

# William C Nichols

## List of Publications by Year in descending order

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Version: 2024-02-01

58  
papers

7,639  
citations

126858

33  
h-index

133188

59  
g-index

62  
all docs

62  
docs citations

62  
times ranked

7836  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in a member of the ADAMTS gene family cause thrombotic thrombocytopenic purpura. Nature, 2001, 413, 488-494.	13.7	1,623
2	Heterozygous germline mutations in BMPR2, encoding a TGF- $\beta$ 2 receptor, cause familial primary pulmonary hypertension. Nature Genetics, 2000, 26, 81-84.	9.4	1,388
3	BMPR2 Haploinsufficiency as the Inherited Molecular Mechanism for Primary Pulmonary Hypertension. American Journal of Human Genetics, 2001, 68, 92-102.	2.6	521
4	Genomewide association study for susceptibility genes contributing to familial Parkinson disease. Human Genetics, 2009, 124, 593-605.	1.8	410
5	A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. JAMA Neurology, 2013, 70, 727.	4.5	374
6	Glucocerebrosidase activity in Parkinson's disease with and without <i>GBA</i> mutations. Brain, 2015, 138, 2648-2658.	3.7	326
7	Genetics and genomics of pulmonary arterial hypertension. European Respiratory Journal, 2019, 53, 1801899.	3.1	306
8	Genetic screening for a single common LRRK2 mutation in familial Parkinson's disease. Lancet, The, 2005, 365, 410-412.	6.3	243
9	Comparison of Parkinson Risk in Ashkenazi Jewish Patients With Gaucher Disease and <i>GBA</i> Heterozygotes. JAMA Neurology, 2014, 71, 752.	4.5	172
10	Genome Screen to Identify Susceptibility Genes for Parkinson Disease in a Sample without parkin Mutations. American Journal of Human Genetics, 2002, 71, 124-135.	2.6	162
11	Significant Linkage of Parkinson Disease to Chromosome 2q36-37. American Journal of Human Genetics, 2003, 72, 1053-1057.	2.6	158
12	Parkinson disease phenotype in Ashkenazi jews with and without <i>LRRK2</i> G2019S mutations. Movement Disorders, 2013, 28, 1966-1971.	2.2	131
13	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
14	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	3.6	112
15	Genomewide association study for onset age in Parkinson disease. BMC Medical Genetics, 2009, 10, 98.	2.1	104
16	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	1.6	104
17	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69.	3.6	86
18	Phenotype characterisation of <i>TBX4</i> mutation and deletion carriers with neonatal and paediatric pulmonary hypertension. European Respiratory Journal, 2019, 54, 1801965.	3.1	77

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19	Novel Mutations and Decreased Expression of the Epigenetic Regulator <i>TET2</i> in Pulmonary Arterial Hypertension. <i>Circulation</i> , 2020, 141, 1986-2000.	1.6	75
20	Copy Number Variation in Familial Parkinson Disease. <i>PLoS ONE</i> , 2011, 6, e20988.	1.1	67
21	Verification of Self-Report of Zygosity Determined via DNA Testing in a Subset of the NAS-NRC Twin Registry 40 Years Later. <i>Twin Research and Human Genetics</i> , 2005, 8, 362-367.	0.3	61
22	Enhancing Insights into Pulmonary Vascular Disease through a Precision Medicine Approach. A Joint NHLBI Cardiovascular Medical Research and Education Fund Workshop Report. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2017, 195, 1661-1670.	2.5	59
23	Multiple step pattern as a biomarker in Parkinson disease. <i>Parkinsonism and Related Disorders</i> , 2009, 15, 506-510.	1.1	58
24	Neuropsychiatric characteristics of GBA-associated Parkinson disease. <i>Journal of the Neurological Sciences</i> , 2016, 370, 63-69.	0.3	50
25	Cognitive and Motor Function in Long-Duration <i>PARKIN</i> -Associated Parkinson Disease. <i>JAMA Neurology</i> , 2014, 71, 62.	4.5	49
26	Cellular sources of interleukin-6 and associations with clinical phenotypes and outcomes in pulmonary arterial hypertension. <i>European Respiratory Journal</i> , 2020, 55, 1901761.	3.1	48
27	Parkinson Disease and Subthalamic Nucleus Deep Brain Stimulation: Cognitive Effects in <i>GBA</i> Mutation Carriers. <i>Annals of Neurology</i> , 2022, 91, 424-435.	2.8	46
28	Mutations in DJ-1 are rare in familial Parkinson disease. <i>Neuroscience Letters</i> , 2006, 408, 209-213.	1.0	45
29	Alpha-synuclein and familial Parkinson's disease. <i>Movement Disorders</i> , 2009, 24, 1125-1131.	2.2	45
30	Whole-Blood RNA Profiles Associated with Pulmonary Arterial Hypertension and Clinical Outcome. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 202, 586-594.	2.5	45
31	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates <i>FBLN2</i> , <i>PDGFD</i> , and rare de novo variants in PAH. <i>Genome Medicine</i> , 2021, 13, 80.	3.6	43
32	Racial and ethnic differences in pulmonary arterial hypertension. <i>Pulmonary Circulation</i> , 2017, 7, 793-796.	0.8	38
33	Clinical correlates of depressive symptoms in familial Parkinson's disease. <i>Movement Disorders</i> , 2008, 23, 2216-2223.	2.2	37
34	Genetic and Clinical Predictors of Deep Brain Stimulation in Young-Onset Parkinson's Disease. <i>Movement Disorders Clinical Practice</i> , 2016, 3, 465-471.	0.8	37
35	Evaluation of the role of <i>Nurr1</i> in a large sample of familial Parkinson's disease. <i>Movement Disorders</i> , 2004, 19, 649-655.	2.2	33
36	Glucocerebrosidase enzyme activity in <i>GBA</i> mutation Parkinson's disease. <i>Journal of Clinical Neuroscience</i> , 2016, 28, 185-186.	0.8	33

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37	Mendelian randomisation and experimental medicine approaches to interleukin-6 as a drug target in pulmonary arterial hypertension. <i>European Respiratory Journal</i> , 2022, 59, 2002463.	3.1	31
38	Reliability of reported age at onset for Parkinson's disease. <i>Movement Disorders</i> , 2003, 18, 275-279.	2.2	29
39	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, .	1.6	29
40	Mutations in <i>LRRK2</i> other than G2019S are rare in a north american-based sample of familial Parkinson's disease. <i>Movement Disorders</i> , 2006, 21, 2257-2260.	2.2	26
41	Mendelian randomisation analysis of red cell distribution width in pulmonary arterial hypertension. <i>European Respiratory Journal</i> , 2020, 55, 1901486.	3.1	26
42	United States Pulmonary Hypertension Scientific Registry. <i>Chest</i> , 2021, 159, 311-327.	0.4	25
43	The <i>EYA3</i> tyrosine phosphatase activity promotes pulmonary vascular remodeling in pulmonary arterial hypertension. <i>Nature Communications</i> , 2019, 10, 4143.	5.8	24
44	Mining the Plasma Proteome for Insights into the Molecular Pathology of Pulmonary Arterial Hypertension. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2022, 205, 1449-1460.	2.5	19
45	Genetic Admixture and Survival in Diverse Populations with Pulmonary Arterial Hypertension. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2020, 201, 1407-1415.	2.5	18
46	Insulin-like growth factor binding protein-2: a new circulating indicator of pulmonary arterial hypertension severity and survival. <i>BMC Medicine</i> , 2020, 18, 268.	2.3	15
47	Cognitive Functioning of Glucocerebrosidase ( <i>GBA</i> ) Non-manifesting Carriers. <i>Frontiers in Neurology</i> , 2021, 12, 635958.	1.1	14
48	Pulmonary arterial hypertension: Specialists' knowledge, practices, and attitudes of genetic counseling and genetic testing in the USA. <i>Pulmonary Circulation</i> , 2017, 7, 372-383.	0.8	12
49	Tandem mass spectrometry assay of $\beta$ -glucocerebrosidase activity in dried blood spots eliminates false positives detected in fluorescence assay. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 135-139.	0.5	12
50	Elevated Interleukin-6 Levels Predict Clinical Worsening in Pediatric Pulmonary Arterial Hypertension. <i>Journal of Pediatrics</i> , 2020, 223, 164-169.e1.	0.9	9
51	R1514Q substitution in <i>Lrrk2</i> is not a pathogenic Parkinson's disease mutation. <i>Movement Disorders</i> , 2007, 22, 254-256.	2.2	8
52	Genomewide linkage study of modifiers of <i>LRRK2</i> -related Parkinson's disease. <i>Movement Disorders</i> , 2011, 26, 2039-2044.	2.2	8
53	United States Pulmonary Hypertension Scientific Registry (USPHSR): rationale, design, and clinical implications. <i>Pulmonary Circulation</i> , 2019, 9, 204589401985169.	0.8	7
54	Pediatric pulmonary hypertension: insulin-like growth factor-binding protein 2 is a novel marker associated with disease severity and survival. <i>Pediatric Research</i> , 2020, 88, 850-856.	1.1	6

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55	A novel BMPR2 mutation with widely disparate heritable pulmonary arterial hypertension clinical phenotype. <i>Pulmonary Circulation</i> , 2020, 10, 1-4.	0.8	6
56	ST2 Is a Biomarker of Pediatric Pulmonary Arterial Hypertension Severity and Clinical Worsening. <i>Chest</i> , 2021, 160, 297-306.	0.4	6
57	Subthalamic Peak Beta Ratio Is Asymmetric in Glucocerebrosidase Mutation Carriers With Parkinson's Disease: A Pilot Study. <i>Frontiers in Neurology</i> , 2021, 12, 723476.	1.1	5
58	Biomarkers of Pulmonary Hypertension Are Altered in Children with Down Syndrome and Pulmonary Hypertension. <i>Journal of Pediatrics</i> , 2022, 241, 68-76.e3.	0.9	3