Sally L Dunwoodie

List of Publications by Year
in descending order

127	7,407	47006 47	62596 80
papers	citations	h-index	g-index

136	136	136	9418
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The Role of Hypoxia in Development of the Mammalian Embryo. Developmental Cell, 2009, 17, 755-773.	7.0	509
2	SmcHD1, containing a structural-maintenance-of-chromosomes hinge domain, has a critical role in X inactivation. Nature Genetics, 2008, 40, 663-669.	21.4	305
3	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
4	Mutation of the LUNATIC FRINGE Gene in Humans Causes Spondylocostal Dysostosis with a Severe Vertebral Phenotype. American Journal of Human Genetics, 2006, 78, 28-37.	6.2	223
5	Murine T-box transcription factor Tbx20 acts as a repressor during heart development, and is essential for adult heart integrity, function and adaptation. Development (Cambridge), 2005, 132, 2451-2462.	2.5	218
6	Axial skeletal defects caused by mutation in the spondylocostal dysplasia/pudgy geneDll3are associated with disruption of the segmentation clock within the presomitic mesoderm. Development (Cambridge), 2002, 129, 1795-1806.	2.5	197
7	A Mechanism for Gene-Environment Interaction in the Etiology of Congenital Scoliosis. Cell, 2012, 149, 295-306.	28.9	188
8	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. New England Journal of Medicine, 2017, 377, 544-552.	27.0	177
9	Notch inhibition by the ligand Delta-Like 3 defines the mechanism of abnormal vertebral segmentation in spondylocostal dysostosis. Human Molecular Genetics, 2011, 20, 905-916.	2.9	159
10	Mutated MESP2 Causes Spondylocostal Dysostosis in Humans. American Journal of Human Genetics, 2004, 74, 1249-1254.	6.2	157
11	Msg1 and Mrg1, founding members of a gene family, show distinct patterns of gene expression during mouse embryogenesis. Mechanisms of Development, 1998, 72, 27-40.	1.7	155
12	Evolution of distinct EGF domains with specific functions. Protein Science, 2005, 14, 1091-1103.	7.6	155
13	Association of the PHACTR1/EDN1 Genetic Locus With Spontaneous Coronary Artery Dissection. Journal of the American College of Cardiology, 2019, 73, 58-66.	2.8	147
14	Folic acid prevents exencephaly in Cited2 deficient mice. Human Molecular Genetics, 2002, 11, 283-293.	2.9	145
15	Abnormal vertebral segmentation and the notch signaling pathway in man. Developmental Dynamics, 2007, 236, 1456-1474.	1.8	143
16	Divergent functions and distinct localization of the Notch ligands DLL1 and DLL3 in vivo. Journal of Cell Biology, 2007, 178, 465-476.	5.2	134
17	Mutation of HAIRY-AND-ENHANCER-OF-SPLIT-7 in humans causes spondylocostal dysostosis. Human Molecular Genetics, 2008, 17, 3761-3766.	2.9	123
18	Transcriptional activating activity of Smad4: Roles of SMAD hetero-oligomerization and enhancement by an associating transactivator. Proceedings of the National Academy of Sciences of the United States of America, 1998, 95, 9785-9790.	7.1	122

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19	Novel mutations in DLL3, a somitogenesis gene encoding a ligand for the Notch signalling pathway, cause a consistent pattern of abnormal vertebral segmentation in spondylocostal dysostosis. Journal of Medical Genetics, 2003, 40, 333-339.	3.2	120
20	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	4.5	118
21	Advances in the Genetics of Congenital HeartÂDisease. Journal of the American College of Cardiology, 2017, 69, 859-870.	2.8	115
22	Cited2is required both for heart morphogenesis and establishment of the left-right axis in mouse development. Development (Cambridge), 2005, 132, 1337-1348.	2.5	113
23	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	28.9	113
24	Developmental regulation of Notch signaling genes in the embryonic pituitary: Prop1 deficiency affects Notch2 expression. Developmental Biology, 2004, 265, 329-340.	2.0	110
25	Sp5, a New Member of the Sp1 Family, Is Dynamically Expressed during Development and Genetically Interacts with Brachyury. Developmental Biology, 2000, 227, 358-372.	2.0	107
26	Loss of Cited2 affects trophoblast formation and vascularization of the mouse placenta. Developmental Biology, 2006, 294, 67-82.	2.0	101
27	Autosomal dominant spondylocostal dysostosis is caused by mutation in TBX6. Human Molecular Genetics, 2013, 22, 1625-1631.	2.9	87
28	Targeted Next-Generation Sequencing Identifies Pathogenic Variants in Familial Congenital Heart Disease. Journal of the American College of Cardiology, 2014, 64, 2498-2506.	2.8	85
29	Cited1 Is Required in Trophoblasts for Placental Development and for Embryo Growth and Survival. Molecular and Cellular Biology, 2004, 24, 228-244.	2.3	80
30	Tinman/Nkx2-5 acts via miR-1 and upstream of Cdc42 to regulate heart function across species. Journal of Cell Biology, 2011, 193, 1181-1196.	5.2	74
31	Edd , the Murine Hyperplastic Disc Gene, Is Essential for Yolk Sac Vascularization and Chorioallantoic Fusion. Molecular and Cellular Biology, 2004, 24, 7225-7234.	2.3	73
32	Hif-1a suppresses ROS-induced proliferation of cardiac fibroblasts following myocardial infarction. Cell Stem Cell, 2022, 29, 281-297.e12.	11.1	71
33	Cited2, a coactivator of HNF4Î±, is essential for liver development. EMBO Journal, 2007, 26, 4445-4456.	7.8	70
34	Progress in the Understanding of the Genetic Etiology of Vertebral Segmentation Disorders in Humans. Annals of the New York Academy of Sciences, 2009, 1151, 38-67.	3.8	70
35	A Screening Approach to Identify Clinically Actionable Variants Causing Congenital Heart Disease in Exome Data. Circulation Genomic and Precision Medicine, 2018, 11, e001978.	3.6	65
36	BMP/SMAD1 signaling sets a threshold for the left/right pathway in lateral plate mesoderm and limits availability of SMAD4. Genes and Development, 2008, 22, 3037-3049.	5.9	63

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37	Axial skeletal defects caused by mutation in the spondylocostal dysplasia/pudgy gene Dll3 are associated with disruption of the segmentation clock within the presomitic mesoderm. Development (Cambridge), 2002, 129, 1795-806.	2.5	63
38	Cited1 Is a Bifunctional Transcriptional Cofactor That Regulates Early Nephronic Patterning. Journal of the American Society of Nephrology: JASN, 2005, 16, 1632-1644.	6.1	58
39	Two novel missense mutations in HAIRY-AND-ENHANCER-OF-SPLIT-7 in a family with spondylocostal dysostosis. European Journal of Human Genetics, 2010, 18, 674-679.	2.8	55
40	HIF-1Î± deletion partially rescues defects of hematopoietic stem cell quiescence caused by Cited2 deficiency. Blood, 2012, 119, 2789-2798.	1.4	55
41	Loss of Cited2 causes congenital heart disease by perturbing leftâ€“right patterning of the body axis. Human Molecular Genetics, 2011, 20, 1097-1110.	2.9	54
42	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. Genetics in Medicine, 2019, 21, 1111-1120.	2.4	54
43	NKX2-5 mutations causative for congenital heart disease retain functionality and are directed to hundreds of targets. ELife, 2015, 4, .	6.0	54
44	Dll3 pudgy mutation differentially disrupts dynamic expression of somite genes. Genesis, 2004, 39, 115-121.	1.6	53
45	Combinatorial signaling in the heart orchestrates cardiac induction, lineage specification and chamber formation. Seminars in Cell and Developmental Biology, 2007, 18, 54-66.	5.0	53
46	â€˜Big issuesâ€™ in neurodevelopment for children and adults with congenital heart disease. Open Heart, 2019, 6, e000998.	2.3	53
47	Coordination of skeletal muscle gene expression occurs late in mammalian development. Developmental Biology, 1991, 146, 167-178.	2.0	51
48	Cited2 is required in trophoblasts for correct placental capillary patterning. Developmental Biology, 2014, 392, 62-79.	2.0	48
49	Cited2 is required for fetal lung maturation. Developmental Biology, 2008, 317, 95-105.	2.0	47
50	Gonadal defects in Cited2 -mutant mice indicate a role for SF1 in both testis and ovary differentiation. International Journal of Developmental Biology, 2010, 54, 683-689.	0.6	46
51	Gestational stress induces the unfolded protein response, resulting in heart defects. Development (Cambridge), 2016, 143, 2561-2572.	2.5	45
52	Cited2 is required for the proper formation of the hyaloid vasculature and for lens morphogenesis. Development (Cambridge), 2008, 135, 2939-2948.	2.5	44
53	Notch4 reveals a novel mechanism regulating Notch signal transduction. Biochimica Et Biophysica Acta - Molecular Cell Research, 2014, 1843, 1272-1284.	4.1	44
54	Cyclical expression of the Notch/Wnt regulator Nrarp requires modulation by Dll3 in somitogenesis. Developmental Biology, 2009, 329, 400-409.	2.0	43

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55	Gene-environment interaction impacts on heart development and embryo survival. Development (Cambridge), 2019, 146, .	2.5	43
56	Spontaneous Coronary Artery Dissection. Circulation Genomic and Precision Medicine, 2020, 13, e003030.	3.6	43
57	De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects. Human Molecular Genetics, 2017, 26, 4849-4860.	2.9	42
58	Cloning of Mouse cited4, a Member of the CITED Family p300/CBP-Binding Transcriptional Coactivators: Induced Expression in Mammary Epithelial Cells. Genomics, 2002, 80, 601-613.	2.9	41
59	Renal developmental defects resulting from in utero hypoxia are associated with suppression of ureteric Î²-catenin signaling. Kidney International, 2015, 87, 975-983.	5.2	39
60	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. Human Molecular Genetics, 2015, 24, 1234-1242.	2.9	39
61	Dll3 and Notch1 genetic interactions model axial segmental and craniofacial malformations of human birth defects. Developmental Dynamics, 2007, 236, 2943-2951.	1.8	38
62	NAD deficiency due to environmental factors or geneâ€“environment interactions causes congenital malformations and miscarriage in mice. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 3738-3747.	7.1	38
63	Complex SUMO-1 Regulation of Cardiac Transcription Factor Nkx2-5. PLoS ONE, 2011, 6, e24812.	2.5	34
64	Mutation of HES7 in a large extended family with spondylocostal dysostosis and dextrocardia with situs inversus. American Journal of Medical Genetics, Part A, 2013, 161, 2244-2249.	1.2	34
65	Cited2 Regulates Neocortical Layer II/III Generation and Somatosensory Callosal Projection Neuron Development and Connectivity. Journal of Neuroscience, 2016, 36, 6403-6419.	3.6	33
66	A radiation hybrid transcript map of the mouse genome. Nature Genetics, 2001, 29, 194-200.	21.4	32
67	Functional genomics and gene-environment interaction highlight the complexity of congenital heart disease caused by Notch pathway variants. Human Molecular Genetics, 2020, 29, 566-579.	2.9	32
68	Cited2 Is Required for the Maintenance of Glycolytic Metabolism in Adult Hematopoietic Stem Cells. Stem Cells and Development, 2014, 23, 83-94.	2.1	31
69	Breaking symmetry: a clinical overview of left-right patterning. Clinical Genetics, 2004, 65, 441-457.	2.0	28
70	A tyrosine-rich domain within homeodomain transcription factor Nkx2-5 is an essential element in the early cardiac transcriptional regulatory machinery. Development (Cambridge), 2006, 133, 1311-1322.	2.5	28
71	The role of Notch in patterning the human vertebral column. Current Opinion in Genetics and Development, 2009, 19, 329-337.	3.3	28
72	The mouse notches up another success: understanding the causes of human vertebral malformation. Mammalian Genome, 2011, 22, 362-376.	2.2	28

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73	Characterizing Embryonic Gene Expression Patterns in the Mouse Using Nonredundant Sequence-Based Selection. Genome Research, 2003, 13, 2609-2620.	5.5	27
74	Bi-allelic Mutations in NADSYN1 Cause Multiple Organ Defects and Expand the Genotypic Spectrum of Congenital NAD Deficiency Disorders. American Journal of Human Genetics, 2020, 106, 129-136.	6.2	27
75	Diverse requirements for Notch signalling in mammals. International Journal of Developmental Biology, 2002, 46, 365-74.	0.6	27
76	Functional characterization of a novel PBX1 de novo missense variant identified in a patient with syndromic congenital heart disease. Human Molecular Genetics, 2020, 29, 1068-1082.	2.9	26
77	Notch1 endocytosis is induced by ligand and is required for signal transduction. Biochimica Et Biophysica Acta - Molecular Cell Research, 2016, 1863, 166-177.	4.1	24
78	Generation of conditionalCited2 null alleles. Genesis, 2006, 44, 579-583.	1.6	23
79	The Transcriptional Activity of CITED1 Is Regulated by Phosphorylation in a Cell Cycle-dependent Manner. Journal of Biological Chemistry, 2006, 281, 27426-27435.	3.4	23
80	Disruption of the somitic molecular clock causes abnormal vertebral segmentation. Birth Defects Research Part C: Embryo Today Reviews, 2007, 81, 93-110.	3.6	23
81	Placental Insufficiency Associated with Loss of Cited1 Causes Renal Medullary Dysplasia. Journal of the American Society of Nephrology: JASN, 2009, 20, 777-786.	6.1	23
82	A cell autonomous role for the Notch ligand Deltaâ€like 3 in Î±Î² Tâ€cell development. Immunology and Cell Biology, 2011, 89, 696-705.	2.3	23
83	Dynamic expression patterns of the pudgy/spondylocostal dysostosis gene Dll3 in the developing nervous system. Mechanisms of Development, 2001, 100, 141-144.	1.7	22
84	Cited2 Gene Controls Pluripotency and Cardiomyocyte Differentiation of Murine Embryonic Stem Cells through Oct4 Gene. Journal of Biological Chemistry, 2012, 287, 29088-29100.	3.4	22
85	Cited2, a Transcriptional Modulator Protein, Regulates Metabolism in Murine Embryonic Stem Cells. Journal of Biological Chemistry, 2014, 289, 251-263.	3.4	21
86	Conditional deletion of Cited2 results in defective corneal epithelial morphogenesis and maintenance. Developmental Biology, 2009, 334, 243-252.	2.0	20
87	Genetic burden and associations with adverse neurodevelopment in neonates with congenital heart disease. American Heart Journal, 2018, 201, 33-39.	2.7	19
88	Mutation of the fucose-specific Î²1,3 N-acetylglucosaminyltransferase LFNG results in abnormal formation of the spine. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 100-111.	3.8	18
89	Insulin Downregulates the Transcriptional Coregulator CITED2, an Inhibitor of Proangiogenic Function in Endothelial Cells. Diabetes, 2016, 65, 3680-3690.	0.6	18
90	Unrestricted lineage differentiation of parthenogenetic ES cells. Development Genes and Evolution, 1997, 206, 377-388.	0.9	17

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91	Autosomal dominant spondylocostal dysostosis in three generations of a Macedonian family: Negative mutation analysis of DLL3, MESP2, HES7, and LFNG. American Journal of Medical Genetics, Part A, 2010, 152A, 1378-1382.	1.2	17
92	CITED2 limits pathogenic inflammatory gene programs in myeloid cells. FASEB Journal, 2020, 34, 12100-12113.	0.5	17
93	A Genetics-First Approach Revealed Monogenic Disorders in Patients With ARM and VACTERL Anomalies. Frontiers in Pediatrics, 2020, 8, 310.	1.9	17
94	The expression of the imprinted gene Ipl is restricted to extra-embryonic tissues and embryonic lateral mesoderm during early mouse development. International Journal of Developmental Biology, 2002, 46, 459-66.	0.6	17
95	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Human Mutation, 2021, 42, 862-876.	2.5	16
96	Precision Medicine in Cardiovascular Disease: Genetics and Impact on Phenotypes. Journal of the American College of Cardiology, 2021, 77, 2517-2530.	2.8	16
97	<scp>KIAA1217</scp>: A novel candidate gene associated with isolated and syndromic vertebral malformations. American Journal of Medical Genetics, Part A, 2020, 182, 1664-1672.	1.2	15
98	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. Human Molecular Genetics, 2021, 29, 3662-3678.	2.9	14
99	Exploring the Genetic Architecture of Spontaneous Coronary Artery Dissection Using Whole-Genome Sequencing. Circulation Genomic and Precision Medicine, 2022, 15, 101161CIRCGEN121003527.	3.6	14
100	Cited1 Deficiency Suppresses Intestinal Tumorigenesis. PLoS Genetics, 2013, 9, e1003638.	3.5	13
101	Geneâ€“environment interaction demonstrates the vulnerability of the embryonic heart. Developmental Biology, 2014, 391, 99-110.	2.0	13
102	Mig-6 regulates endometrial genes involved in cell cycle and progesterone signaling. Biochemical and Biophysical Research Communications, 2015, 462, 409-414.	2.1	11
103	CITED2 inhibits STAT1â€IRF1 signaling and atherogenesis. FASEB Journal, 2021, 35, e21833.	0.5	11
104	The promises and challenges of exome sequencing in familial, non-syndromic congenital heart disease. International Journal of Cardiology, 2017, 230, 155-163.	1.7	10
105	The pro-death role of Cited2 in stroke is regulated by E2F1/4 transcription factors. Journal of Biological Chemistry, 2019, 294, 8617-8629.	3.4	10
106	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. Genomics, Proteomics and Bioinformatics, 2019, 17, 540-545.	6.9	10
107	Whole genome sequencing in transposition of the great arteries and associations with clinically relevant heart, brain and laterality genes. American Heart Journal, 2022, 244, 1-13.	2.7	10
108	SVPV: a structural variant prediction viewer for paired-end sequencing datasets. Bioinformatics, 2017, 33, 2032-2033.	4.1	9

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109	Deletion of HIF-1Î± partially rescues the abnormal hyaloid vascular system in Cited2 conditional knockout mouse eyes. Molecular Vision, 2012, 18, 1260-70.	1.1	8
110	Molecular diagnosis of vertebral segmentation disorders in humans. Expert Opinion on Medical Diagnostics, 2008, 2, 1107-1121.	1.6	7
111	A new era of genetic testing in congenital heart disease: A review. Trends in Cardiovascular Medicine, 2022, 32, 311-319.	4.9	7
112	Simultaneous quantification of 26 NAD-related metabolites in plasma, blood, and liver tissue using UHPLC-MS/MS. Analytical Biochemistry, 2021, 633, 114409.	2.4	7
113	Benchmarking the Effectiveness and Accuracy of Multiple Mitochondrial DNA Variant Callers: Practical Implications for Clinical Application. Frontiers in Genetics, 2022, 13, 692257.	2.3	6
114	An image analysis protocol using CellProfiler for automated quantification of post-ischemic cardiac parameters. STAR Protocols, 2022, 3, 101097.	1.2	5
115	Spondylocostal dysostosis in a pregnancy complicated by confined placental mosaicism for tetrasomy 9p. American Journal of Medical Genetics, Part A, 2008, 146A, 1972-1976.	1.2	4
116	CHDgene: A Curated Database for Congenital Heart Disease Genes. Circulation Genomic and Precision Medicine, 2022, 15, 101161CIRCGEN121003539.	3.6	4
117	Role of Delta-Like-3 in Mammalian Somitogenesis and Vertebral Column Formation. Advances in Experimental Medicine and Biology, 2008, 638, 95-112.	1.6	3
118	Reprint of mutation of the fucose-specific beta1,3 N-acetylglucosaminyltransferase LFNG results in abnormal formation of the spine. Biochimica Et Biophysica Acta, 2009, 1792, 862-73.	1.3	3
119	Cooperation between somatic Ikaros and Notch1 mutations at the inception of T-ALL. Leukemia Research, 2011, 35, 1512-1519.	0.8	2
120	Genetic and Environmental Interaction in Malformation of the Vertebral Column. , 2015, , 131-151.		2
121	Quantitative 3D analysis and visualization of cardiac fibrosis by microcomputed tomography. STAR Protocols, 2022, 3, 101055.	1.2	2
122	Diseases of development: leveraging developmental biology to understand human disease. Development (Cambridge), 2020, 147, .	2.5	1
123	Ways, means and consequences of shaping morphogen gradients. Current Opinion in Genetics and Development, 2013, 23, 361-362.	3.3	0
124	Four-Generation Family With Ebstein Anomaly Highlights Future Challenges in Congenital Heart Disease Genetics. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	0
125	Câ€…Identification of the major genetic contributors to tetralogy of fallot. , 2019, , .		0
126	Divergent functions and distinct localization of the Notch ligands DLL1 and DLL3 in vivo. Journal of Experimental Medicine, 2007, 204, i20-i20.	8.5	0

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127	Tinman/Nkx2-5 acts via miR-1 and upstream of Cdc42 to regulate heart function across species. Journal of Experimental Medicine, 2011, 208, i20-i20.	8.5	0