# 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 | 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 |
| 2 | Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, . | 1.6 | 29 |
| 3 | Pulmonary Arterial Hypertension: A Deeper Evaluation of Genetic Risk in the -Omics Era. Genes, 2021, 12, 1798. | 1.0 | 0 |
| 4 | Molecular genetic framework underlying pulmonary arterial hypertension. Nature Reviews Cardiology, 2020, 17, 85-95. | 6.1 | 181 |
| 5 | 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 |
| 6 | Whole Exome Sequence Analysis Provides Novel Insights into the Genetic Framework of Childhood-Onset Pulmonary Arterial Hypertension. Genes, 2020, 11, 1328. | 1.0 | 14 |
| 7 | 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 |
| 8 | Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. Nature Communications, 2018, 9, 1416. | 5.8 | 279 |
| 9 | De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153. | 2.6 | 36 |
| 10 | 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 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127. | 1.1 | 185 |
| 15 | 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 |
| 16 | Selective enhancement of endothelial BMPR-II with BMP9 reverses pulmonary arterial hypertension. Nature Medicine, 2015, 21, 777-785. | 15.2 | 389 |
| 17 | Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2013, 62, D13-D21. | 1.2 | 367 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Seeking the right targets: gene therapy advances in pulmonary arterial hypertension. European Respiratory Journal, 2012, 39, 235-237. | 3.1 | 7 |
| 20 | Impaired Natural Killer Cell Phenotype and Function in Idiopathic and Heritable Pulmonary Arterial Hypertension. Circulation, 2012, 126, 1099-1109. | 1.6 | 99 |
| 21 | The Molecular Genetics and Cellular Mechanisms Underlying Pulmonary Arterial Hypertension. Scientifica, 2012, 2012, 1-17. | 0.6 | 12 |
| 22 | 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 |
| 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 | 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 |
| 25 | Molecular genetic characterization of SMAD signaling molecules in pulmonary arterial hypertension. Human Mutation, 2011, 32, 1385-1389. | 1.1 | 152 |
| 26 | 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 |
| 27 | Elevated Levels of Inflammatory Cytokines Predict Survival in Idiopathic and Familial Pulmonary Arterial Hypertension. Circulation, 2010, 122, 920-927. | 1.6 | 661 |
| 28 | Stress Doppler Echocardiography in Relatives of Patients With Idiopathic and Familial Pulmonary Arterial Hypertension. Circulation, 2009, 119, 1747-1757. | 1.6 | 205 |
| 29 | Genetics and Genomics of Pulmonary Arterial Hypertension. Journal of the American College of Cardiology, 2009, 54, S32-S42. | 1.2 | 342 |
| 30 | Demographic features, BMPR2 status and outcomes in distal chronic thromboembolic pulmonary hypertension. Thorax, 2007, 62, 617-622. | 2.7 | 43 |
| 31 | 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 |
| 32 | 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 |
| 33 | Mutations of the TGF-Î² type II receptorBMPR2 in pulmonary arterial hypertension. Human Mutation, 2006, 27, 121-132. | 1.1 | 368 |
| 34 | 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 |
| 35 | BMPR2 mutations have short lifetime expectancy in primary pulmonary hypertension. Human Mutation, 2005, 26, 119-124. | 1.1 | 30 |
| 36 | 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 |

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|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | 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 |
| 40 | Pulmonary arterial hypertension and type-I glycogen-storage disease: the serotonin hypothesis. European Respiratory Journal, 2002, 20, 59-65. | 3.1 | 47 |
| 41 | Functional analysis of bone morphogenetic protein type II receptor mutations underlying primary pulmonary hypertension. Human Molecular Genetics, 2002, 11, 1517-1525. | 1.4 | 231 |
| 42 | BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. American Journal of Human Genetics, 2001, 68, 92-102. | 2.6 | 521 |
| 43 | 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 |
| 44 | 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 |
| 45 | 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 |
| 46 | 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 |