Hannah M Mitchison

List of Publications by Year
in descending order

106	9,767	38742	39675
papers	citations	50 h-index	94 g-index

112	112	112	11355
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Primary ciliary dyskinesia: a big data genomics approach. Lancet Respiratory Medicine,the, 2022, , .	10.7	6
2	Motile cilia and airway disease. Seminars in Cell and Developmental Biology, 2021, 110, 19-33.	5.0	87
3	Higher throughput drug screening for rare respiratory diseases: Readthrough therapy in primary ciliary dyskinesia. European Respiratory Journal, 2021, 58, 2000455.	6.7	13
4	Identification of a wide spectrum of ciliary gene mutations in nonsyndromic biliary atresia patients implicates ciliary dysfunction as a novel disease mechanism. EBioMedicine, 2021, 71, 103530.	6.1	32
5	Topological data analysis reveals genotypeâ€“phenotype relationships in primary ciliary dyskinesia. European Respiratory Journal, 2021, 58, 2002359.	6.7	49
6	Development and first results of the BEAT-PCD international Primary Ciliary Dyskinesia gene variant database: CiliaVar. , 2021, , .		1
7	Clinical and genetic spectrum in 33 Egyptian families with suspected primary ciliary dyskinesia. Clinical Genetics, 2020, 97, 509-515.	2.0	20
8	Sperm defects in primary ciliary dyskinesia and related causes of male infertility. Cellular and Molecular Life Sciences, 2020, 77, 2029-2048.	5.4	140
9	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
10	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
11	Hydrocephalus and diffuse choroid plexus hyperplasia in primary ciliary dyskinesia-related MCIDAS mutation. Neurology: Genetics, 2020, 6, e482.	1.9	24
12	Primary ciliary dyskinesia and non-CF bronchiectasis in the 100,000 Genomes Project. , 2020, , .		0
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	Mechanical loading inhibits cartilage inflammatory signalling via an HDAC6 and IFT-dependent mechanism regulating primary cilia elongation. Osteoarthritis and Cartilage, 2019, 27, 1064-1074.	1.3	58
15	Opportunities and Challenges for Molecular Understanding of Ciliopathiesâ€“The 100,000 Genomes Project. Frontiers in Genetics, 2019, 10, 127.	2.3	71
16	Risk factors for situs defects and congenital heart disease in primary ciliary dyskinesia. Thorax, 2019, 74, 203-205.	5.6	52
17	Primary ciliary dyskinesia with normal ultrastructure: three-dimensional tomography detects absence of DNAH11. European Respiratory Journal, 2018, 51, 1701809.	6.7	33
18	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	2.9	45

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19	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
20	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
21	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
22	Severe skeletal abnormalities caused by defects in retrograde intraflagellar transport dyneins. , 2018, , 356-401.		7
23	Altered Cerebellar Short-Term Plasticity but No Change in Postsynaptic AMPA-Type Glutamate Receptors in a Mouse Model of Juvenile Batten Disease. ENeuro, 2018, 5, ENEURO.0387-17.2018.	1.9	5
24	Motile cilia structure and function in patients with mutations in the outer dynein arm heavy chain DNAH9. , 2018, , .		0
25	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
26	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor PIH1D3. Nature Communications, 2017, 8, 14279.	12.8	133
27	Update on primary ciliary dyskinesia. Paediatrics and Child Health (United Kingdom), 2017, 27, 337-342.	0.4	2
28	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
29	Motile and nonâ€motile cilia in human pathology: from function to phenotypes. Journal of Pathology, 2017, 241, 294-309.	4.5	341
30	Glial cells are functionally impaired in juvenile neuronal ceroid lipofuscinosis and detrimental to neurons. Acta Neuropathologica Communications, 2017, 5, 74.	5.2	57
31	A high prevalence CCDC103 p.His154Pro mutation causing primary ciliary dyskinesia is associated with normal diagnostic investigations. , 2017, , .		2
32	Genetic risk factors for laterality defects and congenital heart disease (CHD) in patients with primary ciliary dyskinesia (PCD). , 2017, , .		0
33	A founder CEP120 mutation in Jeune asphyxiating thoracic dystrophy expands the role of centriolar proteins in skeletal ciliopathies. Human Molecular Genetics, 2015, 24, 1410-1419.	2.9	70
34	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215
35	The more we know, the more we have to discover: an exciting future for understanding cilia and ciliopathies. Cilia, 2015, 4, 5.	1.8	8
36	Photoreceptor phagosome processing defects and disturbed autophagy in retinal pigment epithelium ofCln3^{Î”ex1-6}mice modelling juvenile neuronal ceroid lipofuscinosis (Batten) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 5	2.9	81

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37	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	12.8	51
38	The UK10K project identifies rare variants in health and disease. Nature, 2015, 526, 82-90.	27.8	1,014
39	HEATR2 Plays a Conserved Role in Assembly of the Ciliary Motile Apparatus. PLoS Genetics, 2014, 10, e1004577.	3.5	67
40	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
41	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
42	CCDC151 Mutations Cause Primary Ciliary Dyskinesia by Disruption of the Outer Dynein Arm Docking Complex Formation. American Journal of Human Genetics, 2014, 95, 257-274.	6.2	149
43	MCIDAS mutations result in a mucociliary clearance disorder with reduced generation of multiple motile cilia. Nature Communications, 2014, 5, 4418.	12.8	221
44	Diagnosis and management of primary ciliary dyskinesia. Archives of Disease in Childhood, 2014, 99, 850-856.	1.9	216
45	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923.	6.2	79
46	DYX1C1 is required for axonemal dynein assembly and ciliary motility. Nature Genetics, 2013, 45, 995-1003.	21.4	256
47	Short-Rib Polydactyly and Jeune Syndromes Are Caused by Mutations in WDR60. American Journal of Human Genetics, 2013, 93, 515-523.	6.2	116
48	Mutations in the Gene Encoding IFT Dynein Complex Component WDR34 Cause Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2013, 93, 932-944.	6.2	108
49	Bioinformatic perspectives in the neuronal ceroid lipofuscinoses. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2013, 1832, 1831-1841.	3.8	10
50	Combined <scp>NGS</scp> Approaches Identify Mutations in the Intraflagellar Transport Gene IFT140 in Skeletal Ciliopathies with Early Progressive Kidney Disease. Human Mutation, 2013, 34, 714-724.	2.5	120
51	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
52	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
53	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
54	Exome sequencing identifiesDYNC2H1mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. Journal of Medical Genetics, 2013, 50, 309-323.	3.2	127

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55	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
56	CCDC103 mutations cause primary ciliary dyskinesia by disrupting assembly of ciliary dynein arms. Nature Genetics, 2012, 44, 714-719.	21.4	228
57	Mutations in axonemal dynein assembly factor DNAAF3 cause primary ciliary dyskinesia. Nature Genetics, 2012, 44, 381-389.	21.4	231
58	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
59	A metabolomic comparison of mouse models of the Neuronal Ceroid Lipofuscinoses. Journal of Biomolecular NMR, 2011, 49, 175-184.	2.8	11
60	Founder Mutation(s) in theRSPH9Gene Leading to Primary Ciliary Dyskinesia in Two Inbred Bedouin Families. Annals of Human Genetics, 2010, 74, 117-125.	0.8	19
61	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
62	Update in Primary Ciliary Dyskinesia. Clinical Pulmonary Medicine, 2009, 16, 219-225.	0.3	2
63	Genome-wide High-Density SNP-Based Linkage Analysis of Infantile Hypertrophic Pyloric Stenosis Identifies Loci on Chromosomes 11q14-q22 and Xq23. American Journal of Human Genetics, 2008, 82, 756-762.	6.2	51
64	DNAI2 Mutations Cause Primary Ciliary Dyskinesia with Defects in the Outer Dynein Arm. American Journal of Human Genetics, 2008, 83, 547-558.	6.2	242
65	Loss of the Batten Disease Gene CLN3 Prevents Exit from the TGN of the Mannose 6â€Phosphate Receptor. Traffic, 2008, 9, 1905-1914.	2.7	68
66	Transcript and in silico analysis of CLN3 in juvenile neuronal ceroid lipofuscinosis and associated mouse models. Human Molecular Genetics, 2008, 17, 3332-3339.	2.9	31
67	Primary ciliary dyskinesia: current state of the art. Archives of Disease in Childhood, 2007, 92, 1136-1140.	1.9	311
68	Increased expression of lysosomal acid phosphatase in CLN3-defective cells and mouse brain tissue. Journal of Neurochemistry, 2007, 103, 2177-2188.	3.9	30
69	Strategies for data analyses in a high resolution 1H NMR based metabolomics study of a mouse model of Batten disease. Metabolomics, 2007, 3, 121-136.	3.0	9
70	Progress towards understanding disease mechanisms in small vertebrate models of neuronal ceroid lipofuscinosis. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 873-889.	3.8	90
71	Batten disease (JNCL) is linked to disturbances in mitochondrial, cytoskeletal, and synaptic compartments. Journal of Neuroscience Research, 2006, 84, 1124-1138.	2.9	65
72	Mutations ofDNAI1in Primary Ciliary Dyskinesia. American Journal of Respiratory and Critical Care Medicine, 2006, 174, 858-866.	5.6	162

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73	High Resolution 1H NMR-based Metabolomics Indicates a Neurotransmitter Cycling Deficit in Cerebral Tissue from a Mouse Model of Batten Disease. Journal of Biological Chemistry, 2005, 280, 42508-42514.	3.4	135
74	Identification and Analysis of Axonemal Dynein Light Chain 1 in Primary Ciliary Dyskinesia Patients. American Journal of Respiratory Cell and Molecular Biology, 2005, 33, 41-47.	2.9	52
75	CLN3L, a novel protein related to the Batten disease protein, is overexpressed in Cln3-/- mice and in Batten disease. Brain, 2004, 127, 1748-1754.	7.6	12
76	Selectivity and Types of Cell Death in the Neuronal Ceroid Lipofuscinoses (NCLs). Brain Pathology, 2004, 14, 86-96.	4.1	80
77	Late onset neurodegeneration in the Cln3âˆ'/âˆ' mouse model of juvenile neuronal ceroid lipofuscinosis is preceded by low level glial activation. Brain Research, 2004, 1023, 231-242.	2.2	139
78	Primary ciliary dyskinesia (Siewert's / Kartagener's Syndrome): Respiratory symptoms and psycho-social impact. BMC Pulmonary Medicine, 2003, 3, 4.	2.0	57
79	Functional categorization of gene expression changes in the cerebellum of a Cln3-knockout mouse model for Batten disease. Molecular Genetics and Metabolism, 2003, 78, 17-30.	1.1	28
80	Association between p47phox pseudogenes and inflammatory bowel disease. Blood, 2003, 101, 3337-3337.	1.4	13
81	Mutations in the DNAH11 (axonemal heavy chain dynein type 11) gene cause one form of situs inversus totalis and most likely primary ciliary dyskinesia. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 10282-10286.	7.1	329
82	Retinal Pathology and Function in a Cln3 Knockout Mouse Model of Juvenile Neuronal Ceroid Lipofuscinosis (Batten Disease). Molecular and Cellular Neurosciences, 2002, 19, 515-527.	2.2	58
83	Genetics and development. Current Opinion in Genetics and Development, 2002, 12, 373-379.	3.3	0
84	Genetics and development. Current Opinion in Genetics and Development, 2002, 12, 489-495.	3.3	0
85	Genetics and development. Current Opinion in Genetics and Development, 2002, 12, 621-627.	3.3	0
86	Mutations in DNAH5 cause primary ciliary dyskinesia and randomization of leftâ€“right asymmetry. Nature Genetics, 2002, 30, 143-144.	21.4	496
87	Paper alert: Genetics and development. Current Opinion in Genetics and Development, 2001, 11, 601-606.	3.3	0
88	Neurodegenerative disease: the neuronal ceroid lipofuscinoses (Batten disease). Current Opinion in Neurology, 2001, 14, 795-803.	3.6	43
89	High resolution MRI reveals global changes in brains of Cln3 mutant mice. European Journal of Paediatric Neurology, 2001, 5, 103-107.	1.6	13
90	Developmental expression of palmitoyl protein thioesterase in normal mice. Developmental Brain Research, 1999, 118, 1-11.	1.7	45

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91	Molecular basis of the neuronal ceroid lipofuscinoses: Mutations inCLN1,CLN2,CLN3, andCLN5. Human Mutation, 1999, 14, 199-215.	2.5	54
92	Targeted Disruption of the Cln3 Gene Provides a Mouse Model for Batten Disease. Neurobiology of Disease, 1999, 6, 321-334.	4.4	180
93	Molecular basis of the neuronal ceroid lipofuscinoses: Mutations in CLN1, CLN2, CLN3, and CLN5. Human Mutation, 1999, 14, 199.	2.5	4
94	Genetic Linkage Analysis of a Variant of Juvenile Onset Neuronal Ceroid Lipofuscinosis with Granular Osmiophilic Deposits. Neuropediatrics, 1997, 28, 21-22.	0.6	15
95	Structure of the CLN3 Gene and Predicted Structure, Location and Function of CLN3 Protein. Neuropediatrics, 1997, 28, 12-14.	0.6	18
96	Strategy for Mutation Detection in CLN3: Characterisation of Two Finnish Mutations. Neuropediatrics, 1997, 28, 15-17.	0.6	12
97	Genomic Structure and Complete Nucleotide Sequence of the Batten Disease Gene,CLN3. Genomics, 1997, 40, 346-350.	2.9	47
98	Spectrum of Mutations in the Batten Disease Gene, CLN3. American Journal of Human Genetics, 1997, 61, 310-316.	6.2	181
99	NON-CLASSICAL-MHC GENETICS OF IMMUNOLOGICAL DISEASE IN MAN AND MOUSE. THE KEY ROLE OF PRO-INFLAMMATORY CYTOKINE GENES. Cytokine, 1996, 8, 593-597.	3.2	40
100	A model for Batten disease protein CLN3: Functional implications from homology and mutations. FEBS Letters, 1996, 399, 75-77.	2.8	71
101	Physical map of the region containing the gene for Batten disease (CLN3). American Journal of Medical Genetics Part A, 1995, 57, 316-319.	2.4	10
102	Analysis of Batten disease candidate genesSTP andSTM. American Journal of Medical Genetics Part A, 1995, 57, 324-326.	2.4	2
103	Phenol sulfotransferases: Candidate genes for Batten disease. American Journal of Medical Genetics Part A, 1995, 57, 327-332.	2.4	4
104	YAC and Cosmid Contigs Spanning the Batten Disease (CLN3) Region at 16p12.1â€“p11.2. Genomics, 1995, 29, 478-489.	2.9	8
105	Genetic Mapping of the Batten Disease Locus (CLN3) to the Interval D16S288-D16S383 by Analysis of Haplotypes and Allelic Association. Genomics, 1994, 22, 465-468.	2.9	33
106	Fine Genetic Mapping of the Batten Disease Locus (CLN3) by Haplotype Analysis and Demonstration of Allelic Association with Chromosome 16p Microsatellite Loci. Genomics, 1993, 16, 455-460.	2.9	45