Rajiv D Machado

List of Publications by Year
in descending order

46	9,333	136885	233338
papers	citations	32 h-index	45 g-index

51	51	51	6713
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Heterozygous germline mutations in BMPR2, encoding a TGF-Î² receptor, cause familial primary pulmonary hypertension. Nature Genetics, 2000, 26, 81-84.	9.4	1,388
2	Clinical and Molecular Genetic Features of Pulmonary Hypertension in Patients with Hereditary Hemorrhagic Telangiectasia. New England Journal of Medicine, 2001, 345, 325-334.	13.9	676
3	Elevated Levels of Inflammatory Cytokines Predict Survival in Idiopathic and Familial Pulmonary Arterial Hypertension. Circulation, 2010, 122, 920-927.	1.6	661
4	Sporadic primary pulmonary hypertension is associated with germline mutations of the gene encoding BMPR-II, a receptor member of the TGF-beta family. Journal of Medical Genetics, 2000, 37, 741-745.	1.5	645
5	Primary Pulmonary Hypertension Is Associated With Reduced Pulmonary Vascular Expression of Type II Bone Morphogenetic Protein Receptor. Circulation, 2002, 105, 1672-1678.	1.6	587
6	BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. American Journal of Human Genetics, 2001, 68, 92-102.	2.6	521
7	Selective enhancement of endothelial BMPR-II with BMP9 reverses pulmonary arterial hypertension. Nature Medicine, 2015, 21, 777-785.	15.2	389
8	Mutations of the TGF-Î² type II receptorBMPR2 in pulmonary arterial hypertension. Human Mutation, 2006, 27, 121-132.	1.1	368
9	Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2013, 62, D13-D21.	1.2	367
10	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	2.6	343
11	Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2009, 54, S32-S42.	1.2	342
12	Molecular and functional analysis identifies ALK-1 as the predominant cause of pulmonary hypertension related to hereditary haemorrhagic telangiectasia. Journal of Medical Genetics, 2003, 40, 865-871.	1.5	309
13	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. Nature Communications, 2018, 9, 1416.	5.8	279
14	Functional analysis of bone morphogenetic protein type II receptor mutations underlying primary pulmonary hypertension. Human Molecular Genetics, 2002, 11, 1517-1525.	1.4	231
15	Stress Doppler Echocardiography in Relatives of Patients With Idiopathic and Familial Pulmonary Arterial Hypertension. Circulation, 2009, 119, 1747-1757.	1.6	205
16	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	1.1	185
17	Molecular genetic framework underlying pulmonary arterial hypertension. Nature Reviews Cardiology, 2020, 17, 85-95.	6.1	181
18	Mutations in FRMD7, a newly identified member of the FERM family, cause X-linked idiopathic congenital nystagmus. Nature Genetics, 2006, 38, 1242-1244.	9.4	180

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19	Molecular genetic characterization of SMAD signaling molecules in pulmonary arterial hypertension. Human Mutation, 2011, 32, 1385-1389.	1.1	152
20	Genetic determinants of risk in pulmonary arterial hypertension: international genome-wide association studies and meta-analysis. Lancet Respiratory Medicine,the, 2019, 7, 227-238.	5.2	122
21	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
22	Functional interaction between BMPR-II and Tctex-1, a light chain of Dynein, is isoform-specific and disrupted by mutations underlying primary pulmonary hypertension. Human Molecular Genetics, 2003, 12, 3277-3286.	1.4	110
23	Gain-of-Function Mutations of ARHGAP31, a Cdc42/Rac1 GTPase Regulator, Cause Syndromic Cutis Aplasia and Limb Anomalies. American Journal of Human Genetics, 2011, 88, 574-585.	2.6	100
24	Impaired Natural Killer Cell Phenotype and Function in Idiopathic and Heritable Pulmonary Arterial Hypertension. Circulation, 2012, 126, 1099-1109.	1.6	99
25	Investigation of Second Genetic Hits at the BMPR2 Locus as a Modulator of Disease Progression in Familial Pulmonary Arterial Hypertension. Circulation, 2005, 111, 607-613.	1.6	88
26	Genetic Association of the Serotonin Transporter in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2006, 173, 793-797.	2.5	88
27	Haploinsufficiency of the NOTCH1 Receptor as a Cause of Adamsâ€“Oliver Syndrome With Variable Cardiac Anomalies. Circulation: Cardiovascular Genetics, 2015, 8, 572-581.	5.1	84
28	Characterization of GDF2 Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2020, 201, 575-585.	2.5	80
29	Pulmonary arterial hypertension and type-I glycogen-storage disease: the serotonin hypothesis. European Respiratory Journal, 2002, 20, 59-65.	3.1	47
30	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	2.6	46
31	Demographic features, BMPR2 status and outcomes in distal chronic thromboembolic pulmonary hypertension. Thorax, 2007, 62, 617-622.	2.7	43
32	Characterization of theBMPR25â€²-Untranslated Region and a Novel Mutation in Pulmonary Hypertension. American Journal of Respiratory and Critical Care Medicine, 2007, 176, 819-824.	2.5	39
33	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
34	BMPR2 mutations have short lifetime expectancy in primary pulmonary hypertension. Human Mutation, 2005, 26, 119-124.	1.1	30
35	CdGAP is required for transforming growth factor Î²- and Neu/ErbB-2-induced breast cancer cell motility and invasion. Oncogene, 2011, 30, 1032-1045.	2.6	29
36	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	1.6	29

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37	Dymeclin, the gene underlying Dyggve-Melchior-Clausen syndrome, encodes a protein integral to extracellular matrix and golgi organization and is associated with protein secretion pathways critical in bone development. Human Mutation, 2011, 32, 231-239.	1.1	26
38	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	2.6	26
39	A Physical and Transcript Map Based upon Refinement of the Critical Interval for PPH1, a Gene for Familial Primary Pulmonary Hypertension. Genomics, 2000, 68, 220-228.	1.3	25
40	Biallelic variants of ATP13A3 cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. Journal of Medical Genetics, 2022, 59, 906-911.	1.5	22
41	Whole Exome Sequence Analysis Provides Novel Insights into the Genetic Framework of Childhood-Onset Pulmonary Arterial Hypertension. Genes, 2020, 11, 1328.	1.0	14
42	The Molecular Genetics and Cellular Mechanisms Underlying Pulmonary Arterial Hypertension. Scientifica, 2012, 2012, 1-17.	0.6	12
43	Seeking the right targets: gene therapy advances in pulmonary arterial hypertension. European Respiratory Journal, 2012, 39, 235-237.	3.1	7
44	Assessment of a Pulmonary Origin for Blood Outgrowth Endothelial Cells by Examination of Identical Twins Harboring aBMPR2Mutation. American Journal of Respiratory and Critical Care Medicine, 2013, 188, 258-260.	2.5	7
45	Response to Letter Regarding Article, â€œElevated Levels of Inflammatory Cytokines Predict Survival in Idiopathic and Familial Pulmonary Arterial Hypertensionâ€• Circulation, 2011, 123, .	1.6	1
46	Pulmonary Arterial Hypertension: A Deeper Evaluation of Genetic Risk in the -Omics Era. Genes, 2021, 12, 1798.	1.0	0