Stefan GrÃ¤f

List of Publications by Year
in descending order

42 papers	4,197 citations	236925 25 h-index	265206 42 g-index
51 all docs	51 docs citations	51 times ranked	5641 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.	3.2	22
2	Mendelian randomisation and experimental medicine approaches to interleukin-6 as a drug target in pulmonary arterial hypertension. European Respiratory Journal, 2022, 59, 2002463.	6.7	31
3	Using the Plasma Proteome for Risk Stratifying Patients with Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2022, 205, 1102-1111.	5.6	35
4	Different Cytokine Patterns in BMPR2-Mutation-Positive Patients and Patients With Pulmonary Arterial Hypertension Without Mutations and Their Influence on Survival. Chest, 2022, 161, 1651-1656.	0.8	2
5	Autoimmunity Is a Significant Feature of Idiopathic Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2022, 206, 81-93.	5.6	9
6	Mining the Plasma Proteome for Insights into the Molecular Pathology of Pulmonary Arterial Hypertension. American Journal of Respiratory and Critical Care Medicine, 2022, 205, 1449-1460.	5.6	19
7	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
8	NOTCH3 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.	1.9	39
9	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.	8.2	43
10	Severe Pulmonary Arterial Hypertension Is Characterized by Increased Neutrophil Elastase and Relative Elafin Deficiency. Chest, 2021, 160, 1442-1458.	0.8	17
11	Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL). Practical Neurology, 2021, 21, 448-451.	1.1	0
12	Federated learning for predicting clinical outcomes in patients with COVID-19. Nature Medicine, 2021, 27, 1735-1743.	30.7	300
13	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
14	Biological heterogeneity in idiopathic pulmonary arterial hypertension identified through unsupervised transcriptomic profiling of whole blood. Nature Communications, 2021, 12, 7104.	12.8	21
15	Molecular genetic framework underlying pulmonary arterial hypertension. Nature Reviews Cardiology, 2020, 17, 85-95.	13.7	181
16	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.	5.6	80
17	â€˜There and Back Againâ€™â€"Forward Genetics and Reverse Phenotyping in Pulmonary Arterial Hypertension. Genes, 2020, 11, 1408.	2.4	11
18	Expression Quantitative Trait Locus Mapping in Pulmonary Arterial Hypertension. Genes, 2020, 11, 1247.	2.4	3

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19	Whole-Blood RNA Profiles Associated with Pulmonary Arterial Hypertension and Clinical Outcome. American Journal of Respiratory and Critical Care Medicine, 2020, 202, 586-594.	5.6	45
20	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95.	27.8	148
21	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
22	Eukaryotic life without tQCUG: the role of Elongator-dependent tRNA modifications in Dictyostelium discoideum. Nucleic Acids Research, 2020, 48, 7899-7913.	14.5	5
23	Familial pulmonary arterial hypertension by KDR heterozygous loss of function. European Respiratory Journal, 2020, 55, 1902165.	6.7	49
24	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. European Respiratory Journal, 2020, 55, 1901486.	6.7	26
25	The role of genomics and genetics in pulmonary arterial hypertension. Global Cardiology Science & Practice, 2020, 2020, e202013.	0.4	5
26	The ADAMTS13â€“VWF axis is dysregulated in chronic thromboembolic pulmonary hypertension. European Respiratory Journal, 2019, 53, 1801805.	6.7	31
27	Traffic exposures, air pollution and outcomes in pulmonary arterial hypertension: a UK cohort study analysis. European Respiratory Journal, 2019, 53, 1801429.	6.7	31
28	How common are single gene mutations as a cause for lacunar stroke?. Neurology, 2019, 93, e2007-e2020.	1.1	26
29	Genetic determinants of risk in pulmonary arterial hypertension: international genome-wide association studies and meta-analysis. Lancet Respiratory Medicine,the, 2019, 7, 227-238.	10.7	122
30	Identification of rare sequence variation underlying heritable pulmonary arterial hypertension. Nature Communications, 2018, 9, 1416.	12.8	279
31	Loss-of-function nuclear factor ÎºB 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.	2.9	185
32	Loss-of-Function ABCC8 Mutations in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002087.	3.6	62
33	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
34	Response by Hadinnapola et al to Letter Regarding Article, â€œPhenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertensionâ€• Circulation, 2018, 137, 2413-2414.	1.6	3
35	Biased apelin receptor agonists for cardiovascular disease. Proceedings for Annual Meeting of the Japanese Pharmacological Society, 2018, WCP2018, SY85-1.	0.0	0
36	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.	6.2	343

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37	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
38	Plasma Metabolomics Implicates Modified Transfer RNAs and Altered Bioenergetics in the Outcomes of Pulmonary Arterial Hypertension. Circulation, 2017, 135, 460-475.	1.6	154
39	BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. Lancet Respiratory Medicine,the, 2016, 4, 129-137.	10.7	307
40	Genome-wide identification and characterisation of human DNA replication origins by initiation site sequencing (ini-seq). Nucleic Acids Research, 2016, 44, gkw760.	14.5	86
41	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	2.5	185
42	Selective enhancement of endothelial BMPR-II with BMP9 reverses pulmonary arterial hypertension. Nature Medicine, 2015, 21, 777-785.	30.7	389