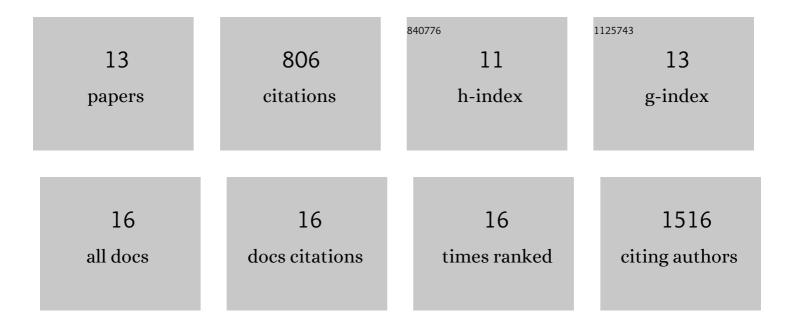


List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/11607071/publications.pdf Version: 2024-02-01



#	Article	IF	CITATIONS
1	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	8.2	112
2	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	3.6	104
3	PGAP2 Mutations, Affecting the GPI-Anchor-Synthesis Pathway, Cause Hyperphosphatasia with Mental Retardation Syndrome. American Journal of Human Genetics, 2013, 92, 584-589.	6.2	98
4	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69.	8.2	86
5	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. PLoS Genetics, 2018, 14, e1007822.	3.5	79
6	Novel Mutations and Decreased Expression of the Epigenetic Regulator <i>TET2</i> in Pulmonary Arterial Hypertension. Circulation, 2020, 141, 1986-2000.	1.6	75
7	Loss-of-Function <i>ABCC8</i> Mutations in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2018, 11, e002087.	3.6	62
8	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
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	Homozygous and Compound-Heterozygous Mutations in TGDS Cause Catel-Manzke Syndrome. American Journal of Human Genetics, 2014, 95, 763-770.	6.2	37
11	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
12	The Genomic Birthday Paradox: How Much Is Enough?. Human Mutation, 2015, 36, 989-997.	2.5	13
13	Strategies to improve the performance of rare variant association studies by optimizing the selection of controls. Bioinformatics, 2015, 31, btv457.	4.1	0