## GisÃ"le Bonne

List of Publications by Year in descending order

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248 papers 15,520 citations

62 h-index 117 g-index

269 all docs 269 docs citations

269 times ranked 12416 citing authors

#	Article	IF	Citations
1	Mutations in the gene encoding lamin A/C cause autosomal dominant Emery-Dreifuss muscular dystrophy. Nature Genetics, 1999, 21, 285-288.	9.4	1,245
2	"Laminopathies― A wide spectrum of human diseases. Experimental Cell Research, 2007, 313, 2121-2133.	1.2	560
3	Identification of mutations in the gene encoding lamins A/C in autosomal dominant limb girdle muscular dystrophy with atrioventricular conduction disturbances (LGMD1B). Human Molecular Genetics, 2000, 9, 1453-1459.	1.4	530
4	Mandibuloacral Dysplasia Is Caused by a Mutation in LMNA-Encoding Lamin A/C. American Journal of Human Genetics, 2002, 71, 426-431.	2.6	509
5	Nuclear Lamins: Laminopathies and Their Role in Premature Ageing. Physiological Reviews, 2006, 86, 967-1008.	13.1	494
6	Cardiac myosin binding protein–C gene splice acceptor site mutation is associated with familial hypertrophic cardiomyopathy. Nature Genetics, 1995, 11, 438-440.	9.4	417
7	Meta-analysis of clinical characteristics of 299 carriers of LMNA gene mutations: do lamin A/C mutations portend a high risk of sudden death?. Journal of Molecular Medicine, 2005, 83, 79-83.	1.7	388
8	Different Mutations in the LMNA Gene Cause Autosomal Dominant and Autosomal Recessive Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2000, 66, 1407-1412.	2.6	384
9	Familial Hypertrophic Cardiomyopathy. Circulation Research, 1998, 83, 580-593.	2.0	354
10	Primary Prevention of Sudden Death in Patients with Lamin A/C Gene Mutations. New England Journal of Medicine, 2006, 354, 209-210.	13.9	323
11	Mouse model carrying H222P- Lmna mutation develops muscular dystrophy and dilated cardiomyopathy similar to human striated muscle laminopathies. Human Molecular Genetics, 2005, 14, 155-169.	1.4	303
12	Activation of MAPK pathways links LMNA mutations to cardiomyopathy in Emery-Dreifuss muscular dystrophy. Journal of Clinical Investigation, 2007, 117, 1282-1293.	3.9	256
13	De novo <i>LMNA</i> mutations cause a new form of congenital muscular dystrophy. Annals of Neurology, 2008, 64, 177-186.	2.8	255
14	The Ig-like Structure of the C-Terminal Domain of Lamin A/C, Mutated in Muscular Dystrophies, Cardiomyopathy, and Partial Lipodystrophy. Structure, 2002, 10, 811-823.	1.6	252
15	Clinical Features and Prognostic Implications of Familial Hypertrophic Cardiomyopathy Related to the Cardiac Myosin-Binding Protein C Gene. Circulation, 1998, 97, 2230-2236.	1.6	241
16	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. Circulation Research, 1997, 80, 427-434.	2.0	240
17	High Incidence of Sudden Death with Conduction System and Myocardial Disease Due to Lamins A and C Gene Mutation. PACE - Pacing and Clinical Electrophysiology, 2000, 23, 1661-1666.	0.5	234
18	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2009, 85, 338-353.	2.6	208

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19	Autophagic degradation of nuclear components in mammalian cells. Autophagy, 2009, 5, 795-804.	4.3	189
20	Zidovudine myopathy: A distinctive disorder associated with mitochondrial dysfunction. Annals of Neurology, 1991, 29, 606-614.	2.8	187
21	Nuclear envelope alterations in fibroblasts from LGMD1B patients carrying nonsense Y259X heterozygous or homozygous mutation in lamin A/C gene. Experimental Cell Research, 2003, 291, 352-362.	1.2	169
22	Nuclear envelope alterations in fibroblasts from patients with muscular dystrophy, cardiomyopathy, and partial lipodystrophy carrying lamin A/C gene mutations. Muscle and Nerve, 2004, 30, 444-450.	1.0	167
23	Clinical Relevance of Atrial Fibrillation/Flutter, Stroke, Pacemaker Implant, and Heart Failure in Emery-Dreifuss Muscular Dystrophy. Stroke, 2003, 34, 901-908.	1.0	158
24	Expanding the phenotype of LMNA mutations in dilated cardiomyopathy and functional consequences of these mutations. Journal of Medical Genetics, 2003, 40, 560-567.	1.5	156
25	Mitogen-Activated Protein Kinase Inhibitors Improve Heart Function and Prevent Fibrosis in Cardiomyopathy Caused by Mutation in Lamin A/C Gene. Circulation, 2011, 123, 53-61.	1.6	154
26	Inhibition of extracellular signal-regulated kinase signaling to prevent cardiomyopathy caused by mutation in the gene encoding A-type lamins. Human Molecular Genetics, 2008, 18, 241-247.	1.4	149
27	Mutant lamins cause nuclear envelope rupture and DNA damage in skeletal muscle cells. Nature Materials, 2020, 19, 464-473.	13.3	148
28	Diagnostic Value of Electrocardiography and Echocardiography for Familial Hypertrophic Cardiomyopathy in a Genotyped Adult Population. Circulation, 1997, 96, 214-219.	1.6	143
29	Primary laminopathy fibroblasts display altered genome organization and apoptosis. Aging Cell, 2007, 6, 139-153.	3.0	140
30	Development and Validation of a New Risk Prediction Score for Life-Threatening Ventricular Tachyarrhythmias in Laminopathies. Circulation, 2019, 140, 293-302.	1.6	131
31	Identification of two novel mutations in the ventricular regulatory myosin light chain gene (MYL2) associated with familial and classical forms of hypertrophic cardiomyopathy. Journal of Molecular Medicine, 1998, 76, 208-214.	1.7	130
32	Activation of MAPK in hearts of EMD null mice: similarities between mouse models of X-linked and autosomal dominant Emery–Dreifuss muscular dystrophy. Human Molecular Genetics, 2007, 16, 1884-1895.	1.4	123
33	Emery-Dreifuss muscular dystrophy. European Journal of Human Genetics, 2002, 10, 157-161.	1.4	121
34	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. Neurology, 2007, 69, 1285-1292.	1.5	120
35	The 13C hyperpolarized pyruvate generated by ParaHydrogen detects the response of the heart to altered metabolism in real time. Scientific Reports, 2018, 8, 8366.	1.6	119
36	Skeletal muscle pathology in autosomal dominant Emery-Dreifuss muscular dystrophy with lamin A/C mutations. Neuropathology and Applied Neurobiology, 2001, 27, 281-290.	1.8	117

#	Article	IF	Citations
37	Extreme Variability of Phenotype in Patients With an Identical Missense Mutation in the Lamin A/C Gene. Archives of Neurology, 2004, 61, 690.	4.9	114
38	Disease severity in dominant Emery Dreifuss is increased by mutations in both emerin and desmin proteins. Brain, 2006, 129, 1260-1268.	3.7	114
39	Lamin and the heart. Heart, 2018, 104, 468-479.	1.2	113
40	Forelimb Treatment in a Large Cohort of Dystrophic Dogs Supports Delivery of a Recombinant AAV for Exon Skipping in Duchenne Patients. Molecular Therapy, 2014, 22, 1923-1935.	3.7	108
41	Clinical and genetic heterogeneity in laminopathies. Biochemical Society Transactions, 2011, 39, 1687-1692.	1.6	107
42	Evidence for FHL1 as a novel disease gene for isolated hypertrophic cardiomyopathy. Human Molecular Genetics, 2012, 21, 3237-3254.	1.4	106
43	Lamin A/C–mediated neuromuscular junction defects in Emery-Dreifuss muscular dystrophy. Journal of Cell Biology, 2009, 184, 31-44.	2.3	105
44	Cellular micro-environments reveal defective mechanosensing responses and elevated YAP signaling in LMNA-mutated muscle precursors. Journal of Cell Science, 2014, 127, 2873-84.	1.2	105
45	Selective Muscle Involvement on Magnetic Resonance Imaging in Autosomal Dominant Emery-Dreifuss Muscular Dystrophy. Neuropediatrics, 2002, 33, 10-14.	0.3	101
46	Codon 102 of the Cardiac Troponin T Gene Is a Putative Hot Spot for Mutations in Familial Hypertrophic Cardiomyopathy. Circulation, 1996, 94, 3069-3073.	1.6	99
47	Distinctive Serum miRNA Profile in Mouse Models of Striated Muscular Pathologies. PLoS ONE, 2013, 8, e55281.	1.1	97
48	Emery–Dreifuss muscular dystrophy, laminopathies, and other nuclear envelopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1367-1376.	1.0	96
49	Genotype–phenotype correlations in familial hypertrophic cardiomyopathy A comparison between mutations in the cardiac protein-C and the beta-myosin heavy chain genes. European Heart Journal, 1998, 19, 139-145.	1.0	92
50	Laminopathies disrupt epigenomic developmental programs and cell fate. Science Translational Medicine, 2016, 8, 335ra58.	5.8	91
51	Treatment with selumetinib preserves cardiac function and improves survival in cardiomyopathy caused by mutation in the lamin A/C gene. Cardiovascular Research, 2012, 93, 311-319.	1.8	86
52	Expression of human cytochrome c, oxidase subunits during fetal development. FEBS Journal, 1993, 217, 1099-1107.	0.2	85
53	ColVI myopathies: where do we stand, where do we go?. Skeletal Muscle, 2011, 1, 30.	1.9	84
54	Complex Interactions between Human Myoblasts and the Surrounding 3D Fibrin-Based Matrix. PLoS ONE, 2012, 7, e36173.	1.1	83

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55	108th ENMC International Workshop, 3rd Workshop of the MYO-CLUSTER project: EUROMEN, 7th International Emery-Dreifuss Muscular Dystrophy (EDMD) Workshop, 13–15 September 2002, Naarden, The Netherlands. Neuromuscular Disorders, 2003, 13, 508-515.	0.3	78
56	DelK32-lamin A/C has abnormal location and induces incomplete tissue maturation and severe metabolic defects leading to premature death. Human Molecular Genetics, 2012, 21, 1037-1048.	1.4	77
57	A new mutation of the lamin A/C gene leading to autosomal dominant axonal neuropathy, muscular dystrophy, cardiac disease, and leuconychia. Journal of Medical Genetics, 2004, 41, 29e-29.	1.5	76
58	ERK1/2 directly acts on CTGF/CCN2 expression to mediate myocardial fibrosis in cardiomyopathy caused by mutations in the lamin A/C gene. Human Molecular Genetics, 2016, 25, 2220-2233.	1.4	76
59	Dystrophin quantification. Neurology, 2014, 83, 2062-2069.	1.5	73
60	Heterozygous LmnadelK32 mice develop dilated cardiomyopathy through a combined pathomechanism of haploinsufficiency and peptide toxicity. Human Molecular Genetics, 2013, 22, 3152-3164.	1.4	72
61	Genetic biochemical and pathophysiological characterization of a familial mitochondrial encephalomyopathy (MERRF). Journal of the Neurological Sciences, 1991, 105, 217-224.	0.3	69
62	Extreme variability of skeletal and cardiac muscle involvement in patients with mutations in exon $11$ of the lamin A/C gene. Muscle and Nerve, 2005, $31$ , $602-609$ .	1.0	68
63	Impaired Presynaptic High-Affinity Choline Transporter Causes a Congenital Myasthenic Syndrome with Episodic Apnea. American Journal of Human Genetics, 2016, 99, 753-761.	2.6	68
64	Contractures and hypertrophic cardiomyopathy in a novel FHL1 mutation. Annals of Neurology, 2010, 67, 136-140.	2.8	64
65	Two patients with â€~Dropped head syndrome' due to mutations in LMNA or SEPN1 genes. Neuromuscular Disorders, 2005, 15, 521-524.	0.3	61
66	â€~State-of-the-heart' of cardiac laminopathies. Current Opinion in Cardiology, 2013, 28, 297-304.	0.8	60
67	Actin scaffolding by clathrin heavy chain is required for skeletal muscle sarcomere organization. Journal of Cell Biology, 2014, 205, 377-393.	2.3	60
68	Decreased WNT/ $\hat{l}^2$ -catenin signalling contributes to the pathogenesis of dilated cardiomyopathy caused by mutations in the lamin a/C gene. Human Molecular Genetics, 2017, 26, ddw389.	1.4	58
69	The 2020 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2019, 29, 980-1018.	0.3	57
70	Structural analysis of four and half LIM protein-2 in dilated cardiomyopathy. Biochemical and Biophysical Research Communications, 2007, 357, 162-167.	1.0	55
71	Frequent low penetrance mutations in the Lamin A/C gene, causing Emery Dreifuss muscular dystrophy. Neuromuscular Disorders, 2002, 12, 958-963.	0.3	52
72	Lamin A N-terminal phosphorylation is associated with myoblast activation: impairment in Emery-Dreifuss muscular dystrophy. Journal of Medical Genetics, 2005, 42, 214-220.	1.5	52

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73	Pharmacological inhibition of c-Jun N-terminal kinase signaling prevents cardiomyopathy caused by mutation in LMNA gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 632-638.	1.8	52
74	A Centronuclear Myopathy – Dynamin 2 Mutation Impairs Autophagy in Mice. Traffic, 2012, 13, 869-879.	1.3	52
75	Diagnostic value of electrocardiography and echocardiography for familial hypertrophic cardiomyopathy in genotyped children. European Heart Journal, 1998, 19, 1377-1382.	1.0	49
76	FHL1 is a major host factor for chikungunya virus infection. Nature, 2019, 574, 259-263.	13.7	49
77	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
78	Striated muscle laminopathies. Seminars in Cell and Developmental Biology, 2014, 29, 107-115.	2.3	48
79	Heart-hand syndrome of Slovenian type: a new kind of laminopathy. Journal of Medical Genetics, 2008, 45, 666-671.	1.5	47
80	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatabolome. Emerging Topics in Life Sciences, 2019, 3, 19-37.	1.1	47
81	The 2021 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2020, 30, 1008-1048.	0.3	45
82	Type B mandibuloacral dysplasia with congenital myopathy due to homozygous ZMPSTE24 missense mutation. European Journal of Human Genetics, 2011, 19, 647-654.	1.4	44
83	A new titinopathy. Neurology, 2015, 85, 2126-2135.	1.5	44
84	Severe dystonia, cerebellar atrophy, and cardiomyopathy likely caused by a missense mutation in TOR1AIP1. Orphanet Journal of Rare Diseases, 2014, 9, 174.	1.2	43
85	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. Neuromuscular Disorders, $2001, 11, 542-546$ .	0.3	42
86	MicroRNA expression profiling in patients with lamin A/Câ€associated muscular dystrophy. FASEB Journal, 2011, 25, 3966-3978.	0.2	42
87	Cofilin-1 phosphorylation catalyzed by ERK1/2 alters cardiac actin dynamics in dilated cardiomyopathy caused by lamin A/C gene mutation. Human Molecular Genetics, 2018, 27, 3060-3078.	1.4	42
88	Nuclear accumulation of androgen receptor in gender difference of dilated cardiomyopathy due to lamin A/C mutations. Cardiovascular Research, 2013, 99, 382-394.	1.8	41
89	The 2018 version of the gene table of monogenic neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2017, 27, 1152-1183.	0.3	41
90	El Pyruvate Dehydrogenase Deficiency in a Child with Motor Neuropathy. Pediatric Research, 1993, 33, 284-288.	1.1	40

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91	Rescue of biosynthesis of nicotinamide adenine dinucleotide protects the heart in cardiomyopathy caused by lamin A/C gene mutation. Human Molecular Genetics, 2018, 27, 3870-3880.	1.4	40
92	Familial hypertrophic cardiomyopathy. Cardiac ultrasonic abnormalities in genetically affected subjects without echocardiographic evidence of left ventricular hypertrophy. European Heart Journal, 1998, 19, 490-499.	1.0	39
93	Apical left ventricular aneurysm without atrio-ventricular block due to a lamin A/C gene mutation. European Journal of Heart Failure, 2003, 5, 821-825.	2.9	39
94	Deletion of the LMNA initiator codon leading to a neurogenic variant of autosomal dominant Emery–Dreifuss muscular dystrophy. Neuromuscular Disorders, 2005, 15, 40-44.	0.3	38
95	Myofibrillar myopathies: State of the art, present and future challenges. Revue Neurologique, 2015, 171, 715-729.	0.6	38
96	The 2022 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2021, 31, 1313-1357.	0.3	38
97	Defect in the lipoyl-bearing protein X subunit of the pyruvate dehydrogenase complex in two patients with encephalomyelopathy. Journal of Pediatrics, 1993, 123, 915-920.	0.9	37
98	LMNA mutations in atypical Werner's syndrome. Lancet, The, 2003, 362, 1585-1586.	6.3	37
99	Skeletal Muscle Biopsy Analysis in Reducing Body Myopathy and Other FHL1-Related Disorders. Journal of Neuropathology and Experimental Neurology, 2013, 72, 833-845.	0.9	36
100	Lamins and nesprin-1 mediate inside-out mechanical coupling in muscle cell precursors through FHOD1. Scientific Reports, 2017, 7, 1253.	1.6	35
101	Microtubule cytoskeleton regulates Connexin 43 localization and cardiac conduction in cardiomyopathy caused by mutation in A-type lamins gene. Human Molecular Genetics, 2019, 28, 4043-4052.	1.4	35
102	The 2019 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2018, 28, 1031-1063.	0.3	33
103	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
104	Modifier locus of the skeletal muscle involvement in Emery–Dreifuss muscular dystrophy. Human Genetics, 2011, 129, 149-159.	1.8	32
105	Proteasome-mediated degradation of integral inner nuclear membrane protein emerin in fibroblasts lacking A-type lamins. Biochemical and Biophysical Research Communications, 2006, 351, 1011-1017.	1.0	31
106	Differentiating Emery-Dreifuss muscular dystrophy and collagen VI-related myopathies using a specific CT scanner pattern. Neuromuscular Disorders, 2010, 20, 517-523.	0.3	31
107	Muscle dystrophy-causing ΔK32 lamin A/C mutant does not impair functions of nucleoplasmic LAP2α - lamin A/C complexes in mice. Journal of Cell Science, 2013, 126, 1753-62.	1.2	31
108	Pediatric laminopathies: Whole-body magnetic resonance imaging fingerprint and comparison with <i>Sepn1</i> myopathy. Muscle and Nerve, 2016, 54, 192-202.	1.0	31

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109	Amelioration of desmin network defects by $\hat{l}\pm B$ -crystallin overexpression confers cardioprotection in a mouse model of dilated cardiomyopathy caused by LMNA gene mutation. Journal of Molecular and Cellular Cardiology, 2018, 125, 73-86.	0.9	31
110	First description of germline mosaicism in familial hypertrophic cardiomyopathy. Journal of Medical Genetics, 2000, 37, 132-134.	1.5	30
111	Laminin α2 Deficiency-Related Muscular Dystrophy Mimicking Emery-Dreifuss andÂCollagen VI related Diseases. Journal of Neuromuscular Diseases, 2015, 2, 229-240.	1.1	30
112	Mutation in lamin A/C sensitizes the myocardium to exercise-induced mechanical stress but has no effect on skeletal muscles in mouse. Neuromuscular Disorders, 2016, 26, 490-499.	0.3	30
113	The Pathogenesis and Therapies of Striated Muscle Laminopathies. Frontiers in Physiology, 2018, 9, 1533.	1.3	30
114	Gene Therapy via Trans-Splicing for LMNA-Related Congenital Muscular Dystrophy. Molecular Therapy - Nucleic Acids, 2018, 10, 376-386.	2.3	29
115	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. Cells, 2020, 9, 844.	1.8	29
116	Kearns-Sayre Syndrome with Sideroblastic Anemia: Molecular Investigations. Neuropediatrics, 1992, 23, 199-205.	0.3	28
117	Primary Myocardial Dysfunction in Autosomal Dominant EDMD. A Tissue Doppler and Cardiovascular Magnetic Resonance Study. Journal of Cardiovascular Magnetic Resonance, 2006, 8, 723-730.	1.6	28
118	The Nuclear muscular dystrophies. Seminars in Pediatric Neurology, 2002, 9, 100-107.	1.0	26
119	Lamin A/C Mutants Disturb Sumo1 Localization and Sumoylation in Vitro and in Vivo. PLoS ONE, 2012, 7, e45918.	1.1	26
120	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in <i>BVES</i> . Neurology: Genetics, 2019, 5, e321.	0.9	26
121	Targeting the histone demethylase LSD1 prevents cardiomyopathy in a mouse model of laminopathy. Journal of Clinical Investigation, 2021, 131, .	3.9	26
122	Elevated TGF $\hat{1}^22$ serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. Nucleus, 2018, 9, 337-349.	0.6	25
123	Germinal mosaicism for LMNA mimics autosomal recessive congenital muscular dystrophy. Neuromuscular Disorders, 2009, 19, 26-28.	0.3	24
124	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. European Journal of Human Genetics, 2015, 23, 1051-1061.	1.4	24
125	What Should the Cardiologist know about Lamin Disease?. Arrhythmia and Electrophysiology Review, 2012, 1, 22.	1.3	24
126	Novel mutations in <i>DNAJB6</i> cause <scp>LGMD</scp> 1D and distal myopathy in French families. European Journal of Neurology, 2018, 25, 790-794.	1.7	23

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127	Loss of Sarcomeric Scaffolding as a Common Baseline Histopathologic Lesion in Titin-Related Myopathies. Journal of Neuropathology and Experimental Neurology, 2018, 77, 1101-1114.	0.9	22
128	Cardiometabolic assessment of lamin A/C gene mutation carriers: a phenotype–genotype correlation. Diabetes and Metabolism, 2019, 45, 382-389.	1.4	22
129	A novel genetic variant in the transcription factor Islet-1 exerts gain of function on myocyte enhancer factor 2C promoter activity. European Journal of Heart Failure, 2013, 15, 267-276.	2.9	21
130	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.	1.4	21
131	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet Journal of Rare Diseases, 2020, 15, 206.	1.2	21
132	Improvement of Left Ventricular Dysfunction and of Survival Prognosis of Dilated Cardiomyopathy by Administration of Calcium Sensitizer SCH00013 in a Mouse Model. Journal of the American College of Cardiology, 2010, 55, 1503-1505.	1.2	20
133	Cardiac manifestations of congenital LMNA-related muscular dystrophy in children: three case reports and recommendations for care. Cardiology in the Young, 2017, 27, 1076-1082.	0.4	20
134	FHL1 mutations that cause clinically distinct human myopathies form protein aggregates and impair myoblast differentiation. Journal of Cell Science, 2014, 127, 2269-81.	1.2	19
135	Cardiac arrhythmia and late-onset muscle weakness caused by a myofibrillar myopathy with unusual histopathological features due to a novel missense mutation in FLNC. Revue Neurologique, 2016, 172, 594-606.	0.6	19
136	Autosomal dominant Emery–Dreifuss muscular dystrophy: a new family with late diagnosis. Neuromuscular Disorders, 2002, 12, 19-25.	0.3	18
137	Genetics of Laminopathies. Novartis Foundation Symposium, 2008, , 81-97.	1.2	18
138	FHL1B Interacts with Lamin A/C andÂEmerin at the Nuclear Lamina andÂisÂMisregulated in Emery-Dreifuss Muscular Dystrophy. Journal of Neuromuscular Diseases, 2016, 3, 497-510.	1.1	17
139	A lamin A/C variant causing striated muscle disease provides insights into filament organization. Journal of Cell Science, 2021, 134, .	1.2	17
140	International retrospective natural history study of <i>LMNA</i> related congenital muscular dystrophy. Brain Communications, 2021, 3, fcab075.	1.5	17
141	A Muscle Hybrid Promoter as a Novel Tool for Gene Therapy. Molecular Therapy - Methods and Clinical Development, 2019, 15, 157-169.	1.8	16
142	Myofiber Degeneration in Autosomal Dominant Emery–Dreifuss Muscular Dystrophy (AD-EDMD) (LGMD1B). Brain Pathology, 2006, 16, 266-272.	2.1	15
143	Anti-HMGCR Antibody–Related Necrotizing Autoimmune Myopathy Mimicking Muscular Dystrophy. Neuropediatrics, 2017, 48, 473-476.	0.3	15
144	Effect of genetic background on the cardiac phenotype in a mouse model of Emery-Dreifuss muscular dystrophy. Biochemistry and Biophysics Reports, 2019, 19, 100664.	0.7	15

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145	Lamin-Related Congenital Muscular Dystrophy Alters Mechanical Signaling and Skeletal Muscle Growth. International Journal of Molecular Sciences, 2021, 22, 306.	1.8	15
146	COOH-terminal truncated human cardiac MyBP-C alters myosin filament organization. Comptes Rendus De L'AcadÃ@mie Des Sciences SÃ@rie 3, Sciences De La Vie, 2001, 324, 251-260.	0.8	14
147	Nuclear envelope and striated muscle diseases. Current Opinion in Cell Biology, 2015, 32, 1-6.	2.6	14
148	Novel role of $\langle i \rangle$ Tieg1 $\langle i \rangle$ in muscle metabolism and mitochondrial oxidative capacities. Acta Physiologica, 2020, 228, e13394.	1.8	14
149	MoBiDiC Prioritization Algorithm, a Free, Accessible, and Efficient Pipeline for Single-Nucleotide Variant Annotation and Prioritization for Next-Generation Sequencing Routine Molecular Diagnosis. Journal of Molecular Diagnostics, 2018, 20, 465-473.	1.2	13
150	Laminopathies' Treatments Systematic Review: A Contribution Towards a â€~Treatabolome'. Journal of Neuromuscular Diseases, 2021, 8, 419-439.	1.1	13
151	Clinical heterogeneity and phenotype/genotype findings in 5 families with <i>GYG1</i> deficiency. Neurology: Genetics, 2017, 3, e208.	0.9	12
152	The mammalian LINC complex component SUN1 regulates muscle regeneration by modulating drosha activity. ELife, 2019, 8, .	2.8	12
153	Cardiac Myosin-Binding Protein C and Hypertrophic Cardiomyopathy. Trends in Cardiovascular Medicine, 1998, 8, 151-157.	2.3	11
154	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. Neuromuscular Disorders, 2007, 17, 655-660.	0.3	11
155	Activation of sarcolipin expression and altered calcium cycling in LMNA cardiomyopathy. Biochemistry and Biophysics Reports, 2020, 22, 100767.	0.7	11
156	The 2017 version of the gene table of monogenic neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2016, 26, 895-929.	0.3	10
157	N-acetyl cysteine alleviates oxidative stress and protects mice from dilated cardiomyopathy caused by mutations in nuclear A-type lamins gene. Human Molecular Genetics, 2018, 27, 3353-3360.	1.4	10
158	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	1.4	10
159	Peripheral nerve lesions associated with a dominant missense mutation, E33D, of the lamin A/C gene. Neuromuscular Disorders, 2005, 15, 618-621.	0.3	9
160	Myoblasts and Embryonic Stem Cells Differentially Engraft in a Mouse Model of Genetic Dilated Cardiomyopathy. Molecular Therapy, 2013, 21, 1064-1075.	3.7	9
161	Deficiency of emerin contributes differently to the pathogenesis of skeletal and cardiac muscles in LmnaH222P/H222P mutant mice. PLoS ONE, 2019, 14, e0221512.	1.1	9
162	The non-muscle ADF/cofilin-1 controls sarcomeric actin filament integrity and force production in striated muscle laminopathies. Cell Reports, 2021, 36, 109601.	2.9	9

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163	High-Throughput Digital Image Analysis Reveals Distinct Patterns of Dystrophin Expression in Dystrophinopathy Patients. Journal of Neuropathology and Experimental Neurology, 2021, 80, 955-965.	0.9	9
164	Upregulation of PPARÎ $^2$  Î $^\prime$ Is Associated with Structural and Functional Changes in the Type I Diabetes Rat Diaphragm. PLoS ONE, 2010, 5, e11494.	1.1	9
165	Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for followâ€up. Muscle and Nerve, 2018, 58, 812-817.	1.0	8
166	Genetics of laminopathies. Novartis Foundation Symposium, 2005, 264, 81-90; discussion 90-97, 227-30.	1.2	8
167	Cardioembolic stroke prompting diagnosis of <i>LMNA</i> â€associated Emery–Dreifuss muscular dystrophy. Muscle and Nerve, 2011, 44, 587-589.	1.0	7
168	Blood glutathione decrease in subjects carrying lamin A/C gene mutations is an early marker of cardiac involvement. Neuromuscular Disorders, 2012, 22, 252-257.	0.3	7
169	Role of dynamin 2 in the disassembly of focal adhesions. Journal of Molecular Medicine, 2013, 91, 803-809.	1.7	7
170	A new case of SMA phenotype without epilepsy due to biallelic variants in ASAH1. European Journal of Human Genetics, 2019, 27, 337-339.	1.4	7
171	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	3.7	7
172	Expression of cytochrome c oxidase subunits encoded by mitochondrial or nuclear DNA in the muscle of patients with zidovudine myopathy. Journal of the Neurological Sciences, 1994, 125, 190-193.	0.3	6
173	Genetic Characterization of a French Cohort of GNEâ€mutation negative inclusion body myopathy patients with exome sequencing. Muscle and Nerve, 2017, 56, 993-997.	1.0	6
174	SMAD6 overexpression leads to accelerated myogenic differentiation of LMNA mutated cells. Scientific Reports, 2018, 8, 5618.	1.6	6
175	Protein Kinase C Alpha Cellular Distribution, Activity, and Proximity with Lamin A/C in Striated Muscle Laminopathies. Cells, 2020, 9, 2388.	1.8	6
176	The Treatabolome, an emerging concept. Journal of Neuromuscular Diseases, 2021, 8, 337-339.	1.1	6
177	Consequences of Lmna Exon 4 Mutations in Myoblast Function. Cells, 2020, 9, 1286.	1.8	6
178	Nuclear envelope proteins in health and diseases. Seminars in Cell and Developmental Biology, 2014, 29, 93-94.	2.3	5
179	EGR2 mutation enhances phenotype spectrum of Dejerine–Sottas syndrome. Journal of Neurology, 2016, 263, 1456-1458.	1.8	5
180	P2.18 A novel missense FLNC mutation causes arrhythmia and late onset myofibrillar myopathy with particular histopathology features. Neuromuscular Disorders, 2010, 20, 623-624.	0.3	4

#	Article	IF	CITATIONS
181	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
182	Laminopathies: One Gene, Two Proteins, Five Diseases…., 2002, , 153-172.		4
183	Preclinical Advances of Therapies for Laminopathies. Journal of Clinical Medicine, 2021, 10, 4834.	1.0	4
184	Hypoplasia of the Aorta in a Patient Diagnosed with LMNAGene Mutation. Congenital Heart Disease, 2013, 8, E127-E129.	0.0	3
185	G.P.18 Muscle pathology and dysfunction in a novel mouse model of COLVI-myopathy. Neuromuscular Disorders, 2012, 22, 827-828.	0.3	2
186	G.P.142. Neuromuscular Disorders, 2014, 24, 843-844.	0.3	2
187	Laminopathies: Why make it simple when it can be complex?. Neuromuscular Disorders, 2016, 26, S150-S151.	0.3	2
188	Novel recessive splice site mutation in POPDC1 (BVES) is associated with first-degree atrioventricular block and muscular dystrophy. Neuromuscular Disorders, 2017, 27, S139-S140.	0.3	2
189	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. Neuromuscular Disorders, 2019, 29, 644-650.	0.3	2
190	P.256Steroid treatment may change natural history in young children with LMNA mutations and dropped head syndrome. Neuromuscular Disorders, 2019, 29, S141.	0.3	2
191	The COX8 gene is not the disease gene of the CMH4 locus in familial hypertrophic cardiomyopathy Journal of Medical Genetics, 1995, 32, 670-671.	1.5	1
192	Muscle diseases with prominent muscle contractures. , 0, , 299-313.		1
193	P1.15 DNA micro-arrays for revisiting molecular pathology in neuromuscular disorders. Neuromuscular Disorders, 2010, 20, 604.	0.3	1
194	P.1.15 Clinical heterogeneity of myopathy related to partial merosin deficiency. Neuromuscular Disorders, 2013, 23, 746.	0.3	1
195	G.P.92. Neuromuscular Disorders, 2014, 24, 822.	0.3	1
196	Mutations in ASAH1 may cause spinal muscular atrophy. Neuromuscular Disorders, 2015, 25, S225.	0.3	1
197	Clinical features and therapeutic strategies for managing the striated muscle laminopathies. Expert Opinion on Orphan Drugs, 2016, 4, 631-638.	0.5	1
198	OPALE: A patient registry for laminopathies and emerinopathies in France. Neuromuscular Disorders, 2016, 26, S138.	0.3	1

#	Article	lF	Citations
199	TIEG1 is a novel regulator of muscle mitochondrial biogenesis. Neuromuscular Disorders, 2017, 27, S117.	0.3	1
200	Corticosteroid treatment in early-onset lamin A/C related muscular dystrophies. Neuromuscular Disorders, 2017, 27, S138.	0.3	1
201	Integrated analysis of the large-scale sequencing project "Myocapture―to identify novel genes for myopathies. Neuromuscular Disorders, 2017, 27, S195.	0.3	1
202	Cardiomyopathy Caused by Mutations in Nuclear A-Type Lamin Gene. , 0, , .		1
203	LGMD AUTOSOMAL RESSESSIVE AND DOMINANT. Neuromuscular Disorders, 2018, 28, S59-S60.	0.3	1
204	MHC Class II Deficiency. , 2009, , 1306-1308.		0
205	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. Neuromuscular Disorders, 2010, 20, 598.	0.3	0
206	P5.62 Garches muscle Whole-Body MRI protocol: Pattern recognition in early onset neuromuscular disorders. Neuromuscular Disorders, 2011, 21, 742-743.	0.3	0
207	P2.10 A new form of congenital muscular dystrophy with subsarcolemmal inclusions arising from disintegrated myonuclei. Neuromuscular Disorders, 2011, 21, 663-664.	0.3	0
208	P2.41 From Emery-Dreifuss muscular dystrophy to striated muscle laminopathies. A 12years retrospective. Neuromuscular Disorders, 2011, 21, 672-673.	0.3	0
209	P2.43 Revisiting X-linked Emery-Dreifuss muscular dystrophy. New insights into an old story. Neuromuscular Disorders, 2011, 21, 673.	0.3	0
210	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.	1.1	0
211	D.P.2 Next generation sequencing after selected DNA capture as a tool for molecular diagnosis of neuromuscular disorders. Neuromuscular Disorders, 2012, 22, 807-808.	0.3	0
212	D.P.25 Mutation in lamin A/C gene causes mechanotransduction defects in human myoblasts. Neuromuscular Disorders, 2012, 22, 825.	0.3	0
213	G.P.120 FHL1-related Reducing Body Myopathy and Emery–Dreifuss muscular dystrophy: A comparative histoenzymological, immunohistochemical and ultrastructural study. Neuromuscular Disorders, 2012, 22, 902.	0.3	0
214	G.P.122 Heterozygous LmnadelK32 mutant mice showed alterations of the ubiquitin–proteasome system and developed dilated cardiomyopathy. Neuromuscular Disorders, 2012, 22, 903.	0.3	0
215	3D Culture of Human Muscle Cells Modulates Cell-Matrix Adhesions and Actin Cytoskeleton Organization. Biophysical Journal, 2012, 102, 417a.	0.2	0
216	Diseases of the Nucleoskeleton. , 2012, , 1003-1012.		O

#	Article	IF	CITATIONS
217	Guiding the molecular diagnosis of hypertrophic cardiomyopathy. Journal of Thoracic and Cardiovascular Surgery, 2012, 143, 1234.	0.4	О
218	P.5.16 Predominant right ventricular involvement in patients with laminopathies. Neuromuscular Disorders, 2013, 23, 768-769.	0.3	0
219	Mutations in Lamin A/C Gene Causes Mechanosensing Defects in Human Myoblasts. Biophysical Journal, 2013, 104, 374a.	0.2	O
220	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.	1.8	0
221	G.P.145. Neuromuscular Disorders, 2014, 24, 844-845.	0.3	O
222	G.P.281. Neuromuscular Disorders, 2014, 24, 901-902.	0.3	0
223	G.P.150. Neuromuscular Disorders, 2014, 24, 846.	0.3	O
224	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.	0.3	0
225	An overview of new translational, clinical and therapeutic perspectives in laminopathies and other nuclear envelope-related diseases Orphanet Journal of Rare Diseases, 2015, 10, I1.	1.2	O
226	A common French-Italian laminopathy registry $\hat{a} \in \text{``update \&'}; future prospects. Orphanet Journal of Rare Diseases, 2015, 10, .$	1.2	0
227	Gene therapy via trans-splicing for LMNA-related congenital muscular dystrophy (L-CMD). Neuromuscular Disorders, 2015, 25, S280.	0.3	O
228	Exome sequencing identifies novel truncating TTN mutations with Emery–Dreifuss like muscular dystrophy and secondary calpain3 deficiency without cardiac abnormality. Neuromuscular Disorders, 2015, 25, S245.	0.3	0
229	Highly variable skeletal muscle histo-immunocytochemical and ultrastructural features in titin-related myopathies. Neuromuscular Disorders, 2015, 25, S287-S288.	0.3	O
230	Interest of whole-body muscle MRI for the diagnosis of Pompe disease in rigid spine syndrome and differential diagnosis. Neuromuscular Disorders, 2016, 26, S109.	0.3	0
231	Mechanosensing Defects in Nuclear Envelope Related Disorders. Biophysical Journal, 2016, 110, 96a.	0.2	О
232	Genetic characterization of a French cohort of GNE -mutation negative inclusion body myopathy patients using exome sequencing. Neuromuscular Disorders, 2017, 27, S149.	0.3	0
233	Welcome to the World Muscle Society Congress in Saint Malo. Neuromuscular Disorders, 2017, 27, S52-S53.	0.3	0
234	Collagen VI deficiency: The heart of the matter. Neuromuscular Disorders, 2017, 27, S106.	0.3	0

#	Article	IF	CITATIONS
235	A novel INPP5K mutation in a sibship from the Reunion Island. Neuromuscular Disorders, 2017, 27, S110-S111.	0.3	0
236	First results from the international LMNA -related congenital and childhood onset muscular dystrophy retrospective natural history study. Neuromuscular Disorders, 2017, 27, S137-S138.	0.3	0
237	Abnormal trafficking of connexin 43: A key element in the development of LMNA cardiomyopathy. Neuromuscular Disorders, 2017, 27, S139.	0.3	0
238	POPDC1 gene mutation screening in patients with LGMD and heart disturbances: a mutation load effect?. Neuromuscular Disorders, 2017, 27, S140.	0.3	0
239	Morphological spectrum of RYR1 recessive myopathies: Clinical and genetic correlation Neuromuscular Disorders, 2017, 27, S239.	0.3	0
240	$\tilde{A}$ %-valuation cardiom $\tilde{A}$ ©taboliques des laminopathies selon le type de mutation (R482 ou NON-R482). Diabetes and Metabolism, 2017, 43, A89-A90.	1.4	0
241	CONGENITAL MYOPATHIES: NEMALINE AND TITINOPATHIES. Neuromuscular Disorders, 2018, 28, S104-S105.	0.3	0
242	Dystrophin quantification in Duchenne and Becker muscular dystrophy: correlation between dystrophin protein and clinical phenotype. Neuromuscular Disorders, 2018, 28, S7-S8.	0.3	0
243	P.143Correlation between dystrophin espression and clinical phenotype using high-throughput digital immunoanalysis in Duchenne and Becker muscular dystrophy patients. Neuromuscular Disorders, 2019, 29, S90.	0.3	0
244	P.245Morphological, ultrastructural and western blot analysis in adult and child with PLEC1-related myopathy. Neuromuscular Disorders, 2019, 29, S138.	0.3	0
245	P.252LGMD, exercise intolerance, ptosis, ophthalmoplegia and dermatologic features: the phenotypic pleiotropy of plectinopathies in 8 French families. Neuromuscular Disorders, 2019, 29, S140.	0.3	0
246	LATE BREAKING NEWS E-POSTER PRESENTATION. Neuromuscular Disorders, 2020, 30, S171.	0.3	0
247	Cardiomyopathy, Dilated (Familial). , 2004, , 216-220.		0
248	La Myologie à l'Académie Nationale de Médecine. Les Cahiers De Myologie, 2016, , 3-4.	0.0	0