Colin A Johnson

List of Publications by Year
in descending order

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87 papers	8,681 citations	50273 46 h-index	54911 84 g-index
100 all docs	100 docs citations	100 times ranked	10125 citing authors

#	Article	IF	Citations
1	Orthopaedic Aspects of SAMS Syndrome. Journal of Pediatric Genetics, 2022, 11, 051-058.	0.7	1
2	Molecular diagnoses in the congenital malformations caused by ciliopathies cohort of the 100,000 Genomes Project. Journal of Medical Genetics, 2022, 59, 737-747.	3.2	11
3	Regulation of canonical Wnt signalling by the ciliopathy protein MKS1 and the E2 ubiquitin-conjugating enzyme UBE2E1. ELife, 2022, 11, .	6.0	4
4	RNA-Seq analysis of a Pax3-expressing myoblast clone in-vitro and effect of culture surface stiffness on differentiation. Scientific Reports, 2022, 12, 2841.	3.3	0
5	Missense mutation of MAL causes a rare leukodystrophy similar to Pelizaeus-Merzbacher disease. European Journal of Human Genetics, 2022, 30, 860-864.	2.8	4
6	Activation of autophagy reverses progressive and deleterious protein aggregation in PRPF31 patientâ€induced pluripotent stem cellâ€derived retinal pigment epithelium cells. Clinical and Translational Medicine, 2022, 12, e759.	4.0	12
7	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.	1.6	2
8	Interpreting ciliopathy-associated missense variants of uncertain significance (VUS) in Caenorhabditis elegans. Human Molecular Genetics, 2022, 31, 1574-1587.	2.9	9
9	Novel loss-of-function mutation in HERC2 is associated with severe developmental delay and paediatric lethality. Journal of Medical Genetics, 2021, 58, 334-341.	3.2	9
10	A Recessively Inherited Risk Locus on Chromosome 13q22-31 Conferring Susceptibility to Schizophrenia. Schizophrenia Bulletin, 2021, 47, 796-802.	4.3	3
11	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	8.1	31
12	Pre-mRNA Processing Factors and Retinitis Pigmentosa: RNA Splicing and Beyond. Frontiers in Cell and Developmental Biology, 2021, 9, 700276.	3.7	14
13	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
14	Primary Cilia, Ciliogenesis and the Actin Cytoskeleton: A Little Less Resorption, A Little More Actin Please. Frontiers in Cell and Developmental Biology, 2020, 8, 622822.	3.7	58
15	CiliaCarta: An integrated and validated compendium of ciliary genes. PLoS ONE, 2019, 14, e0216705.	2.5	104
16	The Nuclear Arsenal of Cilia. Developmental Cell, 2019, 49, 161-170.	7.0	27
17	The ciliary Frizzled-like receptor Tmem67 regulates canonical Wnt/Î²-catenin signalling in the developing cerebellum via Hoxb5. Scientific Reports, 2019, 9, 5446.	3.3	15
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	Characterization of Primary Cilia in Normal Fallopian Tube Epithelium and Serous Tubal Intraepithelial Carcinoma. International Journal of Gynecological Cancer, 2018, 28, 1535-1544.	2.5	8
20	Disrupted alternative splicing for genes implicated in splicing and ciliogenesis causes PRPF31 retinitis pigmentosa. Nature Communications, 2018, 9, 4234.	12.8	158
21	The EORTC CAT Coreâ€"The computer adaptive version of the EORTC QLQ-C30 questionnaire. European Journal of Cancer, 2018, 100, 8-16.	2.8	68
22	Human urine-derived renal epithelial cells provide insights into kidney-specific alternate splicing variants. European Journal of Human Genetics, 2018, 26, 1791-1796.	2.8	22
23	PET-PANC: multicentre prospective diagnostic accuracy and health economic analysis study of the impact of combined modality 18fluorine-2-fluoro-2-deoxy-d-glucose positron emission tomography with computed tomography scanning in the diagnosis and management of pancreatic cancer. Health Technology Assessment, 2018, 22, 1-114.	2.8	82
24	Proâ€migratory and TGFâ€Î²â€activating functions of Î±vÎ²6 integrin in pancreatic cancer are differentially regulated via an Eps8â€dependent GTPase switch. Journal of Pathology, 2017, 243, 37-50.	4.5	27
25	Fifteen years of research on oralâ€"facialâ€"digital syndromes: from 1 to 16 causal genes. Journal of Medical Genetics, 2017, 54, 371-380.	3.2	85
26	The Cilium: Cellular Antenna and Central Processing Unit. Trends in Cell Biology, 2017, 27, 126-140.	7.9	320
27	Meckelâ€"Gruber Syndrome: An Update on Diagnosis, Clinical Management, and Research Advances. Frontiers in Pediatrics, 2017, 5, 244.	1.9	107
28	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	8.8	118
29	Mutations in the pH-Sensing G-protein-Coupled Receptor GPR68 Cause Amelogenesis Imperfecta. American Journal of Human Genetics, 2016, 99, 984-990.	6.2	56
30	Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. American Journal of Human Genetics, 2016, 99, 674-682.	6.2	48
31	Ciliogenesis and the DNA damage response: a stressful relationship. Cilia, 2016, 5, 19.	1.8	44
32	A homozygous STIM1 mutation impairs store-operated calcium entry and natural killer cell effector function without clinical immunodeficiency. Journal of Allergy and Clinical Immunology, 2016, 137, 955-957.e8.	2.9	38
33	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	6.2	71
34	TMEM107 recruits ciliopathy proteins to subdomains of the ciliary transition zone and causes JoubertÂsyndrome. Nature Cell Biology, 2016, 18, 122-131.	10.3	118
35	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72.	3.2	48
36	Congenital Myasthenic Syndrome Type 19 Is Caused by Mutations in COL13A1, Encoding the Atypical Non-fibrillar Collagen Type XIII Î±1 Chain. American Journal of Human Genetics, 2015, 97, 878-885.	6.2	57

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37	Unraveling the genetics of Joubert and Meckel-Gruber syndromes. Journal of Pediatric Genetics, 2015, 03, 065-078.	0.7	35
38	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
39	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.	2.4	40
40	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215
41	Variability of systemic and oro-dental phenotype in two families with non-lethal Raine syndrome with FAM20C mutations. BMC Medical Genetics, 2015, 16, 8.	2.1	67
42	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954.	6.2	42
43	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	12.8	51
44	Health-Related Quality of Life in SCALOP, aÂRandomized Phase 2 Trial Comparing Chemoradiation Therapy Regimens in Locally Advanced Pancreatic Cancer. International Journal of Radiation Oncology Biology Physics, 2015, 93, 810-818.	0.8	32
45	A new case of Fas-associated death domain protein deficiency and update on treatment outcomes. Journal of Allergy and Clinical Immunology, 2015, 136, 502-505.e4.	2.9	14
46	Screen-based identification and validation of four novel ion channels as regulators of renal ciliogenesis. Journal of Cell Science, 2015, 128, 4550-9.	2.0	15
47	Mutation Screening of Retinal Dystrophy Patients by Targeted Capture from Tagged Pooled DNAs and Next Generation Sequencing. PLoS ONE, 2014, 9, e104281.	2.5	20
48	The role of primary cilia in the development and disease of the retina. Organogenesis, 2014, 10, 69-85.	1.2	126
49	ATMIN is a transcriptional regulator of both lung morphogenesis and ciliogenesis. Development (Cambridge), 2014, 141, 3966-3977.	2.5	40
50	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	6.2	77
51	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.	21.4	311
52	IFT27 Links the BBSome to IFT for Maintenance of the Ciliary Signaling Compartment. Developmental Cell, 2014, 31, 279-290.	7.0	225
53	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	21.4	118
54	Mutations in TJP2 cause progressive cholestatic liver disease. Nature Genetics, 2014, 46, 326-328.	21.4	244

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55	SAMS, a Syndrome of Short Stature, Auditory-Canal Atresia, Mandibular Hypoplasia, and Skeletal Abnormalities Is a Unique Neurocristopathy Caused by Mutations in Goosecoid. American Journal of Human Genetics, 2013, 93, 1135-1142.	6.2	30
56	Aberrant Wnt signalling and cellular over-proliferation in a novel mouse model of Meckelâ€“Gruber syndrome. Developmental Biology, 2013, 377, 55-66.	2.0	40
57	Human Homolog of Drosophila Ariadne (HHARI) is a marker of cellular proliferation associated with nuclear bodies. Experimental Cell Research, 2013, 319, 161-172.	2.6	22
58	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.	2.9	94
59	A meckelinâ€“filamin A interaction mediates ciliogenesis. Human Molecular Genetics, 2012, 21, 1272-1286.	2.9	96
60	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
61	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	21.4	157
62	The transition zone: an essential functional compartment of cilia. Cilia, 2012, 1, 10.	1.8	107
63	Founder mutations and genotype-phenotype correlations in Meckel-Gruber syndrome and associated ciliopathies. Cilia, 2012, 1, 18.	1.8	42
64	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
65	Disruption of a Ciliary B9 Protein Complex Causes Meckel Syndrome. American Journal of Human Genetics, 2011, 89, 94-110.	6.2	136
66	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 2011, 89, 713-730.	6.2	178
67	Molecular Genetics and Pathogenic Mechanisms for the Severe Ciliopathies: Insights into Neurodevelopment and Pathogenesis of Neural Tube Defects. Molecular Neurobiology, 2011, 43, 12-26.	4.0	67
68	Renal Cystic Disease Proteins Play Critical Roles in the Organization of the Olfactory Epithelium. PLoS ONE, 2011, 6, e19694.	2.5	20
69	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
70	Planar Cell Polarity Acts Through Septins to Control Collective Cell Movement and Ciliogenesis. Science, 2010, 329, 1337-1340.	12.6	309
71	Nesprin-2 interacts with meckelin and mediates ciliogenesis via remodelling of the actin cytoskeleton. Journal of Cell Science, 2009, 122, 2716-2726.	2.0	119
72	Shadow autozygosity mapping by linkage exclusion (SAMPLE): a simple strategy to identify the genetic basis of lethal autosomal recessive disorders. Human Mutation, 2009, 30, 1642-1649.	2.5	5

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73	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	21.4	255
74	16-P024 Functional characterization of the ciliary proteins lebercilin and MKS1. Mechanisms of Development, 2009, 126, S269.	1.7	0
75	Localization of proteins associated with renal cystic diseases to the olfactory epithelium. FASEB Journal, 2009, 23, 796.11.	0.5	0
76	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	6.2	352
77	The Meckelâ€“Gruber Syndrome proteins MKS1 and meckelin interact and are required for primary cilium formation. Human Molecular Genetics, 2007, 16, 173-186.	2.9	245
78	The Meckel-Gruber Syndrome Gene, MKS3, Is Mutated in Joubert Syndrome. American Journal of Human Genetics, 2007, 80, 186-194.	6.2	217
79	Pleiotropic Effects of CEP290 (NPHP6) Mutations Extend to Meckel Syndrome. American Journal of Human Genetics, 2007, 81, 170-179.	6.2	248
80	Spectrum ofMKS1andMKS3mutations in Meckel syndrome: a genotype-phenotype correlation. Human Mutation, 2007, 28, 523-524.	2.5	92
81	IFT80, which encodes a conserved intraflagellar transport protein, is mutated in Jeune asphyxiating thoracic dystrophy. Nature Genetics, 2007, 39, 727-729.	21.4	310
82	The ciliary gene RPGRIP1L is mutated in cerebello-oculo-renal syndrome (Joubert syndrome type B) and Meckel syndrome. Nature Genetics, 2007, 39, 875-881.	21.4	442
83	Mutations in the Embryonal Subunit of the Acetylcholine Receptor (CHRNG) Cause Lethal and Escobar Variants of Multiple Pterygium Syndrome. American Journal of Human Genetics, 2006, 79, 390-395.	6.2	145
84	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome and the wpk rat. Nature Genetics, 2006, 38, 191-196.	21.4	266
85	A novel locus for Meckel-Gruber syndrome, MKS3 , maps to chromosome 8q24. Human Genetics, 2002, 111, 456-461.	3.8	55
86	Deacetylase Activity Associates with Topoisomerase II and Is Necessary for Etoposide-induced Apoptosis. Journal of Biological Chemistry, 2001, 276, 4539-4542.	3.4	92
87	Uncovering the burden of hidden ciliopathies in the 100 000 Genomes Project: a reverse phenotyping approach. Journal of Medical Genetics, 0, , jmedgenet-2022-108476.	3.2	3