Gabrielle Wheway

List of Publications by Year
in descending order

38 papers	2,513 citations	331259 21 h-index	344852 36 g-index
48 all docs	48 docs citations	48 times ranked	4502 citing authors

#	Article	IF	Citations
1	Molecular diagnoses in the congenital malformations caused by ciliopathies cohort of the 100,000 Genomes Project. Journal of Medical Genetics, 2022, 59, 737-747.	1.5	11
2	Regulation of canonical Wnt signalling by the ciliopathy protein MKS1 and the E2 ubiquitin-conjugating enzyme UBE2E1. ELife, 2022, 11, .	2.8	4
3	Unlocking the potential of the <scp>UK</scp> 100,000 Genomes Projectâ€"lessons learned from analysis of the â€œCongenital Malformations caused by Ciliopathiesâ€•cohort. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 5-8.	0.7	2
4	A CRISPR and high-content imaging assay compliant with ACMG/AMP guidelines for clinical variant interpretation in ciliopathies. Human Genetics, 2021, 140, 593-607.	1.8	6
5	A novel ACE2 isoform is expressed in human respiratory epithelia and is upregulated in response to interferons and RNA respiratory virus infection. Nature Genetics, 2021, 53, 205-214.	9.4	125
6	Biallelic variants in COPB1 cause a novel, severe intellectual disability syndrome with cataracts and variable microcephaly. Genome Medicine, 2021, 13, 34.	3.6	18
7	Whole genome sequencing in the diagnosis of primary ciliary dyskinesia. BMC Medical Genomics, 2021, 14, 234.	0.7	15
8	An updated SYSCILIA gold standard (SCGSv2) of known ciliary genes, revealing the vast progress that has been made in the cilia research field. Molecular Biology of the Cell, 2021, 32, br13.	0.9	23
9	WT1 activates transcription of the splice factor kinase SRPK1 gene in PC3 and K562 cancer cells in the absence of corepressor BASP1. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2020, 1863, 194642.	0.9	14
10	DYNC2H1 hypomorphic or retina-predominant variants cause nonsyndromic retinal degeneration. Genetics in Medicine, 2020, 22, 2041-2051.	1.1	38
11	Genomic programming of IRF4-expressing human Langerhans cells. Nature Communications, 2020, 11, 313.	5.8	22
12	Mutation spectrum of PRPF31, genotype-phenotype correlation in retinitis pigmentosa, and opportunities for therapy. Experimental Eye Research, 2020, 192, 107950.	1.2	36
13	Splicing in the pathogenesis, diagnosis and treatment of ciliopathies. Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms, 2019, 1862, 194433.	0.9	25
14	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	1.1	104
15	661W Photoreceptor Cell Line as a Cell Model for Studying Retinal Ciliopathies. Frontiers in Genetics, 2019, 10, 308.	1.1	37
16	Opportunities and Challenges for Molecular Understanding of Ciliopathiesâ€"The 100,000 Genomes Project. Frontiers in Genetics, 2019, 10, 127.	1.1	71
17	A Combined in silico, in vitro and Clinical Approach to Characterize Novel Pathogenic Missense Variants in PRPF31 in Retinitis Pigmentosa. Frontiers in Genetics, 2019, 10, 248.	1.1	7
18	The ciliary Frizzled-like receptor Tmem67 regulates canonical Wnt/Î²-catenin signalling in the developing cerebellum via Hoxb5. Scientific Reports, 2019, 9, 5446.	1.6	15

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19	Disrupted alternative splicing for genes implicated in splicing and ciliogenesis causes PRPF31 retinitis pigmentosa. Nature Communications, 2018, 9, 4234.	5.8	158
20	Development and biological evaluation of fluorophosphonate-modified hydroxyapatite for orthopaedic applications. Journal of Materials Science: Materials in Medicine, 2018, 29, 122.	1.7	7
21	Signaling through the Primary Cilium. Frontiers in Cell and Developmental Biology, 2018, 6, 8.	1.8	353
22	Particle sorting by Paramecium cilia arrays. BioSystems, 2017, 156-157, 46-52.	0.9	13
23	Meckelâ€“Gruber Syndrome: An Update on Diagnosis, Clinical Management, and Research Advances. Frontiers in Pediatrics, 2017, 5, 244.	0.9	107
24	The Meckel-Gruber syndrome protein TMEM67 controls basal body positioning and epithelial branching morphogenesis in mice via the non-canonical Wnt pathway. DMM Disease Models and Mechanisms, 2015, 8, 527-541.	1.2	40
25	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	4.6	215
26	Screen-based identification and validation of four novel ion channels as regulators of renal ciliogenesis. Journal of Cell Science, 2015, 128, 4550-9.	1.2	15
27	The role of primary cilia in the development and disease of the retina. Organogenesis, 2014, 10, 69-85.	0.4	126
28	ATMIN is a transcriptional regulator of both lung morphogenesis and ciliogenesis. Development (Cambridge), 2014, 141, 3966-3977.	1.2	40
29	Loss-of-function mutations in MICU1 cause a brain and muscle disorder linked to primary alterations in mitochondrial calcium signaling. Nature Genetics, 2014, 46, 188-193.	9.4	311
30	The SYSCILIA gold standard (SCGSv1) of known ciliary components and its applications within a systems biology consortium. Cilia, 2013, 2, 7.	1.8	160
31	Aberrant Wnt signalling and cellular over-proliferation in a novel mouse model of Meckelâ€“Gruber syndrome. Developmental Biology, 2013, 377, 55-66.	0.9	40
32	Human Homolog of Drosophila Ariadne (HHARI) is a marker of cellular proliferation associated with nuclear bodies. Experimental Cell Research, 2013, 319, 161-172.	1.2	22
33	Variable expressivity of ciliopathy neurological phenotypes that encompass Meckelâ€“Gruber syndrome and Joubert syndrome is caused by complex de-regulated ciliogenesis, Shh and Wnt signalling defects. Human Molecular Genetics, 2013, 22, 1358-1372.	1.4	94
34	A meckelinâ€“filamin A interaction mediates ciliogenesis. Human Molecular Genetics, 2012, 21, 1272-1286.	1.4	96
35	Nesprin-2 interacts with meckelin and mediates ciliogenesis via remodelling of the actin cytoskeleton. Journal of Cell Science, 2009, 122, 2716-2726.	1.2	119
36	Human iPSC-Derived RPE and Retinal Organoids Reveal Impaired Alternative Splicing of Genes Involved in Pre-mRNA Splicing in <i>PRPF31</i> Autosomal Dominant Retinitis Pigmentosa Type 11. SSRN Electronic Journal, 0, , .	0.4	0

#	Article	IF	Citations
37	Temporal Whole-Transcriptomic Analysis of Characterized In Vitro and Ex Vivo Primary Nasal Epithelia. Frontiers in Cell and Developmental Biology, 0, 10, .	1.8	1
38	Uncovering the burden of hidden ciliopathies in the 100 000 Genomes Project: a reverse phenotyping approach. Journal of Medical Genetics, 0, , jmedgenet-2022-108476.	1.5	3