

# Gisèle Bonne

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3600549/publications.pdf>

Version: 2024-02-01

248  
papers

15,520  
citations

18482

62  
h-index

19749

117  
g-index

269  
all docs

269  
docs citations

269  
times ranked

12416  
citing authors

#	ARTICLE	IF	CITATIONS
1	A lamin A/C variant causing striated muscle disease provides insights into filament organization. <i>Journal of Cell Science</i> , 2021, 134, .	2.0	17
2	International retrospective natural history study of <i>LMNA</i>-related congenital muscular dystrophy. <i>Brain Communications</i> , 2021, 3, fcab075.	3.3	17
3	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. <i>Brain</i> , 2021, 144, 2427-2442.	7.6	7
4	The Treatabolome, an emerging concept. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 337-339.	2.6	6
5	Laminopathiesâ€™ Treatments Systematic Review: A Contribution Towards a â€™Treatabolomeâ€™. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 419-439.	2.6	13
6	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). <i>European Journal of Human Genetics</i> , 2021, 29, 1348-1353.	2.8	10
7	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. <i>European Journal of Human Genetics</i> , 2021, 29, 1325-1331.	2.8	49
8	The non-muscle ADF/cofilin-1 controls sarcomeric actin filament integrity and force production in striated muscle laminopathies. <i>Cell Reports</i> , 2021, 36, 109601.	6.4	9
9	High-Throughput Digital Image Analysis Reveals Distinct Patterns of Dystrophin Expression in Dystrophinopathy Patients. <i>Journal of Neuropathology and Experimental Neurology</i> , 2021, 80, 955-965.	1.7	9
10	Targeting the histone demethylase LSD1 prevents cardiomyopathy in a mouse model of laminopathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	26
11	Lamin-Related Congenital Muscular Dystrophy Alters Mechanical Signaling and Skeletal Muscle Growth. <i>International Journal of Molecular Sciences</i> , 2021, 22, 306.	4.1	15
12	Preclinical Advances of Therapies for Laminopathies. <i>Journal of Clinical Medicine</i> , 2021, 10, 4834.	2.4	4
13	The 2022 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2021, 31, 1313-1357.	0.6	38
14	Novel role of <i>Tieg1</i> in muscle metabolism and mitochondrial oxidative capacities. <i>Acta Physiologica</i> , 2020, 228, e13394.	3.8	14
15	Mutant lamins cause nuclear envelope rupture and DNA damage in skeletal muscle cells. <i>Nature Materials</i> , 2020, 19, 464-473.	27.5	148
16	LATE BREAKING NEWS E-POSTER PRESENTATION. <i>Neuromuscular Disorders</i> , 2020, 30, S171.	0.6	0
17	The 2021 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2020, 30, 1008-1048.	0.6	45
18	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 206.	2.7	21

#	ARTICLE	IF	CITATIONS
19	Protein Kinase C Alpha Cellular Distribution, Activity, and Proximity with Lamin A/C in Striated Muscle Laminopathies. <i>Cells</i> , 2020, 9, 2388.	4.1	6
20	Activation of sarcolipin expression and altered calcium cycling in LMNA cardiomyopathy. <i>Biochemistry and Biophysics Reports</i> , 2020, 22, 100767.	1.3	11
21	Lamin A/C Assembly Defects in LMNA-Congenital Muscular Dystrophy Is Responsible for the Increased Severity of the Disease Compared with Emery-Dreifuss Muscular Dystrophy. <i>Cells</i> , 2020, 9, 844.	4.1	29
22	Consequences of Lmna Exon 4 Mutations in Myoblast Function. <i>Cells</i> , 2020, 9, 1286.	4.1	6
23	Microtubule cytoskeleton regulates Connexin 43 localization and cardiac conduction in cardiomyopathy caused by mutation in A-type lamins gene. <i>Human Molecular Genetics</i> , 2019, 28, 4043-4052.	2.9	35
24	214th ENMC International Workshop: Establishing an international consortium for gene discovery and clinical research for Congenital Muscle Disease, Heemskerk, the Netherlands, 6-18 October 2015. <i>Neuromuscular Disorders</i> , 2019, 29, 644-650.	0.6	2
25	Effect of genetic background on the cardiac phenotype in a mouse model of Emery-Dreifuss muscular dystrophy. <i>Biochemistry and Biophysics Reports</i> , 2019, 19, 100664.	1.3	15
26	P.143 Correlation between dystrophin expression and clinical phenotype using high-throughput digital immunoanalysis in Duchenne and Becker muscular dystrophy patients. <i>Neuromuscular Disorders</i> , 2019, 29, S90.	0.6	0
27	P.245 Morphological, ultrastructural and western blot analysis in adult and child with PLEC1-related myopathy. <i>Neuromuscular Disorders</i> , 2019, 29, S138.	0.6	0
28	P.252 LGMD, exercise intolerance, ptosis, ophthalmoplegia and dermatologic features: the phenotypic pleiotropy of plectinopathies in 8 French families. <i>Neuromuscular Disorders</i> , 2019, 29, S140.	0.6	0
29	P.256 Steroid treatment may change natural history in young children with LMNA mutations and dropped head syndrome. <i>Neuromuscular Disorders</i> , 2019, 29, S141.	0.6	2
30	The 2020 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2019, 29, 980-1018.	0.6	57
31	Deficiency of emerin contributes differently to the pathogenesis of skeletal and cardiac muscles in LmnaH222P/H222P mutant mice. <i>PLoS ONE</i> , 2019, 14, e0221512.	2.5	9
32	A Muscle Hybrid Promoter as a Novel Tool for Gene Therapy. <i>Molecular Therapy - Methods and Clinical Development</i> , 2019, 15, 157-169.	4.1	16
33	FHL1 is a major host factor for chikungunya virus infection. <i>Nature</i> , 2019, 574, 259-263.	27.8	49
34	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in <i>BVES</i> . <i>Neurology: Genetics</i> , 2019, 5, e321.	1.9	26
35	Development and Validation of a New Risk Prediction Score for Life-Threatening Ventricular Tachyarrhythmias in Laminopathies. <i>Circulation</i> , 2019, 140, 293-302.	1.6	131
36	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatable. <i>Emerging Topics in Life Sciences</i> , 2019, 3, 19-37.	2.6	47

#	ARTICLE	IF	CITATIONS
37	Cardiometabolic assessment of lamin A/C gene mutation carriers: a phenotype-genotype correlation. <i>Diabetes and Metabolism</i> , 2019, 45, 382-389.	2.9	22
38	A new case of SMA phenotype without epilepsy due to biallelic variants in <i>ASAH1</i> . <i>European Journal of Human Genetics</i> , 2019, 27, 337-339.	2.8	7
39	The mammalian LINC complex component <i>SUN1</i> regulates muscle regeneration by modulating <i>drosha</i> activity. <i>ELife</i> , 2019, 8, .	6.0	12
40	<i>SMAD6</i> overexpression leads to accelerated myogenic differentiation of <i>LMNA</i> mutated cells. <i>Scientific Reports</i> , 2018, 8, 5618.	3.3	6
41	Novel mutations in <i>DNAJB6</i> cause <i>LGMD1D</i> and distal myopathy in French families. <i>European Journal of Neurology</i> , 2018, 25, 790-794.	3.3	23
42	Gene Therapy via Trans-Splicing for <i>LMNA</i> -Related Congenital Muscular Dystrophy. <i>Molecular Therapy - Nucleic Acids</i> , 2018, 10, 376-386.	5.1	29
43	Elevated <i>TGF-<math>\beta</math>2</i> serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. <i>Nucleus</i> , 2018, 9, 337-349.	2.2	25
44	MoBiDiC Prioritization Algorithm, a Free, Accessible, and Efficient Pipeline for Single-Nucleotide Variant Annotation and Prioritization for Next-Generation Sequencing Routine Molecular Diagnosis. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 465-473.	2.8	13
45	Lamin and the heart. <i>Heart</i> , 2018, 104, 468-479.	2.9	113
46	<i>LGMD</i> AUTOSOMAL RECESSIVE AND DOMINANT. <i>Neuromuscular Disorders</i> , 2018, 28, S59-S60.	0.6	1
47	CONGENITAL MYOPATHIES: NEMALINE AND TITINOPATHIES. <i>Neuromuscular Disorders</i> , 2018, 28, S104-S105.	0.6	0
48	The Pathogenesis and Therapies of Striated Muscle Laminopathies. <i>Frontiers in Physiology</i> , 2018, 9, 1533.	2.8	30
49	The 2019 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2018, 28, 1031-1063.	0.6	33
50	Loss of Sarcomeric Scaffolding as a Common Baseline Histopathologic Lesion in Titin-Related Myopathies. <i>Journal of Neuropathology and Experimental Neurology</i> , 2018, 77, 1101-1114.	1.7	22
51	Amelioration of desmin network defects by $\beta$ -crystallin overexpression confers cardioprotection in a mouse model of dilated cardiomyopathy caused by <i>LMNA</i> gene mutation. <i>Journal of Molecular and Cellular Cardiology</i> , 2018, 125, 73-86.	1.9	31
52	Dystrophin quantification in Duchenne and Becker muscular dystrophy: correlation between dystrophin protein and clinical phenotype. <i>Neuromuscular Disorders</i> , 2018, 28, S7-S8.	0.6	0
53	<i>N</i> -acetyl cysteine alleviates oxidative stress and protects mice from dilated cardiomyopathy caused by mutations in nuclear A-type lamins gene. <i>Human Molecular Genetics</i> , 2018, 27, 3353-3360.	2.9	10
54	Rescue of biosynthesis of nicotinamide adenine dinucleotide protects the heart in cardiomyopathy caused by lamin A/C gene mutation. <i>Human Molecular Genetics</i> , 2018, 27, 3870-3880.	2.9	40

#	ARTICLE	IF	CITATIONS
55	Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for follow-up. <i>Muscle and Nerve</i> , 2018, 58, 812-817.	2.2	8
56	The <sup>13</sup> C hyperpolarized pyruvate generated by ParaHydrogen detects the response of the heart to altered metabolism in real time. <i>Scientific Reports</i> , 2018, 8, 8366.	3.3	119
57	Cofilin-1 phosphorylation catalyzed by ERK1/2 alters cardiac actin dynamics in dilated cardiomyopathy caused by lamin A/C gene mutation. <i>Human Molecular Genetics</i> , 2018, 27, 3060-3078.	2.9	42
58	Decreased WNT/ $\beta$ -catenin signalling contributes to the pathogenesis of dilated cardiomyopathy caused by mutations in the lamin a/C gene. <i>Human Molecular Genetics</i> , 2017, 26, ddw389.	2.9	58
59	Genetic Characterization of a French Cohort of GNE-mutation negative inclusion body myopathy patients with exome sequencing. <i>Muscle and Nerve</i> , 2017, 56, 993-997.	2.2	6
60	Cardiac manifestations of congenital LMNA-related muscular dystrophy in children: three case reports and recommendations for care. <i>Cardiology in the Young</i> , 2017, 27, 1076-1082.	0.8	20
61	Genetic characterization of a French cohort of GNE-mutation negative inclusion body myopathy patients using exome sequencing. <i>Neuromuscular Disorders</i> , 2017, 27, S149.	0.6	0
62	Welcome to the World Muscle Society Congress in Saint Malo. <i>Neuromuscular Disorders</i> , 2017, 27, S52-S53.	0.6	0
63	Anti-HMGR Antibody-Related Necrotizing Autoimmune Myopathy Mimicking Muscular Dystrophy. <i>Neuropediatrics</i> , 2017, 48, 473-476.	0.6	15
64	The 2018 version of the gene table of monogenic neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2017, 27, 1152-1183.	0.6	41
65	Collagen VI deficiency: The heart of the matter. <i>Neuromuscular Disorders</i> , 2017, 27, S106.	0.6	0
66	A novel INPP5K mutation in a sibship from the Reunion Island. <i>Neuromuscular Disorders</i> , 2017, 27, S110-S111.	0.6	0
67	TIEG1 is a novel regulator of muscle mitochondrial biogenesis. <i>Neuromuscular Disorders</i> , 2017, 27, S117.	0.6	1
68	First results from the international LMNA-related congenital and childhood onset muscular dystrophy retrospective natural history study. <i>Neuromuscular Disorders</i> , 2017, 27, S137-S138.	0.6	0
69	Corticosteroid treatment in early-onset lamin A/C related muscular dystrophies. <i>Neuromuscular Disorders</i> , 2017, 27, S138.	0.6	1
70	Abnormal trafficking of connexin 43: A key element in the development of LMNA cardiomyopathy. <i>Neuromuscular Disorders</i> , 2017, 27, S139.	0.6	0
71	Novel recessive splice site mutation in POPDC1 ( BVES ) is associated with first-degree atrioventricular block and muscular dystrophy. <i>Neuromuscular Disorders</i> , 2017, 27, S139-S140.	0.6	2
72	POPDC1 gene mutation screening in patients with LGMD and heart disturbances: a mutation load effect?. <i>Neuromuscular Disorders</i> , 2017, 27, S140.	0.6	0

#	ARTICLE	IF	CITATIONS
73	Integrated analysis of the large-scale sequencing project "Myocapture" to identify novel genes for myopathies. <i>Neuromuscular Disorders</i> , 2017, 27, S195.	0.6	1
74	Morphological spectrum of RYR1 recessive myopathies: Clinical and genetic correlation.. <i>Neuromuscular Disorders</i> , 2017, 27, S239.	0.6	0
75	Lamins and nesprin-1 mediate inside-out mechanical coupling in muscle cell precursors through FHOD1. <i>Scientific Reports</i> , 2017, 7, 1253.	3.3	35
76	Évaluation cardiométaboliques des laminopathies selon le type de mutation (R482 ou NON-R482). <i>Diabetes and Metabolism</i> , 2017, 43, A89-A90.	2.9	0
77	Clinical heterogeneity and phenotype/genotype findings in 5 families with <i>CYG1</i> deficiency. <i>Neurology: Genetics</i> , 2017, 3, e208.	1.9	12
78	Clinical features and therapeutic strategies for managing the striated muscle laminopathies. <i>Expert Opinion on Orphan Drugs</i> , 2016, 4, 631-638.	0.8	1
79	Laminopathies: Why make it simple when it can be complex?. <i>Neuromuscular Disorders</i> , 2016, 26, S150-S151.	0.6	2
80	FHL1B Interacts with Lamin A/C and Émerin at the Nuclear Lamina and ÉmisÉmisregulated in Emery-Dreifuss Muscular Dystrophy. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 497-510.	2.6	17
81	The 2017 version of the gene table of monogenic neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2016, 26, 895-929.	0.6	10
82	Mutation in lamin A/C sensitizes the myocardium to exercise-induced mechanical stress but has no effect on skeletal muscles in mouse. <i>Neuromuscular Disorders</i> , 2016, 26, 490-499.	0.6	30
83	ERK1/2 directly acts on CTGF/CCN2 expression to mediate myocardial fibrosis in cardiomyopathy caused by mutations in the lamin A/C gene. <i>Human Molecular Genetics</i> , 2016, 25, 2220-2233.	2.9	76
84	Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. <i>American Journal of Human Genetics</i> , 2016, 99, 753-761.	6.2	68
85	Cardiac arrhythmia and late-onset muscle weakness caused by a myofibrillar myopathy with unusual histopathological features due to a novel missense mutation in FLNC. <i>Revue Neurologique</i> , 2016, 172, 594-606.	1.5	19
86	Laminopathies disrupt epigenomic developmental programs and cell fate. <i>Science Translational Medicine</i> , 2016, 8, 335ra58.	12.4	91
87	Interest of whole-body muscle MRI for the diagnosis of Pompe disease in rigid spine syndrome and differential diagnosis. <i>Neuromuscular Disorders</i> , 2016, 26, S109.	0.6	0
88	OPALE: A patient registry for laminopathies and emerinopathies in France. <i>Neuromuscular Disorders</i> , 2016, 26, S138.	0.6	1
89	EGR2 mutation enhances phenotype spectrum of DejerineÉSottas syndrome. <i>Journal of Neurology</i> , 2016, 263, 1456-1458.	3.6	5
90	Pediatric laminopathies: Whole-body magnetic resonance imaging fingerprint and comparison with <i>Sepn1</i> myopathy. <i>Muscle and Nerve</i> , 2016, 54, 192-202.	2.2	31

#	ARTICLE	IF	CITATIONS
91	Mechanosensing Defects in Nuclear Envelope Related Disorders. Biophysical Journal, 2016, 110, 96a.	0.5	0
92	La Myologie ã lâ€™AcadÃ©mie Nationale de MÃ©decine. Les Cahiers De Myologie, 2016, , 3-4.	0.0	0
93	Whole exome sequencing at the Institute of Myology in the context of the Myocapture project to identify novel genes of myopathies. Neuromuscular Disorders, 2015, 25, S299.	0.6	0
94	An overview of new translational, clinical and therapeutic perspectives in laminopathies and other nuclear envelope-related diseases.. Orphanet Journal of Rare Diseases, 2015, 10, 11.	2.7	0
95	A common French-Italian laminopathy registry â€” update & future prospects. Orphanet Journal of Rare Diseases, 2015, 10, .	2.7	0
96	Laminin Î±2 Deficiency-Related Muscular Dystrophy Mimicking Emery-Dreifuss andÃ©Collagen VI related Diseases. Journal of Neuromuscular Diseases, 2015, 2, 229-240.	2.6	30
97	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. European Journal of Human Genetics, 2015, 23, 1051-1061.	2.8	24
98	Myofibrillar myopathies: State of the art, present and future challenges. Revue Neurologique, 2015, 171, 715-729.	1.5	38
99	Gene therapy via trans-splicing for LMNA-related congenital muscular dystrophy (L-CMD). Neuromuscular Disorders, 2015, 25, S280.	0.6	0
100	A new titinopathy. Neurology, 2015, 85, 2126-2135.	1.1	44
101	Mutations in ASAH1 may cause spinal muscular atrophy. Neuromuscular Disorders, 2015, 25, S225.	0.6	1
102	Exome sequencing identifies novel truncating TTN mutations with Emeryâ€™Dreifuss like muscular dystrophy and secondary calpain3 deficiency without cardiac abnormality. Neuromuscular Disorders, 2015, 25, S245.	0.6	0
103	Highly variable skeletal muscle histo-immunocytochemical and ultrastructural features in titin-related myopathies. Neuromuscular Disorders, 2015, 25, S287-S288.	0.6	0
104	Detection of TRIM32 deletions in LGMD patients analyzed by a combined strategy of CGH array and massively parallel sequencing. European Journal of Human Genetics, 2015, 23, 929-934.	2.8	21
105	Nuclear envelope and striated muscle diseases. Current Opinion in Cell Biology, 2015, 32, 1-6.	5.4	14
106	Severe dystonia, cerebellar atrophy, and cardiomyopathy likely caused by a missense mutation in TOR1AIP1. Orphanet Journal of Rare Diseases, 2014, 9, 174.	2.7	43
107	FHL1 mutations that cause clinically distinct human myopathies form protein aggregates and impair myoblast differentiation. Journal of Cell Science, 2014, 127, 2269-81.	2.0	19
108	P330Overexpression of the muscle specific chaperone Melusin delays heart failure and mortality in a mouse model of Emery Dreyfus cardiomyopathy. Cardiovascular Research, 2014, 103, S60.1-S60.	3.8	0

#	ARTICLE	IF	CITATIONS
109	Dystrophin quantification. <i>Neurology</i> , 2014, 83, 2062-2069.	1.1	73
110	Striated muscle laminopathies. <i>Seminars in Cell and Developmental Biology</i> , 2014, 29, 107-115.	5.0	48
111	Actin scaffolding by clathrin heavy chain is required for skeletal muscle sarcomere organization. <i>Journal of Cell Biology</i> , 2014, 205, 377-393.	5.2	60
112	Forelimb Treatment in a Large Cohort of Dystrophic Dogs Supports Delivery of a Recombinant AAV for Exon Skipping in Duchenne Patients. <i>Molecular Therapy</i> , 2014, 22, 1923-1935.	8.2	108
113	G.P.142. <i>Neuromuscular Disorders</i> , 2014, 24, 843-844.	0.6	2
114	G.P.145. <i>Neuromuscular Disorders</i> , 2014, 24, 844-845.	0.6	0
115	G.P.92. <i>Neuromuscular Disorders</i> , 2014, 24, 822.	0.6	1
116	G.P.281. <i>Neuromuscular Disorders</i> , 2014, 24, 901-902.	0.6	0
117	Cellular micro-environments reveal defective mechanosensing responses and elevated YAP signaling in LMNA-mutated muscle precursors. <i>Journal of Cell Science</i> , 2014, 127, 2873-84.	2.0	105
118	G.P.150. <i>Neuromuscular Disorders</i> , 2014, 24, 846.	0.6	0
119	Nuclear envelope proteins in health and diseases. <i>Seminars in Cell and Developmental Biology</i> , 2014, 29, 93-94.	5.0	5
120	P.5.16 Predominant right ventricular involvement in patients with laminopathies. <i>Neuromuscular Disorders</i> , 2013, 23, 768-769.	0.6	0
121	Emeryâ€Dreifuss muscular dystrophy, laminopathies, and other nuclear envelopathies. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , 2013, 113, 1367-1376.	1.8	96
122	A novel genetic variant in the transcription factor Islet-1 exerts gain of function on myocyte enhancer factor 2C promoter activity. <i>European Journal of Heart Failure</i> , 2013, 15, 267-276.	7.1	21
123	Muscle dystrophy-causing $\hat{K}32$ lamin A/C mutant does not impair functions of nucleoplasmic LAP2 $\hat{\pm}$ - lamin A/C complexes in mice. <i>Journal of Cell Science</i> , 2013, 126, 1753-62.	2.0	31
124	P.1.15 Clinical heterogeneity of myopathy related to partial merosin deficiency. <i>Neuromuscular Disorders</i> , 2013, 23, 746.	0.6	1
125	Mutations in Lamin A/C Gene Causes Mechanosensing Defects in Human Myoblasts. <i>Biophysical Journal</i> , 2013, 104, 374a.	0.5	0
126	Role of dynamin 2 in the disassembly of focal adhesions. <i>Journal of Molecular Medicine</i> , 2013, 91, 803-809.	3.9	7



#	ARTICLE	IF	CITATIONS
127	Nuclear accumulation of androgen receptor in gender difference of dilated cardiomyopathy due to lamin A/C mutations. <i>Cardiovascular Research</i> , 2013, 99, 382-394.	3.8	41
128	Myoblasts and Embryonic Stem Cells Differentially Engraft in a Mouse Model of Genetic Dilated Cardiomyopathy. <i>Molecular Therapy</i> , 2013, 21, 1064-1075.	8.2	9
129	Heterozygous LmndelK32 mice develop dilated cardiomyopathy through a combined pathomechanism of haploinsufficiency and peptide toxicity. <i>Human Molecular Genetics</i> , 2013, 22, 3152-3164.	2.9	72
130	Skeletal Muscle Biopsy Analysis in Reducing Body Myopathy and Other FHL1-Related Disorders. <i>Journal of Neuropathology and Experimental Neurology</i> , 2013, 72, 833-845.	1.7	36
131	“State-of-the-heart”™ of cardiac laminopathies. <i>Current Opinion in Cardiology</i> , 2013, 28, 297-304.	1.8	60
132	Hypoplasia of the Aorta in a Patient Diagnosed with LMNA Gene Mutation. <i>Congenital Heart Disease</i> , 2013, 8, E127-E129.	0.2	3
133	Distinctive Serum miRNA Profile in Mouse Models of Striated Muscular Pathologies. <i>PLoS ONE</i> , 2013, 8, e55281.	2.5	97
134	DelK32-lamin A/C has abnormal location and induces incomplete tissue maturation and severe metabolic defects leading to premature death. <i>Human Molecular Genetics</i> , 2012, 21, 1037-1048.	2.9	77
135	Treatment with selumetinib preserves cardiac function and improves survival in cardiomyopathy caused by mutation in the lamin A/C gene. <i>Cardiovascular Research</i> , 2012, 93, 311-319.	3.8	86
136	Evidence for FHL1 as a novel disease gene for isolated hypertrophic cardiomyopathy. <i>Human Molecular Genetics</i> , 2012, 21, 3237-3254.	2.9	106
137	Blood glutathione decrease in subjects carrying lamin A/C gene mutations is an early marker of cardiac involvement. <i>Neuromuscular Disorders</i> , 2012, 22, 252-257.	0.6	7
138	D.P.2 Next generation sequencing after selected DNA capture as a tool for molecular diagnosis of neuromuscular disorders. <i>Neuromuscular Disorders</i> , 2012, 22, 807-808.	0.6	0
139	D.P.25 Mutation in lamin A/C gene causes mechanotransduction defects in human myoblasts. <i>Neuromuscular Disorders</i> , 2012, 22, 825.	0.6	0
140	G.P.18 Muscle pathology and dysfunction in a novel mouse model of COLVI-myopathy. <i>Neuromuscular Disorders</i> , 2012, 22, 827-828.	0.6	2
141	G.P.120 FHL1-related Reducing Body Myopathy and Emery “Dreifuss muscular dystrophy: A comparative histoenzymological, immunohistochemical and ultrastructural study. <i>Neuromuscular Disorders</i> , 2012, 22, 902.	0.6	0
142	G.P.122 Heterozygous LmndelK32 mutant mice showed alterations of the ubiquitin “proteasome system and developed dilated cardiomyopathy. <i>Neuromuscular Disorders</i> , 2012, 22, 903.	0.6	0
143	3D Culture of Human Muscle Cells Modulates Cell-Matrix Adhesions and Actin Cytoskeleton Organization. <i>Biophysical Journal</i> , 2012, 102, 417a.	0.5	0
144	Diseases of the Nucleoskeleton. , 2012, , 1003-1012.		0

#	ARTICLE	IF	CITATIONS
145	Complex Interactions between Human Myoblasts and the Surrounding 3D Fibrin-Based Matrix. PLoS ONE, 2012, 7, e36173.	2.5	83
146	Lamin A/C Mutants Disturb Sumo1 Localization and Sumoylation in Vitro and in Vivo. PLoS ONE, 2012, 7, e45918.	2.5	26
147	A Centronuclear Myopathy â€“ Dynamin 2 Mutation Impairs Autophagy in Mice. Traffic, 2012, 13, 869-879.	2.7	52
148	Guiding the molecular diagnosis of hypertrophic cardiomyopathy. Journal of Thoracic and Cardiovascular Surgery, 2012, 143, 1234.	0.8	0
149	What Should the Cardiologist know about Lamin Disease?. Arrhythmia and Electrophysiology Review, 2012, 1, 22.	2.4	24
150	Distinction Between Two Populations of Islet-1-Positive Cells in Hearts of Different Murine Strains. Stem Cells and Development, 2011, 20, 1043-1052.	2.1	32
151	MicroRNA expression profiling in patients with lamin A/Câ€“associated muscular dystrophy. FASEB Journal, 2011, 25, 3966-3978.	0.5	42
152	N-terminal Pro brain natriuretic peptide is a reliable biomarker of reduced myocardial contractility in patients with lamin A/C gene mutations. International Journal of Cardiology, 2011, 151, 160-163.	1.7	4
153	P5.62 Garches muscle Whole-Body MRI protocol: Pattern recognition in early onset neuromuscular disorders. Neuromuscular Disorders, 2011, 21, 742-743.	0.6	0
154	P2.10 A new form of congenital muscular dystrophy with subsarcolemmal inclusions arising from disintegrated myonuclei. Neuromuscular Disorders, 2011, 21, 663-664.	0.6	0
155	P2.41 From Emery-Dreifuss muscular dystrophy to striated muscle laminopathies. A 12years retrospective. Neuromuscular Disorders, 2011, 21, 672-673.	0.6	0
156	P2.43 Revisiting X-linked Emery-Dreifuss muscular dystrophy. New insights into an old story. Neuromuscular Disorders, 2011, 21, 673.	0.6	0
157	Clinical and genetic heterogeneity in laminopathies. Biochemical Society Transactions, 2011, 39, 1687-1692.	3.4	107
158	Type B mandibuloacral dysplasia with congenital myopathy due to homozygous ZMPSTE24 missense mutation. European Journal of Human Genetics, 2011, 19, 647-654.	2.8	44
159	Association of Adult-Onset Glycogenosis Type II and a Mutation in the LMNA Gene in Two Patients: Different Clinical and Histological Phenotypes. Clinical Therapeutics, 2011, 33, S36.	2.5	0
160	Modifier locus of the skeletal muscle involvement in Emeryâ€“Dreifuss muscular dystrophy. Human Genetics, 2011, 129, 149-159.	3.8	32
161	ColVI myopathies: where do we stand, where do we go?. Skeletal Muscle, 2011, 1, 30.	4.2	84
162	Cardioembolic stroke prompting diagnosis of <i>LMNA</i>-â€“associated Emeryâ€“Dreifuss muscular dystrophy. Muscle and Nerve, 2011, 44, 587-589.	2.2	7

#	ARTICLE	IF	CITATIONS
163	Mitogen-Activated Protein Kinase Inhibitors Improve Heart Function and Prevent Fibrosis in Cardiomyopathy Caused by Mutation in Lamin A/C Gene. <i>Circulation</i> , 2011, 123, 53-61.	1.6	154
164	Contractures and hypertrophic cardiomyopathy in a novel FHL1 mutation. <i>Annals of Neurology</i> , 2010, 67, 136-140.	5.3	64
165	Improvement of Left Ventricular Dysfunction and of Survival Prognosis of Dilated Cardiomyopathy by Administration of Calcium Sensitizer SCH00013 in a Mouse Model. <i>Journal of the American College of Cardiology</i> , 2010, 55, 1503-1505.	2.8	20
166	Pharmacological inhibition of c-Jun N-terminal kinase signaling prevents cardiomyopathy caused by mutation in LMNA gene. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2010, 1802, 632-638.	3.8	52
167	Differentiating Emery-Dreifuss muscular dystrophy and collagen VI-related myopathies using a specific CT scanner pattern. <i>Neuromuscular Disorders</i> , 2010, 20, 517-523.	0.6	31
168	O.4 Mouse model of LMNA-congenital muscular dystrophy shows severe skeletal and cardiac muscle maturation defects associated with major metabolic defects leading to early death. <i>Neuromuscular Disorders</i> , 2010, 20, 598.	0.6	0
169	P1.15 DNA micro-arrays for revisiting molecular pathology in neuromuscular disorders. <i>Neuromuscular Disorders</i> , 2010, 20, 604.	0.6	1
170	P2.18 A novel missense FLNC mutation causes arrhythmia and late onset myofibrillar myopathy with particular histopathology features. <i>Neuromuscular Disorders</i> , 2010, 20, 623-624.	0.6	4
171	Upregulation of PPAR $\alpha$ Is Associated with Structural and Functional Changes in the Type I Diabetes Rat Diaphragm. <i>PLoS ONE</i> , 2010, 5, e11494.	2.5	9
172	MHC Class II Deficiency. , 2009, , 1306-1308.		0
173	Autophagic degradation of nuclear components in mammalian cells. <i>Autophagy</i> , 2009, 5, 795-804.	9.1	189
174	Lamin A-mediated neuromuscular junction defects in Emery-Dreifuss muscular dystrophy. <i>Journal of Cell Biology</i> , 2009, 184, 31-44.	5.2	105
175	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2009, 85, 338-353.	6.2	208
176	Germinal mosaicism for LMNA mimics autosomal recessive congenital muscular dystrophy. <i>Neuromuscular Disorders</i> , 2009, 19, 26-28.	0.6	24
177	De novo LMNA mutations cause a new form of congenital muscular dystrophy. <i>Annals of Neurology</i> , 2008, 64, 177-186.	5.3	255
178	Heart-hand syndrome of Slovenian type: a new kind of laminopathy. <i>Journal of Medical Genetics</i> , 2008, 45, 666-671.	3.2	47
179	Inhibition of extracellular signal-regulated kinase signaling to prevent cardiomyopathy caused by mutation in the gene encoding A-type lamins. <i>Human Molecular Genetics</i> , 2008, 18, 241-247.	2.9	149
180	Genetics of Laminopathies. <i>Novartis Foundation Symposium</i> , 2008, , 81-97.	1.1	18

#	ARTICLE	IF	CITATIONS
181	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. <i>Neurology</i> , 2007, 69, 1285-1292.	1.1	120
182	Activation of MAPK in hearts of EMD null mice: similarities between mouse models of X-linked and autosomal dominant Emery-Dreifuss muscular dystrophy. <i>Human Molecular Genetics</i> , 2007, 16, 1884-1895.	2.9	123
183	141st ENMC International Workshop Inaugural Meeting of the EURO-Laminopathies Project Nuclear Envelope-linked Rare Human Diseases: From Molecular Pathophysiology towards Clinical Applications 10-12 March 2006, Naarden, The Netherlands. <i>Neuromuscular Disorders</i> , 2007, 17, 655-660.	0.6	11
184	Structural analysis of four and half LIM protein-2 in dilated cardiomyopathy. <i>Biochemical and Biophysical Research Communications</i> , 2007, 357, 162-167.	2.1	55
185	Primary laminopathy fibroblasts display altered genome organization and apoptosis. <i>Aging Cell</i> , 2007, 6, 139-153.	6.7	140
186	“Laminopathies” A wide spectrum of human diseases. <i>Experimental Cell Research</i> , 2007, 313, 2121-2133.	2.6	560
187	Activation of MAPK pathways links LMNA mutations to cardiomyopathy in Emery-Dreifuss muscular dystrophy. <i>Journal of Clinical Investigation</i> , 2007, 117, 1282-1293.	8.2	256
188	Primary Prevention of Sudden Death in Patients with Lamin A/C Gene Mutations. <i>New England Journal of Medicine</i> , 2006, 354, 209-210.	27.0	323
189	Nuclear Lamins: Laminopathies and Their Role in Premature Ageing. <i>Physiological Reviews</i> , 2006, 86, 967-1008.	28.8	494
190	Proteasome-mediated degradation of integral inner nuclear membrane protein emerin in fibroblasts lacking A-type lamins. <i>Biochemical and Biophysical Research Communications</i> , 2006, 351, 1011-1017.	2.1	31
191	Myofiber Degeneration in Autosomal Dominant Emery-Dreifuss Muscular Dystrophy (AD-EDMD) (LGMD1B). <i>Brain Pathology</i> , 2006, 16, 266-272.	4.1	15
192	Disease severity in dominant Emery Dreifuss is increased by mutations in both emerin and desmin proteins. <i>Brain</i> , 2006, 129, 1260-1268.	7.6	114
193	Primary Myocardial Dysfunction in Autosomal Dominant EDMD. A Tissue Doppler and Cardiovascular Magnetic Resonance Study. <i>Journal of Cardiovascular Magnetic Resonance</i> , 2006, 8, 723-730.	3.3	28
194	Extreme variability of skeletal and cardiac muscle involvement in patients with mutations in exon 11 of the lamin A/C gene. <i>Muscle and Nerve</i> , 2005, 31, 602-609.	2.2	68
195	Meta-analysis of clinical characteristics of 299 carriers of LMNA gene mutations: do lamin A/C mutations portend a high risk of sudden death?. <i>Journal of Molecular Medicine</i> , 2005, 83, 79-83.	3.9	388
196	Mouse model carrying H222P- Lmna mutation develops muscular dystrophy and dilated cardiomyopathy similar to human striated muscle laminopathies. <i>Human Molecular Genetics</i> , 2005, 14, 155-169.	2.9	303
197	Lamin A N-terminal phosphorylation is associated with myoblast activation: impairment in Emery-Dreifuss muscular dystrophy. <i>Journal of Medical Genetics</i> , 2005, 42, 214-220.	3.2	52
198	Deletion of the LMNA initiator codon leading to a neurogenic variant of autosomal dominant Emery-Dreifuss muscular dystrophy. <i>Neuromuscular Disorders</i> , 2005, 15, 40-44.	0.6	38

#	ARTICLE	IF	CITATIONS
199	Two patients with â€“Dropped head syndromeâ€™ due to mutations in LMNA or SEPN1 genes. <i>Neuromuscular Disorders</i> , 2005, 15, 521-524.	0.6	61
200	Peripheral nerve lesions associated with a dominant missense mutation, E33D, of the lamin A/C gene. <i>Neuromuscular Disorders</i> , 2005, 15, 618-621.	0.6	9
201	Genetics of laminopathies. <i>Novartis Foundation Symposium</i> , 2005, 264, 81-90; discussion 90-97, 227-30.	1.1	8
202	Extreme Variability of Phenotype in Patients With an Identical Missense Mutation in the Lamin A/C Gene. <i>Archives of Neurology</i> , 2004, 61, 690.	4.5	114
203	Nuclear envelope alterations in fibroblasts from patients with muscular dystrophy, cardiomyopathy, and partial lipodystrophy carrying lamin A/C gene mutations. <i>Muscle and Nerve</i> , 2004, 30, 444-450.	2.2	167
204	A new mutation of the lamin A/C gene leading to autosomal dominant axonal neuropathy, muscular dystrophy, cardiac disease, and leuconychia. <i>Journal of Medical Genetics</i> , 2004, 41, 29e-29.	3.2	76
205	Cardiomyopathy, Dilated (Familial). , 2004, , 216-220.		0
206	Nuclear envelope alterations in fibroblasts from LGMD1B patients carrying nonsense Y259X heterozygous or homozygous mutation in lamin A/C gene. <i>Experimental Cell Research</i> , 2003, 291, 352-362.	2.6	169
207	LMNA mutations in atypical Werner's syndrome. <i>Lancet, The</i> , 2003, 362, 1585-1586.	13.7	37
208	108th ENMC International Workshop, 3rd Workshop of the MYO-CLUSTER project: EUROMEN, 7th International Emery-Dreifuss Muscular Dystrophy (EDMD) Workshop, 13â€“15 September 2002, Naarden, The Netherlands. <i>Neuromuscular Disorders</i> , 2003, 13, 508-515.	0.6	78
209	Expanding the phenotype of LMNA mutations in dilated cardiomyopathy and functional consequences of these mutations. <i>Journal of Medical Genetics</i> , 2003, 40, 560-567.	3.2	156
210	Apical left ventricular aneurysm without atrio-ventricular block due to a lamin A/C gene mutation. <i>European Journal of Heart Failure</i> , 2003, 5, 821-825.	7.1	39
211	Clinical Relevance of Atrial Fibrillation/Flutter, Stroke, Pacemaker Implant, and Heart Failure in Emery-Dreifuss Muscular Dystrophy. <i>Stroke</i> , 2003, 34, 901-908.	2.0	158
212	Selective Muscle Involvement on Magnetic Resonance Imaging in Autosomal Dominant Emery-Dreifuss Muscular Dystrophy. <i>Neuropediatrics</i> , 2002, 33, 10-14.	0.6	101
213	Emery-Dreifuss muscular dystrophy. <i>European Journal of Human Genetics</i> , 2002, 10, 157-161.	2.8	121
214	Mandibuloacral Dysplasia Is Caused by a Mutation in LMNA-Encoding Lamin A/C. <i>American Journal of Human Genetics</i> , 2002, 71, 426-431.	6.2	509
215	Autosomal dominant Emeryâ€“Dreifuss muscular dystrophy: a new family with late diagnosis. <i>Neuromuscular Disorders</i> , 2002, 12, 19-25.	0.6	18
216	Frequent low penetrance mutations in the Lamin A/C gene, causing Emery Dreifuss muscular dystrophy. <i>Neuromuscular Disorders</i> , 2002, 12, 958-963.	0.6	52

#	ARTICLE	IF	CITATIONS
217	The Nuclear muscular dystrophies. <i>Seminars in Pediatric Neurology</i> , 2002, 9, 100-107.	2.0	26
218	The Ig-like Structure of the C-Terminal Domain of Lamin A/C, Mutated in Muscular Dystrophies, Cardiomyopathy, and Partial Lipodystrophy. <i>Structure</i> , 2002, 10, 811-823.	3.3	252
219	Laminopathies: One Gene, Two Proteins, Five Diseases. <i>J. Biol. Chem.</i> , 2002, 277, 153-172.		4
220	COOH-terminal truncated human cardiac MyBP-C alters myosin filament organization. <i>Comptes Rendus De L'Académie Des Sciences Série 3, Sciences De La Vie</i> , 2001, 324, 251-260.	0.8	14
221	A missense mutation in the exon 8 of lamin A/C gene in a Japanese case of autosomal dominant limb-girdle muscular dystrophy and cardiac conduction block. <i>Neuromuscular Disorders</i> , 2001, 11, 542-546.	0.6	42
222	Skeletal muscle pathology in autosomal dominant Emery-Dreifuss muscular dystrophy with lamin A/C mutations. <i>Neuropathology and Applied Neurobiology</i> , 2001, 27, 281-290.	3.2	117
223	High Incidence of Sudden Death with Conduction System and Myocardial Disease Due to Lamins A and C Gene Mutation. <i>PACE - Pacing and Clinical Electrophysiology</i> , 2000, 23, 1661-1666.	1.2	234
224	Identification of mutations in the gene encoding lamins A/C in autosomal dominant limb girdle muscular dystrophy with atrioventricular conduction disturbances (LGMD1B). <i>Human Molecular Genetics</i> , 2000, 9, 1453-1459.	2.9	530
225	First description of germline mosaicism in familial hypertrophic cardiomyopathy. <i>Journal of Medical Genetics</i> , 2000, 37, 132-134.	3.2	30
226	Different Mutations in the LMNA Gene Cause Autosomal Dominant and Autosomal Recessive Emery-Dreifuss Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2000, 66, 1407-1412.	6.2	384
227	Mutations in the gene encoding lamin A/C cause autosomal dominant Emery-Dreifuss muscular dystrophy. <i>Nature Genetics</i> , 1999, 21, 285-288.	21.4	1,245
228	Cardiac Myosin-Binding Protein C and Hypertrophic Cardiomyopathy. <i>Trends in Cardiovascular Medicine</i> , 1998, 8, 151-157.	4.9	11
229	Identification of two novel mutations in the ventricular regulatory myosin light chain gene ( MYL2 ) associated with familial and classical forms of hypertrophic cardiomyopathy. <i>Journal of Molecular Medicine</i> , 1998, 76, 208-214.	3.9	130
230	Familial Hypertrophic Cardiomyopathy. <i>Circulation Research</i> , 1998, 83, 580-593.	4.5	354
231	Clinical Features and Prognostic Implications of Familial Hypertrophic Cardiomyopathy Related to the Cardiac Myosin-Binding Protein C Gene. <i>Circulation</i> , 1998, 97, 2230-2236.	1.6	241
232	Familial hypertrophic cardiomyopathy. Cardiac ultrasonic abnormalities in genetically affected subjects without echocardiographic evidence of left ventricular hypertrophy. <i>European Heart Journal</i> , 1998, 19, 490-499.	2.2	39
233	Diagnostic value of electrocardiography and echocardiography for familial hypertrophic cardiomyopathy in genotyped children. <i>European Heart Journal</i> , 1998, 19, 1377-1382.	2.2	49
234	Genotype-phenotype correlations in familial hypertrophic cardiomyopathy A comparison between mutations in the cardiac protein-C and the beta-myosin heavy chain genes. <i>European Heart Journal</i> , 1998, 19, 139-145.	2.2	92

#	ARTICLE	IF	CITATIONS
235	Diagnostic Value of Electrocardiography and Echocardiography for Familial Hypertrophic Cardiomyopathy in a Genotyped Adult Population. <i>Circulation</i> , 1997, 96, 214-219.	1.6	143
236	Organization and Sequence of Human Cardiac Myosin Binding Protein C Gene (MYBPC3) and Identification of Mutations Predicted to Produce Truncated Proteins in Familial Hypertrophic Cardiomyopathy. <i>Circulation Research</i> , 1997, 80, 427-434.	4.5	240
237	Codon 102 of the Cardiac Troponin T Gene Is a Putative Hot Spot for Mutations in Familial Hypertrophic Cardiomyopathy. <i>Circulation</i> , 1996, 94, 3069-3073.	1.6	99
238	Cardiac myosin binding proteinâ€C gene splice acceptor site mutation is associated with familial hypertrophic cardiomyopathy. <i>Nature Genetics</i> , 1995, 11, 438-440.	21.4	417
239	The COX8 gene is not the disease gene of the CMH4 locus in familial hypertrophic cardiomyopathy.. <i>Journal of Medical Genetics</i> , 1995, 32, 670-671.	3.2	1
240	Expression of cytochrome c oxidase subunits encoded by mitochondrial or nuclear DNA in the muscle of patients with zidovudine myopathy. <i>Journal of the Neurological Sciences</i> , 1994, 125, 190-193.	0.6	6
241	Expression of human cytochrome c, oxidase subunits during fetal development. <i>FEBS Journal</i> , 1993, 217, 1099-1107.	0.2	85
242	Defect in the lipoyl-bearing protein X subunit of the pyruvate dehydrogenase complex in two patients with encephalomyopathy. <i>Journal of Pediatrics</i> , 1993, 123, 915-920.	1.8	37
243	El Pyruvate Dehydrogenase Deficiency in a Child with Motor Neuropathy. <i>Pediatric Research</i> , 1993, 33, 284-288.	2.3	40
244	Kearns-Sayre Syndrome with Sideroblastic Anemia: Molecular Investigations. <i>Neuropediatrics</i> , 1992, 23, 199-205.	0.6	28
245	Genetic biochemical and pathophysiological characterization of a familial mitochondrial encephalomyopathy (MERRF). <i>Journal of the Neurological Sciences</i> , 1991, 105, 217-224.	0.6	69
246	Zidovudine myopathy: A distinctive disorder associated with mitochondrial dysfunction. <i>Annals of Neurology</i> , 1991, 29, 606-614.	5.3	187
247	Muscle diseases with prominent muscle contractures. , 0, , 299-313.		1
248	Cardiomyopathy Caused by Mutations in Nuclear A-Type Lamin Gene. , 0, , .		1