SilvÃ¨re M Van Der Maarel

List of Publications by Year
in descending order

201 papers	13,780 citations	22099 59 h-index	29081 104 g-index
212 all docs	212 docs citations	212 times ranked	8614 citing authors

#	Article	IF	Citations
1	Chromosome 10q-linked FSHD identifies DUX4 as principal disease gene. Journal of Medical Genetics, 2022, 59, 180-188.	1.5	18
2	High-resolution breakpoint junction mapping of proximally extended D4Z4 deletions in FSHD1 reveals evidence for a founder effect. Human Molecular Genetics, 2022, 31, 748-760.	1.4	8
3	Facioscapulohumeral dystrophy transcriptome signatures correlate with different stages of disease and are marked by different MRI biomarkers. Scientific Reports, 2022, 12, 1426.	1.6	14
4	Elevated plasma complement components in facioscapulohumeral dystrophy. Human Molecular Genetics, 2022, 31, 1821-1829.	1.4	10
5	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499.	0.6	26
6	Functional monovalency amplifies the pathogenicity of anti-MuSK IgG4 in myasthenia gravis. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	28
7	Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation. JCI Insight, 2021, 6, .	2.3	38
8	p53 convergently activates Dux/DUX4 in embryonic stem cells and in facioscapulohumeral muscular dystrophy cell models. Nature Genetics, 2021, 53, 1207-1220.	9.4	59
9	Adenine base editing of the DUX4 polyadenylation signal for targeted genetic therapy in facioscapulohumeral muscular dystrophy. Molecular Therapy - Nucleic Acids, 2021, 25, 342-354.	2.3	12
10	Identical twins carry a persistent epigenetic signature of early genome programming. Nature Communications, 2021, 12, 5618.	5.8	26
11	Systemic delivery of a DUX4-targeting antisense oligonucleotide to treat facioscapulohumeral muscular dystrophy. Molecular Therapy - Nucleic Acids, 2021, 26, 813-827.	2.3	11
12	Profiling Serum Antibodies Against Muscle Antigens in Facioscapulohumeral Muscular Dystrophy Finds No Disease-Specific Autoantibodies. Journal of Neuromuscular Diseases, 2021, 8, 801-814.	1.1	6
13	A proteomics study identifying interactors of the FSHD2 gene product SMCHD1 reveals RUVBL1-dependent DUX4 repression. Scientific Reports, 2021, 11, 23642.	1.6	2
14	SATB1, genomic instability and Gleason grading constitute a novel risk score for prostate cancer. Scientific Reports, 2021, 11, 24446.	1.6	1
15	Integrating gene delivery and gene-editing technologies by adenoviral vector transfer of optimized CRISPR-Cas9 components. Gene Therapy, 2020, 27, 209-225.	2.3	42
16	Magnetic resonance imaging correlates with electrical impedance myography in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2020, 61, 644-649.	1.0	10
17	Loss of ZBTB24 impairs nonhomologous end-joining and class-switch recombination in patients with ICF syndrome. Journal of Experimental Medicine, 2020, 217, .	4.2	27
18	The prospects of targeting DUX4 in facioscapulohumeral muscular dystrophy. Current Opinion in Neurology, 2020, 33, 635-640.	1.8	7

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19	Genetic testing offer for inherited neuromuscular diseases within the EURO-NMD reference network: A European survey study. PLoS ONE, 2020, 15, e0239329.	1.1	6
20	Homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e2441-e2447.	1.5	84
21	Longitudinal measures of RNA expression and disease activity in FSHD muscle biopsies. Human Molecular Genetics, 2020, 29, 1030-1043.	1.4	38
22	Consequences of epigenetic derepression in facioscapulohumeral muscular dystrophy. Clinical Genetics, 2020, 97, 799-814.	1.0	40
23	Preserved single muscle fiber specific force in facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e1157-e1170.	1.5	8
24	Dnmt3b regulates DUX4 expression in a tissue-dependent manner in transgenic D4Z4 mice. Skeletal Muscle, 2020, 10, 27.	1.9	5
25	Title is missing!. , 2020, 15, e0239329.		0
26	Title is missing!. , 2020, 15, e0239329.		0
27	Title is missing!. , 2020, 15, e0239329.		0
28	Title is missing!. , 2020, 15, e0239329.		0
29	SMCHD1 mutation spectrum for facioscapulohumeral muscular dystrophy type 2 (FSHD2) and Bosma arhinia microphthalmia syndrome (BAMS) reveals disease-specific localisation of variants in the ATPase domain. Journal of Medical Genetics, 2019, 56, 693-700.	1.5	27
30	Ophthalmological findings in facioscapulohumeral dystrophy. Brain Communications, 2019, 1, fcz023.	1.5	14
31	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.	1.4	26
32	Intronic SMCHD1 variants in FSHD: testing the potential for CRISPR-Cas9 genome editing. Journal of Medical Genetics, 2019, 56, 828-837.	1.5	27
33	DUX4-Induced Histone Variants H3.X and H3.Y Mark DUX4 Target Genes for Expression. Cell Reports, 2019, 29, 1812-1820.e5.	2.9	34
34	Generation of genetically matched hiPSC lines from two mosaic facioscapulohumeral dystrophy type 1 patients. Stem Cell Research, 2019, 40, 101560.	0.3	6
35	A functional assay to classify ZBTB24 missense variants of unknown significance. Human Mutation, 2019, 40, 1077-1083.	1.1	6
36	FSHD1 and FSHD2 form a disease continuum. Neurology, 2019, 92, e2273-e2285.	1.5	50

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37	Efgartigimod improves muscle weakness in a mouse model for muscle-specific kinase myasthenia gravis. Experimental Neurology, 2019, 317, 133-143.	2.0	25
38	MuSK myasthenia gravis monoclonal antibodies. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e547.	3.1	64
39	Early onset as a marker for disease severity in facioscapulohumeral muscular dystrophy. Neurology, 2019, 92, e378-e385.	1.5	30
40	Single-cell RNA sequencing in facioscapulohumeral muscular dystrophy disease etiology and development. Human Molecular Genetics, 2019, 28, 1064-1075.	1.4	46
41	IgG4â€mediated autoimmune diseases: a niche of antibodyâ€mediated disorders. Annals of the New York Academy of Sciences, 2018, 1413, 92-103.	1.8	54
42	Passive transfer models of myasthenia gravis with muscleâ€specific kinase antibodies. Annals of the New York Academy of Sciences, 2018, 1413, 111-118.	1.8	4
43	Smchd1 haploinsufficiency exacerbates the phenotype of a transgenic FSHD1 mouse model. Human Molecular Genetics, 2018, 27, 716-731.	1.4	23
44	Monosomy 18p is a risk factor for facioscapulohumeral dystrophy. Journal of Medical Genetics, 2018, 55, 469-478.	1.5	11
45	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.	1.4	22
46	The Effect of Corticosteroids on Human Choroidal Endothelial Cells: A Model to Study Central Serous Chorioretinopathy. , 2018, 59, 5682.		19
47	Autosomal genetic variation is associated with DNA methylation in regions variably escaping X-chromosome inactivation. Nature Communications, 2018, 9, 3738.	5.8	24
48	Facioscapulohumeral Dystrophy in Childhood: A Nationwide Natural History Study. Annals of Neurology, 2018, 84, 627-637.	2.8	21
49	Phenotypeâ€genotype relations in facioscapulohumeral muscular dystrophy type 1. Clinical Genetics, 2018, 94, 521-527.	1.0	25
50	A 22-year follow-up reveals a variable disease severity in early-onset facioscapulohumeral dystrophy. European Journal of Paediatric Neurology, 2018, 22, 782-785.	0.7	8
51	NuRD and CAF-1-mediated silencing of the D4Z4 array is modulated by DUX4-induced MBD3L proteins. ELife, 2018, 7, .	2.8	47
52	Cis D4Z4 repeat duplications associated with facioscapulohumeral muscular dystrophy type 2. Human Molecular Genetics, 2018, 27, 3488-3497.	1.4	27
53	Brain Transcriptomic Analysis of Hereditary Cerebral Hemorrhage With Amyloidosis-Dutch Type. Frontiers in Aging Neuroscience, 2018, 10, 102.	1.7	13
54	A family-based study into penetrance in facioscapulohumeral muscular dystrophy type 1. Neurology, 2018, 91, e444-e454.	1.5	33

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55	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.5	24
56	Small noncoding RNAs in FSHD2 muscle cells reveal both DUX4- and SMCHD1-specific signatures. Human Molecular Genetics, 2018, 27, 2644-2657.	1.4	6
57	225th ENMC international workshop:. Neuromuscular Disorders, 2017, 27, 782-790.	0.3	20
58	IgG4 autoantibodies against muscle-specific kinase undergo Fab-arm exchange in myasthenia gravis patients. Journal of Autoimmunity, 2017, 77, 104-115.	3.0	92
59	Adding quantitative muscle MRI to the FSHD clinical trial toolbox. Neurology, 2017, 89, 2057-2065.	1.5	72
60	SMCHD1 regulates a limited set of gene clusters on autosomal chromosomes. Skeletal Muscle, 2017, 7, 12.	1.9	32
61	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.	1.9	46
62	DUX4-induced dsRNA and MYC mRNA stabilization activate apoptotic pathways in human cell models of facioscapulohumeral dystrophy. PLoS Genetics, 2017, 13, e1006658.	1.5	77
63	DUX4 induces a transcriptome more characteristic of a less-differentiated cell state and inhibits myogenesis. Journal of Cell Science, 2016, 129, 3816-3831.	1.2	77
64	Integrating clinical and genetic observations in facioscapulohumeral muscular dystrophy. Current Opinion in Neurology, 2016, 29, 606-613.	1.8	10
65	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.	2.6	188
66	Myasthenia gravis with muscle specific kinase antibodies mimicking amyotrophic lateral sclerosis. Neuromuscular Disorders, 2016, 26, 350-353.	0.3	24
67	Converging disease genes in ICF syndrome:ZBTB24controls expression ofCDCA7in mammals. Human Molecular Genetics, 2016, 25, 4041-4051.	1.4	49
68	Model systems of DUX4 expression recapitulate the transcriptional profile of FSHD cells. Human Molecular Genetics, 2016, 25, ddw271.	1.4	75
69	Facioscapulohumeral dystrophy in children: design of a prospective, observational study on natural history, predictors and clinical impact (iFocus FSHD). BMC Neurology, 2016, 16, 138.	0.8	15
70	Longitudinal epitope mapping in MuSK myasthenia gravis: implications for disease severity. Journal of Neuroimmunology, 2016, 291, 82-88.	1.1	59
71	Allele-specific DNA hypomethylation characterises FSHD1 and FSHD2. Journal of Medical Genetics, 2016, 53, 348-355.	1.5	54
72	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.	0.3	43

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73	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.	1.4	23
74	DUX4 induces a transcriptome more characteristic of a less-differentiated cell state and inhibits myogenesis. Development (Cambridge), 2016, 143, e1.1-e1.1.	1.2	0
75	Differential myofiber-type transduction preference of adeno-associated virus serotypes 6 and 9. Skeletal Muscle, 2015, 5, 37.	1.9	31
76	Camelid heavy chain only antibody fragment domain against Î²â€site of amyloid precursor protein cleaving enzyme 1 inhibits Î²â€secretase activity inÂvitro and inÂvivo. FEBS Journal, 2015, 282, 3618-3631.	2.2	15
77	Immunohistochemical Characterization ofÂFacioscapulohumeralMuscular DystrophyÂMuscle Biopsies. Journal of Neuromuscular Diseases, 2015, 2, 291-299.	1.1	26
78	A feedback loop between nonsense-mediated decay and the retrogene DUX4 in facioscapulohumeral muscular dystrophy. ELife, 2015, 4, .	2.8	97
79	Muscle pathology grade for facioscapulohumeral muscular dystrophy biopsies. Muscle and Nerve, 2015, 52, 521-526.	1.0	50
80	Milder phenotype in facioscapulohumeral dystrophy with 7â€“10 residual D4Z4 repeats. Neurology, 2015, 85, 2147-2150.	1.5	44
81	Enhanced glutathione PEGylated liposomal brain delivery of an anti-amyloid single domain antibody fragment in a mouse model for Alzheimer's disease. Journal of Controlled Release, 2015, 203, 40-50.	4.8	114
82	Selection and characterization of llama single domain antibodies against N-terminal huntingtin. Neurological Sciences, 2015, 36, 429-434.	0.9	16
83	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.	1.4	37
84	Facioscapulohumeral Dystrophy. , 2015, , 620-630.		0
85	Fusion of hIgG1-Fc to 111In-anti-amyloid single domain antibody fragment VHH-pa2H prolongs blood residential time in APP/PS1 mice but does not increase brain uptake. Nuclear Medicine and Biology, 2015, 42, 695-702.	0.3	47
86	Early-Onset Facioscapulohumeral Muscular Dystrophy Type 1 With Some Atypical Features. Journal of Child Neurology, 2015, 30, 580-587.	0.7	14
87	Genetic and epigenetic contributors to FSHD. Current Opinion in Genetics and Development, 2015, 33, 56-61.	1.5	69
88	Hemizygosity forSMCHD1in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. Human Mutation, 2015, 36, 679-683.	1.1	32
89	Mutations in CDCA7 and HELLS cause immunodeficiencyâ€“centromeric instabilityâ€“facial anomalies syndrome. Nature Communications, 2015, 6, 7870.	5.8	148
90	Genome-wide binding and mechanistic analyses of Smchd1-mediated epigenetic regulation. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E3535-44.	3.3	83

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91	Increased DUX4 expression during muscle differentiation correlates with decreased SMCHD1 protein levels at D4Z4. Epigenetics, 2015, 10, 1133-1142.	1.3	52
92	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. Human Molecular Genetics, 2015, 24, 659-669.	1.4	130
93	Multiplex Screen of Serum Biomarkers in Facioscapulohumeral Muscular Dystrophy. Journal of Neuromuscular Diseases, 2014, 1, 181-190.	1.1	38
94	Centromeric Instability in ICF Syndrome. , 2014, , 427-433.		0
95	DUX4-induced gene expression is the major molecular signature in FSHD skeletal muscle. Human Molecular Genetics, 2014, 23, 5342-5352.	1.4	170
96	DUX4 promotes transcription of FRG2 by directly activating its promoter in facioscapulohumeral muscular dystrophy. Skeletal Muscle, 2014, 4, 19.	1.9	19
97	A Novel Feed-Forward Loop between ARIH2 E3-Ligase and PABPN1 Regulates Aging-Associated Muscle Degeneration. American Journal of Pathology, 2014, 184, 1119-1131.	1.9	27
98	Genetic and Epigenetic Characteristics of FSHD-Associated 4q and 10q D4Z4 that are Distinct from Non-4q/10q D4Z4 Homologs. Human Mutation, 2014, 35, 998-1010.	1.1	42
99	Population-based incidence and prevalence of facioscapulohumeral dystrophy. Neurology, 2014, 83, 1056-1059.	1.5	278
100	Facioscapulohumeral dystrophy: the path to consensus on pathophysiology. Skeletal Muscle, 2014, 4, 12.	1.9	144
101	DNA polymorphism and epigenetic marks modulate the affinity of a scaffold/matrix attachment region to the nuclear matrix. European Journal of Human Genetics, 2014, 22, 1117-1123.	1.4	14
102	High prevalence of incomplete right bundle branch block in facioscapulohumeral muscular dystrophy without cardiac symptoms. Functional Neurology, 2014, 29, 159-65.	1.3	18
103	Genome-wide analysis of macrosatellite repeat copy number variation in worldwide populations: evidence for differences and commonalities in size distributions and size restrictions. BMC Genomics, 2013, 14, 143.	1.2	29
104	Nuclear entrapment and extracellular depletion of PCOLCE is associated with muscle degeneration in oculopharyngeal muscular dystrophy. BMC Neurology, 2013, 13, 70.	0.8	15
105	MuSK IgG4 autoantibodies cause myasthenia gravis by inhibiting binding between MuSK and Lrp4. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 20783-20788.	3.3	234
106	Dysferlin Regulates Cell Adhesion in Human Monocytes. Journal of Biological Chemistry, 2013, 288, 14147-14157.	1.6	49
107	Determining the role of sarcomeric proteins in facioscapulohumeral muscular dystrophy: a study protocol. BMC Neurology, 2013, 13, 144.	0.8	12
108	The FSHD2 Gene SMCHD1 Is a Modifier of Disease Severity in Families Affected by FSHD1. American Journal of Human Genetics, 2013, 93, 744-751.	2.6	154

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109	Antibodies to active zone protein ERC1 in Lambertâ€“Eaton myasthenic syndrome. Human Immunology, 2013, 74, 849-851.	1.2	10
110	Sarcomeric dysfunction contributes to muscle weakness in facioscapulohumeral muscular dystrophy. Neurology, 2013, 80, 733-737.	1.5	36
111	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. European Journal of Human Genetics, 2013, 21, 1219-1225.	1.4	115
112	DUX4 Binding to Retroelements Creates Promoters That Are Active in FSHD Muscle and Testis. PLoS Genetics, 2013, 9, e1003947.	1.5	151
113	Intrinsic Epigenetic Regulation of the D4Z4 Macrosatellite Repeat in a Transgenic Mouse Model for FSHD. PLoS Genetics, 2013, 9, e1003415.	1.5	95
114	Chromatin remodeling of human subtelomeres and TERRA promoters upon cellular senescence. Epigenetics, 2013, 8, 512-521.	1.3	25
115	A focal domain of extreme demethylation within D4Z4 in FSHD2. Neurology, 2013, 80, 392-399.	1.5	67
116	A decline in PABPN1 induces progressive muscle weakness in Oculopharyngeal muscle dystrophy and in muscle aging. Aging, 2013, 5, 412-426.	1.4	49
117	Correlation analysis of clinical parameters with epigenetic modifications in the DUX4 promoter in FSHD. Epigenetics, 2012, 7, 579-584.	1.3	48
118	Epigenetic regulation of the X-chromosomal macrosatellite repeat encoding for the cancer/testis gene CT47. European Journal of Human Genetics, 2012, 20, 185-191.	1.4	15
119	Facioscapulohumeral muscular dystrophy. Current Opinion in Neurology, 2012, 25, 614-620.	1.8	42
120	Poly(A) binding protein nuclear 1 levels affect alternative polyadenylation. Nucleic Acids Research, 2012, 40, 9089-9101.	6.5	148
121	Patients with a phenotype consistent with facioscapulohumeral muscular dystrophy display genetic and epigenetic heterogeneity. Journal of Medical Genetics, 2012, 49, 41-46.	1.5	55
122	Selfâ€regulated alternative splicing at the AHNAK locus. FASEB Journal, 2012, 26, 93-103.	0.2	24
123	Muscle-specific kinase myasthenia gravis IgG4 autoantibodies cause severe neuromuscular junction dysfunction in mice. Brain, 2012, 135, 1081-1101.	3.7	180
124	Generation of Isogenic D4Z4 Contracted and Noncontracted Immortal Muscle Cell Clones from a Mosaic Patient. American Journal of Pathology, 2012, 181, 1387-1401.	1.9	63
125	Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. Nature Genetics, 2012, 44, 1370-1374.	9.4	582
126	Best practice guidelines on genetic diagnostics of Facioscapulohumeral muscular dystrophy: Workshop 9th June 2010, LUMC, Leiden, The Netherlands. Neuromuscular Disorders, 2012, 22, 463-470.	0.3	53

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127	DUX4 Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy. Developmental Cell, 2012, 22, 38-51.	3.1	384
128	A genome-wide signature of glucocorticoid receptor binding in neuronal PC12 cells. BMC Neuroscience, 2012, 13, 118.	0.8	93
129	Pathogenic IgG4 subclass autoantibodies in MuSK myasthenia gravis. Annals of the New York Academy of Sciences, 2012, 1275, 114-122.	1.8	34
130	Selection of VHH Antibody Fragments That Recognize Different AÎ² Depositions Using Complex Immune Libraries. Methods in Molecular Biology, 2012, 911, 241-253.	0.4	4
131	Asymmetric Bidirectional Transcription from the FSHD-Causing D4Z4 Array Modulates DUX4 Production. PLoS ONE, 2012, 7, e35532.	1.1	20
132	Immunodeficiency, centromeric instability, facial anomalies (ICF) syndrome, due to ZBTB24 mutations, presenting with large cerebral cyst. American Journal of Medical Genetics, Part A, 2012, 158A, 2043-2046.	0.7	25
133	In Vivo Detection of Amyloid-Î² Deposits Using Heavy Chain Antibody Fragments in a Transgenic Mouse Model for Alzheimer's Disease. PLoS ONE, 2012, 7, e38284.	1.1	34
134	Clinical Dutch-English Lambert-Eaton Myasthenic Syndrome (LEMS) Tumor Association Prediction Score Accurately Predicts Small-Cell Lung Cancer in the LEMS. Journal of Clinical Oncology, 2011, 29, 902-908.	0.8	210
135	Modeling Oculopharyngeal Muscular Dystrophy in Myotube Cultures Reveals Reduced Accumulation of Soluble Mutant PABPN1 Protein. American Journal of Pathology, 2011, 179, 1988-2000.	1.9	34
136	Facioscapulohumeral muscular dystrophy and DUX4: breaking the silence. Trends in Molecular Medicine, 2011, 17, 252-258.	3.5	180
137	Facioscapulohumeral Muscular Dystrophy Region Gene 1 Is a Dynamic RNA-Associated and Actin-Bundling Protein. Journal of Molecular Biology, 2011, 411, 397-416.	2.0	22
138	Differential recognition of vascular and parenchymal beta amyloid deposition. Neurobiology of Aging, 2011, 32, 1774-1783.	1.5	34
139	Comparison of Dysferlin Expression in Human Skeletal Muscle with That in Monocytes for the Diagnosis of Dysferlin Myopathy. PLoS ONE, 2011, 6, e29061.	1.1	43
140	Distinguishing the 4qA and 4qB variants is essential for the diagnosis of facioscapulohumeral muscular dystrophy in the Chinese population. European Journal of Human Genetics, 2011, 19, 64-69.	1.4	15
141	Deregulation of the ubiquitin-proteasome system is the predominant molecular pathology in OPMD animal models and patients. Skeletal Muscle, 2011, 1, 15.	1.9	40
142	Mutations in ZBTB24 Are Associated with Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome Type 2. American Journal of Human Genetics, 2011, 88, 796-804.	2.6	158
143	Reversible aggregation of PABPN1 pre-inclusion structures. Nucleus, 2011, 2, 208-218.	0.6	20
144	Interspecies Translation of Disease Networks Increases Robustness and Predictive Accuracy. PLoS Computational Biology, 2011, 7, e1002258.	1.5	15

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145	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. Science, 2010, 329, 1650-1653.	6.0	638
146	Worldwide Population Analysis of the 4q and 10q Subtelomeres Identifies Only Four Discrete Interchromosomal Sequence Transfers in Human Evolution. American Journal of Human Genetics, 2010, 86, 364-377.	2.6	93
147	Structural basis for a PABPN1 aggregationâ€preventing antibody fragment in OPMD. FEBS Letters, 2010, 584, 1558-1564.	1.3	5
148	Analysis of allele-specific RNA transcription in FSHD by RNA-DNA FISH in single myonuclei. European Journal of Human Genetics, 2010, 18, 448-456.	1.4	34
149	Therapeutic exon skipping for dysferlinopathies?. European Journal of Human Genetics, 2010, 18, 889-894.	1.4	47
150	Reply to LÃ©vy et al. European Journal of Human Genetics, 2010, 18, 971-971.	1.4	2
151	Minimum information about a protein affinity reagent (MIAPAR). Nature Biotechnology, 2010, 28, 650-653.	9.4	50
152	Calpain 3 Is a Rapid-Action, Unidirectional Proteolytic Switch Central to Muscle Remodeling. PLoS ONE, 2010, 5, e11940.	1.1	23
153	Molecular and phenotypic characterization of a mouse model of oculopharyngeal muscular dystrophy reveals severe muscular atrophy restricted to fast glycolytic fibres. Human Molecular Genetics, 2010, 19, 2191-2207.	1.4	78
154	A Community Standard Format for the Representation of Protein Affinity Reagents. Molecular and Cellular Proteomics, 2010, 9, 1-10.	2.5	35
155	Facioscapulohumeral Dystrophy: Incomplete Suppression of a Retrotransposed Gene. PLoS Genetics, 2010, 6, e1001181.	1.5	394
156	171st ENMC International Workshop: Standards of care and management of facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2010, 20, 471-475.	0.3	88
157	Proteomic Analysis of the Dysferlin Protein Complex Unveils Its Importance for Sarcolemmal Maintenance and Integrity. PLoS ONE, 2010, 5, e13854.	1.1	62
158	Epigenomic Studies of Facioscapulohumeral Muscular Dystrophy (FSHD). FASEB Journal, 2010, 24, 713.8.	0.2	0
159	SOX Antibodies in Small-Cell Lung Cancer and Lambert-Eaton Myasthenic Syndrome: Frequency and Relation With Survival. Journal of Clinical Oncology, 2009, 27, 4260-4267.	0.8	178
160	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.	1.4	182
161	Common epigenetic changes of D4Z4 in contraction-dependent and contraction-independent FSHD. Human Mutation, 2009, 30, 1449-1459.	1.1	172
162	Comprehensive expression analysis of FSHD candidate genes at the mRNA and protein level. European Journal of Human Genetics, 2009, 17, 1615-1624.	1.4	56

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163	Prevention of oculopharyngeal muscular dystrophy by muscular expression of Llama single-chain intrabodies in vivo. Human Molecular Genetics, 2009, 18, 1849-1859.	1.4	49
164	Specific Loss of Histone H3 Lysine 9 Trimethylation and HP1Î³/Cohesin Binding at D4Z4 Repeats Is Associated with Facioscapulohumeral Dystrophy (FSHD). PLoS Genetics, 2009, 5, e1000559.	1.5	234
165	Novel Protein-Protein Interactions Inferred from Literature Context. PLoS ONE, 2009, 4, e7894.	1.1	41
166	Epigenetic mechanisms of facioscapulohumeral muscular dystrophy. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2008, 647, 94-102.	0.4	50
167	Calpain 3 is a modulator of the dysferlin protein complex in skeletal muscle. Human Molecular Genetics, 2008, 17, 1855-1866.	1.4	89
168	AHNAK a novel component of the dysferlin protein complex, redistributes to the cytoplasm with dysferlin during skeletal muscle regeneration. FASEB Journal, 2007, 21, 732-742.	0.2	133
169	Facioscapulohumeral muscular dystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2007, 1772, 186-194.	1.8	86
170	Specific Sequence Variations within the 4q35 Region Are Associated with Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2007, 81, 884-894.	2.6	200
171	ProteomeBinders: planning a European resource of affinity reagents for analysis of the human proteome. Nature Methods, 2007, 4, 13-17.	9.0	231
172	FRG1P-mediated aggregation of proteins involved in pre-mRNA processing. Chromosoma, 2007, 116, 53-64.	1.0	52
173	Prevention of oculopharyngeal muscular dystrophy-associated aggregation of nuclear poly(A)-binding protein with a single-domain intracellular antibody. Human Molecular Genetics, 2006, 15, 105-111.	1.4	78
174	Reliable and controllable antibody fragment selections from Camelid non-immune libraries for target validation. Biochimica Et Biophysica Acta - Proteins and Proteomics, 2006, 1764, 1307-1319.	1.1	62
175	Equal proportions of affected cells in muscle and blood of a mosaic carrier of facioscapulohumeral muscular dystrophy. Human Genetics, 2006, 119, 23-28.	1.8	13
176	Facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2006, 34, 1-15.	1.0	302
177	The Subtelomeric D4Z4 Repeat Instability in Facioscapulohumeral Muscular Dystrophy. , 2006, , 151-162.		0
178	Protein studies in dysferlinopathy patients using llama-derived antibody fragments selected by phage display. European Journal of Human Genetics, 2005, 13, 721-730.	1.4	60
179	Variable hypomethylation of D4Z4 in facioscapulohumeral muscular dystrophy. Annals of Neurology, 2005, 58, 569-576.	2.8	113
180	Genetic confirmation of facioscapulohumeral muscular dystrophy in a case with complex D4Z4 rearrangments. Human Genetics, 2005, 116, 262-266.	1.8	6

#	Article	IF	Citations
181	The D4Z4 Repeatâ€“Mediated Pathogenesis of Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2005, 76, 375-386.	2.6	71
182	Recurrent somatic mosaicism for D4Z4 contractions in a family with facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2005, 15, 471-475.	0.3	8
183	Localization of 4q35.2 to the nuclear periphery: is FSHD a nuclear envelope disease?. Human Molecular Genetics, 2004, 13, 1857-1871.	1.4	90
184	Somatic mosaicism in FSHD often goes undetected. Annals of Neurology, 2004, 55, 845-850.	2.8	69
185	Mechanism and Timing of Mitotic Rearrangements in the Subtelomeric D4Z4 Repeat Involved in Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2004, 75, 44-53.	2.6	45
186	Contractions of D4Z4 on 4qB Subtelomeres Do Not Cause Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2004, 75, 1124-1130.	2.6	109
187	Oculopharyngeal muscular dystrophy with limb girdle weakness as major complaint. Journal of Neurology, 2003, 250, 1307-1312.	1.8	91
188	Facioscapulohumeral muscular dystrophy. Journal of Neurology, 2003, 250, 932-937.	1.8	65
189	Hypomethylation of D4Z4 in 4q-linked and non-4q-linked facioscapulohumeral muscular dystrophy. Nature Genetics, 2003, 35, 315-317.	9.4	349
190	Expression profiling of FSHD muscle supports a defect in specific stages of myogenic differentiation. Human Molecular Genetics, 2003, 12, 2895-2907.	1.4	191
191	Testing the position-effect variegation hypothesis for facioscapulohumeral muscular dystrophy by analysis of histone modification and gene expression in subtelomeric 4q. Human Molecular Genetics, 2003, 12, 2909-2921.	1.4	138
192	Perturbations of chromatin structure in human genetic disease: recent advances. Human Molecular Genetics, 2003, 12, R207-R213.	1.4	51
193	Genomic Analysis of Human Chromosome 10q and 4q Telomeres Suggests a Common Origin. Genomics, 2002, 79, 210-217.	1.3	122
194	A Cascade of Complex Subtelomeric Duplications during the Evolution of the Hominoid and Old World Monkey Genomes. American Journal of Human Genetics, 2002, 70, 269-278.	2.6	27
195	Facioscapulohumeral muscular dystrophy is uniquely associated with one of the two variants of the 4q subtelomere. Nature Genetics, 2002, 32, 235-236.	9.4	255
196	Mutations of MLC1 (KIAA0027), Encoding a Putative Membrane Protein, Cause Megalencephalic Leukoencephalopathy with Subcortical Cysts. American Journal of Human Genetics, 2001, 68, 831-838.	2.6	243
197	Active genes in junk DNA? Characterization of DUX genes embedded within 3.3 kb repeated elements. Gene, 2001, 264, 51-57.	1.0	38
198	Complete allele information in the diagnosis of facioscapulohumeral muscular dystrophy by triple DNA analysis. Annals of Neurology, 2001, 50, 816-819.	2.8	91

#	Article	IF	Citations
199	De Novo Facioscapulohumeral Muscular Dystrophy: Frequent Somatic Mosaicism, Sex-Dependent Phenotype, and the Role of Mitotic Transchromosomal Repeat Interaction between Chromosomes 4 and 10. American Journal of Human Genetics, 2000, 66, 26-35.	2.6	136
200	Somatic pairing between subtelomeric chromosome regions: implications for human genetic disease?. Chromosome Research, 1999, 7, 323-329.	1.0	29
201	FRG1, a gene in the FSH muscular dystrophy region on human chromosome 4q35, is highly conserved in vertebrates and invertebrates. Gene, 1998, 216, 13-19.	1.0	41