Stephen J Tapscott

List of Publications by Year in descending order

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	Chromosome 10q-linked FSHD identifies DUX4 as principal disease gene. Journal of Medical Genetics, 2022, 59, 180-188.	3.2	18
2	Facioscapulohumeral dystrophy transcriptome signatures correlate with different stages of disease and are marked by different MRI biomarkers. Scientific Reports, 2022, 12, 1426.	3.3	14
3	Canine DUXC: implications for DUX4 retrotransposition and preclinical models of FSHD. Human Molecular Genetics, 2022, 31, 1694-1704.	2.9	4
4	Elevated plasma complement components in facioscapulohumeral dystrophy. Human Molecular Genetics, 2022, 31, 1821-1829.	2.9	10
5	Unchecked oxidative stress in skeletal muscle prevents outgrowth of disseminated tumour cells. Nature Cell Biology, 2022, 24, 538-553.	10.3	20
6	Systemic delivery of a DUX4-targeting antisense oligonucleotide to treat facioscapulohumeral muscular dystrophy. Molecular Therapy - Nucleic Acids, 2021, 26, 813-827.	5.1	11
7	A proteomics study identifying interactors of the FSHD2 gene product SMCHD1 reveals RUVBL1-dependent DUX4 repression. Scientific Reports, 2021, 11, 23642.	3.3	2
8	Longitudinal measures of RNA expression and disease activity in FSHD muscle biopsies. Human Molecular Genetics, 2020, 29, 1030-1043.	2.9	38
9	DUX4 Suppresses MHC Class I to Promote Cancer Immune Evasion and Resistance to Checkpoint Blockade. Developmental Cell, 2019, 50, 658-671.e7.	7.0	76
10	DUX4-induced bidirectional HSATII satellite repeat transcripts form intranuclear double-stranded RNA foci in human cell models of FSHD. Human Molecular Genetics, 2019, 28, 3997-4011.	2.9	26
11	Intronic SMCHD1 variants in FSHD: testing the potential for CRISPR-Cas9 genome editing. Journal of Medical Genetics, 2019, 56, 828-837.	3.2	27
12	DUX4-Induced Histone Variants H3.X and H3.Y Mark DUX4 Target Genes for Expression. Cell Reports, 2019, 29, 1812-1820.e5.	6.4	34
13	TWIST1 Heterodimerization with E12 Requires Coordinated Protein Phosphorylation to Regulate Periostin Expression. Cancers, 2019, 11, 1392.	3.7	4
14	Clinically Advanced p38 Inhibitors Suppress DUX4 Expression in Cellular and Animal Models of Facioscapulohumeral Muscular Dystrophy. Journal of Pharmacology and Experimental Therapeutics, 2019, 370, 219-230.	2.5	58
15	Single-cell RNA sequencing in facioscapulohumeral muscular dystrophy disease etiology and development. Human Molecular Genetics, 2019, 28, 1064-1075.	2.9	46
16	MRI-informed muscle biopsies correlate MRI with pathology and DUX4 target gene expression in FSHD. Human Molecular Genetics, 2019, 28, 476-486.	2.9	86
17	Quantitative proteomics reveals key roles for post-transcriptional gene regulation in the molecular pathology of facioscapulohumeral muscular dystrophy. ELife, 2019, 8, .	6.0	34
18	Smchd1 haploinsufficiency exacerbates the phenotype of a transgenic FSHD1 mouse model. Human Molecular Genetics, 2018, 27, 716-731.	2.9	23

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19	Facioscapulohumeral dystrophy: activating an early embryonic transcriptional program in human skeletal muscle. Human Molecular Genetics, 2018, 27, R153-R162.	2.9	46
20	Monosomy 18p is a risk factor for facioscapulohumeral dystrophy. Journal of Medical Genetics, 2018, 55, 469-478.	3.2	11
21	Deep characterization of a common D4Z4 variant identifies biallelic DUX4 expression as a modifier for disease penetrance in FSHD2. European Journal of Human Genetics, 2018, 26, 94-106.	2.8	22
22	NuRD and CAF-1-mediated silencing of the D4Z4 array is modulated by DUX4-induced MBD3L proteins. ELife, 2018, 7, .	6.0	47
23	Identification of SMCHD1 domains for nuclear localization, homo-dimerization, and protein cleavage. Skeletal Muscle, 2018, 8, 24.	4.2	2
24	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24
25	Small noncoding RNAs in FSHD2 muscle cells reveal both DUX4- and SMCHD1-specific signatures. Human Molecular Genetics, 2018, 27, 2644-2657.	2.9	6
26	Conserved roles of mouse DUX and human DUX4 in activating cleavage-stage genes and MERVL/HERVL retrotransposons. Nature Genetics, 2017, 49, 925-934.	21.4	545
27	Conservation and innovation in the DUX4-family gene network. Nature Genetics, 2017, 49, 935-940.	21.4	265
28	Impediment of Replication Forks by Long Non-coding RNA Provokes Chromosomal Rearrangements by Error-Prone Restart. Cell Reports, 2017, 21, 2223-2235.	6.4	13
29	SMCHD1 regulates a limited set of gene clusters on autosomal chromosomes. Skeletal Muscle, 2017, 7, 12.	4.2	32
30	BET bromodomain inhibitors and agonists of the beta-2 adrenergic receptor identified in screens for compounds that inhibit DUX4 expression in FSHD muscle cells. Skeletal Muscle, 2017, 7, 16.	4.2	46
31	DUX4-induced dsRNA and MYC mRNA stabilization activate apoptotic pathways in human cell models of facioscapulohumeral dystrophy. PLoS Genetics, 2017, 13, e1006658.	3.5	77
32	Mutations in DNMT3B Modify Epigenetic Repression of the D4Z4 Repeat and the Penetrance of Facioscapulohumeral Dystrophy. American Journal of Human Genetics, 2016, 98, 1020-1029.	6.2	188
33	Model systems of DUX4 expression recapitulate the transcriptional profile of FSHD cells. Human Molecular Genetics, 2016, 25, ddw271.	2.9	75
34	Distinct Activities of Myf5 and MyoD Indicate Separate Roles in Skeletal Muscle Lineage Specification and Differentiation. Developmental Cell, 2016, 36, 375-385.	7.0	85
35	Clinical trial preparedness in facioscapulohumeral muscular dystrophy: Clinical, tissue, and imaging outcome measures 29â€“30 May 2015, Rochester, New York. Neuromuscular Disorders, 2016, 26, 181-186.	0.6	43
36	Double SMCHD1 variants in FSHD2: the synergistic effect of two SMCHD1 variants on D4Z4 hypomethylation and disease penetrance in FSHD2. European Journal of Human Genetics, 2016, 24, 78-85.	2.8	23

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37	Immunohistochemical Characterization ofÂFacioscapulohumeralMuscular DystrophyÂMuscle Biopsies. Journal of Neuromuscular Diseases, 2015, 2, 291-299.	2.6	26
38	A feedback loop between nonsense-mediated decay and the retrogene DUX4 in facioscapulohumeral muscular dystrophy. ELife, 2015, 4, .	6.0	97
39	Muscle pathology grade for facioscapulohumeral muscular dystrophy biopsies. Muscle and Nerve, 2015, 52, 521-526.	2.2	50
40	Milder phenotype in facioscapulohumeral dystrophy with 7â€“10 residual D4Z4 repeats. Neurology, 2015, 85, 2147-2150.	1.1	44
41	Conversion of MyoD to a Neurogenic Factor: Binding Site Specificity Determines Lineage. Cell Reports, 2015, 10, 1937-1946.	6.4	45
42	DICER/AGO-dependent epigenetic silencing of D4Z4 repeats enhanced by exogenous siRNA suggests mechanisms and therapies for FSHD. Human Molecular Genetics, 2015, 24, 4817-4828.	2.9	37
43	Genetic and epigenetic contributors to FSHD. Current Opinion in Genetics and Development, 2015, 33, 56-61.	3.3	69
44	Hemizygosity forSMCHD1in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. Human Mutation, 2015, 36, 679-683.	2.5	32
45	Increased DUX4 expression during muscle differentiation correlates with decreased SMCHD1 protein levels at D4Z4. Epigenetics, 2015, 10, 1133-1142.	2.7	52
46	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. Human Molecular Genetics, 2015, 24, 659-669.	2.9	130
47	Multiplex Screen of Serum Biomarkers in Facioscapulohumeral Muscular Dystrophy. Journal of Neuromuscular Diseases, 2014, 1, 181-190.	2.6	38
48	DUX4-induced gene expression is the major molecular signature in FSHD skeletal muscle. Human Molecular Genetics, 2014, 23, 5342-5352.	2.9	170
49	DUX4 promotes transcription of FRG2 by directly activating its promoter in facioscapulohumeral muscular dystrophy. Skeletal Muscle, 2014, 4, 19.	4.2	19
50	Facioscapulohumeral dystrophy: the path to consensus on pathophysiology. Skeletal Muscle, 2014, 4, 12.	4.2	144
51	The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. American Journal of Human Genetics, 2013, 93, 744-751.	6.2	154
52	Genome-wide binding of the basic helix-loop-helix myogenic inhibitor musculin has substantial overlap with MyoD: implications for buffering activity. Skeletal Muscle, 2013, 3, 26.	4.2	16
53	DUX4 Binding to Retroelements Creates Promoters That Are Active in FSHD Muscle and Testis. PLoS Genetics, 2013, 9, e1003947.	3.5	151
54	Intrinsic Epigenetic Regulation of the D4Z4 Macrosatellite Repeat in a Transgenic Mouse Model for FSHD. PLoS Genetics, 2013, 9, e1003415.	3.5	95

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55	Fundamental differences in promoter CpG island DNA hypermethylation between human cancer and genetically engineered mouse models of cancer. Epigenetics, 2013, 8, 1254-1260.	2.7	16
56	Correlation analysis of clinical parameters with epigenetic modifications in the DUX4 promoter in FSHD. Epigenetics, 2012, 7, 579-584.	2.7	48
57	Facioscapulohumeral muscular dystrophy. Current Opinion in Neurology, 2012, 25, 614-620.	3.6	42
58	Generation of Isogenic D4Z4 Contracted and Noncontracted Immortal Muscle Cell Clones from a Mosaic Patient. American Journal of Pathology, 2012, 181, 1387-1401.	3.8	63
59	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. Nature Genetics, 2012, 44, 1370-1374.	21.4	582
60	DUX4 Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy. Developmental Cell, 2012, 22, 38-51.	7.0	384
61	Genetic and Epigenetic Determinants of Neurogenesis and Myogenesis. Developmental Cell, 2012, 22, 721-735.	7.0	100
62	Differential genomic targeting of the transcription factor TAL1 in alternate haematopoietic lineages. EMBO Journal, 2011, 30, 494-509.	7.8	120
63	Immunodetection of Human Double Homeobox 4. Hybridoma, 2011, 30, 125-130.	0.4	43
64	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. Science, 2010, 329, 1650-1653.	12.6	638
65	Facioscapulohumeral Dystrophy: Incomplete Suppression of a Retrotransposed Gene. PLoS Genetics, 2010, 6, e1001181.	3.5	394
66	Genome-wide MyoD Binding in Skeletal Muscle Cells: A Potential for Broad Cellular Reprogramming. Developmental Cell, 2010, 18, 662-674.	7.0	434
67	RNA transcripts, miRNA-sized fragments and proteins produced from D4Z4 units: new candidates for the pathophysiology of facioscapulohumeral dystrophy. Human Molecular Genetics, 2009, 18, 2414-2430.	2.9	182
68	MyoD and E-protein heterodimers switch rhabdomyosarcoma cells from an arrested myoblast phase to a differentiated state. Genes and Development, 2009, 23, 694-707.	5.9	84
69	Electronic Detection of Micro RNA Mir206 with Molecularly-Differentiated Nanoelectrodes. , 2007, , .		0
70	Global and gene-specific analyses show distinct roles for Myod and Myog at a common set of promoters. EMBO Journal, 2006, 25, 502-511.	7.8	227
71	The circuitry of a master switch: Myod and the regulation of skeletal muscle gene transcription. Development (Cambridge), 2005, 132, 2685-2695.	2.5	612
72	Abstract of Symposium. Human Cell, 2005, 18, 29-33.	2.7	0

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73	Expression profiling of FSHD muscle supports a defect in specific stages of myogenic differentiation. Human Molecular Genetics, 2003, 12, 2895-2907.	2.9	191
74	CTCF-binding sites flank CTG/CAG repeats and form a methylation-sensitive insulator at the DM1 locus. Nature Genetics, 2001, 28, 335-343.	21.4	301
75	Molecular Distinction between Specification and Differentiation in the Myogenic Basic Helix-Loop-Helix Transcription Factor Family. Molecular and Cellular Biology, 2001, 21, 2404-2412.	2.3	120
76	A fragment of theneurogenin1 gene confers regulated expression of a reporter gene in vitro and in vivo. , 2000, 218, 189-194.		14
77	Trinucleotide repeat expansion at the myotonic dystrophy locus reduces expression of DMAHP. Nature Genetics, 1997, 16, 402-406.	21.4	242
78	Surgical management of superficial siderosis following cervical nerve root avulsion. Annals of Neurology, 1996, 40, 936-940.	5.3	35