

Hannah M Mitchison

List of Publications by Year in descending order

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106
papers

9,767
citations

38742

50
h-index

39675

94
g-index

112
all docs

112
docs citations

112
times ranked

11355
citing authors

#	ARTICLE	IF	CITATIONS
1	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 2015, 526, 82-90.	27.8	1,014
2	Mutations in DNAH5 cause primary ciliary dyskinesia and randomization of left–right asymmetry. <i>Nature Genetics</i> , 2002, 30, 143-144.	21.4	496
3	Motile and non–motile cilia in human pathology: from function to phenotypes. <i>Journal of Pathology</i> , 2017, 241, 294-309.	4.5	341
4	Mutations in the <i>DNAH11</i> (axonemal heavy chain dynein type 11) gene cause one form of situs inversus totalis and most likely primary ciliary dyskinesia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2002, 99, 10282-10286.	7.1	329
5	Primary ciliary dyskinesia: current state of the art. <i>Archives of Disease in Childhood</i> , 2007, 92, 1136-1140.	1.9	311
6	Mutations in Radial Spoke Head Protein Genes RSPH9 and RSPH4A Cause Primary Ciliary Dyskinesia with Central-Microtubular-Pair Abnormalities. <i>American Journal of Human Genetics</i> , 2009, 84, 197-209.	6.2	303
7	DYX1C1 is required for axonemal dynein assembly and ciliary motility. <i>Nature Genetics</i> , 2013, 45, 995-1003.	21.4	256
8	Recessive HYDIN Mutations Cause Primary Ciliary Dyskinesia without Randomization of Left-Right Body Asymmetry. <i>American Journal of Human Genetics</i> , 2012, 91, 672-684.	6.2	252
9	DNAI2 Mutations Cause Primary Ciliary Dyskinesia with Defects in the Outer Dynein Arm. <i>American Journal of Human Genetics</i> , 2008, 83, 547-558.	6.2	242
10	Mutations in axonemal dynein assembly factor DNAAF3 cause primary ciliary dyskinesia. <i>Nature Genetics</i> , 2012, 44, 381-389.	21.4	231
11	CCDC103 mutations cause primary ciliary dyskinesia by disrupting assembly of ciliary dynein arms. <i>Nature Genetics</i> , 2012, 44, 714-719.	21.4	228
12	MCIDAS mutations result in a mucociliary clearance disorder with reduced generation of multiple motile cilia. <i>Nature Communications</i> , 2014, 5, 4418.	12.8	221
13	Diagnosis and management of primary ciliary dyskinesia. <i>Archives of Disease in Childhood</i> , 2014, 99, 850-856.	1.9	216
14	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. <i>Nature Cell Biology</i> , 2015, 17, 1074-1087.	10.3	215
15	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. <i>American Journal of Human Genetics</i> , 2013, 93, 915-925.	6.2	196
16	Spectrum of Mutations in the Batten Disease Gene, CLN3. <i>American Journal of Human Genetics</i> , 1997, 61, 310-316.	6.2	181
17	Targeted Disruption of the Cln3 Gene Provides a Mouse Model for Batten Disease. <i>Neurobiology of Disease</i> , 1999, 6, 321-334.	4.4	180
18	Splice-Site Mutations in the Axonemal Outer Dynein Arm Docking Complex Gene CCDC114 Cause Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 92, 88-98.	6.2	176

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19	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. <i>Human Mutation</i> , 2013, 34, 462-472.	2.5	176
20	Mutations in <i>ZMYND10</i> , a Gene Essential for Proper Axonemal Assembly of Inner and Outer Dynein Arms in Humans and Flies, Cause Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 93, 346-356.	6.2	167
21	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
22	<i>CCDC151</i> Mutations Cause Primary Ciliary Dyskinesia by Disruption of the Outer Dynein Arm Docking Complex Formation. <i>American Journal of Human Genetics</i> , 2014, 95, 257-274.	6.2	149
23	Sperm defects in primary ciliary dyskinesia and related causes of male infertility. <i>Cellular and Molecular Life Sciences</i> , 2020, 77, 2029-2048.	5.4	140
24	Late onset neurodegeneration in the <i>Cln3^{Δ/Δ}</i> mouse model of juvenile neuronal ceroid lipofuscinosis is preceded by low level glial activation. <i>Brain Research</i> , 2004, 1023, 231-242.	2.2	139
25	High Resolution ¹ H NMR-based Metabolomics Indicates a Neurotransmitter Cycling Deficit in Cerebral Tissue from a Mouse Model of Batten Disease. <i>Journal of Biological Chemistry</i> , 2005, 280, 42508-42514.	3.4	135
26	X-linked primary ciliary dyskinesia due to mutations in the cytoplasmic axonemal dynein assembly factor <i>PIH1D3</i> . <i>Nature Communications</i> , 2017, 8, 14279.	12.8	133
27	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
28	Exome sequencing identifies <i>DYNC2H1</i> mutations as a common cause of asphyxiating thoracic dystrophy (Jeune syndrome) without major polydactyly, renal or retinal involvement. <i>Journal of Medical Genetics</i> , 2013, 50, 309-323.	3.2	127
29	Combined <i>sc</i> NGS Approaches Identify Mutations in the Intraflagellar Transport Gene <i>IFT140</i> in Skeletal Ciliopathies with Early Progressive Kidney Disease. <i>Human Mutation</i> , 2013, 34, 714-724.	2.5	120
30	Short-Rib Polydactyly and Jeune Syndromes Are Caused by Mutations in <i>WDR60</i> . <i>American Journal of Human Genetics</i> , 2013, 93, 515-523.	6.2	116
31	Mutations in the Gene Encoding IFT Dynein Complex Component <i>WDR34</i> Cause Jeune Asphyxiating Thoracic Dystrophy. <i>American Journal of Human Genetics</i> , 2013, 93, 932-944.	6.2	108
32	Accuracy of Immunofluorescence in the Diagnosis of Primary Ciliary Dyskinesia. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2017, 196, 94-101.	5.6	97
33	Mutations in Outer Dynein Arm Heavy Chain <i>DNAH9</i> Cause Motile Cilia Defects and Situs Inversus. <i>American Journal of Human Genetics</i> , 2018, 103, 984-994.	6.2	95
34	Progress towards understanding disease mechanisms in small vertebrate models of neuronal ceroid lipofuscinosis. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 873-889.	3.8	90
35	Combined exome and whole-genome sequencing identifies mutations in <i>ARMC4</i> as a cause of primary ciliary dyskinesia with defects in the outer dynein arm. <i>Journal of Medical Genetics</i> , 2014, 51, 61-67.	3.2	88
36	Motile cilia and airway disease. <i>Seminars in Cell and Developmental Biology</i> , 2021, 110, 19-33.	5.0	87

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37	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. <i>Human Molecular Genetics</i> , 2014, 23, 3362-3374.	2.9	82
38	Selectivity and Types of Cell Death in the Neuronal Ceroid Lipofuscinoses (NCLs). <i>Brain Pathology</i> , 2004, 14, 86-96.	4.1	80
39	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. <i>American Journal of Human Genetics</i> , 2014, 94, 915-923.	6.2	79
40	A model for Batten disease protein CLN3: Functional implications from homology and mutations. <i>FEBS Letters</i> , 1996, 399, 75-77.	2.8	71
41	Opportunities and Challenges for Molecular Understanding of Ciliopathies—The 100,000 Genomes Project. <i>Frontiers in Genetics</i> , 2019, 10, 127.	2.3	71
42	A founder CEP120 mutation in Jeune asphyxiating thoracic dystrophy expands the role of centriolar proteins in skeletal ciliopathies. <i>Human Molecular Genetics</i> , 2015, 24, 1410-1419.	2.9	70
43	Loss of the Batten Disease Gene CLN3 Prevents Exit from the TGN of the Mannose 6-Phosphate Receptor. <i>Traffic</i> , 2008, 9, 1905-1914.	2.7	68
44	HEATR2 Plays a Conserved Role in Assembly of the Ciliary Motile Apparatus. <i>PLoS Genetics</i> , 2014, 10, e1004577.	3.5	67
45	Batten disease (JNCL) is linked to disturbances in mitochondrial, cytoskeletal, and synaptic compartments. <i>Journal of Neuroscience Research</i> , 2006, 84, 1124-1138.	2.9	65
46	High prevalence of <i>CCDC103</i> p.His154Pro mutation causing primary ciliary dyskinesia disrupts protein oligomerisation and is associated with normal diagnostic investigations. <i>Thorax</i> , 2018, 73, 157-166.	5.6	63
47	Retinal Pathology and Function in a Cln3 Knockout Mouse Model of Juvenile Neuronal Ceroid Lipofuscinosis (Batten Disease). <i>Molecular and Cellular Neurosciences</i> , 2002, 19, 515-527.	2.2	58
48	Mechanical loading inhibits cartilage inflammatory signalling via an HDAC6 and IFT-dependent mechanism regulating primary cilia elongation. <i>Osteoarthritis and Cartilage</i> , 2019, 27, 1064-1074.	1.3	58
49	Primary ciliary dyskinesia (Siewert's / Kartagener's Syndrome): Respiratory symptoms and psycho-social impact. <i>BMC Pulmonary Medicine</i> , 2003, 3, 4.	2.0	57
50	Glial cells are functionally impaired in juvenile neuronal ceroid lipofuscinosis and detrimental to neurons. <i>Acta Neuropathologica Communications</i> , 2017, 5, 74.	5.2	57
51	Molecular basis of the neuronal ceroid lipofuscinoses: Mutations in CLN1, CLN2, CLN3, and CLN5. <i>Human Mutation</i> , 1999, 14, 199-215.	2.5	54
52	Identification and Analysis of Axonemal Dynein Light Chain 1 in Primary Ciliary Dyskinesia Patients. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 2005, 33, 41-47.	2.9	52
53	Risk factors for situs defects and congenital heart disease in primary ciliary dyskinesia. <i>Thorax</i> , 2019, 74, 203-205.	5.6	52
54	Genome-wide High-Density SNP-Based Linkage Analysis of Infantile Hypertrophic Pyloric Stenosis Identifies Loci on Chromosomes 11q14-q22 and Xq23. <i>American Journal of Human Genetics</i> , 2008, 82, 756-762.	6.2	51

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55	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. <i>Nature Communications</i> , 2015, 6, 7074.	12.8	51
56	C11orf70 Mutations Disrupting the Intraflagellar Transport-Dependent Assembly of Multiple Axonemal Dyneins Cause Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2018, 102, 956-972.	6.2	51
57	Clinical utility of NGS diagnosis and disease stratification in a multiethnic primary ciliary dyskinesia cohort. <i>Journal of Medical Genetics</i> , 2020, 57, 322-330.	3.2	50
58	Topological data analysis reveals genotype-phenotype relationships in primary ciliary dyskinesia. <i>European Respiratory Journal</i> , 2021, 58, 2002359.	6.7	49
59	Genomic Structure and Complete Nucleotide Sequence of the Batten Disease Gene, CLN3. <i>Genomics</i> , 1997, 40, 346-350.	2.9	47
60	Fine Genetic Mapping of the Batten Disease Locus (CLN3) by Haplotype Analysis and Demonstration of Allelic Association with Chromosome 16p Microsatellite Loci. <i>Genomics</i> , 1993, 16, 455-460.	2.9	45
61	Developmental expression of palmitoyl protein thioesterase in normal mice. <i>Developmental Brain Research</i> , 1999, 118, 1-11.	1.7	45
62	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. <i>Human Molecular Genetics</i> , 2018, 27, 529-545.	2.9	45
63	Neurodegenerative disease: the neuronal ceroid lipofuscinoses (Batten disease). <i>Current Opinion in Neurology</i> , 2001, 14, 795-803.	3.6	43
64	NON-CLASSICAL-MHC GENETICS OF IMMUNOLOGICAL DISEASE IN MAN AND MOUSE. THE KEY ROLE OF PRO-INFLAMMATORY CYTOKINE GENES. <i>Cytokine</i> , 1996, 8, 593-597.	3.2	40
65	Genetic Mapping of the Batten Disease Locus (CLN3) to the Interval D16S288-D16S383 by Analysis of Haplotypes and Allelic Association. <i>Genomics</i> , 1994, 22, 465-468.	2.9	33
66	Primary ciliary dyskinesia with normal ultrastructure: three-dimensional tomography detects absence of DNAH11. <i>European Respiratory Journal</i> , 2018, 51, 1701809.	6.7	33
67	Identification of a wide spectrum of ciliary gene mutations in nonsyndromic biliary atresia patients implicates ciliary dysfunction as a novel disease mechanism. <i>EBioMedicine</i> , 2021, 71, 103530.	6.1	32
68	Transcript and in silico analysis of CLN3 in juvenile neuronal ceroid lipofuscinosis and associated mouse models. <i>Human Molecular Genetics</i> , 2008, 17, 3332-3339.	2.9	31
69	Photoreceptor phagosome processing defects and disturbed autophagy in retinal pigment epithelium of <i>Cln3</i> ^{ex1-6} mice modelling juvenile neuronal ceroid lipofuscinosis (Batten) <i>Tj ETQq1 1 0.784314 rgBT / Overlock</i>	3.4	31
70	Increased expression of lysosomal acid phosphatase in CLN3-defective cells and mouse brain tissue. <i>Journal of Neurochemistry</i> , 2007, 103, 2177-2188.	3.9	30
71	Functional categorization of gene expression changes in the cerebellum of a <i>Cln3</i> -knockout mouse model for Batten disease. <i>Molecular Genetics and Metabolism</i> , 2003, 78, 17-30.	1.1	28
72	Hydrocephalus and diffuse choroid plexus hyperplasia in primary ciliary dyskinesia-related MCIDAS mutation. <i>Neurology: Genetics</i> , 2020, 6, e482.	1.9	24

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73	Clinical and genetic spectrum in 33 Egyptian families with suspected primary ciliary dyskinesia. <i>Clinical Genetics</i> , 2020, 97, 509-515.	2.0	20
74	Founder Mutation(s) in the <i>RSPH9</i> Gene Leading to Primary Ciliary Dyskinesia in Two Inbred Bedouin Families. <i>Annals of Human Genetics</i> , 2010, 74, 117-125.	0.8	19
75	Structure of the CLN3 Gene and Predicted Structure, Location and Function of CLN3 Protein. <i>Neuropediatrics</i> , 1997, 28, 12-14.	0.6	18
76	Genetic Linkage Analysis of a Variant of Juvenile Onset Neuronal Ceroid Lipofuscinosis with Granular Osmiophilic Deposits. <i>Neuropediatrics</i> , 1997, 28, 21-22.	0.6	15
77	High resolution MRI reveals global changes in brains of Cln3 mutant mice. <i>European Journal of Paediatric Neurology</i> , 2001, 5, 103-107.	1.6	13
78	Association between p47phox pseudogenes and inflammatory bowel disease. <i>Blood</i> , 2003, 101, 3337-3337.	1.4	13
79	Higher throughput drug screening for rare respiratory diseases: Readthrough therapy in primary ciliary dyskinesia. <i>European Respiratory Journal</i> , 2021, 58, 2000455.	6.7	13
80	Strategy for Mutation Detection in CLN3: Characterisation of Two Finnish Mutations. <i>Neuropediatrics</i> , 1997, 28, 15-17.	0.6	12
81	CLN3L, a novel protein related to the Batten disease protein, is overexpressed in Cln3 ^{-/-} mice and in Batten disease. <i>Brain</i> , 2004, 127, 1748-1754.	7.6	12
82	Motile cilia defects in diseases other than primary ciliary dyskinesia: The contemporary diagnostic and research role for transmission electron microscopy. <i>Ultrastructural Pathology</i> , 2017, 41, 415-427.	0.9	12
83	A metabolomic comparison of mouse models of the Neuronal Ceroid Lipofuscinoses. <i>Journal of Biomolecular NMR</i> , 2011, 49, 175-184.	2.8	11
84	Physical map of the region containing the gene for Batten disease (CLN3). <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 316-319.	2.4	10
85	Bioinformatic perspectives in the neuronal ceroid lipofuscinoses. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2013, 1832, 1831-1841.	3.8	10
86	PCD Detect: enhancing ciliary features through image averaging and classification. <i>American Journal of Physiology - Lung Cellular and Molecular Physiology</i> , 2020, 319, L1048-L1060.	2.9	10
87	Strategies for data analyses in a high resolution 1H NMR based metabolomics study of a mouse model of Batten disease. <i>Metabolomics</i> , 2007, 3, 121-136.	3.0	9
88	YAC and Cosmid Contigs Spanning the Batten Disease (CLN3) Region at 16p12.1â€“p11.2. <i>Genomics</i> , 1995, 29, 478-489.	2.9	8
89	The more we know, the more we have to discover: an exciting future for understanding cilia and ciliopathies. <i>Cilia</i> , 2015, 4, 5.	1.8	8
90	Severe skeletal abnormalities caused by defects in retrograde intraflagellar transport dyneins. , 2018, , 356-401.		7

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91	Primary ciliary dyskinesia: a big data genomics approach. <i>Lancet Respiratory Medicine</i> , 2022, , .	10.7	6
92	Altered Cerebellar Short-Term Plasticity but No Change in Postsynaptic AMPA-Type Glutamate Receptors in a Mouse Model of Juvenile Batten Disease. <i>ENeuro</i> , 2018, 5, ENEURO.0387-17.2018.	1.9	5
93	Phenol sulfotransferases: Candidate genes for Batten disease. <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 327-332.	2.4	4
94	Molecular basis of the neuronal ceroid lipofuscinoses: Mutations in CLN1, CLN2, CLN3, and CLN5. <i>Human Mutation</i> , 1999, 14, 199.	2.5	4
95	Analysis of Batten disease candidate genes STP and STM. <i>American Journal of Medical Genetics Part A</i> , 1995, 57, 324-326.	2.4	2
96	Update in Primary Ciliary Dyskinesia. <i>Clinical Pulmonary Medicine</i> , 2009, 16, 219-225.	0.3	2
97	Update on primary ciliary dyskinesia. <i>Paediatrics and Child Health (United Kingdom)</i> , 2017, 27, 337-342.	0.4	2
98	A high prevalence CCDC103 p.His154Pro mutation causing primary ciliary dyskinesia is associated with normal diagnostic investigations. , 2017, , .		2
99	Development and first results of the BEAT-PCD international Primary Ciliary Dyskinesia gene variant database: CiliaVar. , 2021, , .		1
100	Paper alert: Genetics and development. <i>Current Opinion in Genetics and Development</i> , 2001, 11, 601-606.	3.3	0
101	Genetics and development. <i>Current Opinion in Genetics and Development</i> , 2002, 12, 373-379.	3.3	0
102	Genetics and development. <i>Current Opinion in Genetics and Development</i> , 2002, 12, 489-495.	3.3	0
103	Genetics and development. <i>Current Opinion in Genetics and Development</i> , 2002, 12, 621-627.	3.3	0
104	Genetic risk factors for laterality defects and congenital heart disease (CHD) in patients with primary ciliary dyskinesia (PCD). , 2017, , .		0
105	Motile cilia structure and function in patients with mutations in the outer dynein arm heavy chain DNAH9. , 2018, , .		0
106	Primary ciliary dyskinesia and non-CF bronchiectasis in the 100,000 Genomes Project. , 2020, , .		0