William J Pavan

List of Publications by Year
in descending order

123	10,501	36303	33894
papers	citations	51 h-index	99 g-index

126	126	126	12597
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Niemann-Pick C1 Disease Gene: Homology to Mediators of Cholesterol Homeostasis. Science, 1997, 277, 228-231.	12.6	1,373
2	Murine Model of Niemann-Pick C Disease: Mutation in a Cholesterol Homeostasis Gene. Science, 1997, 277, 232-235.	12.6	766
3	SOX10 mutation disrupts neural crest development in Dom Hirschsprung mouse model. Nature Genetics, 1998, 18, 60-64.	21.4	702
4	Transcription factor hierarchy in Waardenburg syndrome: regulation of MITF expression by SOX10 and PAX3. Human Genetics, 2000, 107, 1-6.	3.8	323
5	Intrathecal 2-hydroxypropyl-Î²-cyclodextrin decreases neurological disease progression in Niemann-Pick disease, type C1: a non-randomised, open-label, phase 1â€“2 trial. Lancet, The, 2017, 390, 1758-1768.	13.7	275
6	Glial-cell-derived neuroregulators control type 3 innate lymphoid cells and gut defence. Nature, 2016, 535, 440-443.	27.8	272
7	Melanoma mouse model implicates metabotropic glutamate signaling in melanocytic neoplasia. Nature Genetics, 2003, 34, 108-112.	21.4	260
8	Loci associated with skin pigmentation identified in African populations. Science, 2017, 358, .	12.6	260
9	The melanomas: a synthesis of epidemiological, clinical, histopathological, genetic, and biological aspects, supporting distinct subtypes, causal pathways, and cells of origin. Pigment Cell and Melanoma Research, 2011, 24, 879-897.	3.3	225
10	The importance of having your SOX on: role of SOX10â€ in the development of neural crest-derived melanocytes and glia. Oncogene, 2003, 22, 3024-3034.	5.9	192
11	Strategic vision for improving human health at The Forefront of Genomics. Nature, 2020, 586, 683-692.	27.8	192
12	A Polymorphism in IRF4 Affects Human Pigmentation through a Tyrosinase-Dependent MITF/TFAP2A Pathway. Cell, 2013, 155, 1022-1033.	28.9	184
13	Analysis of SOX10 Function in Neural Crest-Derived Melanocyte Development: SOX10-Dependent Transcriptional Control of Dopachrome Tautomerase. Developmental Biology, 2001, 237, 245-257.	2.0	169
14	Transcriptional and signaling regulation in neural crest stem cell-derived melanocyte development: do all roads lead to Mitf?. Cell Research, 2008, 18, 1163-1176.	12.0	168
15	The exon junction complex component Magoh controls brain size by regulating neural stem cell division. Nature Neuroscience, 2010, 13, 551-558.	14.8	156
16	Mutation of melanosome protein RAB38 in chocolate mice. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 4471-4476.	7.1	155
17	Targeting a Complex Transcriptome: The Construction of the Mouse Full-Length cDNA Encyclopedia. Genome Research, 2003, 13, 1273-1289.	5.5	154
18	Linear clinical progression, independent of age of onset, in Niemannâ€“Pick disease, type C. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 132-140.	1.7	145

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19	Sox proteins in melanocyte development and melanoma. Pigment Cell and Melanoma Research, 2010, 23, 496-513.	3.3	138
20	Frequent mutations in the MITF pathway in melanoma. Pigment Cell and Melanoma Research, 2009, 22, 435-444.	3.3	132
21	The Sox10^{Dom} Mouse: Modeling the Genetic Variation of Waardenburg-Shah (WS4) Syndrome. Genome Research, 1999, 9, 215-225.	5.5	127
22	Interspecies difference in the regulation of melanocyte development by SOX10 and MITF. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 9081-9085.	7.1	117
23	Oxidative stress in Niemannâ€“Pick disease, type C. Molecular Genetics and Metabolism, 2010, 101, 214-218.	1.1	113
24	Collaborative Development of 2-Hydroxypropyl-β-Cyclodextrin for the Treatment of Niemann-Pick Type C1 Disease. Current Topics in Medicinal Chemistry, 2014, 14, 330-339.	2.1	108
25	Î´-Tocopherol Reduces Lipid Accumulation in Niemann-Pick Type C1 and Wolman Cholesterol Storage Disorders. Journal of Biological Chemistry, 2012, 287, 39349-39360.	3.4	107
26	The Secreted Metalloprotease ADAMTS20 Is Required for Melanoblast Survival. PLoS Genetics, 2008, 4, e1000003.	3.5	102
27	Genomic copy number and expression variation within the C57BL/6J inbred mouse strain. Genome Research, 2008, 18, 60-66.	5.5	100
28	An evolutionarily conserved intronic region controls the spatiotemporal expression of the transcription factor Sox10. BMC Developmental Biology, 2008, 8, 105.	2.1	99
29	Identification of Neural Crest and Glial Enhancers at the Mouse Sox10 Locus through Transgenesis in Zebrafish. PLoS Genetics, 2008, 4, e1000174.	3.5	99
30	The Genetics of Human Skin and Hair Pigmentation. Annual Review of Genomics and Human Genetics, 2019, 20, 41-72.	6.2	98
31	Mutations in TRIOBP, Which Encodes a Putative Cytoskeletal-Organizing Protein, Are Associated with Nonsyndromic Recessive Deafness. American Journal of Human Genetics, 2006, 78, 137-143.	6.2	93
32	Transcription factor hierarchy in Waardenburg syndrome: regulation of MITF expression by SOX10 and PAX3. Human Genetics, 2000, 107, 1-6.	3.8	92
33	Spotlight on Spotted Mice: A Review of White Spotting Mouse Mutants and Associated Human Pigmentation Disorders. Pigment Cell & Melanoma Research, 2004, 17, 215-224.	3.6	91
34	Direct Interaction of Sox10 with the Promoter of Murine Dopachrome Tautomerase (Dct) and Synergistic Activation of Dct Expression with Mitf. Pigment Cell & Melanoma Research, 2004, 17, 352-362.	3.6	89
35	A Dual Role for SOX10 in the Maintenance of the Postnatal Melanocyte Lineage and the Differentiation of Melanocyte Stem Cell Progenitors. PLoS Genetics, 2013, 9, e1003644.	3.5	85
36	Microarray expression analysis and identification of serum biomarkers for Niemannâ€“Pick disease, type C1. Human Molecular Genetics, 2012, 21, 3632-3646.	2.9	84

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37	A defect in a novel ADAMTS family member is the cause of the belted white-spotting mutation. Development (Cambridge), 2003, 130, 4665-4672.	2.5	80
38	Hepatocyte Growth Factor/Scatter Factor-MET Signaling in Neural Crest-Derived Melanocyte Development. Pigment Cell & Melanoma Research, 1999, 12, 13-21.	3.6	79
39	Pmel17 expression is Mitf-dependent and reveals cranial melanoblast migration during murine development. Gene Expression Patterns, 2003, 3, 703-707.	0.8	79
40	TFAP2 paralogs regulate melanocyte differentiation in parallel with MITF. PLoS Genetics, 2017, 13, e1006636.	3.5	78
41	WNT1 and WNT3a promote expansion of melanocytes through distinct modes of action. Pigment Cell & Melanoma Research, 2005, 18, 167-180.	3.6	76
42	Rescue of neurodegeneration in Niemann-Pick C mice by a prion-promoter-driven Npc1 cDNA transgene. Human Molecular Genetics, 2002, 11, 3107-3114.	2.9	73
43	A curated gene list for expanding the horizons of pigmentation biology. Pigment Cell and Melanoma Research, 2019, 32, 348-358.	3.3	72
44	NRG1â€Š/â€ŠERBB3 signaling in melanocyte development and melanoma: inhibition of differentiation and promotion of proliferation. Pigment Cell and Melanoma Research, 2009, 22, 773-784.	3.3	70
45	SOX10 Ablation Arrests Cell Cycle, Induces Senescence, and Suppresses Melanomagenesis. Cancer Research, 2013, 73, 5709-5718.	0.9	70
46	Cell-typeâ€“specific eQTL of primary melanocytes facilitates identification of melanoma susceptibility genes. Genome Research, 2018, 28, 1621-1635.	5.5	67
47	The mouse neurological mutant weaver maps within the region of chromosome 16 that is homologous to human chromosome 21. Genomics, 1989, 5, 522-526.	2.9	66
48	Integration of ChIP-seq and machine learning reveals enhancers and a predictive regulatory sequence vocabulary in melanocytes. Genome Research, 2012, 22, 2290-2301.	5.5	64
49	Endothelin signalling in the development of neural crest-derived melanocytes. Biochemistry and Cell Biology, 1998, 76, 1093-1099.	2.0	62
50	Isolation, genomic organization, and expression analysis of Men1, the murine homolog of the MEN1 gene. Mammalian Genome, 1999, 10, 592-596.	2.2	62
51	Cell-autonomous and cell non-autonomous signaling through endothelin receptor B during melanocyte development. Development (Cambridge), 2004, 131, 3239-3247.	2.5	62
52	Deletion of long-range sequences at Sox10 compromises developmental expression in a mouse model of Waardenburgâ€“Shah (WS4) syndrome. Human Molecular Genetics, 2006, 15, 259-271.	2.9	60
53	Specification of neural crest into sensory neuron and melanocyte lineages. Developmental Biology, 2012, 366, 55-63.	2.0	56
54	A sensitized mutagenesis screen identifies Gli3 as a modifier of Sox10 neurocristopathy. Human Molecular Genetics, 2008, 17, 2118-2131.	2.9	53

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55	High-efficiency yeast artificial chromosome fragmentation vectors. Gene, 1991, 106, 125-127.	2.2	51
56	Gpnmb is a melanoblastâ€expressed, MITFâ€dependent gene. Pigment Cell and Melanoma Research, 2009, 22, 99-110.	3.3	51
57	SOX10 directly modulates ERBB3 transcription via an intronic neural crest enhancer. BMC Developmental Biology, 2011, 11, 40.	2.1	51
58	BRG1 interacts with SOX10 to establish the melanocyte lineage and to promote differentiation. Nucleic Acids Research, 2017, 45, 6442-6458.	14.5	51
59	Systemic AAV9 gene therapy improves the lifespan of mice with Niemann-Pick disease, type C1. Human Molecular Genetics, 2017, 26, ddw367.	2.9	50
60	Rescue of an In Vitro Neuron Phenotype Identified in Niemann-Pick Disease, Type C1 Induced Pluripotent Stem Cell-Derived Neurons by Modulating the WNT Pathway and Calcium Signaling. Stem Cells Translational Medicine, 2015, 4, 230-238.	3.3	48
61	Mutation of the Diamond-Blackfan Anemia Gene Rps7 in Mouse Results in Morphological and Neuroanatomical Phenotypes. PLoS Genetics, 2013, 9, e1003094.	3.5	47
62	A direct link between MITF, innate immunity, and hair graying. PLoS Biology, 2018, 16, e2003648.	5.6	47
63	Modeling Smith-Lemli-Opitz syndrome with induced pluripotent stem cells reveals a causal role for Wnt/Î²-catenin defects in neuronal cholesterol synthesis phenotypes. Nature Medicine, 2016, 22, 388-396.	30.7	46
64	The etiology and molecular genetics of human pigmentation disorders. Wiley Interdisciplinary Reviews: Developmental Biology, 2013, 2, 379-392.	5.9	44
65	Modeling Niemann-Pick disease type C1 in zebrafish: a robust platform for in vivo screening of candidate therapeutic compounds. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	38
66	The EJC component Magoh regulates proliferation and expansion of neural crest-derived melanocytes. Developmental Biology, 2013, 375, 172-181.	2.0	36
67	Genomic analysis reveals distinct mechanisms and functional classes of SOX10-regulated genes in melanocytes. Human Molecular Genetics, 2015, 24, 5433-5450.	2.9	34
68	A unique missense allele of BAF155, a core BAF chromatin remodeling complex protein, causes neural tube closure defects in mice. Developmental Neurobiology, 2014, 74, 483-497.	3.0	33
69	Analysis of Ocular Hypopigmentation inRab38^{cht/cht}Mice. , 2007, 48, 3905.		31
70	The oculocutaneous albinism type IV gene Matp is a new marker of pigment cell precursors during mouse embryonic development. Mechanisms of Development, 2002, 116, 209-212.	1.7	30
71	Hypoxiaâ€induced <scp>HIF</scp>1Î± targets in melanocytes reveal a molecular profile associated with poor melanoma prognosis. Pigment Cell and Melanoma Research, 2017, 30, 339-352.	3.3	29
72	The Genetic Regulation of Pigment Cell Development. , 2006, 589, 155-169.		28

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73	Efficacy of N-acetylcysteine in phenotypic suppression of mouse models of Niemannâ€“Pick disease, type C1. Human Molecular Genetics, 2013, 22, 3508-3523.	2.9	27
74	Genomic Organization of the Murine Millerâ€“Dieker/Lissencephaly Region: Conservation of Linkage with the Humanâ€‰Region. Genome Research, 1997, 7, 625-634.	5.5	26
75	Highly Efficient Cpf1-Mediated Gene Targeting in Mice Following High Concentration Pronuclear Injection. G3: Genes, Genomes, Genetics, 2017, 7, 719-722.	1.8	25
76	Identification and functional analysis of SOX10 phosphorylation sites in melanoma. PLoS ONE, 2018, 13, e0190834.	2.5	24
77	Induction of RET Dependent and Independent Pro-Inflammatory Programs in Human Peripheral Blood Mononuclear Cells from Hirschsprung Patients. PLoS ONE, 2013, 8, e59066.	2.5	24
78	Genetic evidence does not support direct regulation of EDNRB by SOX10 in migratory neural crest and the melanocyte lineage. Mechanisms of Development, 2006, 123, 124-134.	1.7	23
79	Stem cells of the melanocyte lineage. Cancer Biomarkers, 2007, 3, 203-209.	1.7	23
80	Networks and pathways in pigmentation, health, and disease. Wiley Interdisciplinary Reviews: Systems Biology and Medicine, 2009, 1, 359-371.	6.6	23
81	Defective Cytochrome P450-Catalysed Drug Metabolism in Niemann-Pick Type C Disease. PLoS ONE, 2016, 11, e0152007.	2.5	22
82	Oligodendroglial and panâ€neural crest expression of Cre recombinase directed by Sox10 enhancer. Genesis, 2009, 47, 765-770.	1.6	21
83	Acinar Cell Apoptosis in Serpini2-Deficient Mice Models Pancreatic Insufficiency. PLoS Genetics, 2005, 1, e38.	3.5	20
84	A Sox10 Expression Screen Identifies an Amino Acid Essential for Erbb3 Function. PLoS Genetics, 2008, 4, e1000177.	3.5	20
85	Plasma and Tissue Concentrations of Î±-Tocopherol and Î´-Tocopherol Following High Dose Dietary Supplementation in Mice. Nutrients, 2012, 4, 467-490.	4.1	20
86	A new glucocerebrosidase deficient neuronal cell model provides a tool to probe pathophysiology and therapeutics for Gaucher disease. DMM Disease Models and Mechanisms, 2016, 9, 769-78.	2.4	20
87	The RhoJ-BAD signaling network: An Achillesâ€™ heel for BRAF mutant melanomas. PLoS Genetics, 2017, 13, e1006913.	3.5	20
88	Cloning and tissue expression of the mouse ortholog of AIM1, a Î²Î³-crystallin superfamily member. Mammalian Genome, 1998, 9, 715-720.	2.2	19
89	A Metagenomics Study on Hirschsprung's Disease Associated Enterocolitis: Biodiversity and Gut Microbial Homeostasis Depend on Resection Length and Patient's Clinical History. Frontiers in Pediatrics, 2019, 7, 326.	1.9	19
90	[49] Yeast artificial chromosome modification and manipulation. Methods in Enzymology, 1992, 216, 584-603.	1.0	18

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91	Complementation of melanocyte development in SOX10 mutant neural crest using lineage-directed gene transfer. Developmental Dynamics, 2004, 229, 54-62.	1.8	17
92	In Niemann-Pick C1 mouse models, glial-only expression of the normal gene extends survival much further than do changes in genetic background or treatment with hydroxypropyl-beta-cyclodextrin. Gene, 2018, 643, 117-123.	2.2	17
93	Genetic background modifies phenotypic severity and longevity in a mouse model of Niemann-Pick disease type C1. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	17
94	Comparison of melanoblast expression patterns identifies distinct classes of genes. Pigment Cell and Melanoma Research, 2009, 22, 611-622.	3.3	16
95	NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. Journal of Clinical Medicine, 2020, 9, 12.	2.4	16
96	Comparative analyses of the Dominant megacolon-SOX10 genomic interval in mouse and human. Mammalian Genome, 1999, 10, 744-749.	2.2	15
97	The Use of Expression Profiling to Study Pigment Cell Biology and Dysfunction. Pigment Cell & Melanoma Research, 2000, 13, 141-146.	3.6	15
98	Transcriptome of HPÎ²CD-treated Niemann-Pick disease type C1 cells highlights GPNMB as a biomarker for therapeutics. Human Molecular Genetics, 2021, 30, 2456-2468.	2.9	15
99	The transcription factors Ets1 and Sox10 interact during murine melanocyte development. Developmental Biology, 2015, 407, 300-312.	2.0	14
100	A Rare Myelin Protein Zero (MPZ) Variant Alters Enhancer Activity In Vitro and In Vivo. PLoS ONE, 2010, 5, e14346.	2.5	14
101	Localization of a neural crest transcription factor, Slug, to mouse Chromosome 16 and human Chromosome 8. Mammalian Genome, 1997, 8, 872-873.	2.2	13
102	MEK inhibition remodels the active chromatin landscape and induces SOX10 genomic recruitment in BRAF(V600E) mutant melanoma cells. Epigenetics and Chromatin, 2019, 12, 50.	3.9	12
103	Modification and manipulation of mammalian DNA cloned as YACs. Gene Analysis Techniques, 1990, 7, 107-113.	1.0	11
104	The pleiotropic mouse phenotype extraâ€toes spotting is caused by translation initiation factorEif3cmutations and is associated with disrupted sonic hedgehog signaling. FASEB Journal, 2011, 25, 1596-1605.	0.5	11
105	Postnatal lineage mapping of follicular melanocytes with the <scp>T</scp>yr::<scp>C</scp>re<scp>ER^T</scp>² transgene. Pigment Cell and Melanoma Research, 2013, 26, 269-274.	3.3	10
106	A mouse model of Waardenburg syndrome type IV resulting from an ENU-induced mutation in endothelin 3. Pigment Cell & Melanoma Research, 2007, 20, 210-215.	3.6	8
107	A curated online resource for SOX10 and pigment cell molecular genetic pathways. Database: the Journal of Biological Databases and Curation, 2010, 2010, baq025-baq025.	3.0	8
108	Human Disease Genes and Their Cloned Mouse Orthologs: Exploration of the FANTOM2 cDNA Sequence Data Set. Genome Research, 2003, 13, 1496-1500.	5.5	7

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109	A somatic cell defect is associated with the onset of neurological symptoms in a lysosomal storage disease. Molecular Genetics and Metabolism, 2013, 110, 188-190.	1.1	7
110	Ectopic differentiation of melanocyte stem cells is influenced by genetic background. Pigment Cell and Melanoma Research, 2015, 28, 223-228.	3.3	7
111	A custom capture sequence approach for oculocutaneous albinism identifies structural variant alleles at the OCA2 locus. Human Mutation, 2021, 42, 1239-1253.	2.5	7
112	Improved systemic AAV gene therapy with a neurotrophic capsid in Niemannâ€“Pick disease type C1 mice. Life Science Alliance, 2021, 4, e202101040.	2.8	6
113	Maternal immune activation modifies the course of Niemann-pick disease, type C1 in a gender specific manner. Molecular Genetics and Metabolism, 2020, 129, 165-170.	1.1	5
114	The Origin and Development of Neural Crest-Derived Melanocytes. , 2006, , 3-26.		5
115	Identification of Gene Variants Associated with Melanocyte Stem Cell Differentiation in Mice Predisposed for Hair Graying. G3: Genes, Genomes, Genetics, 2019, 9, 817-827.	1.8	4
116	Informatic and genomic analysis of melanocyte cDNA libraries as a resource for the study of melanocyte development and function. Pigment Cell & Melanoma Research, 2007, 20, 201-209.	3.6	3
117	Correlation of age of onset and clinical severity in Niemannâ€“Pick disease type C1 with lysosomal abnormalities and gene expression. Scientific Reports, 2022, 12, 2162.	3.3	3
118	Melanoma to Vitiligo: The Melanocyte in Biology & Medicineâ€“Joint Montagna Symposium on the Biology of Skin/PanAmerican Society for Pigment Cell Research Annual Meeting. Journal of Investigative Dermatology, 2020, 140, 269-274.	0.7	2
119	Axing the cancer loop. Pigment Cell and Melanoma Research, 2014, 27, 691-693.	3.3	1
120	Purification and Characterization of YACs Containing Large Inserts. , 2001, Chapter 5, Unit 5.7.		0
121	Î´-Tocopherol reduces lipid accumulation in Niemann-Pick type C1 and Wolman cholesterol storage disorders.. Journal of Biological Chemistry, 2013, 288, 296.	3.4	0
122	Gene therapy for the treatment of Niemann-Pick disease type C1: Comparison of AAV9 to a novel serotype, AAV-PHP.B. Molecular Genetics and Metabolism, 2018, 123, S36-S37.	1.1	0
123	Improved disease amelioration with combination therapy for Niemann-Pick disease type C1. Molecular Genetics and Metabolism, 2019, 126, S46.	1.1	0