

Daryl A Scott

List of Publications by Year in descending order

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Version: 2024-02-01

86
papers

3,348
citations

186265

28
h-index

168389

53
g-index

90
all docs

90
docs citations

90
times ranked

6476
citing authors

#	ARTICLE	IF	CITATIONS
1	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. <i>Journal of Medical Genetics</i> , 2010, 47, 332-341.	3.2	447
2	Use of Exome Sequencing for Infants in Intensive Care Units. <i>JAMA Pediatrics</i> , 2017, 171, e173438.	6.2	348
3	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. <i>American Journal of Human Genetics</i> , 2017, 100, 843-853.	6.2	181
4	Chromosome 8p23.1 deletions as a cause of complex congenital heart defects and diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1661-1677.	1.2	147
5	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	6.2	142
6	1p36 deletion syndrome: an update. <i>The Application of Clinical Genetics</i> , 2015, 8, 189.	3.0	104
7	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. <i>American Journal of Human Genetics</i> , 2019, 104, 213-228.	6.2	90
8	Genomic alterations that contribute to the development of isolated and non-isolated congenital diaphragmatic hernia. <i>Journal of Medical Genetics</i> , 2011, 48, 299-307.	3.2	82
9	Clinical spectrum of individuals with pathogenic <i>NF1</i> missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. <i>Human Mutation</i> , 2020, 41, 299-315.	2.5	80
10	Genome-wide oligonucleotide-based array comparative genome hybridization analysis of non-isolated congenital diaphragmatic hernia. <i>Human Molecular Genetics</i> , 2007, 16, 424-430.	2.9	79
11	Mouse model reveals the role of SOX7 in the development of congenital diaphragmatic hernia associated with recurrent deletions of 8p23.1. <i>Human Molecular Genetics</i> , 2012, 21, 4115-4125.	2.9	78
12	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. <i>Genetics in Medicine</i> , 2017, 19, 412-420.	2.4	73
13	Stromal <i>Fat4</i> acts non-autonomously with <i>Dachsous1/2</i> to restrict the nephron progenitor pool. <i>Development (Cambridge)</i> , 2015, 142, 2564-73.	2.5	70
14	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. <i>American Journal of Human Genetics</i> , 2016, 98, 963-970.	6.2	67
15	Schaaf-Yang syndrome overview: Report of 78 individuals. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2564-2574.	1.2	66
16	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotype-phenotype correlation. <i>Genetics in Medicine</i> , 2019, 21, 867-876.	2.4	62
17	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	6.2	59
18	A 1q42 deletion involving <i>DISC1</i> , <i>DISC2</i> , and <i>TSNAX</i> in an autism spectrum disorder. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1758-1762.	1.2	57

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19	Identification of Critical Regions and Candidate Genes for Cardiovascular Malformations and Cardiomyopathy Associated with Deletions of Chromosome 1p36. PLoS ONE, 2014, 9, e85600.	2.5	51
20	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
21	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45
22	Review of the phenotypic spectrum associated with haploinsufficiency of <i>MYRF</i> . American Journal of Medical Genetics, Part A, 2019, 179, 1376-1382.	1.2	44
23	Deficiency of FRAS1-related extracellular matrix 1 (FREM1) causes congenital diaphragmatic hernia in humans and mice. Human Molecular Genetics, 2013, 22, 1026-1038.	2.9	42
24	Recurrent microdeletions of 15q25.2 are associated with increased risk of congenital diaphragmatic hernia, cognitive deficits and possibly Diamond-Blackfan anaemia. Journal of Medical Genetics, 2010, 47, 777-781.	3.2	40
25	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	3.8	40
26	An Allelic Series of Mice Reveals a Role for RERE in the Development of Multiple Organs Affected in Chromosome 1p36 Deletions. PLoS ONE, 2013, 8, e57460.	2.5	35
27	Disruptive variants of <i>CSDE1</i> associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
28	Genotype-phenotype correlations in individuals with pathogenic <i>RERE</i> variants. Human Mutation, 2018, 39, 666-675.	2.5	34
29	BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. American Journal of Human Genetics, 2020, 107, 1096-1112.	6.2	32
30	Disruption of PHF21A causes syndromic intellectual disability with craniofacial anomalies, epilepsy, hypotonia, and neurobehavioral problems including autism. Molecular Autism, 2019, 10, 35.	4.9	30
31	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
32	Copy number variations in 375 patients with oesophageal atresia and/or tracheoesophageal fistula. European Journal of Human Genetics, 2016, 24, 1715-1723.	2.8	27
33	Clinical exome sequencing data reveal high diagnostic yields for congenital diaphragmatic hernia plus (CDH+) and new phenotypic expansions involving CDH. Journal of Medical Genetics, 2022, 59, 270-278.	3.2	27
34	Mouse Model Reveals the Role of RERE in Cerebellar Foliation and the Migration and Maturation of Purkinje Cells. PLoS ONE, 2014, 9, e87518.	2.5	26
35	<i>FBN1</i> contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836.	1.2	24
36	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants in <i>NONO</i> . Journal of Medical Genetics, 2017, 54, 47-53.	3.2	24

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37	SOX7 expression is critically required in FLK1-expressing cells for vasculogenesis and angiogenesis during mouse embryonic development. <i>Mechanisms of Development</i> , 2017, 146, 31-41.	1.7	24
38	Genetics of congenital diaphragmatic hernia. <i>Seminars in Pediatric Surgery</i> , 2007, 16, 88-93.	1.1	23
39	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€™s Magenis syndrome. <i>Genome Medicine</i> , 2019, 11, 12.	8.2	23
40	Widening of the genetic and clinical spectrum of Lambâ€™s Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. <i>Genetics in Medicine</i> , 2020, 22, 524-537.	2.4	21
41	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. <i>American Journal of Human Genetics</i> , 2021, 108, 1710-1724.	6.2	18
42	Further delineation of the phenotypic spectrum associated with hemizygous loss of function variants in <i>NONO</i> . <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 652-658.	1.2	17
43	The role of <i>FREM2</i> and <i>FRAS1</i> in the development of congenital diaphragmatic hernia. <i>Human Molecular Genetics</i> , 2018, 27, 2064-2075.	2.9	16
44	Novel <i>Frem1</i> -Related Mouse Phenotypes and Evidence of Genetic Interactions with <i>Gata4</i> and <i>Slit3</i> . <i>PLoS ONE</i> , 2013, 8, e58830.	2.5	15
45	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. <i>Journal of Pediatric Genetics</i> , 2018, 07, 164-173.	0.7	15
46	Delineation of a less than 200â€‰kb minimal deleted region for cardiac malformations on chromosome 7p22. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1729-1734.	1.2	14
47	Expanding the phenotypic spectrum of Succinyl-CoA ligase deficiency through functional validation of a new <i>SUCLG1</i> variant. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 68-74.	1.1	14
48	Xp11.22 deletions encompassing <i>CENPVL1</i> , <i>CENPVL2</i> , <i>MAGED1</i> and <i>GSPT2</i> as a cause of syndromic X-linked intellectual disability. <i>PLoS ONE</i> , 2017, 12, e0175962.	2.5	14
49	Loss of <i>CLTRN</i> function produces a neuropsychiatric disorder and a biochemical phenotype that mimics Hartnup disease. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2459-2468.	1.2	14
50	Novel Missense Variants in <i>ADAT3</i> as a Cause of Syndromic Intellectual Disability. <i>Journal of Pediatric Genetics</i> , 2019, 08, 244-251.	0.7	13
51	Variants in <i>SCAF4</i> Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. <i>American Journal of Human Genetics</i> , 2020, 107, 544-554.	6.2	13
52	Co-occurring defect analysis: A platform for analyzing birth defect co-occurrence in registries. <i>Birth Defects Research</i> , 2019, 111, 1356-1364.	1.5	12
53	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate disease-associated loci for BAFopathies. <i>Genetics in Medicine</i> , 2022, 24, 364-373.	2.4	12
54	Deletions of Xp provide evidence for the role of holocytochrome C-type synthase (<i>HCCS</i>) in congenital diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1588-1590.	1.2	11

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55	<i>BAZ2B</i> haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
56	Contribution of <i>LPP</i> copy number and sequence changes to esophageal atresia, tracheoesophageal fistula, and VACTERL association. American Journal of Medical Genetics, Part A, 2012, 158A, 1785-1787.	1.2	10
57	Duplication of <i>HEY2</i> in cardiac and neurologic development. American Journal of Medical Genetics, Part A, 2015, 167, 2145-2149.	1.2	10
58	Delineation of a 1.65â€‰Mb critical region for hemihyperplasia and digital anomalies on Xq25. American Journal of Medical Genetics, Part A, 2010, 152A, 453-458.	1.2	9
59	Syndromic congenital myelofibrosis associated with a loss-of-function variant in RBSN. Blood, 2018, 132, 658-662.	1.4	9
60	Birth defects that co-occur with non-syndromic gastroschisis and omphalocele. American Journal of Medical Genetics, Part A, 2020, 182, 2581-2593.	1.2	9
61	<i>NUBPL</i> mitochondrial disease: new patients and review of the genetic and clinical spectrum. Journal of Medical Genetics, 2021, 58, 314-325.	3.2	9
62	Underlying genetic etiologies of congenital diaphragmatic hernia. Prenatal Diagnosis, 2022, 42, 373-386.	2.3	9
63	A dominant negative variant of <i>RAB5B</i> disrupts maturation of surfactant protein B and surfactant protein C. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.1	9
64	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L -related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925.	1.2	8
65	Phenotypic expansion in <i>KIF1A</i> -related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	2.5	8
66	Recessive ACO2 variants as a cause of isolated ophthalmologic phenotypes. American Journal of Medical Genetics, Part A, 2020, 182, 1960-1966.	1.2	8
67	Multiple mitochondrial dysfunctions syndrome 1: An unusual cause of developmental pulmonary hypertension. American Journal of Medical Genetics, Part A, 2020, 182, 755-761.	1.2	8
68	Evidence that <sc><i>FGFRL1</i></sc> contributes to congenital diaphragmatic hernia development in humans. American Journal of Medical Genetics, Part A, 2021, 185, 836-840.	1.2	8
69	Birth defect co-occurrence patterns in the Texas Birth Defects Registry. Pediatric Research, 2022, 91, 1278-1285.	2.3	8
70	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. Npj Genomic Medicine, 2021, 6, 104.	3.8	7
71	Chromosome 5q33 deletions associated with congenital heart defects. American Journal of Medical Genetics, Part A, 2016, 170, 3338-3342.	1.2	6
72	Delineation of a novel neurodevelopmental syndrome associated with <i>PAX5</i> haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5

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73	SOX7 Is Required for Muscle Satellite Cell Development and Maintenance. <i>Stem Cell Reports</i> , 2017, 9, 1139-1151.	4.8	4
74	RERE deficiency leads to decreased expression of GATA4 and the development of ventricular septal defects. <i>DMM Disease Models and Mechanisms</i> , 2018, 11, .	2.4	4
75	Patterns of co-occurring birth defects among infants with hypospadias. <i>Journal of Pediatric Urology</i> , 2021, 17, 64.e1-64.e8.	1.1	4
76	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. <i>Genetics in Medicine</i> , 2021, 23, 384-395.	2.4	4
77	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 631428.	3.7	4
78	A Comprehensive Assessment of Co-occurring Birth Defects among Infants with Non-Syndromic Anophthalmia or Microphthalmia. <i>Ophthalmic Epidemiology</i> , 2021, 28, 428-435.	1.7	4
79	A novel, de novo intronic variant in <i>POGZ</i> causes Whiteâ€“Sutton syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2198-2203.	1.2	4
80	The frequency and efficacy of genetic testing in individuals with scimitar syndrome. <i>Cardiology in the Young</i> , 2022, 32, 550-557.	0.8	3
81	Evidence for an association between Coffinâ€“Siris syndrome and congenital diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2718-2723.	1.2	3
82	Patterns of congenital anomalies among individuals with trisomy 13 in Texas. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1787-1793.	1.2	2
83	RERE deficiency contributes to the development of orofacial clefts in humans and mice. <i>Human Molecular Genetics</i> , 2021, 30, 595-602.	2.9	2
84	<i>RERE</i> deficiency causes retinal and optic nerve atrophy through degeneration of retinal cells. <i>Developmental Dynamics</i> , 2021, 250, 1398-1409.	1.8	1
85	Evidence-Based Genetic Testing for Individuals with Congenital Diaphragmatic Hernia. <i>Journal of Pediatrics</i> , 2022, 248, 13-14.	1.8	1
86	Human Studies and Mouse Models Provide Insight into Diaphragm Development. <i>FASEB Journal</i> , 2011, 25, 305.2.	0.5	0