

Daniel A Doherty

List of Publications by Year in descending order

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Version: 2024-02-01

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	Multiplex Targeted Sequencing Identifies Recurrently Mutated Genes in Autism Spectrum Disorders. <i>Science</i> , 2012, 338, 1619-1622.	12.6	1,133
2	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. <i>Cell</i> , 2011, 145, 513-528.	28.9	531
3	A human cell atlas of fetal gene expression. <i>Science</i> , 2020, 370, .	12.6	436
4	Mutations in the gene encoding the basal body protein RPGRIP1L, a nephrocystin-4 interactor, cause Joubert syndrome. <i>Nature Genetics</i> , 2007, 39, 882-888.	21.4	285
5	A human cell atlas of fetal chromatin accessibility. <i>Science</i> , 2020, 370, .	12.6	265
6	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
7	CC2D2A Is Mutated in Joubert Syndrome and Interacts with the Ciliopathy-Associated Basal Body Protein CEP290. <i>American Journal of Human Genetics</i> , 2008, 83, 559-571.	6.2	202
8	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
9	Joubert syndrome (and related disorders) (OMIM 213300). <i>European Journal of Human Genetics</i> , 2007, 15, 511-521.	2.8	189
10	OFD1 Is Mutated in X-Linked Joubert Syndrome and Interacts with LCA5-Encoded Lebercilin. <i>American Journal of Human Genetics</i> , 2009, 85, 465-481.	6.2	180
11	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. <i>American Journal of Human Genetics</i> , 2011, 89, 713-730.	6.2	178
12	Joubert Syndrome: Insights Into Brain Development, Cilium Biology, and Complex Disease. <i>Seminars in Pediatric Neurology</i> , 2009, 16, 143-154.	2.0	158
13	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. <i>Nature Genetics</i> , 2012, 44, 193-199.	21.4	157
14	Hypomorphism for RPGRIP1L, a Ciliary Gene Vicinal to the FTO Locus, Causes Increased Adiposity in Mice. <i>Cell Metabolism</i> , 2014, 19, 767-779.	16.2	145
15	Super-resolution microscopy reveals that disruption of ciliary transition-zone architecture causes Joubert syndrome. <i>Nature Cell Biology</i> , 2017, 19, 1178-1188.	10.3	138
16	Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. <i>Brain</i> , 2012, 135, 1370-1386.	7.6	131
17	Identification of Mutations in TRAPPC9, which Encodes the NIK- and IKK- β -Binding Protein, in Nonsyndromic Autosomal-Recessive Mental Retardation. <i>American Journal of Human Genetics</i> , 2009, 85, 909-915.	6.2	120
18	Midbrain and hindbrain malformations: advances in clinical diagnosis, imaging, and genetics. <i>Lancet Neurology</i> , The, 2013, 12, 381-393.	10.2	110

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19	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
20	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
21	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.2	105
22	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
23	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
24	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
25	Spatial and cell type transcriptional landscape of human cerebellar development. Nature Neuroscience, 2021, 24, 1163-1175.	14.8	98
26	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
27	GCC Repeat Expansion and Exon 1 Methylation of XYL1 Is a Common Pathogenic Variant in Baratela-Scott Syndrome. American Journal of Human Genetics, 2019, 104, 35-44.	6.2	81
28	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
29	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Human Molecular Genetics, 2015, 24, 5313-5325.	2.9	77
30	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
31	Prenatal diagnosis in pregnancies at risk for Joubert syndrome by ultrasound and MRI. Prenatal Diagnosis, 2005, 25, 442-447.	2.3	69
32	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
33	The Ciliopathy Protein CC2D2A Associates with NINL and Functions in RAB8-MICAL3-Regulated Vesicle Trafficking. PLoS Genetics, 2015, 11, e1005575.	3.5	64
34	<i>KIAA0586</i> is Mutated in Joubert Syndrome. Human Mutation, 2015, 36, 831-835.	2.5	62
35	Redefining the Etiologic Landscape of Cerebellar Malformations. American Journal of Human Genetics, 2019, 105, 606-615.	6.2	61
36	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

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37	Prospective Evaluation of Kidney Disease in Joubert Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2017, 12, 1962-1973.	4.5	56
38	Joubert syndrome: neuroimaging findings in 110 patients in correlation with cognitive function and genetic cause. <i>Journal of Medical Genetics</i> , 2017, 54, 521-529.	3.2	53
39	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. <i>Journal of Medical Genetics</i> , 2016, 53, 62-72.	3.2	48
40	The genetics of cerebellar malformations. <i>Seminars in Fetal and Neonatal Medicine</i> , 2016, 21, 321-332.	2.3	47
41	Recessive Inactivating Mutations in TBCK, Encoding a Rab GTPase-Activating Protein, Cause Severe Infantile Syndromic Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 98, 772-781.	6.2	43
42	Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. <i>Ophthalmology</i> , 2018, 125, 1937-1952.	5.2	43
43	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. <i>Journal of Clinical Investigation</i> , 2020, 130, 4423-4439.	8.2	43
44	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. <i>Journal of Medical Genetics</i> , 2016, 53, 208-214.	3.2	39
45	Eye Movement Abnormalities in Joubert Syndrome. , 2009, 50, 4669.		38
46	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. <i>Brain</i> , 2020, 143, 55-68.	7.6	38
47	Mutations in TRAPPC12 Manifest in Progressive Childhood Encephalopathy and Golgi Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 101, 291-299.	6.2	37
48	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. <i>American Journal of Human Genetics</i> , 2017, 101, 824-832.	6.2	36
49	MKS3-Related Ciliopathy with Features of Autosomal Recessive Polycystic Kidney Disease, Nephronophthisis, and Joubert Syndrome. <i>Journal of Pediatrics</i> , 2009, 155, 386-392.e1.	1.8	35
50	The Joubert Syndrome-associated Missense Mutation (V443D) in the Abelson-helper Integration Site 1 (AHI1) Protein Alters Its Localization and Protein-Protein Interactions. <i>Journal of Biological Chemistry</i> , 2013, 288, 13676-13694.	3.4	31
51	Rapid identification and recovery of ENU-induced mutations with next-generation sequencing and Paired-End Low-Error analysis. <i>BMC Genomics</i> , 2015, 16, 83.	2.8	30
52	Two Hundred Thirty-Six Children With Developmental Hydrocephalus: Causes and Clinical Consequences. <i>Journal of Child Neurology</i> , 2016, 31, 309-320.	1.4	30
53	Neuropsychological phenotypes of 76 individuals with Joubert syndrome evaluated at a single center. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1796-1812.	1.2	26
54	Rhombencephalosynapsis: Fused cerebellum, confused geneticists. , 2018, 178, 432-439.		26

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55	Mortality in Joubert syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1237-1242.	1.2	22
56	Interpreting the clinical significance of combined variants in multiple recessive disease genes: systematic investigation of Joubert syndrome yields little support for oligogenicity. <i>Genetics in Medicine</i> , 2018, 20, 223-233.	2.4	22
57	Characteristics of Liver Disease in 100 Individuals With Joubert Syndrome Prospectively Evaluated at a Single Center. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2018, 66, 428-435.	1.8	21
58	Persistent figure-eight and side-to-side head shaking is a marker for rhombencephalosynapsis. <i>Movement Disorders</i> , 2013, 28, 2019-2023.	3.9	20
59	<i>SUFU</i> haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. <i>Journal of Medical Genetics</i> , 2022, 59, 888-894.	3.2	19
60	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. <i>American Journal of Human Genetics</i> , 2020, 106, 623-631.	6.2	18
61	Temporal bone and cranial nerve findings in pontine tegmental cap dysplasia. <i>Neuroradiology</i> , 2016, 58, 179-187.	2.2	17
62	The Joubert–Meckel–Nephronophthisis Spectrum of Ciliopathies. <i>Annual Review of Genomics and Human Genetics</i> , 2022, 23, 301-329.	6.2	17
63	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. <i>Human Mutation</i> , 2020, 41, 2179-2194.	2.5	16
64	Abnormal glycosylation in Joubert syndrome type 10. <i>Cilia</i> , 2017, 6, 2.	1.8	14
65	Expanding phenotype with severe midline brain anomalies and missense variant supports a causal role for <i>FOXA2</i> in 20p11.2 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1783-1790.	1.2	10
66	Cerebellar Watershed Injury in Children. <i>American Journal of Neuroradiology</i> , 2020, 41, 923-928.	2.4	10
67	Brainstem Disconnection: Two Additional Patients and Expansion of the Phenotype. <i>Neuropediatrics</i> , 2015, 46, 139-144.	0.6	9
68	Prenatal diagnosis of Chudley–McCullough syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2426-2430.	1.2	9
69	TMEM218 dysfunction causes ciliopathies, including Joubert and Meckel syndromes. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100016.	1.7	7
70	Rescuing human fetal tissue research in the United States: A call for additional regulatory reform. <i>Stem Cell Reports</i> , 2021, 16, 2839-2843.	4.8	6
71	Discovering a new part of the phenotypic spectrum of Coffin–Siris syndrome in a fetal cohort. <i>Genetics in Medicine</i> , 2022, 24, 1753-1760.	2.4	6
72	Intellectual and developmental disabilities research centers: Fifty years of scientific accomplishments. <i>Annals of Neurology</i> , 2019, 86, 332-343.	5.3	5

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73	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. <i>Genes</i> , 2021, 12, 1275.	2.4	5
74	Congenital Malformations of the Central Nervous System. , 2018, , 857-878.e5.		4
75	Genetics of cerebellar disorders. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 154, 267-286.	1.8	3
76	Reply: <i>MN1</i> gene loss-of-function mutation causes cleft palate in a pedigree. <i>Brain</i> , 2021, 144, e19-e19.	7.6	3
77	<i>De novo</i> heterozygous variants in <i>SLC30A7</i> are a candidate cause for Joubert syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2360-2366.	1.2	3
78	Genetic and phenotypic heterogeneity in KIAA0753 related ciliopathies. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	1.2	2
79	Casting a wide net to find the molar tooth. <i>Neurology</i> , 2020, 94, 337-338.	1.1	1
80	Disorders of Cerebellar and Brainstem Development. , 2017, , 199-207.		0
81	Reply. <i>American Journal of Neuroradiology</i> , 2020, 41, E61-E61.	2.4	0