Daryl A Scott

List of Publications by Year
in descending order

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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	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
2	Birth defect co-occurrence patterns in the Texas Birth Defects Registry. Pediatric Research, 2022, 91, 1278-1285.	2.3	8
3	The frequency and efficacy of genetic testing in individuals with scimitar syndrome. Cardiology in the Young, 2022, 32, 550-557.	0.8	3
4	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	2.4	12
5	Underlying genetic etiologies of congenital diaphragmatic hernia. Prenatal Diagnosis, 2022, 42, 373-386.	2.3	9
6	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
7	A dominant negative variant of RAB5B 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
8	A novel, de novo intronic variant in <scp>POGZ</scp> causes <scp>Whiteâ€“Sutton</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2198-2203.	1.2	4
9	Evidence-Based Genetic Testing for Individuals with Congenital Diaphragmatic Hernia. Journal of Pediatrics, 2022, 248, 13-14.	1.8	1
10	Evidence for an association between <scp>Coffinâ€Siris</scp> syndrome and congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2022, 188, 2718-2723.	1.2	3
11	NUBPL mitochondrial disease: new patients and review of the genetic and clinical spectrum. Journal of Medical Genetics, 2021, 58, 314-325.	3.2	9
12	Patterns of co-occurring birth defects among infants with hypospadias. Journal of Pediatric Urology, 2021, 17, 64.e1-64.e8.	1.1	4
13	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	2.4	4
14	Evidence that <scp>FGFRL1</scp> contributes to congenital diaphragmatic hernia development in humans. American Journal of Medical Genetics, Part A, 2021, 185, 836-840.	1.2	8
15	<scp>RERE</scp> deficiency causes retinal and optic nerve atrophy through degeneration of retinal cells. Developmental Dynamics, 2021, 250, 1398-1409.	1.8	1
16	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	3.7	4
17	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
18	Patterns of congenital anomalies among individuals with trisomy 13 in Texas. American Journal of Medical Genetics, Part A, 2021, 185, 1787-1793.	1.2	2

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19	RERE deficiency contributes to the development of orofacial clefts in humans and mice. Human Molecular Genetics, 2021, 30, 595-602.	2.9	2
20	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	6.2	18
21	A Comprehensive Assessment of Co-occurring Birth Defects among Infants with Non-Syndromic Anophthalmia or Microphthalmia. Ophthalmic Epidemiology, 2021, 28, 428-435.	1.7	4
22	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
23	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
24	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
25	Further delineation of the phenotypic spectrum associated with hemizygous lossâ€ofâ€function variants in NONO. American Journal of Medical Genetics, Part A, 2020, 182, 652-658.	1.2	17
26	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925.	1.2	8
27	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
28	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
29	Birth defects that coâ€occur with nonâ€syndromic gastroschisis and omphalocele. American Journal of Medical Genetics, Part A, 2020, 182, 2581-2593.	1.2	9
30	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	2.5	8
31	Recessive ACO2 variants as a cause of isolated ophthalmologic phenotypes. American Journal of Medical Genetics, Part A, 2020, 182, 1960-1966.	1.2	8
32	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
33	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
34	Coâ€occurring defect analysis: A platform for analyzing birth defect coâ€occurrence in registries. Birth Defects Research, 2019, 111, 1356-1364.	1.5	12
35	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
36	Loss of CLTRN function produces a neuropsychiatric disorder and a biochemical phenotype that mimics Hartnup disease. American Journal of Medical Genetics, Part A, 2019, 179, 2459-2468.	1.2	14

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37	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
38	Review of the phenotypic spectrum associated with haploinsufficiency of MYRF. American Journal of Medical Genetics, Part A, 2019, 179, 1376-1382.	1.2	44
39	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23
40	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
41	Novel Missense Variants in ADAT3 as a Cause of Syndromic Intellectual Disability. Journal of Pediatric Genetics, 2019, 08, 244-251.	0.7	13
42	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
43	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
44	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
45	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075.	2.9	16
46	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34
47	Schaafâ€“Yang syndrome overview: Report of 78 individuals. American Journal of Medical Genetics, Part A, 2018, 176, 2564-2574.	1.2	66
48	RERE deficiency leads to decreased expression of GATA4 and the development of ventricular septal defects. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	4
49	Prioritization of Candidate Genes for Congenital Diaphragmatic Hernia in a Critical Region on Chromosome 4p16 using a Machine-Learning Algorithm. Journal of Pediatric Genetics, 2018, 07, 164-173.	0.7	15
50	Syndromic congenital myelofibrosis associated with a loss-of-function variant in RBSN. Blood, 2018, 132, 658-662.	1.4	9
51	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
52	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants inNONO. Journal of Medical Genetics, 2017, 54, 47-53.	3.2	24
53	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
54	SOX7 Is Required for Muscle Satellite Cell Development and Maintenance. Stem Cell Reports, 2017, 9, 1139-1151.	4.8	4

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55	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
56	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. Genetics in Medicine, 2017, 19, 412-420.	2.4	73
57	Xp11.22 deletions encompassing CENPVL1, CENPVL2, MAGED1 and GSPT2 as a cause of syndromic X-linked intellectual disability. PLoS ONE, 2017, 12, e0175962.	2.5	14
58	SOX7 expression is critically required in FLK1-expressing cells for vasculogenesis and angiogenesis during mouse embryonic development. Mechanisms of Development, 2017, 146, 31-41.	1.7	24
59	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. American Journal of Human Genetics, 2016, 98, 963-970.	6.2	67
60	Chromosome 5q33 deletions associated with congenital heart defects. American Journal of Medical Genetics, Part A, 2016, 170, 3338-3342.	1.2	6
61	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
62	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	3.8	40
63	Expanding the phenotypic spectrum of Succinyl-CoA ligase deficiency through functional validation of a new SUCLG1 variant. Molecular Genetics and Metabolism, 2016, 119, 68-74.	1.1	14
64	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
65	Duplication of HEY2 in cardiac and neurologic development. American Journal of Medical Genetics, Part A, 2015, 167, 2145-2149.	1.2	10
66	1p36 deletion syndrome: an update. The Application of Clinical Genetics, 2015, 8, 189.	3.0	104
67	Stromal Fat4 acts non-autonomously with Dachsous1/2 to restrict the nephron progenitor pool. Development (Cambridge), 2015, 142, 2564-73.	2.5	70
68	FBN1 contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836.	1.2	24
69	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
70	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
71	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
72	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

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73	Novel Frem1-Related Mouse Phenotypes and Evidence of Genetic Interactions with Gata4 and Slit3. PLoS ONE, 2013, 8, e58830.	2.5	15
74	Mouse model reveals the role of SOX7 in the development of congenital diaphragmatic hernia associated with recurrent deletions of 8p23.1. Human Molecular Genetics, 2012, 21, 4115-4125.	2.9	78
75	Contribution of LPP 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
76	Genomic alterations that contribute to the development of isolated and non-isolated congenital diaphragmatic hernia. Journal of Medical Genetics, 2011, 48, 299-307.	3.2	82
77	Delineation of a less than 200â€‰kb minimal deleted region for cardiac malformations on chromosome 7p22. American Journal of Medical Genetics, Part A, 2011, 155, 1729-1734.	1.2	14
78	Human Studies and Mouse Models Provide Insight into Diaphragm Development. FASEB Journal, 2011, 25, 305.2.	0.5	0
79	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
80	Deletions of Xp provide evidence for the role of holocytochrome Câ€type synthase (HCCS) in congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2010, 152A, 1588-1590.	1.2	11
81	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	3.2	447
82	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
83	Chromosome 8p23.1 deletions as a cause of complex congenital heart defects and diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2009, 149A, 1661-1677.	1.2	147
84	A 1q42 deletion involving DISC1, DISC2, and TSNAX in an autism spectrum disorder. American Journal of Medical Genetics, Part A, 2009, 149A, 1758-1762.	1.2	57
85	Genome-wide oligonucleotide-based array comparative genome hybridization analysis of non-isolated congenital diaphragmatic hernia. Human Molecular Genetics, 2007, 16, 424-430.	2.9	79
86	Genetics of congenital diaphragmatic hernia. Seminars in Pediatric Surgery, 2007, 16, 88-93.	1.1	23