William C Nichols

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

59 papers	5,794 citations	28 h-index	62 g-index
62 ext. papers	6,859 ext. citations	9.8 avg, IF	4.63 L-index

#	Paper	IF	Citations
59	Mutations in a member of the ADAMTS gene family cause thrombotic thrombocytopenic purpura. *Nature*, **2001**, 413, 488-94	50.4	1394
58	Heterozygous germline mutations in BMPR2, encoding a TGF-beta receptor, cause familial primary pulmonary hypertension. *Nature Genetics*, **2000**, 26, 81-4	36.3	1167
57	BMPR2 haploinsufficiency as the inherited molecular mechanism for primary pulmonary hypertension. *American Journal of Human Genetics*, **2001**, 68, 92-102	11	447
56	Genomewide association study for susceptibility genes contributing to familial Parkinson disease. *Human Genetics*, **2009**, 124, 593-605	6.3	363
55	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. *JAMA Neurology*, **2013**, 70, 727-35	17.2	285
54	Glucocerebrosidase activity in Parkinson's disease with and without GBA mutations. *Brain*, **2015**, 138, 2648-58	11.2	234
53	Genetics and genomics of pulmonary arterial hypertension. *European Respiratory Journal*, **2019**, 53,	13.6	179
52	Genome screen to identify susceptibility genes for Parkinson disease in a sample without parkin mutations. *American Journal of Human Genetics*, **2002**, 71, 124-35	11	150
51	Genetic screening for a single common LRRK2 mutation in familial Parkinson's disease. *Lancet, The*, **2005**, 365, 410-2	40	145
50	Significant linkage of Parkinson disease to chromosome 2q36-37. *American Journal of Human Genetics*, **2003**, 72, 1053-7	11	138
49	Comparison of Parkinson risk in Ashkenazi Jewish patients with Gaucher disease and GBA heterozygotes. *JAMA Neurology*, **2014**, 71, 752-7	17.2	132
48	Parkinson disease phenotype in Ashkenazi Jews with and without LRRK2 G2019S mutations. *Movement Disorders*, **2013**, 28, 1966-71	7	98
47	Genomewide association study for onset age in Parkinson disease. *BMC Medical Genetics*, **2009**, 10, 98	2.1	78
46	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. *Genome Medicine*, **2018**, 10, 56	14.4	66
45	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. *Circulation Genomic and Precision Medicine*, **2018**, 11, e001887	5.2	65
44	Verification of Self-Report of Zygosity Determined via DNA Testing in a Subset of the NAS-NRC Twin Registry 40 Years Later. *Twin Research and Human Genetics*, **2005**, 8, 362-367	2.2	58
43	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

42	Copy number variation in familial Parkinson disease. *PLoS ONE*, **2011**, 6, e20988	3.7	53
41	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. *Genome Medicine*, **2019**, 11, 69	14.4	45
40	Cognitive and motor function in long-duration PARKIN-associated Parkinson disease. *JAMA Neurology*, **2014**, 71, 62-7	17.2	43
39	Multiple step pattern as a biomarker in Parkinson disease. *Parkinsonism and Related Disorders*, **2009** , 15, 506-10	3.6	39
38	Enhancing Insights into Pulmonary Vascular Disease through a Precision Medicine Approach. A Joint NHLBI-Cardiovascular Medical Research and Education Fund Workshop Report. *American Journal of Respiratory and Critical Care Medicine*, **2017**, 195, 1661-1670	10.2	38
37	Neuropsychiatric characteristics of GBA-associated Parkinson disease. *Journal of the Neurological Sciences*, **2016**, 370, 63-69	3.2	38
36	Mutations in DJ-1 are rare in familial Parkinson disease. *Neuroscience Letters*, **2006**, 408, 209-13	3.3	37
35	Phenotype characterisation of mutation and deletion carriers with neonatal and paediatric pulmonary hypertension. *European Respiratory Journal*, **2019**, 54,	13.6	36
34	Evaluation of the role of Nurr1 in a large sample of familial Parkinsonâ disease. *Movement Disorders*, **2004**, 19, 649-55	7	33
33	Alpha-synuclein and familial Parkinsonâ disease. *Movement Disorders*, **2009**, 24, 1125-31	7	32
32	Novel Mutations and Decreased Expression of the Epigenetic Regulator in Pulmonary Arterial Hypertension. *Circulation*, **2020**, 141, 1986-2000	16.7	28
31	Reliability of reported age at onset for Parkinsonâ disease. *Movement Disorders*, **2003**, 18, 275-279	7	27
30	Racial and ethnic differences in pulmonary arterial hypertension. *Pulmonary Circulation*, **2017**, 7, 793-796	2.7	25
29	Clinical correlates of depressive symptoms in familial Parkinsonâ disease. *Movement Disorders*, **2008**, 23, 2216-23	7	25
28	Mutations in LRRK2 other than G2019S are rare in a north American-based sample of familial Parkinsonâ disease. *Movement Disorders*, **2006**, 21, 2257-60	7	23
27	Glucocerebrosidase enzyme activity in GBA mutation Parkinsonâ disease. *Journal of Clinical Neuroscience*, **2016**, 28, 185-6	2.2	22
26	Cellular sources of interleukin-6 and associations with clinical phenotypes and outcomes in pulmonary arterial hypertension. *European Respiratory Journal*, **2020**, 55,	13.6	21
25	Genetic and Clinical Predictors of Deep Brain Stimulation in Young-Onset Parkinsonâ Disease. *Movement Disorders Clinical Practice*, **2016**, 3, 465-471	2.2	21

24	The EYA3 tyrosine phosphatase activity promotes pulmonary vascular remodeling in pulmonary arterial hypertension. *Nature Communications*, **2019**, 10, 4143	17.4	19
23	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
22	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. *European Respiratory Journal*, **2020**, 55,	13.6	12
21	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
20	Pulmonary arterial hypertension: SpecialistsUknowledge, practices, and attitudes of genetic counseling and genetic testing in the USA. *Pulmonary Circulation*, **2017**, 7, 372-383	2.7	10
19	Tandem mass spectrometry assay of Eglucocerebrosidase activity in dried blood spots eliminates false positives detected in fluorescence assay. *Molecular Genetics and Metabolism*, **2018**, 123, 135-139	3.7	10
18	Bayesian Inference Associates Rare Variants with Specific Phenotypes in Pulmonary Arterial Hypertension. *Circulation Genomic and Precision Medicine*, **2020**,	5.2	9
17	R1514Q substitution in Lrrk2 is not a pathogenic Parkinsonʼs disease mutation. *Movement Disorders* , **2007**, 22, 254-7	7	8
16	Genetic Admixture and Survival in Diverse Populations with Pulmonary Arterial Hypertension. *American Journal of Respiratory and Critical Care Medicine*, **2020**, 201, 1407-1415	10.2	7
15	Genomewide linkage study of modifiers of LRRK2-related Parkinsonʼs disease. *Movement Disorders*, **2011**, 26, 2039-44	7	7
14	United States Pulmonary Hypertension Scientific Registry: Baseline Characteristics. *Chest*, **2021**, 159, 311-327	5.3	7
13	Cognitive Functioning of Glucocerebrosidase () Non-manifesting Carriers. *Frontiers in Neurology*, **2021**, 12, 635958	4.1	6
12	Mendelian randomisation and experimental medicine approaches to IL-6 as a drug target in PAH. *European Respiratory Journal*, **2021**,	13.6	6
11	United States Pulmonary Hypertension Scientific Registry (USPHSR): rationale, design, and clinical implications. *Pulmonary Circulation*, **2019**, 9, 2045894019851696	2.7	5
10	Parkinson disease and STN-DBS: cognitive effects in GBA mutation carriers.. *Annals of Neurology*, **2022**,	9.4	4
9	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
8	Insulin-like growth factor binding protein-2: a new circulating indicator of pulmonary arterial hypertension severity and survival. *BMC Medicine*, **2020**, 18, 268	11.4	4
7	Bayesian inference associates rare KDR variants with specific phenotypes in pulmonary arterial hypertension		3

6	A novel BMPR2 mutation with widely disparate heritable pulmonary arterial hypertension clinical phenotype. *Pulmonary Circulation*, **2020**, 10, 2045894020931315	2.7	2
5	Elevated Interleukin-6 Levels Predict Clinical Worsening in Pediatric Pulmonary Arterial Hypertension. *Journal of Pediatrics*, **2020**, 223, 164-169.e1	3.6	2
4	Biomarkers of Pulmonary Hypertension Are Altered in Children with Down Syndrome and Pulmonary Hypertension. *Journal of Pediatrics*, **2021**,	3.6	1
3	ST2 Is a Biomarker of Pediatric Pulmonary Arterial Hypertension Severity and Clinical Worsening. *Chest*, **2021**, 160, 297-306	5.3	1
2	Subthalamic Peak Beta Ratio Is Asymmetric in Glucocerebrosidase Mutation Carriers With Parkinson's Disease: A Pilot Study. *Frontiers in Neurology*, **2021**, 12, 723476	4.1	1
1	Pediatric pulmonary hypertension: insulin-like growth factor-binding protein 2 is a novel marker associated with disease severity and survival. *Pediatric Research*, **2020**, 88, 850-856	3.2	0