

# Maimoona A Zariwala

## List of Publications by Year in descending order

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Version: 2024-02-01

41  
papers

4,849  
citations

201674

27  
h-index

265206

42  
g-index

43  
all docs

43  
docs citations

43  
times ranked

3731  
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.	1.9	19
2	Hereditary Mucin Deficiency Caused by Biallelic Loss of Function of <i>MUC5B</i> . <i>American Journal of Respiratory and Critical Care Medicine</i> , 2022, 205, 761-768.	5.6	12
3	The global prevalence and ethnic heterogeneity of primary ciliary dyskinesia gene variants: a genetic database analysis. <i>Lancet Respiratory Medicine</i> , 2022, 10, 459-468.	10.7	63
4	Going beyond the chest X-ray: Investigating laterality defects in primary ciliary dyskinesia. <i>Pediatric Pulmonology</i> , 2022, 57, 1318-1324.	2.0	3
5	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.	1.2	22
6	Use caution interpreting nasal nitric oxide: Overlap in primary ciliary dyskinesia and primary immunodeficiency. <i>Pediatric Pulmonology</i> , 2021, 56, 4045-4047.	2.0	4
7	Identification of genetic variants in <i>CFAP221</i> as a cause of primary ciliary dyskinesia. <i>Journal of Human Genetics</i> , 2020, 65, 175-180.	2.3	27
8	Cytoplasmic ciliary inclusions in isolation are not sufficient for the diagnosis of primary ciliary dyskinesia. <i>Pediatric Pulmonology</i> , 2020, 55, 130-135.	2.0	2
9	Mutation of <i>CFAP57</i> , 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.	3.5	36
10	Motile ciliopathies. <i>Nature Reviews Disease Primers</i> , 2020, 6, 77.	30.5	191
11	A human ciliopathy reveals essential functions for <i>NEK10</i> in airway mucociliary clearance. <i>Nature Medicine</i> , 2020, 26, 244-251.	30.7	45
12	The expanding phenotype of <i>OFD1</i> -related disorders: Hemizygous loss of function variants in three patients with primary ciliary dyskinesia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e911.	1.2	31
13	Primary ciliary dyskinesia (PCD): A genetic disorder of motile cilia. <i>Translational Science of Rare Diseases</i> , 2019, 4, 51-75.	1.5	49
14	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.	1.2	30
15	De Novo Mutations in <i>FOXJ1</i> 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.	6.2	129
16	Lack of <i>GAS2L2</i> Causes PCD by Impairing Cilia Orientation and Mucociliary Clearance. <i>American Journal of Human Genetics</i> , 2019, 104, 229-245.	6.2	74
17	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.	3.8	21
18	Primary ciliary dyskinesia: keep it on your radar. <i>Thorax</i> , 2018, 73, 101-102.	5.6	5

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19	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.	2.0	4
20	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.	5.6	285
21	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.	3.2	138
22	Primary Ciliary Dyskinesia. <i>Clinics in Chest Medicine</i> , 2016, 37, 449-461.	2.1	168
23	Diagnosis, monitoring, and treatment of primary ciliary dyskinesia: PCD foundation consensus recommendations based on state of the art review. <i>Pediatric Pulmonology</i> , 2016, 51, 115-132.	2.0	297
24	Mutation of Growth Arrest Specific 8 Reveals a Role in Motile Cilia Function and Human Disease. <i>PLoS Genetics</i> , 2016, 12, e1006220.	3.5	33
25	The prevalence of clinical features associated with primary ciliary dyskinesia in a heterotaxy population: results of a web-based survey. <i>Cardiology in the Young</i> , 2015, 25, 752-759.	0.8	22
26	Clinical Features of Childhood Primary Ciliary Dyskinesia by Genotype and Ultrastructural Phenotype. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2015, 191, 316-324.	5.6	214
27	Whole-Exome Sequencing and Targeted Copy Number Analysis in Primary Ciliary Dyskinesia. <i>G3: Genes, Genomes, Genetics</i> , 2015, 5, 1775-1781.	1.8	53
28	Mutations in <i>RSPH1</i> Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2014, 189, 707-717.	5.6	191
29	The Role of Molecular Genetic Analysis in the Diagnosis of Primary Ciliary Dyskinesia. <i>Annals of the American Thoracic Society</i> , 2014, 11, 351-359.	3.2	47
30	Laterality Defects Other Than Situs Inversus Totalis in Primary Ciliary Dyskinesia. <i>Chest</i> , 2014, 146, 1176-1186.	0.8	192
31	Cryo-electron tomography reveals ciliary defects underlying human <i>RSPH1</i> primary ciliary dyskinesia. <i>Nature Communications</i> , 2014, 5, 5727.	12.8	135
32	Cri du Chat Syndrome and Primary Ciliary Dyskinesia: A Common Genetic Cause on Chromosome 5p. <i>Journal of Pediatrics</i> , 2014, 165, 858-861.	1.8	15
33	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. <i>American Journal of Human Genetics</i> , 2013, 93, 336-345.	6.2	183
34	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 93, 672-686.	6.2	184
35	Primary Ciliary Dyskinesia. Recent Advances in Diagnostics, Genetics, and Characterization of Clinical Disease. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2013, 188, 913-922.	5.6	419
36	Standardizing Nasal Nitric Oxide Measurement as a Test for Primary Ciliary Dyskinesia. <i>Annals of the American Thoracic Society</i> , 2013, 10, 574-581.	3.2	222

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37	Primary Ciliary Dyskinesia in Amish Communities. <i>Journal of Pediatrics</i> , 2010, 156, 1023-1025.	1.8	13
38	A multicenter study of the frequency and distribution of GJB2 and GJB6 mutations in a large North American cohort. <i>Genetics in Medicine</i> , 2007, 9, 413-426.	2.4	134
39	Genetic Defects in Ciliary Structure and Function. <i>Annual Review of Physiology</i> , 2007, 69, 423-450.	13.1	270
40	Mutations of <i>DNAI1</i> in Primary Ciliary Dyskinesia. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2006, 174, 858-866.	5.6	162
41	Primary Ciliary Dyskinesia. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2004, 169, 459-467.	5.6	701