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List of Publications by Year in Descending Order

40 papers	2,073 citations	18 h-index	45 g-index
51 ext. papers	3,173 ext. citations	15.9 avg, IF	4.25 L-index

#	Paper	IF	Citations
40	Biological heterogeneity in idiopathic pulmonary arterial hypertension identified through unsupervised transcriptomic profiling of whole blood. *Nature Communications*, **2021**, 12, 7104	17.4	1
39	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
38	variants are more common than expected in the general population and associated with stroke and vascular dementia: an analysis of 200 000 participants. *Journal of Neurology, Neurosurgery and Psychiatry*, **2021**, 92, 694-701	5.5	13
37	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. *Genome Medicine*, **2021**, 13, 80	14.4	11
36	Severe Pulmonary Arterial Hypertension Is Characterized by Increased Neutrophil Elastase and Relative Elafin Deficiency. *Chest*, **2021**, 160, 1442-1458	5.3	4
35	Federated Learning used for predicting outcomes in SARS-COV-2 patients **2021**,		6
34	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL). *Practical Neurology*, **2021**, 21, 448-451	2.4	
33	Biallelic variants of cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. *Journal of Medical Genetics*, **2021**,	5.8	3
32	Mendelian randomisation and experimental medicine approaches to IL-6 as a drug target in PAH. *European Respiratory Journal*, **2021**,	13.6	6
31	Federated learning for predicting clinical outcomes in patients with COVID-19. *Nature Medicine*, **2021**, 27, 1735-1743	50.5	41
30	Whole-Blood RNA Profiles Associated with Pulmonary Arterial Hypertension and Clinical Outcome. *American Journal of Respiratory and Critical Care Medicine*, **2020**, 202, 586-594	10.2	14
29	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. *Nature*, **2020**, 583, 90-95	50.4	69
28	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
27	Eukaryotic life without tQCUG: the role of Elongator-dependent tRNA modifications in Dictyostelium discoideum. *Nucleic Acids Research*, **2020**, 48, 7899-7913	20.1	0
26	Familial pulmonary arterial hypertension by heterozygous loss of function. *European Respiratory Journal*, **2020**, 55,	13.6	23
25	The role of genomics and genetics in pulmonary arterial hypertension. *Global Cardiology Science & Practice*, **2020**, 2020, e202013	0.7	3
24	Characterization of Mutations and Levels of BMP9 and BMP10 in Pulmonary Arterial Hypertension. *American Journal of Respiratory and Critical Care Medicine*, **2020**, 201, 575-585	10.2	46

23	TThere and Back AgainTForward Genetics and Reverse Phenotyping in Pulmonary Arterial Hypertension. *Genes*, **2020**, 11,	4.2	3
22	Bayesian Inference Associates Rare Variants with Specific Phenotypes in Pulmonary Arterial Hypertension. *Circulation Genomic and Precision Medicine*, **2020**,	5.2	9
21	Molecular genetic framework underlying pulmonary arterial hypertension. *Nature Reviews Cardiology*, **2020**, 17, 85-95	14.8	86
20	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. *European Respiratory Journal*, **2020**, 55,	13.6	12
19	The ADAMTS13-VWF axis is dysregulated in chronic thromboembolic pulmonary hypertension. *European Respiratory Journal*, **2019**, 53,	13.6	16
18	Traffic exposures, air pollution and outcomes in pulmonary arterial hypertension: a UK cohort study analysis. *European Respiratory Journal*, **2019**, 53,	13.6	17
17	How common are single gene mutations as a cause for lacunar stroke? A targeted gene panel study. *Neurology*, **2019**, 93, e2007-e2020	6.5	10
16	Genetic determinants of risk in pulmonary arterial hypertension: international genome-wide association studies and meta-analysis. *Lancet Respiratory Medicine,the*, **2019**, 7, 227-238	35.1	55
15	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. *Nature Communications*, **2018**, 9, 1416	17.4	182
14	Loss-of-function nuclear factor B subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 1285-1296	11.5	109
13	Response by Hadinnapola et al to Letter Regarding Article, "Phenotypic Characterization of Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension". *Circulation*, **2018**, 137, 2413-2414	16.7	1
12	Biased apelin receptor agonists for cardiovascular disease. *Proceedings for Annual Meeting of the Japanese Pharmacological Society*, **2018**, WCP2018, SY85-1	0	
11	Loss-of-Function ABCC8 Mutations in Pulmonary Arterial Hypertension. *Circulation Genomic and Precision Medicine*, **2018**, 11, e002087	5.2	33
10	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
9	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	11	235
8	Phenotypic Characterization of Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. *Circulation*, **2017**, 136, 2022-2033	16.7	75
7	Plasma Metabolomics Implicates Modified Transfer RNAs and Altered Bioenergetics in the Outcomes of Pulmonary Arterial Hypertension. *Circulation*, **2017**, 135, 460-475	16.7	96
6	BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. *Lancet Respiratory Medicine,the*, **2016**, 4, 129-37	35.1	202

5	Genome-wide identification and characterisation of human DNA replication origins by initiation site sequencing (ini-seq). *Nucleic Acids Research*, **2016**, 44, 10230-10247	20.1	55
4	Selective enhancement of endothelial BMPR-II with BMP9 reverses pulmonary arterial hypertension. *Nature Medicine*, **2015**, 21, 777-85	50.5	292
3	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. *Human Mutation*, **2015**, 36, 1113-27	4.7	142
2	Bayesian inference associates rare KDR variants with specific phenotypes in pulmonary arterial hypertension		3
1	Rare variant analysis of 4,241 pulmonary arterial hypertension cases from an international consortium implicate FBLN2, PDGFD and rare de novo variants in PAH		4