GisÃ¨le Bonne

List of Publications by Year
in descending order

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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	Mutations in the gene encoding lamin A/C cause autosomal dominant Emery-Dreifuss muscular dystrophy. Nature Genetics, 1999, 21, 285-288.	21.4	1,245
2	â€œLaminopathiesâ€: A wide spectrum of human diseases. Experimental Cell Research, 2007, 313, 2121-2133.	2.6	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.	2.9	530
4	Mandibuloacral Dysplasia Is Caused by a Mutation in LMNA-Encoding Lamin A/C. American Journal of Human Genetics, 2002, 71, 426-431.	6.2	509
5	Nuclear Lamins: Laminopathies and Their Role in Premature Ageing. Physiological Reviews, 2006, 86, 967-1008.	28.8	494
6	Cardiac myosin binding proteinâ€“C gene splice acceptor site mutation is associated with familial hypertrophic cardiomyopathy. Nature Genetics, 1995, 11, 438-440.	21.4	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.	3.9	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.	6.2	384
9	Familial Hypertrophic Cardiomyopathy. Circulation Research, 1998, 83, 580-593.	4.5	354
10	Primary Prevention of Sudden Death in Patients with Lamin A/C Gene Mutations. New England Journal of Medicine, 2006, 354, 209-210.	27.0	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.	2.9	303
12	Activation of MAPK pathways links LMNA mutations to cardiomyopathy in Emery-Dreifuss muscular dystrophy. Journal of Clinical Investigation, 2007, 117, 1282-1293.	8.2	256
13	De novo LMNA mutations cause a new form of congenital muscular dystrophy. Annals of Neurology, 2008, 64, 177-186.	5.3	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.	3.3	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.	4.5	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.	1.2	234
18	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2009, 85, 338-353.	6.2	208

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19	Autophagic degradation of nuclear components in mammalian cells. Autophagy, 2009, 5, 795-804.	9.1	189
20	Zidovudine myopathy: A distinctive disorder associated with mitochondrial dysfunction. Annals of Neurology, 1991, 29, 606-614.	5.3	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.	2.6	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.	2.2	167
23	Clinical Relevance of Atrial Fibrillation/Flutter, Stroke, Pacemaker Implant, and Heart Failure in Emery-Dreifuss Muscular Dystrophy. Stroke, 2003, 34, 901-908.	2.0	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.	3.2	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.	2.9	149
27	Mutant lamins cause nuclear envelope rupture and DNA damage in skeletal muscle cells. Nature Materials, 2020, 19, 464-473.	27.5	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.	6.7	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.	3.9	130
32	Activation of MAPK in hearts of EMD null mice: similarities between mouse models of X-linked and autosomal dominant Emeryâ€“Dreifuss muscular dystrophy. Human Molecular Genetics, 2007, 16, 1884-1895.	2.9	123
33	Emery-Dreifuss muscular dystrophy. European Journal of Human Genetics, 2002, 10, 157-161.	2.8	121
34	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. Neurology, 2007, 69, 1285-1292.	1.1	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.	3.3	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.	3.2	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.	4.5	114
38	Disease severity in dominant Emery Dreifuss is increased by mutations in both emerin and desmin proteins. Brain, 2006, 129, 1260-1268.	7.6	114
39	Lamin and the heart. Heart, 2018, 104, 468-479.	2.9	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.	8.2	108
41	Clinical and genetic heterogeneity in laminopathies. Biochemical Society Transactions, 2011, 39, 1687-1692.	3.4	107
42	Evidence for FHL1 as a novel disease gene for isolated hypertrophic cardiomyopathy. Human Molecular Genetics, 2012, 21, 3237-3254.	2.9	106
43	Lamin A/Câ€“mediated neuromuscular junction defects in Emery-Dreifuss muscular dystrophy. Journal of Cell Biology, 2009, 184, 31-44.	5.2	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.	2.0	105
45	Selective Muscle Involvement on Magnetic Resonance Imaging in Autosomal Dominant Emery-Dreifuss Muscular Dystrophy. Neuropediatrics, 2002, 33, 10-14.	0.6	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.	2.5	97
48	Emeryâ€“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.	1.8	96
49	Genotypeâ€“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.	2.2	92
50	Laminopathies disrupt epigenomic developmental programs and cell fate. Science Translational Medicine, 2016, 8, 335ra58.	12.4	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.	3.8	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.	4.2	84
54	Complex Interactions between Human Myoblasts and the Surrounding 3D Fibrin-Based Matrix. PLoS ONE, 2012, 7, e36173.	2.5	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, 13â€“15 September 2002, Naarden, The Netherlands. Neuromuscular Disorders, 2003, 13, 508-515.	0.6	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.	2.9	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.	3.2	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.	2.9	76
59	Dystrophin quantification. Neurology, 2014, 83, 2062-2069.	1.1	73
60	Heterozygous LmnadelK32 mice develop dilated cardiomyopathy through a combined pathomechanism of haploinsufficiency and peptide toxicity. Human Molecular Genetics, 2013, 22, 3152-3164.	2.9	72
61	Genetic biochemical and pathophysiological characterization of a familial mitochondrial encephalomyopathy (MERRF). Journal of the Neurological Sciences, 1991, 105, 217-224.	0.6	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.	2.2	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.	6.2	68
64	Contractures and hypertrophic cardiomyopathy in a novel FHL1 mutation. Annals of Neurology, 2010, 67, 136-140.	5.3	64
65	Two patients with â€˜Dropped head syndromeâ€™ due to mutations in LMNA or SEPN1 genes. Neuromuscular Disorders, 2005, 15, 521-524.	0.6	61
66	â€˜State-of-the-heartâ€™ of cardiac laminopathies. Current Opinion in Cardiology, 2013, 28, 297-304.	1.8	60
67	Actin scaffolding by clathrin heavy chain is required for skeletal muscle sarcomere organization. Journal of Cell Biology, 2014, 205, 377-393.	5.2	60
68	Decreased WNT/Î²-catenin signalling contributes to the pathogenesis of dilated cardiomyopathy caused by mutations in the lamin a/C gene. Human Molecular Genetics, 2017, 26, ddw389.	2.9	58
69	The 2020 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2019, 29, 980-1018.	0.6	57
70	Structural analysis of four and half LIM protein-2 in dilated cardiomyopathy. Biochemical and Biophysical Research Communications, 2007, 357, 162-167.	2.1	55
71	Frequent low penetrance mutations in the Lamin A/C gene, causing Emery Dreifuss muscular dystrophy. Neuromuscular Disorders, 2002, 12, 958-963.	0.6	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.	3.2	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.	3.8	52
74	A Centronuclear Myopathy â€“ Dynamin 2 Mutation Impairs Autophagy in Mice. Traffic, 2012, 13, 869-879.	2.7	52
75	Diagnostic value of electrocardiography and echocardiography for familial hypertrophic cardiomyopathy in genotyped children. European Heart Journal, 1998, 19, 1377-1382.	2.2	49
76	FHL1 is a major host factor for chikungunya virus infection. Nature, 2019, 574, 259-263.	27.8	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.	2.8	49
78	Striated muscle laminopathies. Seminars in Cell and Developmental Biology, 2014, 29, 107-115.	5.0	48
79	Heart-hand syndrome of Slovenian type: a new kind of laminopathy. Journal of Medical Genetics, 2008, 45, 666-671.	3.2	47
80	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatabolome. Emerging Topics in Life Sciences, 2019, 3, 19-37.	2.6	47
81	The 2021 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2020, 30, 1008-1048.	0.6	45
82	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
83	A new titinopathy. Neurology, 2015, 85, 2126-2135.	1.1	44
84	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
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.	0.6	42
86	MicroRNA expression profiling in patients with lamin A/Câ€associated muscular dystrophy. FASEB Journal, 2011, 25, 3966-3978.	0.5	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.	2.9	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.	3.8	41
89	The 2018 version of the gene table of monogenic neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2017, 27, 1152-1183.	0.6	41
90	El Pyruvate Dehydrogenase Deficiency in a Child with Motor Neuropathy. Pediatric Research, 1993, 33, 284-288.	2.3	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.	2.9	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.	2.2	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.	7.1	39
94	Deletion of the LMNA initiator codon leading to a neurogenic variant of autosomal dominant Emeryâ€“Dreifuss muscular dystrophy. Neuromuscular Disorders, 2005, 15, 40-44.	0.6	38
95	Myofibrillar myopathies: State of the art, present and future challenges. Revue Neurologique, 2015, 171, 715-729.	1.5	38
96	The 2022 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2021, 31, 1313-1357.	0.6	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.	1.8	37
98	LMNA mutations in atypical Werner's syndrome. Lancet, The, 2003, 362, 1585-1586.	13.7	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.	1.7	36
100	Lamins and nesprin-1 mediate inside-out mechanical coupling in muscle cell precursors through FHOD1. Scientific Reports, 2017, 7, 1253.	3.3	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.	2.9	35
102	The 2019 version of the gene table of neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2018, 28, 1031-1063.	0.6	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.	2.1	32
104	Modifier locus of the skeletal muscle involvement in Emeryâ€“Dreifuss muscular dystrophy. Human Genetics, 2011, 129, 149-159.	3.8	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.	2.1	31
106	Differentiating Emery-Dreifuss muscular dystrophy and collagen VI-related myopathies using a specific CT scanner pattern. Neuromuscular Disorders, 2010, 20, 517-523.	0.6	31
107	Muscle dystrophy-causing Î”K32 lamin A/C mutant does not impair functions of nucleoplasmic LAP2Î± - lamin A/C complexes in mice. Journal of Cell Science, 2013, 126, 1753-62.	2.0	31
108	Pediatric laminopathies: Whole-body magnetic resonance imaging fingerprint and comparison with Sepn1 myopathy. Muscle and Nerve, 2016, 54, 192-202.	2.2	31

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109	Amelioration of desmin network defects by Î±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.	1.9	31
110	First description of germline mosaicism in familial hypertrophic cardiomyopathy. Journal of Medical Genetics, 2000, 37, 132-134.	3.2	30
111	Laminin Î±2 Deficiency-Related Muscular Dystrophy Mimicking Emery-Dreifuss andÂCollagen VI related Diseases. Journal of Neuromuscular Diseases, 2015, 2, 229-240.	2.6	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.	0.6	30
113	The Pathogenesis and Therapies of Striated Muscle Laminopathies. Frontiers in Physiology, 2018, 9, 1533.	2.8	30
114	Gene Therapy via Trans-Splicing for LMNA-Related Congenital Muscular Dystrophy. Molecular Therapy - Nucleic Acids, 2018, 10, 376-386.	5.1	29
115	Lamin A/C Assembly Defects in LMNA-Congenital Muscular Dystrophy Is Responsible for the Increased Severity of the Disease Compared with Emeryâ€“Dreifuss Muscular Dystrophy. Cells, 2020, 9, 844.	4.1	29
116	Kearns-Sayre Syndrome with Sideroblastic Anemia: Molecular Investigations. Neuropediatrics, 1992, 23, 199-205.	0.6	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.	3.3	28
118	The Nuclear muscular dystrophies. Seminars in Pediatric Neurology, 2002, 9, 100-107.	2.0	26
119	Lamin A/C Mutants Disturb Sumo1 Localization and Sumoylation in Vitro and in Vivo. PLoS ONE, 2012, 7, e45918.	2.5	26
120	Muscular dystrophy with arrhythmia caused by loss-of-function mutations in BVES. Neurology: Genetics, 2019, 5, e321.	1.9	26
121	Targeting the histone demethylase LSD1 prevents cardiomyopathy in a mouse model of laminopathy. Journal of Clinical Investigation, 2021, 131, .	8.2	26
122	Elevated TGF Î²2 serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. Nucleus, 2018, 9, 337-349.	2.2	25
123	Germinal mosaicism for LMNA mimics autosomal recessive congenital muscular dystrophy. Neuromuscular Disorders, 2009, 19, 26-28.	0.6	24
124	Truncated prelamin A expression in HGPS-like patients: a transcriptional study. European Journal of Human Genetics, 2015, 23, 1051-1061.	2.8	24
125	What Should the Cardiologist know about Lamin Disease?. Arrhythmia and Electrophysiology Review, 2012, 1, 22.	2.4	24
126	Novel mutations in DNAJB6 cause <scp>LGMD</scp>1D and distal myopathy in French families. European Journal of Neurology, 2018, 25, 790-794.	3.3	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.	1.7	22
128	Cardiometabolic assessment of lamin A/C gene mutation carriers: a phenotypeâ€“genotype correlation. Diabetes and Metabolism, 2019, 45, 382-389.	2.9	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.	7.1	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.	2.8	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.	2.7	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.	2.8	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.	0.8	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.	2.0	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.	1.5	19
136	Autosomal dominant Emeryâ€“Dreifuss muscular dystrophy: a new family with late diagnosis. Neuromuscular Disorders, 2002, 12, 19-25.	0.6	18
137	Genetics of Laminopathies. Novartis Foundation Symposium, 2008, , 81-97.	1.1	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.	2.6	17
139	A lamin A/C variant causing striated muscle disease provides insights into filament organization. Journal of Cell Science, 2021, 134, .	2.0	17
140	International retrospective natural history study of LMNA-related congenital muscular dystrophy. Brain Communications, 2021, 3, fcab075.	3.3	17
141	A Muscle Hybrid Promoter as a Novel Tool for Gene Therapy. Molecular Therapy - Methods and Clinical Development, 2019, 15, 157-169.	4.1	16
142	Myofiber Degeneration in Autosomal Dominant Emeryâ€“Dreifuss Muscular Dystrophy (AD-EDMD) (LGMD1B). Brain Pathology, 2006, 16, 266-272.	4.1	15
143	Anti-HMGCR Antibodyâ€“Related Necrotizing Autoimmune Myopathy Mimicking Muscular Dystrophy. Neuropediatrics, 2017, 48, 473-476.	0.6	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.	1.3	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.	4.1	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.	5.4	14
148	Novel role of Tieg1 in muscle metabolism and mitochondrial oxidative capacities. Acta Physiologica, 2020, 228, e13394.	3.8	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.	2.8	13
150	Laminopathiesâ€™ Treatments Systematic Review: A Contribution Towards a â€˜Treatabolomeâ€™. Journal of Neuromuscular Diseases, 2021, 8, 419-439.	2.6	13
151	Clinical heterogeneity and phenotype/genotype findings in 5 families with GYG1 deficiency. Neurology: Genetics, 2017, 3, e208.	1.9	12
152	The mammalian LINC complex component SUN1 regulates muscle regeneration by modulating drosha activity. ELife, 2019, 8, .	6.0	12
153	Cardiac Myosin-Binding Protein C and Hypertrophic Cardiomyopathy. Trends in Cardiovascular Medicine, 1998, 8, 151-157.	4.9	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 10â€“12 March 2006, Naarden, The Netherlands. Neuromuscular Disorders, 2007, 17, 655-660.	0.6	11
155	Activation of sarcolipin expression and altered calcium cycling in LMNA cardiomyopathy. Biochemistry and Biophysics Reports, 2020, 22, 100767.	1.3	11
156	The 2017 version of the gene table of monogenic neuromuscular disorders (nuclear genome). Neuromuscular Disorders, 2016, 26, 895-929.	0.6	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.	2.9	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.	2.8	10
159	Peripheral nerve lesions associated with a dominant missense mutation, E33D, of the lamin A/C gene. Neuromuscular Disorders, 2005, 15, 618-621.	0.6	9
160	Myoblasts and Embryonic Stem Cells Differentially Engraft in a Mouse Model of Genetic Dilated Cardiomyopathy. Molecular Therapy, 2013, 21, 1064-1075.	8.2	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.	2.5	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.	6.4	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.	1.7	9
164	Upregulation of PPARÎ²/Î´ Is Associated with Structural and Functional Changes in the Type I Diabetes Rat Diaphragm. PLoS ONE, 2010, 5, e11494.	2.5	9
165	Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for followâ€up. Muscle and Nerve, 2018, 58, 812-817.	2.2	8
166	Genetics of laminopathies. Novartis Foundation Symposium, 2005, 264, 81-90; discussion 90-97, 227-30.	1.1	8
167	Cardioembolic stroke prompting diagnosis of LMNAâ€associated Emeryâ€“Dreifuss muscular dystrophy. Muscle and Nerve, 2011, 44, 587-589.	2.2	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.	0.6	7
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