

# Gisèle Bonne

## List of Publications by Year in descending order

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Version: 2024-02-01

248  
papers

15,520  
citations

18465

62  
h-index

19726

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, .	1.2	17
2	International retrospective natural history study of <i>LMNA</i>-related congenital muscular dystrophy. <i>Brain Communications</i> , 2021, 3, fcab075.	1.5	17
3	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. <i>Brain</i> , 2021, 144, 2427-2442.	3.7	7
4	The Treatabolome, an emerging concept. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 337-339.	1.1	6
5	Laminopathiesâ€™ Treatments Systematic Review: A Contribution Towards a â€™Treatabolomeâ€™. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 419-439.	1.1	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.	1.4	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.	1.4	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.	2.9	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.	0.9	9
10	Targeting the histone demethylase LSD1 prevents cardiomyopathy in a mouse model of laminopathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	26
11	Lamin-Related Congenital Muscular Dystrophy Alters Mechanical Signaling and Skeletal Muscle Growth. <i>International Journal of Molecular Sciences</i> , 2021, 22, 306.	1.8	15
12	Preclinical Advances of Therapies for Laminopathies. <i>Journal of Clinical Medicine</i> , 2021, 10, 4834.	1.0	4
13	The 2022 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2021, 31, 1313-1357.	0.3	38
14	Novel role of <i>Tieg1</i> in muscle metabolism and mitochondrial oxidative capacities. <i>Acta Physiologica</i> , 2020, 228, e13394.	1.8	14
15	Mutant lamins cause nuclear envelope rupture and DNA damage in skeletal muscle cells. <i>Nature Materials</i> , 2020, 19, 464-473.	13.3	148
16	LATE BREAKING NEWS E-POSTER PRESENTATION. <i>Neuromuscular Disorders</i> , 2020, 30, S171.	0.3	0
17	The 2021 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2020, 30, 1008-1048.	0.3	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.	1.2	21

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19	Protein Kinase C Alpha Cellular Distribution, Activity, and Proximity with Lamin A/C in Striated Muscle Laminopathies. <i>Cells</i> , 2020, 9, 2388.	1.8	6
20	Activation of sarcolipin expression and altered calcium cycling in LMNA cardiomyopathy. <i>Biochemistry and Biophysics Reports</i> , 2020, 22, 100767.	0.7	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.	1.8	29
22	Consequences of Lmna Exon 4 Mutations in Myoblast Function. <i>Cells</i> , 2020, 9, 1286.	1.8	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.	1.4	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.	0.3	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.	0.7	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.	0.3	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.	0.3	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.	0.3	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.	0.3	2
30	The 2020 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2019, 29, 980-1018.	0.3	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.	1.1	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.	1.8	16
33	FHL1 is a major host factor for chikungunya virus infection. <i>Nature</i> , 2019, 574, 259-263.	13.7	49
34	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in <i>BVES</i> . <i>Neurology: Genetics</i> , 2019, 5, e321.	0.9	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.	1.1	47

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37	Cardiometabolic assessment of lamin A/C gene mutation carriers: a phenotypeâ€“genotype correlation. <i>Diabetes and Metabolism</i> , 2019, 45, 382-389.	1.4	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.	1.4	7
39	The mammalian LINC complex component <i>SUN1</i> regulates muscle regeneration by modulating <i>drosha</i> activity. <i>ELife</i> , 2019, 8, .	2.8	12
40	<i>SMAD6</i> overexpression leads to accelerated myogenic differentiation of <i>LMNA</i> mutated cells. <i>Scientific Reports</i> , 2018, 8, 5618.	1.6	6
41	Novel mutations in <i>DNAJB6</i> cause <i>LGMD</i> 1D and distal myopathy in French families. <i>European Journal of Neurology</i> , 2018, 25, 790-794.	1.7	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.	2.3	29
43	Elevated <i>TGF Î²2</i> serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. <i>Nucleus</i> , 2018, 9, 337-349.	0.6	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.	1.2	13
45	Lamin and the heart. <i>Heart</i> , 2018, 104, 468-479.	1.2	113
46	<i>LGMD</i> AUTOSOMAL RESESSIVE AND DOMINANT. <i>Neuromuscular Disorders</i> , 2018, 28, S59-S60.	0.3	1
47	CONGENITAL MYOPATHIES: NEMALINE AND TITINOPATHIES. <i>Neuromuscular Disorders</i> , 2018, 28, S104-S105.	0.3	0
48	The Pathogenesis and Therapies of Striated Muscle Laminopathies. <i>Frontiers in Physiology</i> , 2018, 9, 1533.	1.3	30
49	The 2019 version of the gene table of neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2018, 28, 1031-1063.	0.3	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.	0.9	22
51	Amelioration of desmin network defects by Î±B-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.	0.9	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.	0.3	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.	1.4	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.	1.4	40

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55	Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for follow-up. <i>Muscle and Nerve</i> , 2018, 58, 812-817.	1.0	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.	1.6	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.	1.4	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.	1.4	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.	1.0	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.	0.4	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.	0.3	0
62	Welcome to the World Muscle Society Congress in Saint Malo. <i>Neuromuscular Disorders</i> , 2017, 27, S52-S53.	0.3	0
63	Anti-HMGR Antibody-Related Necrotizing Autoimmune Myopathy Mimicking Muscular Dystrophy. <i>Neuropediatrics</i> , 2017, 48, 473-476.	0.3	15
64	The 2018 version of the gene table of monogenic neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2017, 27, 1152-1183.	0.3	41
65	Collagen VI deficiency: The heart of the matter. <i>Neuromuscular Disorders</i> , 2017, 27, S106.	0.3	0
66	A novel INPP5K mutation in a sibship from the Reunion Island. <i>Neuromuscular Disorders</i> , 2017, 27, S110-S111.	0.3	0
67	TIEG1 is a novel regulator of muscle mitochondrial biogenesis. <i>Neuromuscular Disorders</i> , 2017, 27, S117.	0.3	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.	0.3	0
69	Corticosteroid treatment in early-onset lamin A/C related muscular dystrophies. <i>Neuromuscular Disorders</i> , 2017, 27, S138.	0.3	1
70	Abnormal trafficking of connexin 43: A key element in the development of LMNA cardiomyopathy. <i>Neuromuscular Disorders</i> , 2017, 27, S139.	0.3	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.	0.3	2
72	POPDC1 gene mutation screening in patients with LGMD and heart disturbances: a mutation load effect?. <i>Neuromuscular Disorders</i> , 2017, 27, S140.	0.3	0

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73	Integrated analysis of the large-scale sequencing project "Myocapture" to identify novel genes for myopathies. <i>Neuromuscular Disorders</i> , 2017, 27, S195.	0.3	1
74	Morphological spectrum of RYR1 recessive myopathies: Clinical and genetic correlation.. <i>Neuromuscular Disorders</i> , 2017, 27, S239.	0.3	0
75	Lamins and nesprin-1 mediate inside-out mechanical coupling in muscle cell precursors through FHOD1. <i>Scientific Reports</i> , 2017, 7, 1253.	1.6	35
76	Évaluation cardiométaboliques des laminopathies selon le type de mutation (R482 ou NON-R482). <i>Diabetes and Metabolism</i> , 2017, 43, A89-A90.	1.4	0
77	Clinical heterogeneity and phenotype/genotype findings in 5 families with <i>CYG1</i> deficiency. <i>Neurology: Genetics</i> , 2017, 3, e208.	0.9	12
78	Clinical features and therapeutic strategies for managing the striated muscle laminopathies. <i>Expert Opinion on Orphan Drugs</i> , 2016, 4, 631-638.	0.5	1
79	Laminopathies: Why make it simple when it can be complex?. <i>Neuromuscular Disorders</i> , 2016, 26, S150-S151.	0.3	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.	1.1	17
81	The 2017 version of the gene table of monogenic neuromuscular disorders (nuclear genome). <i>Neuromuscular Disorders</i> , 2016, 26, 895-929.	0.3	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.	0.3	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.	1.4	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.	2.6	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.	0.6	19
86	Laminopathies disrupt epigenomic developmental programs and cell fate. <i>Science Translational Medicine</i> , 2016, 8, 335ra58.	5.8	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.	0.3	0
88	OPALE: A patient registry for laminopathies and emerinopathies in France. <i>Neuromuscular Disorders</i> , 2016, 26, S138.	0.3	1
89	EGR2 mutation enhances phenotype spectrum of DejerineÉSottas syndrome. <i>Journal of Neurology</i> , 2016, 263, 1456-1458.	1.8	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.	1.0	31

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91	Mechanosensing Defects in Nuclear Envelope Related Disorders. <i>Biophysical Journal</i> , 2016, 110, 96a.	0.2	0
92	La Myologie ã lâ€™AcadÃ©mie Nationale de MÃ©decine. <i>Les Cahiers De Myologie</i> , 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. <i>Neuromuscular Disorders</i> , 2015, 25, S299.	0.3	0
94	An overview of new translational, clinical and therapeutic perspectives in laminopathies and other nuclear envelope-related diseases.. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 11.	1.2	0
95	A common French-Italian laminopathy registry â€“ update & future prospects. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, .	1.2	0
96	Laminin Î±2 Deficiency-Related Muscular Dystrophy Mimicking Emery-Dreifuss and Collagen VI related Diseases. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 229-240.	1.1	30
97	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. <i>European Journal of Human Genetics</i> , 2015, 23, 1051-1061.	1.4	24
98	Myofibrillar myopathies: State of the art, present and future challenges. <i>Revue Neurologique</i> , 2015, 171, 715-729.	0.6	38
99	Gene therapy via trans-splicing for LMNA-related congenital muscular dystrophy (L-CMD). <i>Neuromuscular Disorders</i> , 2015, 25, S280.	0.3	0
100	A new titinopathy. <i>Neurology</i> , 2015, 85, 2126-2135.	1.5	44
101	Mutations in <i>ASAH1</i> may cause spinal muscular atrophy. <i>Neuromuscular Disorders</i> , 2015, 25, S225.	0.3	1
102	Exome sequencing identifies novel truncating <i>TTN</i> mutations with Emeryâ€™Dreifuss like muscular dystrophy and secondary calpain3 deficiency without cardiac abnormality. <i>Neuromuscular Disorders</i> , 2015, 25, S245.	0.3	0
103	Highly variable skeletal muscle histo-immunocytochemical and ultrastructural features in titin-related myopathies. <i>Neuromuscular Disorders</i> , 2015, 25, S287-S288.	0.3	0
104	Detection of <i>TRIM32</i> deletions in LGMD patients analyzed by a combined strategy of CGH array and massively parallel sequencing. <i>European Journal of Human Genetics</i> , 2015, 23, 929-934.	1.4	21
105	Nuclear envelope and striated muscle diseases. <i>Current Opinion in Cell Biology</i> , 2015, 32, 1-6.	2.6	14
106	Severe dystonia, cerebellar atrophy, and cardiomyopathy likely caused by a missense mutation in <i>TOR1AIP1</i> . <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 174.	1.2	43
107	<i>FHL1</i> mutations that cause clinically distinct human myopathies form protein aggregates and impair myoblast differentiation. <i>Journal of Cell Science</i> , 2014, 127, 2269-81.	1.2	19
108	P330Overexpression of the muscle specific chaperone Melusin delays heart failure and mortality in a mouse model of Emery Dreyfus cardiomyopathy. <i>Cardiovascular Research</i> , 2014, 103, S60.1-S60.	1.8	0

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109	Dystrophin quantification. <i>Neurology</i> , 2014, 83, 2062-2069.	1.5	73
110	Striated muscle laminopathies. <i>Seminars in Cell and Developmental Biology</i> , 2014, 29, 107-115.	2.3	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.	2.3	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.	3.7	108
113	G.P.142. <i>Neuromuscular Disorders</i> , 2014, 24, 843-844.	0.3	2
114	G.P.145. <i>Neuromuscular Disorders</i> , 2014, 24, 844-845.	0.3	0
115	G.P.92. <i>Neuromuscular Disorders</i> , 2014, 24, 822.	0.3	1
116	G.P.281. <i>Neuromuscular Disorders</i> , 2014, 24, 901-902.	0.3	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.	1.2	105
118	G.P.150. <i>Neuromuscular Disorders</i> , 2014, 24, 846.	0.3	0
119	Nuclear envelope proteins in health and diseases. <i>Seminars in Cell and Developmental Biology</i> , 2014, 29, 93-94.	2.3	5
120	P.5.16 Predominant right ventricular involvement in patients with laminopathies. <i>Neuromuscular Disorders</i> , 2013, 23, 768-769.	0.3	0
121	Emeryâ€Dreifuss muscular dystrophy, laminopathies, and other nuclear envelopathies. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 113, 1367-1376.	1.0	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.	2.9	21
123	Muscle dystrophy-causing K32 lamin A/C mutant does not impair functions of nucleoplasmic LAP2 - lamin A/C complexes in mice. <i>Journal of Cell Science</i> , 2013, 126, 1753-62.	1.2	31
124	P.1.15 Clinical heterogeneity of myopathy related to partial merosin deficiency. <i>Neuromuscular Disorders</i> , 2013, 23, 746.	0.3	1
125	Mutations in Lamin A/C Gene Causes Mechanosensing Defects in Human Myoblasts. <i>Biophysical Journal</i> , 2013, 104, 374a.	0.2	0
126	Role of dynamin 2 in the disassembly of focal adhesions. <i>Journal of Molecular Medicine</i> , 2013, 91, 803-809.	1.7	7



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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.	1.8	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.	3.7	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.	1.4	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.	0.9	36
131	“State-of-the-heart”™ of cardiac laminopathies. <i>Current Opinion in Cardiology</i> , 2013, 28, 297-304.	0.8	60
132	Hypoplasia of the Aorta in a Patient Diagnosed with LMNA Gene Mutation. <i>Congenital Heart Disease</i> , 2013, 8, E127-E129.	0.0	3
133	Distinctive Serum miRNA Profile in Mouse Models of Striated Muscular Pathologies. <i>PLoS ONE</i> , 2013, 8, e55281.	1.1	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.	1.4	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.	1.8	86
136	Evidence for FHL1 as a novel disease gene for isolated hypertrophic cardiomyopathy. <i>Human Molecular Genetics</i> , 2012, 21, 3237-3254.	1.4	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.	0.3	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.	0.3	0
139	D.P.25 Mutation in lamin A/C gene causes mechanotransduction defects in human myoblasts. <i>Neuromuscular Disorders</i> , 2012, 22, 825.	0.3	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.	0.3	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.	0.3	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.	0.3	0
143	3D Culture of Human Muscle Cells Modulates Cell-Matrix Adhesions and Actin Cytoskeleton Organization. <i>Biophysical Journal</i> , 2012, 102, 417a.	0.2	0
144	Diseases of the Nucleoskeleton. , 2012, , 1003-1012.		0

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145	Complex Interactions between Human Myoblasts and the Surrounding 3D Fibrin-Based Matrix. PLoS ONE, 2012, 7, e36173.	1.1	83
146	Lamin A/C Mutants Disturb Sumo1 Localization and Sumoylation in Vitro and in Vivo. PLoS ONE, 2012, 7, e45918.	1.1	26
147	A Centronuclear Myopathy â€“ Dynamin 2 Mutation Impairs Autophagy in Mice. Traffic, 2012, 13, 869-879.	1.3	52
148	Guiding the molecular diagnosis of hypertrophic cardiomyopathy. Journal of Thoracic and Cardiovascular Surgery, 2012, 143, 1234.	0.4	0
149	What Should the Cardiologist know about Lamin Disease?. Arrhythmia and Electrophysiology Review, 2012, 1, 22.	1.3	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.	1.1	32
151	MicroRNA expression profiling in patients with lamin A/Câ€“associated muscular dystrophy. FASEB Journal, 2011, 25, 3966-3978.	0.2	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.	0.8	4
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