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	A new era of genetic testing in congenital heart disease: A review. Trends in Cardiovascular Medicine, 2022, 32, 311-319.	4.9	7
2	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
3	Hif-1a suppresses ROS-induced proliferation of cardiac fibroblasts following myocardial infarction. Cell Stem Cell, 2022, 29, 281-297.e12.	11.1	71
4	Quantitative 3D analysis and visualization of cardiac fibrosis by microcomputed tomography. STAR Protocols, 2022, 3, 101055.	1.2	2
5	An image analysis protocol using CellProfiler for automated quantification of post-ischemic cardiac parameters. STAR Protocols, 2022, 3, 101097.	1.2	5
6	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
7	CHDgene: A Curated Database for Congenital Heart Disease Genes. Circulation Genomic and Precision Medicine, 2022, 15, 101161CIRCGEN121003539.	3.6	4
8	Exploring the Genetic Architecture of Spontaneous Coronary Artery Dissection Using Whole-Genome Sequencing. Circulation Genomic and Precision Medicine, 2022, 15, 101161CIRCGEN121003527.	3.6	14
9	Heterozygous loss of WBP11 function causes multiple congenital defects in humans and mice. Human Molecular Genetics, 2021, 29, 3662-3678.	2.9	14
10	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Human Mutation, 2021, 42, 862-876.	2.5	16
11	Precision Medicine in Cardiovascular Disease: Genetics and Impact on Phenotypes. Journal of the American College of Cardiology, 2021, 77, 2517-2530.	2.8	16
12	CITED2 inhibits STAT1â€IRF1 signaling and atherogenesis. FASEB Journal, 2021, 35, e21833.	0.5	11
13	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
14	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
15	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
16	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
17	Diseases of development: leveraging developmental biology to understand human disease. Development (Cambridge), 2020, 147, .	2.5	1
18	CITED2 limits pathogenic inflammatory gene programs in myeloid cells. FASEB Journal, 2020, 34, 12100-12113.	0.5	17

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19	Spontaneous Coronary Artery Dissection. Circulation Genomic and Precision Medicine, 2020, 13, e003030.	3.6	43
20	<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
21	A Genetics-First Approach Revealed Monogenic Disorders in Patients With ARM and VACTERL Anomalies. Frontiers in Pediatrics, 2020, 8, 310.	1.9	17
22	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
23	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	28.9	113
24	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
25	Gene-environment interaction impacts on heart development and embryo survival. Development (Cambridge), 2019, 146, .	2.5	43
26	Câ€¦Identification of the major genetic contributors to tetralogy of fallot. , 2019, , .		0
27	â€˜Big issuesâ€™ in neurodevelopment for children and adults with congenital heart disease. Open Heart, 2019, 6, e000998.	2.3	53
28	VPOT: A Customizable Variant Prioritization Ordering Tool for Annotated Variants. Genomics, Proteomics and Bioinformatics, 2019, 17, 540-545.	6.9	10
29	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
30	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	4.5	118
31	Identification of clinically actionable variants from genome sequencing of families with congenital heart disease. Genetics in Medicine, 2019, 21, 1111-1120.	2.4	54
32	Genetic burden and associations with adverse neurodevelopment in neonates with congenital heart disease. American Heart Journal, 2018, 201, 33-39.	2.7	19
33	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
34	Advances in the Genetics of Congenital HeartÂDisease. Journal of the American College of Cardiology, 2017, 69, 859-870.	2.8	115
35	SVPV: a structural variant prediction viewer for paired-end sequencing datasets. Bioinformatics, 2017, 33, 2032-2033.	4.1	9
36	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

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37	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
38	NAD Deficiency, Congenital Malformations, and Niacin Supplementation. New England Journal of Medicine, 2017, 377, 544-552.	27.0	177
39	Four-Generation Family With Ebstein Anomaly Highlights Future Challenges in Congenital Heart Disease Genetics. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	0
40	Insulin Downregulates the Transcriptional Coregulator CITED2, an Inhibitor of Proangiogenic Function in Endothelial Cells. Diabetes, 2016, 65, 3680-3690.	0.6	18
41	Gestational stress induces the unfolded protein response, resulting in heart defects. Development (Cambridge), 2016, 143, 2561-2572.	2.5	45
42	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
43	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
44	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
45	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
46	Mig-6 regulates endometrial genes involved in cell cycle and progesterone signaling. Biochemical and Biophysical Research Communications, 2015, 462, 409-414.	2.1	11
47	Compound heterozygous mutations in RIPPLY2 associated with vertebral segmentation defects. Human Molecular Genetics, 2015, 24, 1234-1242.	2.9	39
48	Genetic and Environmental Interaction in Malformation of the Vertebral Column. , 2015, , 131-151.		2
49	NKX2-5 mutations causative for congenital heart disease retain functionality and are directed to hundreds of targets. ELife, 2015, 4, .	6.0	54
50	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
51	Geneâ€“environment interaction demonstrates the vulnerability of the embryonic heart. Developmental Biology, 2014, 391, 99-110.	2.0	13
52	Cited2 is required in trophoblasts for correct placental capillary patterning. Developmental Biology, 2014, 392, 62-79.	2.0	48
53	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
54	Notch4 reveals a novel mechanism regulating Notch signal transduction. Biochimica Et Biophysica Acta - Molecular Cell Research, 2014, 1843, 1272-1284.	4.1	44

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55	Cited2, a Transcriptional Modulator Protein, Regulates Metabolism in Murine Embryonic Stem Cells. Journal of Biological Chemistry, 2014, 289, 251-263.	3.4	21
56	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
57	Ways, means and consequences of shaping morphogen gradients. Current Opinion in Genetics and Development, 2013, 23, 361-362.	3.3	0
58	Cited1 Deficiency Suppresses Intestinal Tumorigenesis. PLoS Genetics, 2013, 9, e1003638.	3.5	13
59	Autosomal dominant spondylocostal dysostosis is caused by mutation in TBX6. Human Molecular Genetics, 2013, 22, 1625-1631.	2.9	87
60	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
61	HIF-1Î± deletion partially rescues defects of hematopoietic stem cell quiescence caused by Cited2 deficiency. Blood, 2012, 119, 2789-2798.	1.4	55
62	A Mechanism for Gene-Environment Interaction in the Etiology of Congenital Scoliosis. Cell, 2012, 149, 295-306.	28.9	188
63	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
64	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
65	Complex SUMO-1 Regulation of Cardiac Transcription Factor Nkx2-5. PLoS ONE, 2011, 6, e24812.	2.5	34
66	Cooperation between somatic Ikaros and Notch1 mutations at the inception of T-ALL. Leukemia Research, 2011, 35, 1512-1519.	0.8	2
67	The mouse notches up another success: understanding the causes of human vertebral malformation. Mammalian Genome, 2011, 22, 362-376.	2.2	28
68	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
69	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
70	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
71	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
72	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

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73	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
74	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
75	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
76	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
77	The Role of Hypoxia in Development of the Mammalian Embryo. Developmental Cell, 2009, 17, 755-773.	7.0	509
78	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
79	The role of Notch in patterning the human vertebral column. Current Opinion in Genetics and Development, 2009, 19, 329-337.	3.3	28
80	Cyclical expression of the Notch/Wnt regulator Nrarp requires modulation by Dll3 in somitogenesis. Developmental Biology, 2009, 329, 400-409.	2.0	43
81	Conditional deletion of Cited2 results in defective corneal epithelial morphogenesis and maintenance. Developmental Biology, 2009, 334, 243-252.	2.0	20
82	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
83	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
84	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
85	Cited2 is required for fetal lung maturation. Developmental Biology, 2008, 317, 95-105.	2.0	47
86	Molecular diagnosis of vertebral segmentation disorders in humans. Expert Opinion on Medical Diagnostics, 2008, 2, 1107-1121.	1.6	7
87	Cited2 is required for the proper formation of the hyaloid vasculature and for lens morphogenesis. Development (Cambridge), 2008, 135, 2939-2948.	2.5	44
88	Mutation of HAIRY-AND-ENHANCER-OF-SPLIT-7 in humans causes spondylocostal dysostosis. Human Molecular Genetics, 2008, 17, 3761-3766.	2.9	123
89	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
90	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

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91	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
92	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
93	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
94	Abnormal vertebral segmentation and the notch signaling pathway in man. Developmental Dynamics, 2007, 236, 1456-1474.	1.8	143
95	Dll3 and Notch1 genetic interactions model axial segmental and craniofacial malformations of human birth defects. Developmental Dynamics, 2007, 236, 2943-2951.	1.8	38
96	Cited2, a coactivator of HNF4Î±, is essential for liver development. EMBO Journal, 2007, 26, 4445-4456.	7.8	70
97	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
98	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
99	Loss of Cited2 affects trophoblast formation and vascularization of the mouse placenta. Developmental Biology, 2006, 294, 67-82.	2.0	101
100	Generation of conditionalCited2 null alleles. Genesis, 2006, 44, 579-583.	1.6	23
101	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
102	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
103	Evolution of distinct EGF domains with specific functions. Protein Science, 2005, 14, 1091-1103.	7.6	155
104	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
105	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
106	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
107	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
108	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

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109	Breaking symmetry: a clinical overview of left-right patterning. Clinical Genetics, 2004, 65, 441-457.	2.0	28
110	Dll3 pudgy mutation differentially disrupts dynamic expression of somite genes. Genesis, 2004, 39, 115-121.	1.6	53
111	Developmental regulation of Notch signaling genes in the embryonic pituitary: Prop1 deficiency affects Notch2 expression. Developmental Biology, 2004, 265, 329-340.	2.0	110
112	Mutated MESP2 Causes Spondylocostal Dysostosis in Humans. American Journal of Human Genetics, 2004, 74, 1249-1254.	6.2	157
113	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
114	Characterizing Embryonic Gene Expression Patterns in the Mouse Using Nonredundant Sequence-Based Selection. Genome Research, 2003, 13, 2609-2620.	5.5	27
115	Folic acid prevents exencephaly in Cited2 deficient mice. Human Molecular Genetics, 2002, 11, 283-293.	2.9	145
116	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
117	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
118	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
119	Diverse requirements for Notch signalling in mammals. International Journal of Developmental Biology, 2002, 46, 365-74.	0.6	27
120	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
121	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
122	A radiation hybrid transcript map of the mouse genome. Nature Genetics, 2001, 29, 194-200.	21.4	32
123	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
124	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
125	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
126	Unrestricted lineage differentiation of parthenogenetic ES cells. Development Genes and Evolution, 1997, 206, 377-388.	0.9	17

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127	Coordination of skeletal muscle gene expression occurs late in mammalian development. Developmental Biology, 1991, 146, 167-178.	2.0	51