Daniel A Doherty

List of Publications by Year
in descending order

81 papers	7,249 citations	76326 40 h-index	71685 76 g-index
85 all docs	85 docs citations	85 times ranked	11130 citing authors

#	Article	IF	Citations
1	SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	3.2	19
2	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
3	The Joubertâ€“Meckelâ€“Nephronophthisis Spectrum of Ciliopathies. Annual Review of Genomics and Human Genetics, 2022, 23, 301-329.	6.2	17
4	De novo heterozygous variants in <scp>SLC30A7</scp> are a candidate cause for Joubert syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2360-2366.	1.2	3
5	TMEM218 dysfunction causes ciliopathies, including Joubert and Meckel syndromes. Human Genetics and Genomics Advances, 2021, 2, 100016.	1.7	7
6	Reply: MN1 gene loss-of-function mutation causes cleft palate in a pedigree. Brain, 2021, 144, e19-e19.	7.6	3
7	Spatial and cell type transcriptional landscape of human cerebellar development. Nature Neuroscience, 2021, 24, 1163-1175.	14.8	98
8	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	2.4	5
9	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.2	105
10	Genetic and phenotypic heterogeneity in KIAA0753 â€related ciliopathies. American Journal of Medical Genetics, Part A, 2021, , .	1.2	2
11	Rescuing human fetal tissue research in the United States: A call for additional regulatory reform. Stem Cell Reports, 2021, 16, 2839-2843.	4.8	6
12	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
13	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
14	A human cell atlas of fetal chromatin accessibility. Science, 2020, 370, .	12.6	265
15	A human cell atlas of fetal gene expression. Science, 2020, 370, .	12.6	436
16	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. Human Mutation, 2020, 41, 2179-2194.	2.5	16
17	Casting a wide net to find the molar tooth. Neurology, 2020, 94, 337-338.	1.1	1
18	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	6.2	18

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19	Cerebellar Watershed Injury in Children. American Journal of Neuroradiology, 2020, 41, 923-928.	2.4	10
20	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	8.2	43
21	Reply. American Journal of Neuroradiology, 2020, 41, E61-E61.	2.4	0
22	Expanding phenotype with severe midline brain anomalies and missense variant supports a causal role for FOXA2 in 20p11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1783-1790.	1.2	10
23	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	61
24	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. Annals of Neurology, 2019, 86, 332-343.	5.3	5
25	GGC Repeat Expansion and Exon 1 Methylation of XYLT1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. American Journal of Human Genetics, 2019, 104, 35-44.	6.2	81
26	Interpreting the clinical significance of combined variants in multiple recessive disease genes: systematic investigation of Joubert syndrome yields little support for oligogenicity. Genetics in Medicine, 2018, 20, 223-233.	2.4	22
27	Characteristics of Liver Disease in 100 Individuals With Joubert Syndrome Prospectively Evaluated at a Single Center. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, 428-435.	1.8	21
28	Rhombencephalosynapsis: Fused cerebellum, confused geneticists. , 2018, 178, 432-439.		26
29	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. American Journal of Human Genetics, 2018, 103, 1009-1021.	6.2	57
30	Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. Ophthalmology, 2018, 125, 1937-1952.	5.2	43
31	Genetics of cerebellar disorders. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 154, 267-286.	1.8	3
32	Congenital Malformations of the Central Nervous System. , 2018, , 857-878.e5.		4
33	Joubert syndrome: neuroimaging findings in 110 patients in correlation with cognitive function and genetic cause. Journal of Medical Genetics, 2017, 54, 521-529.	3.2	53
34	Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and related disorders prospectively evaluated at a single center. Genetics in Medicine, 2017, 19, 875-882.	2.4	100
35	Neuropsychological phenotypes of 76 individuals with Joubert syndrome evaluated at a single center. American Journal of Medical Genetics, Part A, 2017, 173, 1796-1812.	1.2	26
36	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36.	6.2	74

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37	Mortality in Joubert syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1237-1242.	1.2	22
38	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. American Journal of Human Genetics, 2017, 101, 824-832.	6.2	36
39	Mutations in TRAPPC12 Manifest in Progressive Childhood Encephalopathy and Golgi Dysfunction. American Journal of Human Genetics, 2017, 101, 291-299.	6.2	37
40	Prospective Evaluation of Kidney Disease in Joubert Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1962-1973.	4.5	56
41	Abnormal glycosylation in Joubert syndrome type 10. Cilia, 2017, 6, 2.	1.8	14
42	Super-resolution microscopy reveals that disruption of ciliary transition-zone architecture causes JoubertÂsyndrome. Nature Cell Biology, 2017, 19, 1178-1188.	10.3	138
43	Disorders of Cerebellar and Brainstem Development. , 2017, , 199-207.		0
44	Prenatal diagnosis of Chudleyâ€“McCullough syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2426-2430.	1.2	9
45	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. American Journal of Human Genetics, 2016, 98, 772-781.	6.2	43
46	The genetics of cerebellar malformations. Seminars in Fetal and Neonatal Medicine, 2016, 21, 321-332.	2.3	47
47	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	3.2	39
48	Two Hundred Thirty-Six Children With Developmental Hydrocephalus: Causes and Clinical Consequences. Journal of Child Neurology, 2016, 31, 309-320.	1.4	30
49	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72.	3.2	48
50	Temporal bone and cranial nerve findings in pontine tegmental cap dysplasia. Neuroradiology, 2016, 58, 179-187.	2.2	17
51	KIAA0586is Mutated in Joubert Syndrome. Human Mutation, 2015, 36, 831-835.	2.5	62
52	The Ciliopathy Protein CC2D2A Associates with NINL and Functions in RAB8-MICAL3-Regulated Vesicle Trafficking. PLoS Genetics, 2015, 11, e1005575.	3.5	64
53	Brainstem Disconnection: Two Additional Patients and Expansion of the Phenotype. Neuropediatrics, 2015, 46, 139-144.	0.6	9
54	An siRNA-based functional genomics screen for theÂidentification of regulators of ciliogenesis and ciliopathyÂgenes. Nature Cell Biology, 2015, 17, 1074-1087.	10.3	215

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55	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Human Molecular Genetics, 2015, 24, 5313-5325.	2.9	77
56	Rapid identification and recovery of ENU-induced mutations with next-generation sequencing and Paired-End Low-Error analysis. BMC Genomics, 2015, 16, 83.	2.8	30
57	Mutations in CSPP1 Cause Primary Cilia Abnormalities and Joubert Syndrome with or without Jeune Asphyxiating Thoracic Dystrophy. American Journal of Human Genetics, 2014, 94, 62-72.	6.2	104
58	Cerebellar hypoplasia: Differential diagnosis and diagnostic approach. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 211-226.	1.6	107
59	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. American Journal of Human Genetics, 2014, 95, 227-234.	6.2	92
60	Hypomorphism for RPGRIP1L, a Ciliary Gene Vicinal to the FTO Locus, Causes Increased Adiposity in Mice. Cell Metabolism, 2014, 19, 767-779.	16.2	145
61	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
62	Midbrain and hindbrain malformations: advances in clinical diagnosis, imaging, and genetics. Lancet Neurology, The, 2013, 12, 381-393.	10.2	110
63	The Joubert Syndrome-associated Missense Mutation (V443D) in the Abelson-helper Integration Site 1 (AHI1) Protein Alters Its Localization and Protein-Protein Interactions. Journal of Biological Chemistry, 2013, 288, 13676-13694.	3.4	31
64	Persistent figureâ€eight and sideâ€toâ€side head shaking is a marker for rhombencephalosynapsis. Movement Disorders, 2013, 28, 2019-2023.	3.9	20
65	Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. Brain, 2012, 135, 1370-1386.	7.6	131
66	Multiplex Targeted Sequencing Identifies Recurrently Mutated Genes in Autism Spectrum Disorders. Science, 2012, 338, 1619-1622.	12.6	1,133
67	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	21.4	157
68	Joubert syndrome: brain and spinal cord malformations in genotyped cases and implications for neurodevelopmental functions of primary cilia. Acta Neuropathologica, 2012, 123, 695-709.	7.7	78
69	GPSM2 Mutations Cause the Brain Malformations and Hearing Loss in Chudley-McCullough Syndrome. American Journal of Human Genetics, 2012, 90, 1088-1093.	6.2	103
70	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. Cell, 2011, 145, 513-528.	28.9	531
71	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
72	The ciliopathy gene cc2d2a controls zebrafish photoreceptor outer segment development through a role in Rab8-dependent vesicle trafficking. Human Molecular Genetics, 2011, 20, 4041-4055.	2.9	106

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73	Eye Movement Abnormalities in Joubert Syndrome. , 2009, 50, 4669.		38
74	MKS3-Related Ciliopathy with Features of Autosomal Recessive Polycystic Kidney Disease, Nephronophthisis, and Joubert Syndrome. Journal of Pediatrics, 2009, 155, 386-392.e1.	1.8	35
75	Joubert Syndrome: Insights Into Brain Development, Cilium Biology, and Complex Disease. Seminars in Pediatric Neurology, 2009, 16, 143-154.	2.0	158
76	OFD1 Is Mutated in X-Linked Joubert Syndrome and Interacts with LCA5-Encoded Lebercilin. American Journal of Human Genetics, 2009, 85, 465-481.	6.2	180
77	Identification of Mutations in TRAPPC9, which Encodes the NIK- and IKK-Î²-Binding Protein, in Nonsyndromic Autosomal-Recessive Mental Retardation. American Journal of Human Genetics, 2009, 85, 909-915.	6.2	120
78	CC2D2A Is Mutated in Joubert Syndrome and Interacts with the Ciliopathy-Associated Basal Body Protein CEP290. American Journal of Human Genetics, 2008, 83, 559-571.	6.2	202
79	Mutations in the gene encoding the basal body protein RPGRIP1L, a nephrocystin-4 interactor, cause Joubert syndrome. Nature Genetics, 2007, 39, 882-888.	21.4	285
80	Joubert syndrome (and related disorders) (OMIM 213300). European Journal of Human Genetics, 2007, 15, 511-521.	2.8	189
81	Prenatal diagnosis in pregnancies at risk for Joubert syndrome by ultrasound and MRI. Prenatal Diagnosis, 2005, 25, 442-447.	2.3	69