

Stephen J Tapscott

List of Publications by Year in descending order

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Version: 2024-02-01

78
papers

8,326
citations

76326

40
h-index

74163

75
g-index

85
all docs

85
docs citations

85
times ranked

6551
citing authors

#	ARTICLE	IF	CITATIONS
1	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. <i>Science</i> , 2010, 329, 1650-1653.	12.6	638
2	The circuitry of a master switch: Myod and the regulation of skeletal muscle gene transcription. <i>Development (Cambridge)</i> , 2005, 132, 2685-2695.	2.5	612
3	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. <i>Nature Genetics</i> , 2012, 44, 1370-1374.	21.4	582
4	Conserved roles of mouse DUX and human DUX4 in activating cleavage-stage genes and MERVL/HERVL retrotransposons. <i>Nature Genetics</i> , 2017, 49, 925-934.	21.4	545
5	Genome-wide MyoD Binding in Skeletal Muscle Cells: A Potential for Broad Cellular Reprogramming. <i>Developmental Cell</i> , 2010, 18, 662-674.	7.0	434
6	Facioscapulohumeral Dystrophy: Incomplete Suppression of a Retrotransposed Gene. <i>PLoS Genetics</i> , 2010, 6, e1001181.	3.5	394
7	DUX4 Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy. <i>Developmental Cell</i> , 2012, 22, 38-51.	7.0	384
8	CTCF-binding sites flank CTG/CAG repeats and form a methylation-sensitive insulator at the DM1 locus. <i>Nature Genetics</i> , 2001, 28, 335-343.	21.4	301
9	Conservation and innovation in the DUX4-family gene network. <i>Nature Genetics</i> , 2017, 49, 935-940.	21.4	265
10	Trinucleotide repeat expansion at the myotonic dystrophy locus reduces expression of DMAHP. <i>Nature Genetics</i> , 1997, 16, 402-406.	21.4	242
11	Global and gene-specific analyses show distinct roles for Myod and Myog at a common set of promoters. <i>EMBO Journal</i> , 2006, 25, 502-511.	7.8	227
12	Expression profiling of FSHD muscle supports a defect in specific stages of myogenic differentiation. <i>Human Molecular Genetics</i> , 2003, 12, 2895-2907.	2.9	191
13	Mutations in DNMT3B Modify Epigenetic Repression of the D4Z4 Repeat and the Penetrance of Facioscapulohumeral Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 98, 1020-1029.	6.2	188
14	RNA transcripts, miRNA-sized fragments and proteins produced from D4Z4 units: new candidates for the pathophysiology of facioscapulohumeral dystrophy. <i>Human Molecular Genetics</i> , 2009, 18, 2414-2430.	2.9	182
15	DUX4-induced gene expression is the major molecular signature in FSHD skeletal muscle. <i>Human Molecular Genetics</i> , 2014, 23, 5342-5352.	2.9	170
16	The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. <i>American Journal of Human Genetics</i> , 2013, 93, 744-751.	6.2	154
17	DUX4 Binding to Retroelements Creates Promoters That Are Active in FSHD Muscle and Testis. <i>PLoS Genetics</i> , 2013, 9, e1003947.	3.5	151
18	Facioscapulohumeral dystrophy: the path to consensus on pathophysiology. <i>Skeletal Muscle</i> , 2014, 4, 12.	4.2	144

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19	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. <i>Human Molecular Genetics</i> , 2015, 24, 659-669.	2.9	130
20	Molecular Distinction between Specification and Differentiation in the Myogenic Basic Helix-Loop-Helix Transcription Factor Family. <i>Molecular and Cellular Biology</i> , 2001, 21, 2404-2412.	2.3	120
21	Differential genomic targeting of the transcription factor TAL1 in alternate haematopoietic lineages. <i>EMBO Journal</i> , 2011, 30, 494-509.	7.8	120
22	Genetic and Epigenetic Determinants of Neurogenesis and Myogenesis. <i>Developmental Cell</i> , 2012, 22, 721-735.	7.0	100
23	A feedback loop between nonsense-mediated decay and the retrogene DUX4 in facioscapulohumeral muscular dystrophy. <i>ELife</i> , 2015, 4, .	6.0	97
24	Intrinsic Epigenetic Regulation of the D4Z4 Macrosatellite Repeat in a Transgenic Mouse Model for FSHD. <i>PLoS Genetics</i> , 2013, 9, e1003415.	3.5	95
25	MRI-informed muscle biopsies correlate MRI with pathology and DUX4 target gene expression in FSHD. <i>Human Molecular Genetics</i> , 2019, 28, 476-486.	2.9	86
26	Distinct Activities of Myf5 and MyoD Indicate Separate Roles in Skeletal Muscle Lineage Specification and Differentiation. <i>Developmental Cell</i> , 2016, 36, 375-385.	7.0	85
27	MyoD and E-protein heterodimers switch rhabdomyosarcoma cells from an arrested myoblast phase to a differentiated state. <i>Genes and Development</i> , 2009, 23, 694-707.	5.9	84
28	DUX4-induced dsRNA and MYC mRNA stabilization activate apoptotic pathways in human cell models of facioscapulohumeral dystrophy. <i>PLoS Genetics</i> , 2017, 13, e1006658.	3.5	77
29	DUX4 Suppresses MHC Class I to Promote Cancer Immune Evasion and Resistance to Checkpoint Blockade. <i>Developmental Cell</i> , 2019, 50, 658-671.e7.	7.0	76
30	Model systems of DUX4 expression recapitulate the transcriptional profile of FSHD cells. <i>Human Molecular Genetics</i> , 2016, 25, ddw271.	2.9	75
31	Genetic and epigenetic contributors to FSHD. <i>Current Opinion in Genetics and Development</i> , 2015, 33, 56-61.	3.3	69
32	Generation of Isogenic D4Z4 Contracted and Noncontracted Immortal Muscle Cell Clones from a Mosaic Patient. <i>American Journal of Pathology</i> , 2012, 181, 1387-1401.	3.8	63
33	Clinically Advanced p38 Inhibitors Suppress DUX4 Expression in Cellular and Animal Models of Facioscapulohumeral Muscular Dystrophy. <i>Journal of Pharmacology and Experimental Therapeutics</i> , 2019, 370, 219-230.	2.5	58
34	Increased DUX4 expression during muscle differentiation correlates with decreased SMCHD1 protein levels at D4Z4. <i>Epigenetics</i> , 2015, 10, 1133-1142.	2.7	52
35	Muscle pathology grade for facioscapulohumeral muscular dystrophy biopsies. <i>Muscle and Nerve</i> , 2015, 52, 521-526.	2.2	50
36	Correlation analysis of clinical parameters with epigenetic modifications in the DUX4 promoter in FSHD. <i>Epigenetics</i> , 2012, 7, 579-584.	2.7	48

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37	NuRD and CAF-1-mediated silencing of the D4Z4 array is modulated by DUX4-induced MBD3L proteins. <i>ELife</i> , 2018, 7, .	6.0	47
38	BET bromodomain inhibitors and agonists of the beta-2 adrenergic receptor identified in screens for compounds that inhibit DUX4 expression in FSHD muscle cells. <i>Skeletal Muscle</i> , 2017, 7, 16.	4.2	46
39	Facioscapulohumeral dystrophy: activating an early embryonic transcriptional program in human skeletal muscle. <i>Human Molecular Genetics</i> , 2018, 27, R153-R162.	2.9	46
40	Single-cell RNA sequencing in facioscapulohumeral muscular dystrophy disease etiology and development. <i>Human Molecular Genetics</i> , 2019, 28, 1064-1075.	2.9	46
41	Conversion of MyoD to a Neurogenic Factor: Binding Site Specificity Determines Lineage. <i>Cell Reports</i> , 2015, 10, 1937-1946.	6.4	45
42	Milder phenotype in facioscapulohumeral dystrophy with 7â€“10 residual D4Z4 repeats. <i>Neurology</i> , 2015, 85, 2147-2150.	1.1	44
43	Immunodetection of Human Double Homeobox 4. <i>Hybridoma</i> , 2011, 30, 125-130.	0.4	43
44	Clinical trial preparedness in facioscapulohumeral muscular dystrophy: Clinical, tissue, and imaging outcome measures 29â€“30 May 2015, Rochester, New York. <i>Neuromuscular Disorders</i> , 2016, 26, 181-186.	0.6	43
45	Facioscapulohumeral muscular dystrophy. <i>Current Opinion in Neurology</i> , 2012, 25, 614-620.	3.6	42
46	Multiplex Screen of Serum Biomarkers in Facioscapulohumeral Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 181-190.	2.6	38
47	Longitudinal measures of RNA expression and disease activity in FSHD muscle biopsies. <i>Human Molecular Genetics</i> , 2020, 29, 1030-1043.	2.9	38
48	DICER/AGO-dependent epigenetic silencing of D4Z4 repeats enhanced by exogenous siRNA suggests mechanisms and therapies for FSHD. <i>Human Molecular Genetics</i> , 2015, 24, 4817-4828.	2.9	37
49	Surgical management of superficial siderosis following cervical nerve root avulsion. <i>Annals of Neurology</i> , 1996, 40, 936-940.	5.3	35
50	DUX4-Induced Histone Variants H3.X and H3.Y Mark DUX4 Target Genes for Expression. <i>Cell Reports</i> , 2019, 29, 1812-1820.e5.	6.4	34
51	Quantitative proteomics reveals key roles for post-transcriptional gene regulation in the molecular pathology of facioscapulohumeral muscular dystrophy. <i>ELife</i> , 2019, 8, .	6.0	34
52	Hemizyosity for <i>SMCHD1</i> in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. <i>Human Mutation</i> , 2015, 36, 679-683.	2.5	32
53	<i>SMCHD1</i> regulates a limited set of gene clusters on autosomal chromosomes. <i>Skeletal Muscle</i> , 2017, 7, 12.	4.2	32
54	Intronic <i>SMCHD1</i> variants in FSHD: testing the potential for CRISPR-Cas9 genome editing. <i>Journal of Medical Genetics</i> , 2019, 56, 828-837.	3.2	27

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55	Immunohistochemical Characterization of Facioscapulohumeral Muscular Dystrophy Muscle Biopsies. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 291-299.	2.6	26
56	DUX4-induced bidirectional HSATII satellite repeat transcripts form intranuclear double-stranded RNA foci in human cell models of FSHD. <i>Human Molecular Genetics</i> , 2019, 28, 3997-4011.	2.9	26
57	FSHD type 2 and Bosma arhinia microphthalmia syndrome. <i>Neurology</i> , 2018, 91, e562-e570.	1.1	24
58	Double SMCHD1 variants in FSHD2: the synergistic effect of two SMCHD1 variants on D4Z4 hypomethylation and disease penetrance in FSHD2. <i>European Journal of Human Genetics</i> , 2016, 24, 78-85.	2.8	23
59	Smchd1 haploinsufficiency exacerbates the phenotype of a transgenic FSHD1 mouse model. <i>Human Molecular Genetics</i> , 2018, 27, 716-731.	2.9	23
60	Deep characterization of a common D4Z4 variant identifies biallelic DUX4 expression as a modifier for disease penetrance in FSHD2. <i>European Journal of Human Genetics</i> , 2018, 26, 94-106.	2.8	22
61	Unchecked oxidative stress in skeletal muscle prevents outgrowth of disseminated tumour cells. <i>Nature Cell Biology</i> , 2022, 24, 538-553.	10.3	20
62	DUX4 promotes transcription of FRG2 by directly activating its promoter in facioscapulohumeral muscular dystrophy. <i>Skeletal Muscle</i> , 2014, 4, 19.	4.2	19
63	Chromosome 10q-linked FSHD identifies DUX4 as principal disease gene. <i>Journal of Medical Genetics</i> , 2022, 59, 180-188.	3.2	18
64	Genome-wide binding of the basic helix-loop-helix myogenic inhibitor myocardin has substantial overlap with MyoD: implications for buffering activity. <i>Skeletal Muscle</i> , 2013, 3, 26.	4.2	16
65	Fundamental differences in promoter CpG island DNA hypermethylation between human cancer and genetically engineered mouse models of cancer. <i>Epigenetics</i> , 2013, 8, 1254-1260.	2.7	16
66	A fragment of the neurogenin1 gene confers regulated expression of a reporter gene in vitro and in vivo. <i>Development</i> , 2000, 127, 189-194.		14
67	Facioscapulohumeral dystrophy transcriptome signatures correlate with different stages of disease and are marked by different MRI biomarkers. <i>Scientific Reports</i> , 2022, 12, 1426.	3.3	14
68	Impediment of Replication Forks by Long Non-coding RNA Provokes Chromosomal Rearrangements by Error-Prone Restart. <i>Cell Reports</i> , 2017, 21, 2223-2235.	6.4	13
69	Monosomy 18p is a risk factor for facioscapulohumeral dystrophy. <i>Journal of Medical Genetics</i> , 2018, 55, 469-478.	3.2	11
70	Systemic delivery of a DUX4-targeting antisense oligonucleotide to treat facioscapulohumeral muscular dystrophy. <i>Molecular Therapy - Nucleic Acids</i> , 2021, 26, 813-827.	5.1	11
71	Elevated plasma complement components in facioscapulohumeral dystrophy. <i>Human Molecular Genetics</i> , 2022, 31, 1821-1829.	2.9	10
72	Small noncoding RNAs in FSHD2 muscle cells reveal both DUX4- and SMCHD1-specific signatures. <i>Human Molecular Genetics</i> , 2018, 27, 2644-2657.	2.9	6

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73	TWIST1 Heterodimerization with E12 Requires Coordinated Protein Phosphorylation to Regulate Periostin Expression. <i>Cancers</i> , 2019, 11, 1392.	3.7	4
74	Canine DUXC: implications for DUX4 retrotransposition and preclinical models of FSHD. <i>Human Molecular Genetics</i> , 2022, 31, 1694-1704.	2.9	4
75	Identification of SMCHD1 domains for nuclear localization, homo-dimerization, and protein cleavage. <i>Skeletal Muscle</i> , 2018, 8, 24.	4.2	2
76	A proteomics study identifying interactors of the FSHD2 gene product SMCHD1 reveals RUVBL1-dependent DUX4 repression. <i>Scientific Reports</i> , 2021, 11, 23642.	3.3	2
77	Abstract of Symposium. <i>Human Cell</i> , 2005, 18, 29-33.	2.7	0
78	Electronic Detection of Micro RNA Mir206 with Molecularly-Differentiated Nanoelectrodes. , 2007, , .		0