Michael R Knowles

List of Publications by Year
in descending order

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73 papers	8,401 citations	66343 42 h-index	82547 72 g-index
74 all docs	74 docs citations	74 times ranked	7245 citing authors

#	Article	IF	Citations
1	Primary Ciliary Dyskinesia. American Journal of Respiratory and Critical Care Medicine, 2004, 169, 459-467.	5.6	701
2	Genetic Modifiers of Lung Disease in Cystic Fibrosis. New England Journal of Medicine, 2005, 353, 1443-1453.	27.0	442
3	Primary Ciliary Dyskinesia. Recent Advances in Diagnostics, Genetics, and Characterization of Clinical Disease. American Journal of Respiratory and Critical Care Medicine, 2013, 188, 913-922.	5.6	419
4	Congenital Heart Disease and Other Heterotaxic Defects in a Large Cohort of Patients With Primary Ciliary Dyskinesia. Circulation, 2007, 115, 2814-2821.	1.6	379
5	Longitudinal analysis of pulmonary function decline in patients with cystic fibrosis. Journal of Pediatrics, 1997, 131, 809-814.	1.8	325
6	Diagnosis, monitoring, and treatment of primary ciliary dyskinesia: PCD foundation consensus recommendations based on state of the art review. Pediatric Pulmonology, 2016, 51, 115-132.	2.0	297
7	Diagnosis of Primary Ciliary Dyskinesia. An Official American Thoracic Society Clinical Practice Guideline. American Journal of Respiratory and Critical Care Medicine, 2018, 197, e24-e39.	5.6	285
8	Adult Patients With Bronchiectasis. Chest, 2017, 151, 982-992.	0.8	282
9	DYX1C1 is required for axonemal dynein assembly and ciliary motility. Nature Genetics, 2013, 45, 995-1003.	21.4	256
10	Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. Nature Communications, 2015, 6, 8382.	12.8	242
11	Standardizing Nasal Nitric Oxide Measurement as a Test for Primary Ciliary Dyskinesia. Annals of the American Thoracic Society, 2013, 10, 574-581.	3.2	222
12	Clinical Features of Childhood Primary Ciliary Dyskinesia by Genotype and Ultrastructural Phenotype. American Journal of Respiratory and Critical Care Medicine, 2015, 191, 316-324.	5.6	214
13	Exome sequencing of extreme phenotypes identifies DCTN4 as a modifier of chronic Pseudomonas aeruginosa infection in cystic fibrosis. Nature Genetics, 2012, 44, 886-889.	21.4	211
14	Genome-wide association and linkage identify modifier loci of lung disease severity in cystic fibrosis at 11p13 and 20q13.2. Nature Genetics, 2011, 43, 539-546.	21.4	209
15	Mutations ofDNAH11in patients with primary ciliary dyskinesia with normal ciliary ultrastructure. Thorax, 2012, 67, 433-441.	5.6	198
16	Laterality Defects Other Than Situs Inversus Totalis in Primary Ciliary Dyskinesia. Chest, 2014, 146, 1176-1186.	0.8	192
17	Mutations in RSPH1 Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 707-717.	5.6	191
18	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 672-686.	6.2	184

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19	Multiple apical plasma membrane constituents are associated with susceptibility to meconium ileus in individuals with cystic fibrosis. Nature Genetics, 2012, 44, 562-569.	21.4	177
20	Mutations inCCDC39andCCDC40are the Major Cause of Primary Ciliary Dyskinesia with Axonemal Disorganization and Absent Inner Dynein Arms. Human Mutation, 2013, 34, 462-472.	2.5	176
21	Primary Ciliary Dyskinesia. Clinics in Chest Medicine, 2016, 37, 449-461.	2.1	168
22	Whole-Exome Capture and Sequencing Identifies HEATR2 Mutation as a Cause of Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2012, 91, 685-693.	6.2	163
23	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	6.2	150
24	Genetic Modifiers of Cystic Fibrosisâ€“Related Diabetes. Diabetes, 2013, 62, 3627-3635.	0.6	148
25	Exome Sequencing Identifies Mutations in CCDC114 as a Cause of Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 92, 99-106.	6.2	138
26	Clinical Features and Associated Likelihood of Primary Ciliary Dyskinesia in Children and Adolescents. Annals of the American Thoracic Society, 2016, 13, 1305-1313.	3.2	138
27	Pulmonary Nontuberculous Mycobacterial Infection. A Multisystem, Multigenic Disease. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 618-628.	5.6	136
28	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	6.2	135
29	Cryo-electron tomography reveals ciliary defects underlying human RSPH1 primary ciliary dyskinesia. Nature Communications, 2014, 5, 5727.	12.8	135
30	De Novo Mutations in FOXJ1 Result in a Motile Ciliopathy with Hydrocephalus and Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2019, 105, 1030-1039.	6.2	129
31	Cystic fibrosis foundation consensus conference report on pulmonary complications of cystic fibrosis. Pediatric Pulmonology, 1993, 15, 187-198.	2.0	124
32	Primary Ciliary Dyskinesia: Longitudinal Study of Lung Disease by Ultrastructure Defect and Genotype. American Journal of Respiratory and Critical Care Medicine, 2019, 199, 190-198.	5.6	116
33	Features of Severe Liver Disease With Portal Hypertension inÂPatients With Cystic Fibrosis. Clinical Gastroenterology and Hepatology, 2016, 14, 1207-1215.e3.	4.4	94
34	Identification of a splice site mutation (2789+5 G>A) associated with small amounts of normal CFTRmRNA and mild cystic fibrosis. Human Mutation, 1997, 9, 332-338.	2.5	77
35	Lack of GAS2L2 Causes PCD by Impairing Cilia Orientation and Mucociliary Clearance. American Journal of Human Genetics, 2019, 104, 229-245.	6.2	74
36	Sources of Variation in Sweat Chloride Measurements in Cystic Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2016, 194, 1375-1382.	5.6	62

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37	A quality-of-life measure for adults with primary ciliary dyskinesia: QOLâ€“PCD. European Respiratory Journal, 2015, 46, 375-383.	6.7	60
38	Discordant organ laterality in monozygotic twins with primary ciliary dyskinesia. , 1999, 82, 155-160.		57
39	Genetic association and transcriptome integration identify contributing genes and tissues at cystic fibrosis modifier loci. PLoS Genetics, 2019, 15, e1008007.	3.5	56
40	Whole-Exome Sequencing and Targeted Copy Number Analysis in Primary Ciliary Dyskinesia. G3: Genes, Genomes, Genetics, 2015, 5, 1775-1781.	1.8	53
41	Diagnostic yield of nasal scrape biopsies in primary ciliary dyskinesia: A multicenter experience. Pediatric Pulmonology, 2011, 46, 483-488.	2.0	52
42	Primary ciliary dyskinesia (PCD): A genetic disorder of motile cilia. Translational Science of Rare Diseases, 2019, 4, 51-75.	1.5	49
43	Airway Mucosal Host Defense Is Key to Genomic Regulation of Cystic Fibrosis Lung Disease Severity. American Journal of Respiratory and Critical Care Medicine, 2018, 197, 79-93.	5.6	46
44	Variants in Solute Carrier SLC26A9 Modify Prenatal Exocrine Pancreatic Damage in Cystic Fibrosis. Journal of Pediatrics, 2015, 166, 1152-1157.e6.	1.8	45
45	Pharmacotherapy for Non-Cystic Fibrosis Bronchiectasis. Chest, 2017, 152, 1120-1127.	0.8	36
46	Mutation of CFAP57, a protein required for the asymmetric targeting of a subset of inner dynein arms in Chlamydomonas, causes primary ciliary dyskinesia. PLoS Genetics, 2020, 16, e1008691.	3.5	36
47	Genetic Modifiers of Cystic Fibrosis-Related Diabetes Have Extensive Overlap With Type 2 Diabetes and Related Traits. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1401-1415.	3.6	34
48	Mutation of Growth Arrest Specific 8 Reveals a Role in Motile Cilia Function and Human Disease. PLoS Genetics, 2016, 12, e1006220.	3.5	33
49	Recurring large deletion in DRC1 (CCDC164) identified as causing primary ciliary dyskinesia in two Asian patients. Molecular Genetics & Genomic Medicine, 2019, 7, e838.	1.2	30
50	Gene Expression in Transformed Lymphocytes Reveals Variation in Endomembrane and HLA Pathways Modifying Cystic Fibrosis Pulmonary Phenotypes. American Journal of Human Genetics, 2015, 96, 318-328.	6.2	28
51	Autosomal dominant variants in FOXJ1 causing primary ciliary dyskinesia in two patients with obstructive hydrocephalus. Molecular Genetics & Genomic Medicine, 2021, 9, e1726.	1.2	22
52	Nasal Nitric Oxide in Primary Immunodeficiency and Primary Ciliary Dyskinesia: Helping to Distinguish Between Clinically Similar Diseases. Journal of Clinical Immunology, 2019, 39, 216-224.	3.8	21
53	Otolaryngology Manifestations of Primary Ciliary Dyskinesia: A Multicenter Study. Otolaryngology - Head and Neck Surgery, 2022, 166, 540-547.	1.9	19
54	Analysis of a large cohort of cystic fibrosis patients with severe liver disease indicates lung function decline does not significantly differ from that of the general cystic fibrosis population. PLoS ONE, 2018, 13, e0205257.	2.5	16

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55	ACTR2 absence or antagonism prevents cystic fibrosis pulmonary manifestations. Journal of Cystic Fibrosis, 2019, 18, 127-134.	0.7	15
56	Primary ciliary dyskinesia in Japan: systematic review and meta-analysis. BMC Pulmonary Medicine, 2019, 19, 135.	2.0	14
57	Complete CFTR gene sequencing in 5,058 individuals with cystic fibrosis informs variant-specific treatment. Journal of Cystic Fibrosis, 2022, 21, 463-470.	0.7	13
58	Novel variation at chr11p13 associated with cystic fibrosis lung disease severity. Human Genome Variation, 2016, 3, 16020.	0.7	9
59	Improving imputation in disease-relevant regions: lessons from cystic fibrosis. Npj Genomic Medicine, 2018, 3, 8.	3.8	9
60	Mining GWAS and eQTL data for CF lung disease modifiers by gene expression imputation. PLoS ONE, 2020, 15, e0239189.	2.5	9
61	Enlarged Dural Sac in Idiopathic Bronchiectasis Implicates Heritable Connective Tissue Gene Variants. Annals of the American Thoracic Society, 2016, 13, 1712-1720.	3.2	8
62	Leveraging TOPMed imputation server and constructing a cohort-specific imputation reference panel to enhance genotype imputation among cystic fibrosis patients. Human Genetics and Genomics Advances, 2022, 3, 100090.	1.7	6
63	Errors in Methodology Affect Diagnostic Accuracy of High-Speed Videomicroscopy Analysis in Primary Ciliary Dyskinesia. Chest, 2019, 156, 1032-1033.	0.8	5
64	Frequency of untreated hypogammaglobulinemia in bronchiectasis. Annals of Allergy, Asthma and Immunology, 2017, 119, 83-85.	1.0	4
65	Primary Ciliary Dyskinesia Diagnosis. Is Color Better Than Black and White?. American Journal of Respiratory and Critical Care Medicine, 2017, 196, 9-10.	5.6	4
66	The prevalence of the defining features of primary ciliary dyskinesia within a cri du chat syndrome cohort. Pediatric Pulmonology, 2018, 53, 1565-1573.	2.0	4
67	Use caution interpreting nasal nitric oxide: Overlap in primary ciliary dyskinesia and primary immunodeficiency. Pediatric Pulmonology, 2021, 56, 4045-4047.	2.0	4
68	Assessment of Ciliary Beat Pattern. Chest, 2017, 151, 958-959.	0.8	3
69	Comparing encounter-based and annualized chronic pseudomonas infection definitions in cystic fibrosis. Journal of Cystic Fibrosis, 2022, 21, 40-44.	0.7	3
70	Nutrition and Markers of Disease Severity in Patients With Bronchiectasis. Chronic Obstructive Pulmonary Diseases (Miami, Fla), 2020, 7, 390-403.	0.7	3
71	Going beyond the chest Xâ€ray:Âlnvestigating laterality defects in primary ciliary dyskinesia. Pediatric Pulmonology, 2022, 57, 1318-1324.	2.0	3
72	Cytoplasmic â€œciliary inclusionsâ€•in isolation are not sufficient for the diagnosis of primary ciliary dyskinesia. Pediatric Pulmonology, 2020, 55, 130-135.	2.0	2

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73	Accounting for population structure in genetic studies of cystic fibrosis. Human Genetics and Genomics Advances, 2022, 3, 100117.	1.7	1