

# Michael R Knowles

## List of Publications by Year in descending order

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73  
papers

8,401  
citations

70961

41  
h-index

85405

71  
g-index

74  
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74  
docs citations

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times ranked

7245  
citing authors

#	ARTICLE	IF	CITATIONS
1	Otolaryngology Manifestations of Primary Ciliary Dyskinesia: A Multicenter Study. <i>Otolaryngology - Head and Neck Surgery</i> , 2022, 166, 540-547.	1.1	19
2	Comparing encounter-based and annualized chronic pseudomonas infection definitions in cystic fibrosis. <i>Journal of Cystic Fibrosis</i> , 2022, 21, 40-44.	0.3	3
3	Complete CFTR gene sequencing in 5,058 individuals with cystic fibrosis informs variant-specific treatment. <i>Journal of Cystic Fibrosis</i> , 2022, 21, 463-470.	0.3	13
4	Leveraging TOPMed imputation server and constructing a cohort-specific imputation reference panel to enhance genotype imputation among cystic fibrosis patients. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100090.	1.0	6
5	Going beyond the chest X-ray: Investigating laterality defects in primary ciliary dyskinesia. <i>Pediatric Pulmonology</i> , 2022, 57, 1318-1324.	1.0	3
6	Accounting for population structure in genetic studies of cystic fibrosis. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100117.	1.0	1
7	Autosomal dominant variants in <i>FOXJ1</i> causing primary ciliary dyskinesia in two patients with obstructive hydrocephalus. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1726.	0.6	22
8	Use caution interpreting nasal nitric oxide: Overlap in primary ciliary dyskinesia and primary immunodeficiency. <i>Pediatric Pulmonology</i> , 2021, 56, 4045-4047.	1.0	4
9	Genetic Modifiers of Cystic Fibrosis-Related Diabetes Have Extensive Overlap With Type 2 Diabetes and Related Traits. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 1401-1415.	1.8	34
10	Cytoplasmic ciliary inclusions in isolation are not sufficient for the diagnosis of primary ciliary dyskinesia. <i>Pediatric Pulmonology</i> , 2020, 55, 130-135.	1.0	2
11	Mutation of CFAP57, a protein required for the asymmetric targeting of a subset of inner dynein arms in <i>Chlamydomonas</i> , causes primary ciliary dyskinesia. <i>PLoS Genetics</i> , 2020, 16, e1008691.	1.5	36
12	Mining GWAS and eQTL data for CF lung disease modifiers by gene expression imputation. <i>PLoS ONE</i> , 2020, 15, e0239189.	1.1	9
13	Nutrition and Markers of Disease Severity in Patients With Bronchiectasis. <i>Chronic Obstructive Pulmonary Diseases (Miami, Fla )</i> , 2020, 7, 390-403.	0.5	3
14	Primary Ciliary Dyskinesia: Longitudinal Study of Lung Disease by Ultrastructure Defect and Genotype. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2019, 199, 190-198.	2.5	116
15	AGTR2 absence or antagonism prevents cystic fibrosis pulmonary manifestations. <i>Journal of Cystic Fibrosis</i> , 2019, 18, 127-134.	0.3	15
16	Primary ciliary dyskinesia in Japan: systematic review and meta-analysis. <i>BMC Pulmonary Medicine</i> , 2019, 19, 135.	0.8	14
17	Primary ciliary dyskinesia (PCD): A genetic disorder of motile cilia. <i>Translational Science of Rare Diseases</i> , 2019, 4, 51-75.	1.6	49
18	Recurring large deletion in <i>DRC1</i> ( <i>CCDC164</i> ) identified as causing primary ciliary dyskinesia in two Asian patients. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e838.	0.6	30

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19	De Novo Mutations in FOXJ1 Result in a Motile Ciliopathy with Hydrocephalus and Randomization of Left/Right Body Asymmetry. <i>American Journal of Human Genetics</i> , 2019, 105, 1030-1039.	2.6	129
20	Lack of GAS2L2 Causes PCD by Impairing Cilia Orientation and Mucociliary Clearance. <i>American Journal of Human Genetics</i> , 2019, 104, 229-245.	2.6	74
21	Nasal Nitric Oxide in Primary Immunodeficiency and Primary Ciliary Dyskinesia: Helping to Distinguish Between Clinically Similar Diseases. <i>Journal of Clinical Immunology</i> , 2019, 39, 216-224.	2.0	21
22	Genetic association and transcriptome integration identify contributing genes and tissues at cystic fibrosis modifier loci. <i>PLoS Genetics</i> , 2019, 15, e1008007.	1.5	56
23	Errors in Methodology Affect Diagnostic Accuracy of High-Speed Videomicroscopy Analysis in Primary Ciliary Dyskinesia. <i>Chest</i> , 2019, 156, 1032-1033.	0.4	5
24	Improving imputation in disease-relevant regions: lessons from cystic fibrosis. <i>Npj Genomic Medicine</i> , 2018, 3, 8.	1.7	9
25	Airway Mucosal Host Defense Is Key to Genomic Regulation of Cystic Fibrosis Lung Disease Severity. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2018, 197, 79-93.	2.5	46
26	The prevalence of the defining features of primary ciliary dyskinesia within a cri du chat syndrome cohort. <i>Pediatric Pulmonology</i> , 2018, 53, 1565-1573.	1.0	4
27	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. <i>PLoS ONE</i> , 2018, 13, e0205257.	1.1	16
28	Diagnosis of Primary Ciliary Dyskinesia. An Official American Thoracic Society Clinical Practice Guideline. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2018, 197, e24-e39.	2.5	285
29	Assessment of Ciliary Beat Pattern. <i>Chest</i> , 2017, 151, 958-959.	0.4	3
30	Pharmacotherapy for Non-Cystic Fibrosis Bronchiectasis. <i>Chest</i> , 2017, 152, 1120-1127.	0.4	36
31	Frequency of untreated hypogammaglobulinemia in bronchiectasis. <i>Annals of Allergy, Asthma and Immunology</i> , 2017, 119, 83-85.	0.5	4
32	Primary Ciliary Dyskinesia Diagnosis. Is Color Better Than Black and White?. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2017, 196, 9-10.	2.5	4
33	Adult Patients With Bronchiectasis. <i>Chest</i> , 2017, 151, 982-992.	0.4	282
34	Novel variation at chr11p13 associated with cystic fibrosis lung disease severity. <i>Human Genome Variation</i> , 2016, 3, 16020.	0.4	9
35	Features of Severe Liver Disease With Portal Hypertension in Patients With Cystic Fibrosis. <i>Clinical Gastroenterology and Hepatology</i> , 2016, 14, 1207-1215.e3.	2.4	94
36	Clinical Features and Associated Likelihood of Primary Ciliary Dyskinesia in Children and Adolescents. <i>Annals of the American Thoracic Society</i> , 2016, 13, 1305-1313.	1.5	138

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37	Sources of Variation in Sweat Chloride Measurements in Cystic Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2016, 194, 1375-1382.	2.5	62
38	Enlarged Dural Sac in Idiopathic Bronchiectasis Implicates Heritable Connective Tissue Gene Variants. Annals of the American Thoracic Society, 2016, 13, 1712-1720.	1.5	8
39	Primary Ciliary Dyskinesia. Clinics in Chest Medicine, 2016, 37, 449-461.	0.8	168
40	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.	1.0	297
41	Mutation of Growth Arrest Specific 8 Reveals a Role in Motile Cilia Function and Human Disease. PLoS Genetics, 2016, 12, e1006220.	1.5	33
42	A quality-of-life measure for adults with primary ciliary dyskinesia: QOLâ€“PCD. European Respiratory Journal, 2015, 46, 375-383.	3.1	60
43	Clinical Features of Childhood Primary Ciliary Dyskinesia by Genotype and Ultrastructural Phenotype. American Journal of Respiratory and Critical Care Medicine, 2015, 191, 316-324.	2.5	214
44	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.	2.6	28
45	Whole-Exome Sequencing and Targeted Copy Number Analysis in Primary Ciliary Dyskinesia. G3: Genes, Genomes, Genetics, 2015, 5, 1775-1781.	0.8	53
46	Pulmonary Nontuberculous Mycobacterial Infection. A Multisystem, Multigenic Disease. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 618-628.	2.5	136
47	Variants in Solute Carrier SLC26A9 Modify Prenatal Exocrine Pancreatic Damage in Cystic Fibrosis. Journal of Pediatrics, 2015, 166, 1152-1157.e6.	0.9	45
48	Genome-wide association meta-analysis identifies five modifier loci of lung disease severity in cystic fibrosis. Nature Communications, 2015, 6, 8382.	5.8	242
49	Mutations in <i>RSPH1</i> Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 707-717.	2.5	191
50	Laterality Defects Other Than Situs Inversus Totalis in Primary Ciliary Dyskinesia. Chest, 2014, 146, 1176-1186.	0.4	192
51	Cryo-electron tomography reveals ciliary defects underlying human RSPH1 primary ciliary dyskinesia. Nature Communications, 2014, 5, 5727.	5.8	135
52	Genetic Modifiers of Cystic Fibrosisâ€“Related Diabetes. Diabetes, 2013, 62, 3627-3635.	0.3	148
53	DYX1C1 is required for axonemal dynein assembly and ciliary motility. Nature Genetics, 2013, 45, 995-1003.	9.4	256
54	Exome Sequencing Identifies Mutations in CCDC114 as a Cause of Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 92, 99-106.	2.6	138

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55	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.	2.6	184
56	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	2.6	135
57	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.	2.5	419
58	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	2.6	150
59	Standardizing Nasal Nitric Oxide Measurement as a Test for Primary Ciliary Dyskinesia. Annals of the American Thoracic Society, 2013, 10, 574-581.	1.5	222
60	Mutations in <i>CCDC39</i> and <i>CCDC40</i> are the Major Cause of Primary Ciliary Dyskinesia with Axonemal Disorganization and Absent Inner Dynein Arms. Human Mutation, 2013, 34, 462-472.	1.1	176
61	Mutations of <i>DNAH11</i> in patients with primary ciliary dyskinesia with normal ciliary ultrastructure. Thorax, 2012, 67, 433-441.	2.7	198
62	Exome sequencing of extreme phenotypes identifies DCTN4 as a modifier of chronic Pseudomonas aeruginosa infection in cystic fibrosis. Nature Genetics, 2012, 44, 886-889.	9.4	211
63	Whole-Exome Capture and Sequencing Identifies HEATR2 Mutation as a Cause of Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2012, 91, 685-693.	2.6	163
64	Multiple apical plasma membrane constituents are associated with susceptibility to meconium ileus in individuals with cystic fibrosis. Nature Genetics, 2012, 44, 562-569.	9.4	177
65	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.	9.4	209
66	Diagnostic yield of nasal scrape biopsies in primary ciliary dyskinesia: A multicenter experience. Pediatric Pulmonology, 2011, 46, 483-488.	1.0	52
67	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
68	Genetic Modifiers of Lung Disease in Cystic Fibrosis. New England Journal of Medicine, 2005, 353, 1443-1453.	13.9	442
69	Primary Ciliary Dyskinesia. American Journal of Respiratory and Critical Care Medicine, 2004, 169, 459-467.	2.5	701
70	Discordant organ laterality in monozygotic twins with primary ciliary dyskinesia. , 1999, 82, 155-160.		57
71	Longitudinal analysis of pulmonary function decline in patients with cystic fibrosis. Journal of Pediatrics, 1997, 131, 809-814.	0.9	325
72	Identification of a splice site mutation (2789+5 G>A) associated with small amounts of normal CFTRmRNA and mild cystic fibrosis. , 1997, 9, 332-338.		77

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73	Cystic fibrosis foundation consensus conference report on pulmonary complications of cystic fibrosis. <i>Pediatric Pulmonology</i> , 1993, 15, 187-198.	1.0	124