Amelia Shoemark

List of Publications by Year
in descending order

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137 papers	5,693 citations	81900 39 h-index	85541 71 g-index
144 all docs	144 docs citations	144 times ranked	4943 citing authors

#	Article	IF	Citations
1	European Respiratory Society guidelines for the diagnosis of primary ciliary dyskinesia. European Respiratory Journal, 2017, 49, 1601090.	6.7	465
2	Mutations in Radial Spoke Head Protein Genes RSPH9 and RSPH4A Cause Primary Ciliary Dyskinesia with Central-Microtubular-Pair Abnormalities. American Journal of Human Genetics, 2009, 84, 197-209.	6.2	303
3	Recessive HYDIN Mutations Cause Primary Ciliary Dyskinesia without Randomization of Left-Right Body Asymmetry. American Journal of Human Genetics, 2012, 91, 672-684.	6.2	252
4	Aetiology in adult patients with bronchiectasis. Respiratory Medicine, 2007, 101, 1163-1170.	2.9	250
5	Diagnosis and management of primary ciliary dyskinesia. Archives of Disease in Childhood, 2014, 99, 850-856.	1.9	216
6	Impaired innate interferon induction in severe therapy resistant atopic asthmatic children. Mucosal Immunology, 2013, 6, 797-806.	6.0	198
7	Primary ciliary dyskinesia in the genomics age. Lancet Respiratory Medicine,the, 2020, 8, 202-216.	10.7	182
8	Splice-Site Mutations in the Axonemal Outer Dynein Arm Docking Complex Gene CCDC114 Cause Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 92, 88-98.	6.2	176
9	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
10	Mutations in ZMYND10, a Gene Essential for Proper Axonemal Assembly of Inner and Outer Dynein Arms in Humans and Flies, Cause Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 346-356.	6.2	167
11	Sperm defects in primary ciliary dyskinesia and related causes of male infertility. Cellular and Molecular Life Sciences, 2020, 77, 2029-2048.	5.4	140
12	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3. Nature Communications, 2017, 8, 14279.	12.8	133
13	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
14	Pseudomonas aeruginosa, cyanide accumulation and lung function in CF and non-CF bronchiectasis patients. European Respiratory Journal, 2008, 32, 740-747.	6.7	105
15	A longitudinal study characterising a large adult primary ciliary dyskinesia population. European Respiratory Journal, 2016, 48, 441-450.	6.7	101
16	Neutrophil extracellular traps, disease severity, and antibiotic response in bronchiectasis: an international, observational, multicohort study. Lancet Respiratory Medicine,the, 2021, 9, 873-884.	10.7	99
17	Twenty-year review of quantitative transmission electron microscopy for the diagnosis of primary ciliary dyskinesia. Journal of Clinical Pathology, 2012, 65, 267-271.	2.0	97
18	Accuracy of Immunofluorescence in the Diagnosis of Primary Ciliary Dyskinesia. American Journal of Respiratory and Critical Care Medicine, 2017, 196, 94-101.	5.6	97

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19	Mutations in Outer Dynein Arm Heavy Chain DNAH9 Cause Motile Cilia Defects and Situs Inversus. American Journal of Human Genetics, 2018, 103, 984-994.	6.2	95
20	Increased nuclear suppressor of cytokine signaling 1 in asthmatic bronchial epithelium suppresses rhinovirus induction of innate interferons. Journal of Allergy and Clinical Immunology, 2015, 136, 177-188.e11.	2.9	89
21	Combined exome and whole-genome sequencing identifies mutations inARMC4as a cause of primary ciliary dyskinesia with defects in the outer dynein arm. Journal of Medical Genetics, 2014, 51, 61-67.	3.2	88
22	Assessment of F/HN-Pseudotyped Lentivirus as a Clinically Relevant Vector for Lung Gene Therapy. American Journal of Respiratory and Critical Care Medicine, 2012, 186, 846-856.	5.6	86
23	Targeted NGS gene panel identifies mutations in RSPH1 causing primary ciliary dyskinesia and a common mechanism for ciliary central pair agenesis due to radial spoke defects. Human Molecular Genetics, 2014, 23, 3362-3374.	2.9	82
24	Primary Cilia Mediate Diverse Kinase Inhibitor Resistance Mechanisms in Cancer. Cell Reports, 2018, 23, 3042-3055.	6.4	77
25	International consensus guideline for reporting transmission electron microscopy results in the diagnosis of primary ciliary dyskinesia (BEAT PCD TEM Criteria). European Respiratory Journal, 2020, 55, 1900725.	6.7	77
26	The efficacy and safety of inhaled antibiotics for the treatment of bronchiectasis in adults: a systematic review and meta-analysis. Lancet Respiratory Medicine,the, 2019, 7, 855-869.	10.7	75
27	Airway Bacterial Load and Inhaled Antibiotic Response in Bronchiectasis. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 33-41.	5.6	70
28	Mutations in ARL2BP, Encoding ADP-Ribosylation-Factor-Like 2 Binding Protein, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2013, 93, 321-329.	6.2	67
29	HEATR2 Plays a Conserved Role in Assembly of the Ciliary Motile Apparatus. PLoS Genetics, 2014, 10, e1004577.	3.5	67
30	Characterization of Eosinophilic Bronchiectasis: A European Multicohort Study. American Journal of Respiratory and Critical Care Medicine, 2022, 205, 894-902.	5.6	67
31	High prevalence of CCDC103 p.His154Pro mutation causing primary ciliary dyskinesia disrupts protein oligomerisation and is associated with normal diagnostic investigations. Thorax, 2018, 73, 157-166.	5.6	63
32	Accuracy of High-Speed Video Analysis toÂDiagnose Primary Ciliary Dyskinesia. Chest, 2019, 155, 1008-1017.	0.8	59
33	Risk factors for situs defects and congenital heart disease in primary ciliary dyskinesia. Thorax, 2019, 74, 203-205.	5.6	52
34	C11orf70 Mutations Disrupting the Intraflagellar Transport-Dependent Assembly of Multiple Axonemal Dyneins Cause Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2018, 102, 956-972.	6.2	51
35	A point-of-care neutrophil elastase activity assay identifies bronchiectasis severity, airway infection and riskÂofÂexacerbation. European Respiratory Journal, 2019, 53, 1900303.	6.7	50
36	Clinical utility of NGS diagnosis and disease stratification in a multiethnic primary ciliary dyskinesia cohort. Journal of Medical Genetics, 2020, 57, 322-330.	3.2	50

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37	Topological data analysis reveals genotypeâ€“phenotype relationships in primary ciliary dyskinesia. European Respiratory Journal, 2021, 58, 2002359.	6.7	49
38	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	2.9	45
39	ZMYND10 functions in a chaperone relay during axonemal dynein assembly. ELife, 2018, 7, .	6.0	44
40	Antimicrobial peptides, disease severity and exacerbations in bronchiectasis. Thorax, 2019, 74, 835-842.	5.6	43
41	Pregnancy Zone Protein Is Associated with Airway Infection, Neutrophil Extracellular Trap Formation, and Disease Severity in Bronchiectasis. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 992-1001.	5.6	42
42	ERS and ATS diagnostic guidelines for primary ciliary dyskinesia: similarities and differences in approach to diagnosis. European Respiratory Journal, 2019, 54, 1901066.	6.7	41
43	BMI-1 extends proliferative potential of human bronchial epithelial cells while retaining their mucociliary differentiation capacity. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2017, 312, L258-L267.	2.9	40
44	Standardised clinical data from patients with primary ciliary dyskinesia: FOLLOW-PCD. ERJ Open Research, 2020, 6, 00237-2019.	2.6	36
45	Primary ciliary dyskinesia with normal ultrastructure: three-dimensional tomography detects absence of DNAH11. European Respiratory Journal, 2018, 51, 1701809.	6.7	33
46	An extracellular matrix fragment drives epithelial remodeling and airway hyperresponsiveness. Science Translational Medicine, 2018, 10, .	12.4	33
47	The Impact of the COVID-19 Pandemic on Exacerbations and Symptoms in Bronchiectasis: A Prospective Study. American Journal of Respiratory and Critical Care Medicine, 2021, 204, 857-859.	5.6	33
48	Bronchial and peripheral airway nitric oxide in primary ciliary dyskinesia and bronchiectasis. Respiratory Medicine, 2009, 103, 700-706.	2.9	30
49	Characterizing the ultrastructure of primary ciliary dyskinesia transposition defect using electron tomography. Cytoskeleton, 2014, 71, 294-301.	2.0	29
50	Endotyping Chronic Obstructive Pulmonary Disease, Bronchiectasis, and the â€œChronic Obstructive Pulmonary Diseaseâ€“Bronchiectasis Associationâ€• American Journal of Respiratory and Critical Care Medicine, 2022, 206, 417-426.	5.6	29
51	Clinical features and management of children with primary ciliary dyskinesia in England. Archives of Disease in Childhood, 2020, 105, 724-729.	1.9	28
52	Primary ciliary dyskinesia: evaluation using cilia beat frequency assessment via spectral analysis of digital microscopy images. Journal of Applied Physiology, 2011, 111, 295-302.	2.5	27
53	Why, when and how to investigate primary ciliary dyskinesia in adult patients with bronchiectasis. Multidisciplinary Respiratory Medicine, 2018, 13, 26.	1.5	27
54	Cyanide levels found in infected cystic fibrosis sputum inhibit airway ciliary function. European Respiratory Journal, 2014, 44, 1253-1261.	6.7	26

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55	The European Multicentre Bronchiectasis Audit and Research Collaboration (EMBARC) ERS Clinical Research Collaboration. European Respiratory Journal, 2018, 52, 1802074.	6.7	26
56	Electron tomography of respiratory cilia. Thorax, 2013, 68, 190-191.	5.6	25
57	Neutrophil dysfunction in bronchiectasis: an emerging role for immunometabolism. European Respiratory Journal, 2021, 58, 2003157.	6.7	25
58	Diagnosis of Primary Ciliary Dyskinesia. Clinics in Chest Medicine, 2022, 43, 127-140.	2.1	25
59	Bardet Biedl Syndrome. Chest, 2015, 147, 764-770.	0.8	24
60	Primary Ciliary Dyskinesia Due to Microtubular Defects is Associated with Worse Lung Clearance Index. Lung, 2018, 196, 231-238.	3.3	22
61	Inhaled aztreonam improves symptoms of cough and sputum production in patients with bronchiectasis: a post hoc analysis of the AIR-BX studies. European Respiratory Journal, 2020, 56, 2000608.	6.7	22
62	Heterogeneity of treatment response in bronchiectasis clinical trials. European Respiratory Journal, 2022, 59, 2100777.	6.7	21
63	Procalcitonin in stable and unstable patients with bronchiectasis. Chronic Respiratory Disease, 2008, 5, 155-160.	2.4	19
64	CXCL-8-dependent and -independent neutrophil activation in COPD: experiences from a pilot study of the CXCR2 antagonist danirixin. ERJ Open Research, 2020, 6, 00583-2020.	2.6	19
65	Generation of a Three-Dimensional Ultrastructural Model of Human Respiratory Cilia. American Journal of Respiratory Cell and Molecular Biology, 2012, 47, 800-806.	2.9	18
66	Time trends in diagnostic testing for primary ciliary dyskinesia in Europe. European Respiratory Journal, 2019, 54, 1900528.	6.7	17
67	Genome sequencing reveals underdiagnosis of primary ciliary dyskinesia in bronchiectasis. European Respiratory Journal, 2022, 60, 2200176.	6.7	17
68	The BEAT-PCD (Better Experimental Approaches to Treat Primary Ciliary Dyskinesia) Clinical Research Collaboration. European Respiratory Journal, 2021, 57, 2004601.	6.7	16
69	Elevated peripheral airway nitric oxide in bronchiectasis reflects disease severity. Respiratory Medicine, 2011, 105, 885-891.	2.9	15
70	Secondary defects detected by transmission electron microscopy in primary ciliary dyskinesia diagnostics. Ultrastructural Pathology, 2017, 41, 390-398.	0.9	15
71	Validation of the Bronchiectasis Impact Measure (BIM): a novel patient-reported outcome measure. European Respiratory Journal, 2021, 57, 2003156.	6.7	14
72	Motile cilia defects in diseases other than primary ciliary dyskinesia: The contemporary diagnostic and research role for transmission electron microscopy. Ultrastructural Pathology, 2017, 41, 415-427.	0.9	12

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73	The Controversies and Difficulties of Diagnosing Primary Ciliary Dyskinesia. American Journal of Respiratory and Critical Care Medicine, 2020, 201, 120-122.	5.6	12
74	Primary Ciliary Dyskinesia. Seminars in Respiratory and Critical Care Medicine, 2021, 42, 537-548.	2.1	12
75	Lung Clearance Index (LCI) is Stable in Most Primary Ciliary Dyskinesia (PCD) Patients Managed in a Specialist Centre: a Pilot Study. Lung, 2017, 195, 441-443.	3.3	11
76	PCD Detect: enhancing ciliary features through image averaging and classification. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2020, 319, L1048-L1060.	2.9	10
77	Impact of T2R38 Receptor Polymorphisms on Pseudomonas aeruginosa Infection in Cystic Fibrosis. American Journal of Respiratory and Critical Care Medicine, 2018, 197, 1635-1638.	5.6	9
78	Exploring the Art of Ciliary Beating. Chest, 2017, 152, 1348-1349.	0.8	7
79	Physiological and Pathophysiological Aspects of Primary Ciliaâ€"A Literature Review with View on Functional and Structural Relationships in Cartilage. International Journal of Molecular Sciences, 2020, 21, 4959.	4.1	6
80	Applications of emerging transmission electron microscopy technology in PCD research and diagnosis. Ultrastructural Pathology, 2017, 41, 408-414.	0.9	5
81	Inflammatory molecular endotypes in bronchiectasis. , 2019, , .		5
82	Primary ciliary dyskinesia in adults with bronchiectasis: Data from the Embarc registry. , 2018, , .		4
83	A point of care neutrophil elastase activity assay identifies bronchiectasis severity, airway infection and risk of exacerbation. , 2019, , .		4
84	Response. Chest, 2019, 156, 1033-1034.	0.8	3
85	SPLUNC1 is a novel marker of disease severity and airway infection in bronchiectasis. European Respiratory Journal, 2021, 58, 2101840.	6.7	3
86	Airway clearance techniques in patients with bronchiectasis. Data from the EMBARC Registry. , 2020, , .		3
87	When to Think of Bronchiectasis and the Investigations to Perform. Clinical Pulmonary Medicine, 2010, 17, 7-13.	0.3	2
88	S88 Electron Tomography Detects Ultrastructural Abnormalities In Patients With Pcd Due To A Dnah11 Defect. Thorax, 2014, 69, A48-A49.	5.6	2
89	UA-Zero as a Uranyl Acetate Replacement When Diagnosing Primary Ciliary Dyskinesia by Transmission Electron Microscopy. Diagnostics, 2021, 11, 1063.	2.6	2
90	International consensus guideline for reporting transmission electron microscopy results in the diagnosis of Primary Ciliary Dyskinesia. , 2019, , .		2

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91	Diagnosis of primary ciliary dyskinesia: current practice and future perspectives. , 0, , 267-281.		2
92	Neutrophil Extracellular Traps are Increased in Severe Bronchiectasis and Reduced by Long-Term Azithromycin Treatment. SSRN Electronic Journal, 0, , .	0.4	2
93	A high prevalence CCDC103 p.His154Pro mutation causing primary ciliary dyskinesia is associated with normal diagnostic investigations. , 2017, , .		2
94	The efficacy and safety of inhaled antibiotics for the treatment of bronchiectasis in adults: a systematic review and meta-analysis. , 2019, , .		2
95	Proteinase-3 as a biomarker of exacerbations in bronchiectasis. , 2020, , .		2
96	The Study Of Primary Ciliary Dyskinesia In Difficult Cases Using Electron Tomography. , 2010, , .		1
97	S68â€…A longitudinal study characterising a large adult primary ciliary dyskinesia cohort. Thorax, 2015, 70, A40.2-A40.	5.6	1
98	Haemophilus influenzae biofilms in primary ciliary dyskinesia: a moving story. European Respiratory Journal, 2017, 50, 1701369.	6.7	1
99	Models of Ciliary Dysfunction: Time to Expand. American Journal of Respiratory Cell and Molecular Biology, 2018, 59, 285-286.	2.9	1
100	Airway Bacterial Load and Response to Inhaled Aztreonam in Bronchiectasis. , 2019, , .		1
101	AMPK is inhibited in severe bronchiectasis and may relate to reduced ciliary beat frequency. , 2018, , .		1
102	Endotyping of COPD, bronchiectasis and their overlap syndrome by integrated sputum proteome/microbiome. , 2019, , .		1
103	Characterisation of sputum pregnancy zone protein in bronchiectasis. , 2019, , .		1
104	Heparin Binding Protein in sputum compromises epithelial defence and relates to bronchiectasis severity. , 2019, , .		1
105	Exhaled Breath Condensate pH as a Non-invasive Measure of Inflammation in Non-CF Bronchiectasis. ISRN Pulmonology, 2011, 2011, 1-6.	0.3	1
106	Whatâ€™s important for people with NTM? An EMBARC-ELF patient survey. , 2020, , .		1
107	Placebo effects in pharmaceutical clinical trials in bronchiectasis: an EMBARC study. , 2020, , .		1
108	Inhaled antibiotics improve symptoms of cough and sputum in patients with bronchiectasis: a post-hoc analysis of the AIR-BX studies. , 2020, , .		1

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109	Heterogeneity of treatment response in bronchiectasis clinical trials. , 2020, , .		1
110	Yellow nail syndrome and bronchiectasis. Nigerian Journal of Surgical Research, 2002, 4, 115.	0.1	0
111	Diagnosis Of Inner Dynein Arm Defects As A Cause Of Primary Ciliary Dyskinesia. , 2010, , .		0
112	A Comparison Between Chlamydomonas Flagella And Human Cilia By Electron Tomography. , 2010, , .		0
113	P95â€…Assessment of F/HN-pseudotyped Lentivirus as a Clinically Relevant Vector For Lung Gene Therapy. Thorax, 2012, 67, A105.1-A105.	5.6	0
114	P239â€…A role for polycystins in airway mucociliary clearance?. Thorax, 2013, 68, A185.2-A185.	5.6	0
115	S125â€…A retrospective study characterising ciliary ultrastructure, light microscopy and sputum microbiology associations with lung function decline in a large adult primary ciliary dyskinesia cohort. Thorax, 2013, 68, A65.1-A65.	5.6	0
116	P82â€…Lung clearance index (LCI) and genotype-phenotype correlations in Primary Ciliary Dyskinesia (PCD). Thorax, 2015, 70, A116.2-A117.	5.6	0
117	S69â€…Development of anin vitroassay to detect chemically-induced changes in ciliary beat frequency. Thorax, 2015, 70, A40.3-A41.	5.6	0
118	S75â€…The T2R38 bitter taste receptor as a modifier of host response to pseudomonas aeruginosa in cystic fibrosis: does T2R38 genotype impact on clinical infection?. Thorax, 2016, 71, A44.2-A44.	5.6	0
119	S69â€…Genetic and structural characterisation of outer dynein arm variants causing primary ciliary dyskinesia. , 2017, , .		0
120	S42â€…Sex differences in reported quality of life in bronchiectasis: an analysis of the embarc registry. , 2017, , .		0
121	S88â€…Neutrophil elastase increases ciliary beat frequency ex-vivo: implications for the bronchiectasis airway. , 2018, , .		0
122	Improving Primary Ciliary Dyskinesia Diagnosis Using Artificial Intelligence. Microscopy and Microanalysis, 2020, 26, 2132-2132.	0.4	0
123	Lung clearance index is stable in most primary ciliary dyskinesia (PCD) patients managed in a specialist centre: A pilot study. , 2016, , .		0
124	Use of electron tomography to confirm the diagnosis of primary ciliary dyskinesia. , 2017, , .		0
125	Nasal cavity inflammation in patients with primary ciliary dyskinesia (PCD) is associated with bacterial infection. , 2017, , .		0
126	Genetic risk factors for laterality defects and congenital heart disease (CHD) in patients with primary ciliary dyskinesia (PCD). , 2017, , .		0

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127	Chest Physiotherapy in patients with Bronchiectasis â€“ what is the current practice in Europe?. , 2018, , .		0
128	Neutrophil elastase increases ciliary beat frequency ex-vivo: implications for the bronchiectasis airway. , 2018, , .		0
129	Validity of COPD diagnosis in Bronchiectasis patients: data from the EMBARC registry. , 2018, , .		0
130	Motile cilia structure and function in patients with mutations in the outer dynein arm heavy chain DNAH9. , 2018, , .		0
131	Accuracy of high-speed video microscopy to diagnose primary ciliary dyskinesia. , 2018, , .		0
132	Improving Primary Ciliary Dyskinesia diagnosis using Artificial Intelligence. , 2020, , .		0
133	Endotyping bronchiectasis through multi-omic profiling. , 2020, , .		0
134	Immunodeficiency associated bronchiectasis in the European Bronchiectasis Registry (EMBARC). , 2020, , .		0
135	Primary ciliary dyskinesia and non-CF bronchiectasis in the 100,000 Genomes Project. , 2020, , .		0
136	Sex related differences in aetiology, severity and quality of life in bronchiectasis: data from the EMBARC, EMBARC-India and Australian bronchiectasis registries. , 2020, , .		0
137	Alpha-1 antitrypsin deficiency in patients with bronchiectasis: data from the European Bronchiectasis Registry EMBARC. , 2020, , .		0