
46	Skeletal Muscle-Specific Genetic Determinants Contribute to the Differential Strain-Dependent Effects of Hindlimb Ischemia in Mice. <i>American Journal of Pathology</i> , 2012, 180, 2156-2169.	1.9	66
47	A novel mouse model of cerebral cavernous malformations based on the two-hit mutation hypothesis recapitulates the human disease. <i>Human Molecular Genetics</i> , 2011, 20, 211-222.	1.4	120
48	A founder mutation in the Ashkenazi Jewish population affecting messenger RNA splicing of the CCM2 gene causes cerebral cavernous malformations. <i>Genetics in Medicine</i> , 2011, 13, 662-666.	1.1	12
49	Overlapping spectra of <i>SMAD4</i> mutations in juvenile polyposis (JP) and JP-HHT syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 333-339.	0.7	128
50	Biallelic somatic and germline mutations in cerebral cavernous malformations (CCMs): evidence for a two-hit mechanism of CCM pathogenesis. <i>Human Molecular Genetics</i> , 2009, 18, 919-930.	1.4	244
51	A Locus Mapping to Mouse Chromosome 7 Determines Infarct Volume in a Mouse Model of Ischemic Stroke. <i>Circulation: Cardiovascular Genetics</i> , 2009, 2, 591-598.	5.1	54
52	The cerebral cavernous malformation signaling pathway promotes vascular integrity via Rho GTPases. <i>Nature Medicine</i> , 2009, 15, 177-184.	15.2	340
53	A Quantitative Trait Locus (LSq-1) on Mouse Chromosome 7 Is Linked to the Absence of Tissue Loss After Surgical Hindlimb Ischemia. <i>Circulation</i> , 2008, 117, 1207-1215.	1.6	105
54	Deletions in CCM2 Are a Common Cause of Cerebral Cavernous Malformations. <i>American Journal of Human Genetics</i> , 2007, 80, 69-75.	2.6	80

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55	Genetic analysis of a family with hereditary glomuvenous malformations. <i>Australasian Journal of Dermatology</i> , 2007, 48, 170-173.	0.4	8
56	Redefining Heart Failure. <i>Journal of the American College of Cardiology</i> , 2006, 48, 1289-1298.	1.2	50
57	SMAD4 mutations found in unselected HHT patients. <i>Journal of Medical Genetics</i> , 2006, 43, 793-797.	1.5	212
58	Neuronal expression of the Ccm2 gene in a new mouse model of cerebral cavernous malformations. <i>Mammalian Genome</i> , 2006, 17, 119-128.	1.0	68
59	Genetics of cerebral cavernous malformations. <i>Current Neurology and Neuroscience Reports</i> , 2005, 5, 391-396.	2.0	62
60	CCM1 and CCM2 protein interactions in cell signaling: implications for cerebral cavernous malformations pathogenesis. <i>Human Molecular Genetics</i> , 2005, 14, 2521-2531.	1.4	238
61	Human Retroviral gag- and gag-pol-like Proteins Interact with the Transforming Growth Factor- β 2 Receptor Activin Receptor-like Kinase 1. <i>Journal of Biological Chemistry</i> , 2005, 280, 8482-8493.	1.6	54
62	Molecular Classification of Patients With Unexplained Hamartomatous and Hyperplastic Polyposis. <i>JAMA - Journal of the American Medical Association</i> , 2005, 294, 2465.	3.8	218
63	The Molecular Basis of Cerebrovascular Malformations. , 2005, , 437-450.		0
64	Ccm1 is required for arterial morphogenesis: implications for the etiology of human cavernous malformations. <i>Development (Cambridge)</i> , 2004, 131, 1437-1448.	1.2	180
65	Loss of p53 Sensitizes Mice with a Mutation in Ccm1 (KRIT1) to Development of Cerebral Vascular Malformations. <i>American Journal of Pathology</i> , 2004, 165, 1509-1518.	1.9	110
66	A combined syndrome of juvenile polyposis and hereditary haemorrhagic telangiectasia associated with mutations in MADH4 (SMAD4). <i>Lancet, The</i> , 2004, 363, 852-859.	6.3	667
67	Polymorphisms in Genes Involved in Inflammatory and Angiogenic Pathways and the Risk of Hemorrhagic Presentation of Brain Arteriovenous Malformations. <i>Stroke</i> , 2004, 35, 2294-2300.	1.0	134
68	Mutations in a Gene Encoding a Novel Protein Containing a Phosphotyrosine-Binding Domain Cause Type 2 Cerebral Cavernous Malformations. <i>American Journal of Human Genetics</i> , 2003, 73, 1459-1464.	2.6	319
69	Vascular morphogenesis: tales of two syndromes. <i>Human Molecular Genetics</i> , 2003, 12, 97R-112.	1.4	134
70	A mouse model for hereditary hemorrhagic telangiectasia (HHT) type 2. <i>Human Molecular Genetics</i> , 2003, 12, 473-482.	1.4	172
71	KRIT1 association with the integrin-binding protein ICAP-1: a new direction in the elucidation of cerebral cavernous malformations (CCM1) pathogenesis. <i>Human Molecular Genetics</i> , 2002, 11, 389-396.	1.4	176
72	Fine mapping and genetic heterogeneity in the pure form of autosomal dominant familial spastic paraplegia. <i>Neurogenetics</i> , 2001, 3, 91-97.	0.7	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.	1.8	6
75	Expression analysis of endoglin missense and truncation mutations: insights into protein structure and disease mechanisms. Human Molecular Genetics, 2000, 9, 745-755.	1.4	59
76	Endoglin, an Ancillary TGF β Receptor, Is Required for Extraembryonic Angiogenesis and Plays a Key Role in Heart Development. Developmental Biology, 2000, 217, 42-53.	0.9	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.	1.8	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.	1.4	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.	1.4	373
80	Assignment of Transforming Growth Factor β 1 and β 3 and a Third New Ligand to the Type I Receptor ALK-1. Journal of Biological Chemistry, 1999, 274, 9984-9992.	1.6	132
81	Genetic mapping of a novel familial form of infantile hemangioma. , 1999, 82, 77-83.		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.	1.1	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.	1.1	25
85	Cloning of the Promoter Region of Human Endoglin, the Target Gene for Hereditary Hemorrhagic Telangiectasia Type 1. Blood, 1998, 92, 4677-4690.	0.6	98
86	Cloning of the Promoter Region of Human Endoglin, the Target Gene for Hereditary Hemorrhagic Telangiectasia Type 1. Blood, 1998, 92, 4677-4690.	0.6	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.	2.6	225
88	Endoglin gene polymorphism as a risk factor for sporadic intracerebral hemorrhage. Annals of Neurology, 1997, 41, 683-686.	2.8	52
89	Mutations in the activin receptor-like kinase 1 gene in hereditary haemorrhagic telangiectasia type 2. Nature Genetics, 1996, 13, 189-195.	9.4	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.	1.5	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. <i>Human Molecular Genetics</i> , 1995, 4, 1983-1985.	1.4	110
92	Hereditary Hemorrhagic Telangiectasia. <i>New England Journal of Medicine</i> , 1995, 333, 918-924.	13.9	986
93	A second locus for hereditary hemorrhagic telangiectasia maps to chromosome 12.. <i>Genome Research</i> , 1995, 5, 21-28.	2.4	143
94	Genetic heterogeneity in hereditary haemorrhagic telangiectasia: possible correlation with clinical phenotype.. <i>Journal of Medical Genetics</i> , 1994, 31, 927-932.	1.5	80
95	Circulating Plasma miRNA Homologs in Mice and Humans Reflect Familial Cerebral Cavernous Malformation Disease. <i>Translational Stroke Research</i> , 0, , .	2.3	0