

# Silvãre M Van Der Maarel

## List of Publications by Year in descending order

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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	Chromosome 10q-linked FSHD identifies <i>DUX4</i> as principal disease gene. <i>Journal of Medical Genetics</i> , 2022, 59, 180-188.	3.2	18
2	High-resolution breakpoint junction mapping of proximally extended D4Z4 deletions in FSHD1 reveals evidence for a founder effect. <i>Human Molecular Genetics</i> , 2022, 31, 748-760.	2.9	8
3	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
4	Elevated plasma complement components in facioscapulohumeral dystrophy. <i>Human Molecular Genetics</i> , 2022, 31, 1821-1829.	2.9	10
5	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. <i>Blood</i> , 2021, 137, 493-499.	1.4	26
6	Functional monovalency amplifies the pathogenicity of anti-MuSK IgG4 in myasthenia gravis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	7.1	28
7	Characterization of HNRNPA1 mutations defines diversity in pathogenic mechanisms and clinical presentation. <i>JCI Insight</i> , 2021, 6, .	5.0	38
8	p53 convergently activates Dux/DUX4 in embryonic stem cells and in facioscapulohumeral muscular dystrophy cell models. <i>Nature Genetics</i> , 2021, 53, 1207-1220.	21.4	59
9	Adenine base editing of the DUX4 polyadenylation signal for targeted genetic therapy in facioscapulohumeral muscular dystrophy. <i>Molecular Therapy - Nucleic Acids</i> , 2021, 25, 342-354.	5.1	12
10	Identical twins carry a persistent epigenetic signature of early genome programming. <i>Nature Communications</i> , 2021, 12, 5618.	12.8	26
11	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
12	Profiling Serum Antibodies Against Muscle Antigens in Facioscapulohumeral Muscular Dystrophy Finds No Disease-Specific Autoantibodies. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 801-814.	2.6	6
13	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
14	SATB1, genomic instability and Gleason grading constitute a novel risk score for prostate cancer. <i>Scientific Reports</i> , 2021, 11, 24446.	3.3	1
15	Integrating gene delivery and gene-editing technologies by adenoviral vector transfer of optimized CRISPR-Cas9 components. <i>Gene Therapy</i> , 2020, 27, 209-225.	4.5	42
16	Magnetic resonance imaging correlates with electrical impedance myography in facioscapulohumeral muscular dystrophy. <i>Muscle and Nerve</i> , 2020, 61, 644-649.	2.2	10
17	Loss of ZBTB24 impairs nonhomologous end-joining and class-switch recombination in patients with ICF syndrome. <i>Journal of Experimental Medicine</i> , 2020, 217, .	8.5	27
18	The prospects of targeting DUX4 in facioscapulohumeral muscular dystrophy. <i>Current Opinion in Neurology</i> , 2020, 33, 635-640.	3.6	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.	2.5	6
20	Homozygous nonsense variant in <i>LRIF1</i> associated with facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e2441-e2447.	1.1	84
21	Longitudinal measures of RNA expression and disease activity in FSHD muscle biopsies. Human Molecular Genetics, 2020, 29, 1030-1043.	2.9	38
22	Consequences of epigenetic derepression in facioscapulohumeral muscular dystrophy. Clinical Genetics, 2020, 97, 799-814.	2.0	40
23	Preserved single muscle fiber specific force in facioscapulohumeral muscular dystrophy. Neurology, 2020, 94, e1157-e1170.	1.1	8
24	Dnmt3b regulates DUX4 expression in a tissue-dependent manner in transgenic D4Z4 mice. Skeletal Muscle, 2020, 10, 27.	4.2	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.	3.2	27
30	Ophthalmological findings in facioscapulohumeral dystrophy. Brain Communications, 2019, 1, fcz023.	3.3	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.	2.9	26
32	Intronic <i>SMCHD1</i> variants in FSHD: testing the potential for CRISPR-Cas9 genome editing. Journal of Medical Genetics, 2019, 56, 828-837.	3.2	27
33	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
34	Generation of genetically matched hiPSC lines from two mosaic facioscapulohumeral dystrophy type 1 patients. Stem Cell Research, 2019, 40, 101560.	0.7	6
35	A functional assay to classify <i>ZBTB24</i> missense variants of unknown significance. Human Mutation, 2019, 40, 1077-1083.	2.5	6
36	FSHD1 and FSHD2 form a disease continuum. Neurology, 2019, 92, e2273-e2285.	1.1	50

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37	Efgartigimod improves muscle weakness in a mouse model for muscle-specific kinase myasthenia gravis. <i>Experimental Neurology</i> , 2019, 317, 133-143.	4.1	25
38	MuSK myasthenia gravis monoclonal antibodies. <i>Neurology: Neuroimmunology and NeuroInflammation</i> , 2019, 6, e547.	6.0	64
39	Early onset as a marker for disease severity in facioscapulohumeral muscular dystrophy. <i>Neurology</i> , 2019, 92, e378-e385.	1.1	30
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	IgG4-mediated autoimmune diseases: a niche of antibody-mediated disorders. <i>Annals of the New York Academy of Sciences</i> , 2018, 1413, 92-103.	3.8	54
42	Passive transfer models of myasthenia gravis with muscle-specific kinase antibodies. <i>Annals of the New York Academy of Sciences</i> , 2018, 1413, 111-118.	3.8	4
43	Smchd1 haploinsufficiency exacerbates the phenotype of a transgenic FSHD1 mouse model. <i>Human Molecular Genetics</i> , 2018, 27, 716-731.	2.9	23
44	Monosomy 18p is a risk factor for facioscapulohumeral dystrophy. <i>Journal of Medical Genetics</i> , 2018, 55, 469-478.	3.2	11
45	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
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. <i>Nature Communications</i> , 2018, 9, 3738.	12.8	24
48	Facioscapulohumeral Dystrophy in Childhood: A Nationwide Natural History Study. <i>Annals of Neurology</i> , 2018, 84, 627-637.	5.3	21
49	Phenotype-genotype relations in facioscapulohumeral muscular dystrophy type 1. <i>Clinical Genetics</i> , 2018, 94, 521-527.	2.0	25
50	A 22-year follow-up reveals a variable disease severity in early-onset facioscapulohumeral dystrophy. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 782-785.	1.6	8
51	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
52	Cis D4Z4 repeat duplications associated with facioscapulohumeral muscular dystrophy type 2. <i>Human Molecular Genetics</i> , 2018, 27, 3488-3497.	2.9	27
53	Brain Transcriptomic Analysis of Hereditary Cerebral Hemorrhage With Amyloidosis-Dutch Type. <i>Frontiers in Aging Neuroscience</i> , 2018, 10, 102.	3.4	13
54	A family-based study into penetrance in facioscapulohumeral muscular dystrophy type 1. <i>Neurology</i> , 2018, 91, e444-e454.	1.1	33

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55	FSHD type 2 and Bosma arhinia microphthalmia syndrome. <i>Neurology</i> , 2018, 91, e562-e570.	1.1	24
56	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
57	225th ENMC international workshop. <i>Neuromuscular Disorders</i> , 2017, 27, 782-790.	0.6	20
58	IgG4 autoantibodies against muscle-specific kinase undergo Fab-arm exchange in myasthenia gravis patients. <i>Journal of Autoimmunity</i> , 2017, 77, 104-115.	6.5	92
59	Adding quantitative muscle MRI to the FSHD clinical trial toolbox. <i>Neurology</i> , 2017, 89, 2057-2065.	1.1	72
60	SMCHD1 regulates a limited set of gene clusters on autosomal chromosomes. <i>Skeletal Muscle</i> , 2017, 7, 12.	4.2	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. <i>Skeletal Muscle</i> , 2017, 7, 16.	4.2	46
62	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
63	DUX4 induces a transcriptome more characteristic of a less-differentiated cell state and inhibits myogenesis. <i>Journal of Cell Science</i> , 2016, 129, 3816-3831.	2.0	77
64	Integrating clinical and genetic observations in facioscapulohumeral muscular dystrophy. <i>Current Opinion in Neurology</i> , 2016, 29, 606-613.	3.6	10
65	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
66	Myasthenia gravis with muscle specific kinase antibodies mimicking amyotrophic lateral sclerosis. <i>Neuromuscular Disorders</i> , 2016, 26, 350-353.	0.6	24
67	Converging disease genes in ICF syndrome: <i>ZBTB24</i> controls expression of <i>CDCA7</i> in mammals. <i>Human Molecular Genetics</i> , 2016, 25, 4041-4051.	2.9	49
68	Model systems of DUX4 expression recapitulate the transcriptional profile of FSHD cells. <i>Human Molecular Genetics</i> , 2016, 25, ddw271.	2.9	75
69	Facioscapulohumeral dystrophy in children: design of a prospective, observational study on natural history, predictors and clinical impact (iFocus FSHD). <i>BMC Neurology</i> , 2016, 16, 138.	1.8	15
70	Longitudinal epitope mapping in MuSK myasthenia gravis: implications for disease severity. <i>Journal of Neuroimmunology</i> , 2016, 291, 82-88.	2.3	59
71	Allele-specific DNA hypomethylation characterises FSHD1 and FSHD2. <i>Journal of Medical Genetics</i> , 2016, 53, 348-355.	3.2	54
72	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

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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. <i>European Journal of Human Genetics</i> , 2016, 24, 78-85.	2.8	23
74	DUX4 induces a transcriptome more characteristic of a less-differentiated cell state and inhibits myogenesis. <i>Development (Cambridge)</i> , 2016, 143, e1.1-e1.1.	2.5	0
75	Differential myofiber-type transduction preference of adeno-associated virus serotypes 6 and 9. <i>Skeletal Muscle</i> , 2015, 5, 37.	4.2	31
76	Camelid heavy chain only antibody fragment domain against $\beta$ -site of amyloid precursor protein cleaving enzyme 1 inhibits $\beta$ -secretase activity <i>in vitro</i> and <i>in vivo</i> . <i>FEBS Journal</i> , 2015, 282, 3618-3631.	4.7	15
77	Immunohistochemical Characterization of Facioscapulohumeral Muscular Dystrophy Muscle Biopsies. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 291-299.	2.6	26
78	A feedback loop between nonsense-mediated decay and the retrogene DUX4 in facioscapulohumeral muscular dystrophy. <i>ELife</i> , 2015, 4, .	6.0	97
79	Muscle pathology grade for facioscapulohumeral muscular dystrophy biopsies. <i>Muscle and Nerve</i> , 2015, 52, 521-526.	2.2	50
80	Milder phenotype in facioscapulohumeral dystrophy with 7-10 residual D4Z4 repeats. <i>Neurology</i> , 2015, 85, 2147-2150.	1.1	44
81	Enhanced glutathione PEGylated liposomal brain delivery of an anti-amyloid single domain antibody fragment in a mouse model for Alzheimer's disease. <i>Journal of Controlled Release</i> , 2015, 203, 40-50.	9.9	114
82	Selection and characterization of llama single domain antibodies against N-terminal huntingtin. <i>Neurological Sciences</i> , 2015, 36, 429-434.	1.9	16
83	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
84	Facioscapulohumeral Dystrophy. , 2015, , 620-630.		0
85	Fusion of hlgG1-Fc to <sup>111</sup> In-anti-amyloid single domain antibody fragment VHH-pa2H prolongs blood residential time in APP/PS1 mice but does not increase brain uptake. <i>Nuclear Medicine and Biology</i> , 2015, 42, 695-702.	0.6	47
86	Early-Onset Facioscapulohumeral Muscular Dystrophy Type 1 With Some Atypical Features. <i>Journal of Child Neurology</i> , 2015, 30, 580-587.	1.4	14
87	Genetic and epigenetic contributors to FSHD. <i>Current Opinion in Genetics and Development</i> , 2015, 33, 56-61.	3.3	69
88	Hemizyosity for SMCHD1 in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. <i>Human Mutation</i> , 2015, 36, 679-683.	2.5	32
89	Mutations in CDCA7 and HELLS cause immunodeficiency-centromeric instability-facial anomalies syndrome. <i>Nature Communications</i> , 2015, 6, 7870.	12.8	148
90	Genome-wide binding and mechanistic analyses of Smchd1-mediated epigenetic regulation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E3535-44.	7.1	83

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91	Increased DUX4 expression during muscle differentiation correlates with decreased SMCHD1 protein levels at D4Z4. <i>Epigenetics</i> , 2015, 10, 1133-1142.	2.7	52
92	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
93	Multiplex Screen of Serum Biomarkers in Facioscapulohumeral Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2014, 1, 181-190.	2.6	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. <i>Human Molecular Genetics</i> , 2014, 23, 5342-5352.	2.9	170
96	DUX4 promotes transcription of FRG2 by directly activating its promoter in facioscapulohumeral muscular dystrophy. <i>Skeletal Muscle</i> , 2014, 4, 19.	4.2	19
97	A Novel Feed-Forward Loop between ARIH2 E3-Ligase and PABPN1 Regulates Aging-Associated Muscle Degeneration. <i>American Journal of Pathology</i> , 2014, 184, 1119-1131.	3.8	27
98	Genetic and Epigenetic Characteristics of FSHD-Associated 4q and 10q D4Z4 that are Distinct from Non-4q/10q D4Z4 Homologs. <i>Human Mutation</i> , 2014, 35, 998-1010.	2.5	42
99	Population-based incidence and prevalence of facioscapulohumeral dystrophy. <i>Neurology</i> , 2014, 83, 1056-1059.	1.1	278
100	Facioscapulohumeral dystrophy: the path to consensus on pathophysiology. <i>Skeletal Muscle</i> , 2014, 4, 12.	4.2	144
101	DNA polymorphism and epigenetic marks modulate the affinity of a scaffold/matrix attachment region to the nuclear matrix. <i>European Journal of Human Genetics</i> , 2014, 22, 1117-1123.	2.8	14
102	High prevalence of incomplete right bundle branch block in facioscapulohumeral muscular dystrophy without cardiac symptoms. <i>Functional Neurology</i> , 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. <i>BMC Genomics</i> , 2013, 14, 143.	2.8	29
104	Nuclear entrapment and extracellular depletion of PCOLCE is associated with muscle degeneration in oculopharyngeal muscular dystrophy. <i>BMC Neurology</i> , 2013, 13, 70.	1.8	15
105	MuSK IgG4 autoantibodies cause myasthenia gravis by inhibiting binding between MuSK and Lrp4. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 20783-20788.	7.1	234
106	Dysferlin Regulates Cell Adhesion in Human Monocytes. <i>Journal of Biological Chemistry</i> , 2013, 288, 14147-14157.	3.4	49
107	Determining the role of sarcomeric proteins in facioscapulohumeral muscular dystrophy: a study protocol. <i>BMC Neurology</i> , 2013, 13, 144.	1.8	12
108	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

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109	Antibodies to active zone protein ERC1 in Lambert-Eaton myasthenic syndrome. <i>Human Immunology</i> , 2013, 74, 849-851.	2.4	10
110	Sarcomeric dysfunction contributes to muscle weakness in facioscapulohumeral muscular dystrophy. <i>Neurology</i> , 2013, 80, 733-737.	1.1	36
111	Heterogeneous clinical presentation in ICF syndrome: correlation with underlying gene defects. <i>European Journal of Human Genetics</i> , 2013, 21, 1219-1225.	2.8	115
112	DUX4 Binding to Retroelements Creates Promoters That Are Active in FSHD Muscle and Testis. <i>PLoS Genetics</i> , 2013, 9, e1003947.	3.5	151
113	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
114	Chromatin remodeling of human subtelomeres and TERRA promoters upon cellular senescence. <i>Epigenetics</i> , 2013, 8, 512-521.	2.7	25
115	A focal domain of extreme demethylation within D4Z4 in FSHD2. <i>Neurology</i> , 2013, 80, 392-399.	1.1	67
116	A decline in PABPN1 induces progressive muscle weakness in Oculopharyngeal muscle dystrophy and in muscle aging. <i>Aging</i> , 2013, 5, 412-426.	3.1	49
117	Correlation analysis of clinical parameters with epigenetic modifications in the DUX4 promoter in FSHD. <i>Epigenetics</i> , 2012, 7, 579-584.	2.7	48
118	Epigenetic regulation of the X-chromosomal macrosatellite repeat encoding for the cancer/testis gene CT47. <i>European Journal of Human Genetics</i> , 2012, 20, 185-191.	2.8	15
119	Facioscapulohumeral muscular dystrophy. <i>Current Opinion in Neurology</i> , 2012, 25, 614-620.	3.6	42
120	Poly(A) binding protein nuclear 1 levels affect alternative polyadenylation. <i>Nucleic Acids Research</i> , 2012, 40, 9089-9101.	14.5	148
121	Patients with a phenotype consistent with facioscapulohumeral muscular dystrophy display genetic and epigenetic heterogeneity. <i>Journal of Medical Genetics</i> , 2012, 49, 41-46.	3.2	55
122	Self-regulated alternative splicing at the AHNAK locus. <i>FASEB Journal</i> , 2012, 26, 93-103.	0.5	24
123	Muscle-specific kinase myasthenia gravis IgG4 autoantibodies cause severe neuromuscular junction dysfunction in mice. <i>Brain</i> , 2012, 135, 1081-1101.	7.6	180
124	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
125	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
126	Best practice guidelines on genetic diagnostics of Facioscapulohumeral muscular dystrophy: Workshop 9th June 2010, LUMC, Leiden, The Netherlands. <i>Neuromuscular Disorders</i> , 2012, 22, 463-470.	0.6	53



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127	DUX4 Activates Germline Genes, Retroelements, and Immune Mediators: Implications for Facioscapulohumeral Dystrophy. <i>Developmental Cell</i> , 2012, 22, 38-51.	7.0	384
128	A genome-wide signature of glucocorticoid receptor binding in neuronal PC12 cells. <i>BMC Neuroscience</i> , 2012, 13, 118.	1.9	93
129	Pathogenic IgG4 subclass autoantibodies in MuSK myasthenia gravis. <i>Annals of the New York Academy of Sciences</i> , 2012, 1275, 114-122.	3.8	34
130	Selection of VHH Antibody Fragments That Recognize Different A $\beta$ 2 Deposits Using Complex Immune Libraries. <i>Methods in Molecular Biology</i> , 2012, 911, 241-253.	0.9	4
131	Asymmetric Bidirectional Transcription from the FSHD-Causing D4Z4 Array Modulates DUX4 Production. <i>PLoS ONE</i> , 2012, 7, e35532.	2.5	20
132	Immunodeficiency, centromeric instability, facial anomalies (ICF) syndrome, due to <i>ZBTB24</i> mutations, presenting with large cerebral cyst. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2043-2046.	1.2	25
133	In Vivo Detection of Amyloid- $\beta$ Deposits Using Heavy Chain Antibody Fragments in a Transgenic Mouse Model for Alzheimer's Disease. <i>PLoS ONE</i> , 2012, 7, e38284.	2.5	34
134	Clinical Dutch-English Lambert-Eaton Myasthenic Syndrome (LEMS) Tumor Association Prediction Score Accurately Predicts Small-Cell Lung Cancer in the LEMS. <i>Journal of Clinical Oncology</i> , 2011, 29, 902-908.	1.6	210
135	Modeling Oculopharyngeal Muscular Dystrophy in Myotube Cultures Reveals Reduced Accumulation of Soluble Mutant PABPN1 Protein. <i>American Journal of Pathology</i> , 2011, 179, 1988-2000.	3.8	34
136	Facioscapulohumeral muscular dystrophy and DUX4: breaking the silence. <i>Trends in Molecular Medicine</i> , 2011, 17, 252-258.	6.7	180
137	Facioscapulohumeral Muscular Dystrophy Region Gene 1 Is a Dynamic RNA-Associated and Actin-Bundling Protein. <i>Journal of Molecular Biology</i> , 2011, 411, 397-416.	4.2	22
138	Differential recognition of vascular and parenchymal beta amyloid deposition. <i>Neurobiology of Aging</i> , 2011, 32, 1774-1783.	3.1	34
139	Comparison of Dysferlin Expression in Human Skeletal Muscle with That in Monocytes for the Diagnosis of Dysferlin Myopathy. <i>PLoS ONE</i> , 2011, 6, e29061.	2.5	43
140	Distinguishing the 4qA and 4qB variants is essential for the diagnosis of facioscapulohumeral muscular dystrophy in the Chinese population. <i>European Journal of Human Genetics</i> , 2011, 19, 64-69.	2.8	15
141	Deregulation of the ubiquitin-proteasome system is the predominant molecular pathology in OPMD animal models and patients. <i>Skeletal Muscle</i> , 2011, 1, 15.	4.2	40
142	Mutations in ZBTB24 Are Associated with Immunodeficiency, Centromeric Instability, and Facial Anomalies Syndrome Type 2. <i>American Journal of Human Genetics</i> , 2011, 88, 796-804.	6.2	158
143	Reversible aggregation of PABPN1 pre-inclusion structures. <i>Nucleus</i> , 2011, 2, 208-218.	2.2	20
144	Interspecies Translation of Disease Networks Increases Robustness and Predictive Accuracy. <i>PLoS Computational Biology</i> , 2011, 7, e1002258.	3.2	15

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145	A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. <i>Science</i> , 2010, 329, 1650-1653.	12.6	638
146	Worldwide Population Analysis of the 4q and 10q Subtelomeres Identifies Only Four Discrete Interchromosomal Sequence Transfers in Human Evolution. <i>American Journal of Human Genetics</i> , 2010, 86, 364-377.	6.2	93
147	Structural basis for a PABPN1 aggregation-preventing antibody fragment in OPMD. <i>FEBS Letters</i> , 2010, 584, 1558-1564.	2.8	5
148	Analysis of allele-specific RNA transcription in FSHD by RNA-DNA FISH in single myonuclei. <i>European Journal of Human Genetics</i> , 2010, 18, 448-456.	2.8	34
149	Therapeutic exon skipping for dysferlinopathies?. <i>European Journal of Human Genetics</i> , 2010, 18, 889-894.	2.8	47
150	Reply to LÃ©vy et al. <i>European Journal of Human Genetics</i> , 2010, 18, 971-971.	2.8	2
151	Minimum information about a protein affinity reagent (MIAPAR). <i>Nature Biotechnology</i> , 2010, 28, 650-653.	17.5	50
152	Calpain 3 Is a Rapid-Action, Unidirectional Proteolytic Switch Central to Muscle Remodeling. <i>PLoS ONE</i> , 2010, 5, e11940.	2.5	23
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