SilvÃ¨re M Van Der Maarel

List of Publications by Year
in descending order

201 papers	13,780 citations	22153 59 h-index	29157 104 g-index
212 all docs	212 docs citations	212 times ranked	8614 citing authors

#	Article	IF	Citations
1	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. Science, 2010, 329, 1650-1653.	12.6	638
2	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
3	Facioscapulohumeral Dystrophy: Incomplete Suppression of a Retrotransposed Gene. PLoS Genetics, 2010, 6, e1001181.	3.5	394
4	DUX4 Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy. Developmental Cell, 2012, 22, 38-51.	7.0	384
5	Hypomethylation of D4Z4 in 4q-linked and non-4q-linked facioscapulohumeral muscular dystrophy. Nature Genetics, 2003, 35, 315-317.	21.4	349
6	Facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2006, 34, 1-15.	2.2	302
7	Population-based incidence and prevalence of facioscapulohumeral dystrophy. Neurology, 2014, 83, 1056-1059.	1.1	278
8	Facioscapulohumeral muscular dystrophy is uniquely associated with one of the two variants of the 4q subtelomere. Nature Genetics, 2002, 32, 235-236.	21.4	255
9	Mutations of MLC1 (KIAA0027), Encoding a Putative Membrane Protein, Cause Megalencephalic Leukoencephalopathy with Subcortical Cysts. American Journal of Human Genetics, 2001, 68, 831-838.	6.2	243
10	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.	7.1	234
11	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.	3.5	234
12	ProteomeBinders: planning a European resource of affinity reagents for analysis of the human proteome. Nature Methods, 2007, 4, 13-17.	19.0	231
13	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.	1.6	210
14	Specific Sequence Variations within the 4q35 Region Are Associated with Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2007, 81, 884-894.	6.2	200
15	Expression profiling of FSHD muscle supports a defect in specific stages of myogenic differentiation. Human Molecular Genetics, 2003, 12, 2895-2907.	2.9	191
16	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
17	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
18	Facioscapulohumeral muscular dystrophy and DUX4: breaking the silence. Trends in Molecular Medicine, 2011, 17, 252-258.	6.7	180

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19	Muscle-specific kinase myasthenia gravis IgG4 autoantibodies cause severe neuromuscular junction dysfunction in mice. Brain, 2012, 135, 1081-1101.	7.6	180
20	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.	1.6	178
21	Common epigenetic changes of D4Z4 in contraction-dependent and contraction-independent FSHD. Human Mutation, 2009, 30, 1449-1459.	2.5	172
22	DUX4-induced gene expression is the major molecular signature in FSHD skeletal muscle. Human Molecular Genetics, 2014, 23, 5342-5352.	2.9	170
23	Mutations in ZBTB24 Are Associated with Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome Type 2. American Journal of Human Genetics, 2011, 88, 796-804.	6.2	158
24	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
25	DUX4 Binding to Retroelements Creates Promoters That Are Active in FSHD Muscle and Testis. PLoS Genetics, 2013, 9, e1003947.	3.5	151
26	Poly(A) binding protein nuclear 1 levels affect alternative polyadenylation. Nucleic Acids Research, 2012, 40, 9089-9101.	14.5	148
27	Mutations in CDCA7 and HELLS cause immunodeficiencyâ€“centromeric instabilityâ€“facial anomalies syndrome. Nature Communications, 2015, 6, 7870.	12.8	148
28	Facioscapulohumeral dystrophy: the path to consensus on pathophysiology. Skeletal Muscle, 2014, 4, 12.	4.2	144
29	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.	2.9	138
30	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.	6.2	136
31	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.	0.5	133
32	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
33	Genomic Analysis of Human Chromosome 10q and 4q Telomeres Suggests a Common Origin. Genomics, 2002, 79, 210-217.	2.9	122
34	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. European Journal of Human Genetics, 2013, 21, 1219-1225.	2.8	115
35	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.	9.9	114
36	Variable hypomethylation of D4Z4 in facioscapulohumeral muscular dystrophy. Annals of Neurology, 2005, 58, 569-576.	5.3	113

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37	Contractions of D4Z4 on 4qB Subtelomeres Do Not Cause Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2004, 75, 1124-1130.	6.2	109
38	A feedback loop between nonsense-mediated decay and the retrogene DUX4 in facioscapulohumeral muscular dystrophy. ELife, 2015, 4, .	6.0	97
39	Intrinsic Epigenetic Regulation of the D4Z4 Macrosatellite Repeat in a Transgenic Mouse Model for FSHD. PLoS Genetics, 2013, 9, e1003415.	3.5	95
40	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.	6.2	93
41	A genome-wide signature of glucocorticoid receptor binding in neuronal PC12 cells. BMC Neuroscience, 2012, 13, 118.	1.9	93
42	IgG4 autoantibodies against muscle-specific kinase undergo Fab-arm exchange in myasthenia gravis patients. Journal of Autoimmunity, 2017, 77, 104-115.	6.5	92
43	Complete allele information in the diagnosis of facioscapulohumeral muscular dystrophy by triple DNA analysis. Annals of Neurology, 2001, 50, 816-819.	5.3	91
44	Oculopharyngeal muscular dystrophy with limb girdle weakness as major complaint. Journal of Neurology, 2003, 250, 1307-1312.	3.6	91
45	Localization of 4q35.2 to the nuclear periphery: is FSHD a nuclear envelope disease?. Human Molecular Genetics, 2004, 13, 1857-1871.	2.9	90
46	Calpain 3 is a modulator of the dysferlin protein complex in skeletal muscle. Human Molecular Genetics, 2008, 17, 1855-1866.	2.9	89
47	171st ENMC International Workshop: Standards of care and management of facioscapulohumeral muscular dystrophy. Neuromuscular Disorders, 2010, 20, 471-475.	0.6	88
48	Facioscapulohumeral muscular dystrophy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2007, 1772, 186-194.	3.8	86
49	Homozygous nonsense variant in LRIF1 associated with facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e2441-e2447.	1.1	84
50	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.	7.1	83
51	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.	2.9	78
52	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.	2.9	78
53	DUX4 induces a transcriptome more characteristic of a less-differentiated cell state and inhibits myogenesis. Journal of Cell Science, 2016, 129, 3816-3831.	2.0	77
54	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

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55	Model systems of DUX4 expression recapitulate the transcriptional profile of FSHD cells. Human Molecular Genetics, 2016, 25, ddw271.	2.9	75
56	Adding quantitative muscle MRI to the FSHD clinical trial toolbox. Neurology, 2017, 89, 2057-2065.	1.1	72
57	The D4Z4 Repeatâ€“Mediated Pathogenesis of Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 2005, 76, 375-386.	6.2	71
58	Somatic mosaicism in FSHD often goes undetected. Annals of Neurology, 2004, 55, 845-850.	5.3	69
59	Genetic and epigenetic contributors to FSHD. Current Opinion in Genetics and Development, 2015, 33, 56-61.	3.3	69
60	A focal domain of extreme demethylation within D4Z4 in FSHD2. Neurology, 2013, 80, 392-399.	1.1	67
61	Facioscapulohumeral muscular dystrophy. Journal of Neurology, 2003, 250, 932-937.	3.6	65
62	MuSK myasthenia gravis monoclonal antibodies. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e547.	6.0	64
63	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
64	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.	2.3	62
65	Proteomic Analysis of the Dysferlin Protein Complex Unveils Its Importance for Sarcolemmal Maintenance and Integrity. PLoS ONE, 2010, 5, e13854.	2.5	62
66	Protein studies in dysferlinopathy patients using llama-derived antibody fragments selected by phage display. European Journal of Human Genetics, 2005, 13, 721-730.	2.8	60
67	Longitudinal epitope mapping in MuSK myasthenia gravis: implications for disease severity. Journal of Neuroimmunology, 2016, 291, 82-88.	2.3	59
68	p53 convergently activates Dux/DUX4 in embryonic stem cells and in facioscapulohumeral muscular dystrophy cell models. Nature Genetics, 2021, 53, 1207-1220.	21.4	59
69	Comprehensive expression analysis of FSHD candidate genes at the mRNA and protein level. European Journal of Human Genetics, 2009, 17, 1615-1624.	2.8	56
70	Patients with a phenotype consistent with facioscapulohumeral muscular dystrophy display genetic and epigenetic heterogeneity. Journal of Medical Genetics, 2012, 49, 41-46.	3.2	55
71	Allele-specific DNA hypomethylation characterises FSHD1 and FSHD2. Journal of Medical Genetics, 2016, 53, 348-355.	3.2	54
72	IgG4â€mediated autoimmune diseases: a niche of antibodyâ€mediated disorders. Annals of the New York Academy of Sciences, 2018, 1413, 92-103.	3.8	54

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73	Best practice guidelines on genetic diagnostics of Facioscapulohumeral muscular dystrophy: Workshop 9th June 2010, LUMC, Leiden, The Netherlands. Neuromuscular Disorders, 2012, 22, 463-470.	0.6	53
74	FRG1P-mediated aggregation of proteins involved in pre-mRNA processing. Chromosoma, 2007, 116, 53-64.	2.2	52
75	Increased DUX4 expression during muscle differentiation correlates with decreased SMCHD1 protein levels at D4Z4. Epigenetics, 2015, 10, 1133-1142.	2.7	52
76	Perturbations of chromatin structure in human genetic disease: recent advances. Human Molecular Genetics, 2003, 12, R207-R213.	2.9	51
77	Epigenetic mechanisms of facioscapulohumeral muscular dystrophy. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2008, 647, 94-102.	1.0	50
78	Minimum information about a protein affinity reagent (MIAPAR). Nature Biotechnology, 2010, 28, 650-653.	17.5	50
79	Muscle pathology grade for facioscapulohumeral muscular dystrophy biopsies. Muscle and Nerve, 2015, 52, 521-526.	2.2	50
80	FSHD1 and FSHD2 form a disease continuum. Neurology, 2019, 92, e2273-e2285.	1.1	50
81	Prevention of oculopharyngeal muscular dystrophy by muscular expression of Llama single-chain intrabodies in vivo. Human Molecular Genetics, 2009, 18, 1849-1859.	2.9	49
82	Dysferlin Regulates Cell Adhesion in Human Monocytes. Journal of Biological Chemistry, 2013, 288, 14147-14157.	3.4	49
83	Converging disease genes in ICF syndrome:ZBTB24controls expression ofCDCA7in mammals. Human Molecular Genetics, 2016, 25, 4041-4051.	2.9	49
84	A decline in PABPN1 induces progressive muscle weakness in Oculopharyngeal muscle dystrophy and in muscle aging. Aging, 2013, 5, 412-426.	3.1	49
85	Correlation analysis of clinical parameters with epigenetic modifications in the DUX4 promoter in FSHD. Epigenetics, 2012, 7, 579-584.	2.7	48
86	Therapeutic exon skipping for dysferlinopathies?. European Journal of Human Genetics, 2010, 18, 889-894.	2.8	47
87	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.	0.6	47
88	NuRD and CAF-1-mediated silencing of the D4Z4 array is modulated by DUX4-induced MBD3L proteins. ELife, 2018, 7, .	6.0	47
89	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
90	Single-cell RNA sequencing in facioscapulohumeral muscular dystrophy disease etiology and development. Human Molecular Genetics, 2019, 28, 1064-1075.	2.9	46

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91	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.	6.2	45
92	Milder phenotype in facioscapulohumeral dystrophy with 7â€“10 residual D4Z4 repeats. Neurology, 2015, 85, 2147-2150.	1.1	44
93	Comparison of Dysferlin Expression in Human Skeletal Muscle with That in Monocytes for the Diagnosis of Dysferlin Myopathy. PLoS ONE, 2011, 6, e29061.	2.5	43
94	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
95	Facioscapulohumeral muscular dystrophy. Current Opinion in Neurology, 2012, 25, 614-620.	3.6	42
96	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.	2.5	42
97	Integrating gene delivery and gene-editing technologies by adenoviral vector transfer of optimized CRISPR-Cas9 components. Gene Therapy, 2020, 27, 209-225.	4.5	42
98	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.	2.2	41
99	Novel Protein-Protein Interactions Inferred from Literature Context. PLoS ONE, 2009, 4, e7894.	2.5	41
100	Deregulation of the ubiquitin-proteasome system is the predominant molecular pathology in OPMD animal models and patients. Skeletal Muscle, 2011, 1, 15.	4.2	40
101	Consequences of epigenetic derepression in facioscapulohumeral muscular dystrophy. Clinical Genetics, 2020, 97, 799-814.	2.0	40
102	Active genes in junk DNA? Characterization of DUX genes embedded within 3.3 kb repeated elements. Gene, 2001, 264, 51-57.	2.2	38
103	Multiplex Screen of Serum Biomarkers in Facioscapulohumeral Muscular Dystrophy. Journal of Neuromuscular Diseases, 2014, 1, 181-190.	2.6	38
104	Longitudinal measures of RNA expression and disease activity in FSHD muscle biopsies. Human Molecular Genetics, 2020, 29, 1030-1043.	2.9	38
105	Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation. JCI Insight, 2021, 6, .	5.0	38
106	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
107	Sarcomeric dysfunction contributes to muscle weakness in facioscapulohumeral muscular dystrophy. Neurology, 2013, 80, 733-737.	1.1	36
108	A Community Standard Format for the Representation of Protein Affinity Reagents. Molecular and Cellular Proteomics, 2010, 9, 1-10.	3.8	35

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109	Analysis of allele-specific RNA transcription in FSHD by RNA-DNA FISH in single myonuclei. European Journal of Human Genetics, 2010, 18, 448-456.	2.8	34
110	Modeling Oculopharyngeal Muscular Dystrophy in Myotube Cultures Reveals Reduced Accumulation of Soluble Mutant PABPN1 Protein. American Journal of Pathology, 2011, 179, 1988-2000.	3.8	34
111	Differential recognition of vascular and parenchymal beta amyloid deposition. Neurobiology of Aging, 2011, 32, 1774-1783.	3.1	34
112	Pathogenic IgG4 subclass autoantibodies in MuSK myasthenia gravis. Annals of the New York Academy of Sciences, 2012, 1275, 114-122.	3.8	34
113	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
114	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.	2.5	34
115	A family-based study into penetrance in facioscapulohumeral muscular dystrophy type 1. Neurology, 2018, 91, e444-e454.	1.1	33
116	Hemizygosity forSMCHD1in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. Human Mutation, 2015, 36, 679-683.	2.5	32
117	SMCHD1 regulates a limited set of gene clusters on autosomal chromosomes. Skeletal Muscle, 2017, 7, 12.	4.2	32
118	Differential myofiber-type transduction preference of adeno-associated virus serotypes 6 and 9. Skeletal Muscle, 2015, 5, 37.	4.2	31
119	Early onset as a marker for disease severity in facioscapulohumeral muscular dystrophy. Neurology, 2019, 92, e378-e385.	1.1	30
120	Somatic pairing between subtelomeric chromosome regions: implications for human genetic disease?. Chromosome Research, 1999, 7, 323-329.	2.2	29
121	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.	2.8	29
122	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, .	7.1	28
123	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.	6.2	27
124	A Novel Feed-Forward Loop between ARIH2 E3-Ligase and PABPN1 Regulates Aging-Associated Muscle Degeneration. American Journal of Pathology, 2014, 184, 1119-1131.	3.8	27
125	Cis D4Z4 repeat duplications associated with facioscapulohumeral muscular dystrophy type 2. Human Molecular Genetics, 2018, 27, 3488-3497.	2.9	27
126	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.	3.2	27

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127	Intronic SMCHD1 variants in FSHD: testing the potential for CRISPR-Cas9 genome editing. Journal of Medical Genetics, 2019, 56, 828-837.	3.2	27
128	Loss of ZBTB24 impairs nonhomologous end-joining and class-switch recombination in patients with ICF syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	27
129	Immunohistochemical Characterization ofÂFacioscapulohumeralMuscular DystrophyÂMuscle Biopsies. Journal of Neuromuscular Diseases, 2015, 2, 291-299.	2.6	26
130	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
131	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499.	1.4	26
132	Identical twins carry a persistent epigenetic signature of early genome programming. Nature Communications, 2021, 12, 5618.	12.8	26
133	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.	1.2	25
134	Chromatin remodeling of human subtelomeres and TERRA promoters upon cellular senescence. Epigenetics, 2013, 8, 512-521.	2.7	25
135	Phenotypeâ€genotype relations in facioscapulohumeral muscular dystrophy type 1. Clinical Genetics, 2018, 94, 521-527.	2.0	25
136	Efgartigimod improves muscle weakness in a mouse model for muscle-specific kinase myasthenia gravis. Experimental Neurology, 2019, 317, 133-143.	4.1	25
137	Selfâ€regulated alternative splicing at the AHNAK locus. FASEB Journal, 2012, 26, 93-103.	0.5	24
138	Myasthenia gravis with muscle specific kinase antibodies mimicking amyotrophic lateral sclerosis. Neuromuscular Disorders, 2016, 26, 350-353.	0.6	24
139	Autosomal genetic variation is associated with DNA methylation in regions variably escaping X-chromosome inactivation. Nature Communications, 2018, 9, 3738.	12.8	24
140	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24
141	Calpain 3 Is a Rapid-Action, Unidirectional Proteolytic Switch Central to Muscle Remodeling. PLoS ONE, 2010, 5, e11940.	2.5	23
142	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
143	Smchd1 haploinsufficiency exacerbates the phenotype of a transgenic FSHD1 mouse model. Human Molecular Genetics, 2018, 27, 716-731.	2.9	23
144	Facioscapulohumeral Muscular Dystrophy Region Gene 1 Is a Dynamic RNA-Associated and Actin-Bundling Protein. Journal of Molecular Biology, 2011, 411, 397-416.	4.2	22

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145	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
146	Facioscapulohumeral Dystrophy in Childhood: A Nationwide Natural History Study. Annals of Neurology, 2018, 84, 627-637.	5.3	21
147	Reversible aggregation of PABPN1 pre-inclusion structures. Nucleus, 2011, 2, 208-218.	2.2	20
148	Asymmetric Bidirectional Transcription from the FSHD-Causing D4Z4 Array Modulates DUX4 Production. PLoS ONE, 2012, 7, e35532.	2.5	20
149	225th ENMC international workshop:. Neuromuscular Disorders, 2017, 27, 782-790.	0.6	20
150	DUX4 promotes transcription of FRG2 by directly activating its promoter in facioscapulohumeral muscular dystrophy. Skeletal Muscle, 2014, 4, 19.	4.2	19
151	The Effect of Corticosteroids on Human Choroidal Endothelial Cells: A Model to Study Central Serous Chorioretinopathy. , 2018, 59, 5682.		19
152	Chromosome 10q-linked FSHD identifies DUX4 as principal disease gene. Journal of Medical Genetics, 2022, 59, 180-188.	3.2	18
153	High prevalence of incomplete right bundle branch block in facioscapulohumeral muscular dystrophy without cardiac symptoms. Functional Neurology, 2014, 29, 159-65.	1.3	18
154	Selection and characterization of llama single domain antibodies against N-terminal huntingtin. Neurological Sciences, 2015, 36, 429-434.	1.9	16
155	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.	2.8	15
156	Interspecies Translation of Disease Networks Increases Robustness and Predictive Accuracy. PLoS Computational Biology, 2011, 7, e1002258.	3.2	15
157	Epigenetic regulation of the X-chromosomal macrosatellite repeat encoding for the cancer/testis gene CT47. European Journal of Human Genetics, 2012, 20, 185-191.	2.8	15
158	Nuclear entrapment and extracellular depletion of PCOLCE is associated with muscle degeneration in oculopharyngeal muscular dystrophy. BMC Neurology, 2013, 13, 70.	1.8	15
159	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.	4.7	15
160	Facioscapulohumeral dystrophy in children: design of a prospective, observational study on natural history, predictors and clinical impact (iFocus FSHD). BMC Neurology, 2016, 16, 138.	1.8	15
161	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.	2.8	14
162	Early-Onset Facioscapulohumeral Muscular Dystrophy Type 1 With Some Atypical Features. Journal of Child Neurology, 2015, 30, 580-587.	1.4	14

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163	Ophthalmological findings in facioscapulohumeral dystrophy. Brain Communications, 2019, 1, fcz023.	3.3	14
164	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
165	Equal proportions of affected cells in muscle and blood of a mosaic carrier of facioscapulohumeral muscular dystrophy. Human Genetics, 2006, 119, 23-28.	3.8	13
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