Patrizia Sabatelli

List of Publications by Year
in descending order

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157	6,472	70961	71532
papers	citations	41 h-index	76 g-index

163	163	163	8662
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mitochondrial dysfunction and apoptosis in myopathic mice with collagen VI deficiency. Nature Genetics, 2003, 35, 367-371.	9.4	469
2	Autophagy is defective in collagen VI muscular dystrophies, and its reactivation rescues myofiber degeneration. Nature Medicine, 2010, 16, 1313-1320.	15.2	457
3	POMT2 mutations cause Â-dystroglycan hypoglycosylation and Walker-Warburg syndrome. Journal of Medical Genetics, 2005, 42, 907-912.	1.5	374
4	Cyclosporin A corrects mitochondrial dysfunction and muscle apoptosis in patients with collagen VI myopathies. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 5225-5229.	3.3	195
5	Mitochondrial dysfunction in the pathogenesis of Ullrich congenital muscular dystrophy and prospective therapy with cyclosporins. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 991-996.	3.3	183
6	Morphology of the Peritoneal Membrane during Continuous Ambulatory Peritoneal Dialysis. Nephron, 1986, 44, 204-211.	0.9	172
7	EMILIN-1 Deficiency Induces Elastogenesis and Vascular Cell Defects. Molecular and Cellular Biology, 2004, 24, 638-650.	1.1	166
8	Mutations in COL6A3 Cause Severe and Mild Phenotypes of Ullrich Congenital Muscular Dystrophy. American Journal of Human Genetics, 2002, 70, 1446-1458.	2.6	165
9	Dystrophin levels as low as 30% are sufficient to avoid muscular dystrophy in the human. Neuromuscular Disorders, 2007, 17, 913-918.	0.3	145
10	Heart-specific localization of emerin: new insights into Emery-Dreifuss muscular dystrophy. Human Molecular Genetics, 1997, 6, 2257-2264.	1.4	138
11	Preclinical PK and PD Studies on 2â€²-O-Methyl-phosphorothioate RNA Antisense Oligonucleotides in the mdx Mouse Model. Molecular Therapy, 2010, 18, 1210-1217.	3.7	132
12	Muscle Interstitial Fibroblasts Are the Main Source of Collagen VI Synthesis in Skeletal Muscle: Implications for Congenital Muscular Dystrophy Types Ullrich and Bethlem. Journal of Neuropathology and Experimental Neurology, 2008, 67, 144-154.	0.9	119
13	The cyclophilin inhibitor Debio 025 normalizes mitochondrial function, muscle apoptosis and ultrastructural defects in Col6a1^{â^'/â^'} myopathic mice. British Journal of Pharmacology, 2009, 157, 1045-1052.	2.7	117
14	Genetic ablation of cyclophilin D rescues mitochondrial defects and prevents muscle apoptosis in collagen VI myopathic mice. Human Molecular Genetics, 2009, 18, 2024-2031.	1.4	116
15	Collagen VI deficiency affects the organization of fibronectin in the extracellular matrix of cultured fibroblasts. Matrix Biology, 2001, 20, 475-486.	1.5	115
16	Emilin1 Deficiency Causes Structural and Functional Defects of Lymphatic Vasculature. Molecular and Cellular Biology, 2008, 28, 4026-4039.	1.1	113
17	Alterations of nuclear envelope and chromatin organization in mandibuloacral dysplasia, a rare form of laminopathy. Physiological Genomics, 2005, 23, 150-158.	1.0	112
18	Autosomal recessive myosclerosis myopathy is a collagen VI disorder. Neurology, 2008, 71, 1245-1253.	1.5	112

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19	The 180-kDa isoform of topoisomerase II is localized in the nucleolus and belongs to the structural elements of the nucleolar remnant. Experimental Cell Research, 1992, 200, 460-466.	1.2	94
20	Nuclear changes in a case of X-linked Emery-Dreifuss muscular dystrophy. , 1999, 22, 864-869.		92
21	Nuclear alterations in autosomal-dominant Emery-Dreifuss muscular dystrophy. Muscle and Nerve, 2001, 24, 826-829.	1.0	80
22	Expression of the Collagen VI Î±5 and Î±6 Chains in Normal Human Skin and in Skin of Patients with Collagen VI-Related Myopathies. Journal of Investigative Dermatology, 2011, 131, 99-107.	0.3	78
23	Failure of lamin A/C to functionally assemble in R482L mutated familial partial lipodystrophy fibroblasts: altered intermolecular interaction with emerin and implications for gene transcription. Experimental Cell Research, 2003, 291, 122-134.	1.2	77
24	Expression of collagen VI Î±5 and Î±6 chains in human muscle and in Duchenne muscular dystrophy-related muscle fibrosis. Matrix Biology, 2012, 31, 187-196.	1.5	73
25	Dominant and recessive COL6A1 mutations in Ullrich scleroatonic muscular dystrophy. Annals of Neurology, 2005, 58, 400-410.	2.8	72
26	Cationic PMMA Nanoparticles Bind and Deliver Antisense Oligoribonucleotides Allowing Restoration of Dystrophin Expression in the mdx Mouse. Molecular Therapy, 2009, 17, 820-827.	3.7	70
27	NIM811, a cyclophilin inhibitor without immunosuppressive activity, is beneficial in collagen VI congenital muscular dystrophy models. Human Molecular Genetics, 2014, 23, 5353-5363.	1.4	64
28	Genetic characterization in symptomatic female DMD carriers: lack of relationship between X-inactivation, transcriptional DMD allele balancing and phenotype. BMC Medical Genetics, 2012, 13, 73.	2.1	63
29	EMILIN1/Î±9Î²1 Integrin Interaction Is Crucial in Lymphatic Valve Formation and Maintenance. Molecular and Cellular Biology, 2013, 33, 4381-4394.	1.1	62
30	Effects on Collagen VI mRNA Stability and Microfibrillar Assembly of Three COL6A2Mutations in Two Families with Ullrich Congenital Muscular Dystrophy. Journal of Biological Chemistry, 2002, 277, 43557-43564.	1.6	61
31	Familial isolated hyperCKaemia associated with a new mutation in the caveolin-3 (CAV-3) gene. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 73, 65-67.	0.9	55
32	Differential and restricted expression of novel collagen VI chains in mouse. Matrix Biology, 2011, 30, 248-257.	1.5	55
33	Properties of Ca2+ Transport in Mitochondria of Drosophila melanogaster. Journal of Biological Chemistry, 2011, 286, 41163-41170.	1.6	53
34	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
35	Cyclosporine A in Ullrich Congenital Muscular Dystrophy: Long-Term Results. Oxidative Medicine and Cellular Longevity, 2011, 2011, 1-10.	1.9	51
36	Alisporivir rescues defective mitochondrial respiration in Duchenne muscular dystrophy. Pharmacological Research, 2017, 125, 122-131.	3.1	51

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37	Dystrophin restoration in skeletal, heart and skin arrector pili smooth muscle of mdx mice by ZM2 NPâ€“AON complexes. Gene Therapy, 2010, 17, 432-438.	2.3	49
38	Autophagy activation in COL6 myopathic patients by a low-protein-diet pilot trial. Autophagy, 2016, 12, 2484-2495.	4.3	48
39	Immunocytochemical detection of emerin within the nuclear matrix. Neuromuscular Disorders, 1998, 8, 338-344.	0.3	44
40	Collagen VI is required for the structural and functional integrity of the neuromuscular junction. Acta Neuropathologica, 2018, 136, 483-499.	3.9	44
41	Oral exfoliative cytology for the non-invasive diagnosis in X-linked Emeryâ€“Dreifuss muscular dystrophy patients and carriers. Neuromuscular Disorders, 1998, 8, 67-71.	0.3	43
42	Monoamine oxidase inhibition prevents mitochondrial dysfunction and apoptosis in myoblasts from patients with collagen VI myopathies. Free Radical Biology and Medicine, 2014, 75, 40-47.	1.3	42
43	Identification and characterization of novel collagen VI non-canonical splicing mutations causing ullrich congenital muscular dystrophy. Human Mutation, 2009, 30, E662-E672.	1.1	40
44	A novel murine model for arrhythmogenic cardiomyopathy points to a pathogenic role of Wnt signalling and miRNA dysregulation. Cardiovascular Research, 2019, 115, 739-751.	1.8	40
45	Intranuclear localization of phospholipids by ultrastructural cytochemistry.. Journal of Histochemistry and Cytochemistry, 1992, 40, 1383-1392.	1.3	38
46	Localization of laminin Î±2 chain in normal human central nervous system: an immunofluorescence and ultrastructural study. Acta Neuropathologica, 1997, 94, 567-571.	3.9	38
47	Congenital muscular dystrophy associated with calf hypertrophy, microcephaly and severe mental retardation in three Italian families: evidence for a novel CMD syndrome. Neuromuscular Disorders, 2000, 10, 541-547.	0.3	36
48	Extracellular matrix and nuclear abnormalities in skeletal muscle of a patient with Walkerâ€“Warburg syndrome caused by POMT1 mutation. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2003, 1638, 57-62.	1.8	36
49	Dysferlin in a hyperCKaemic patient with caveolin 3 mutation and in C2C12 cells after p38 MAP kinase inhibition. Experimental and Molecular Medicine, 2003, 35, 538-544.	3.2	36
50	Exon skipping-mediated dystrophin reading frame restoration for small mutations. Human Mutation, 2009, 30, 1527-1534.	1.1	36
51	Collagen VI Status and Clinical Severity in Ullrich Congenital Muscular Dystrophy: Phenotype Analysis of 11 Families Linked to theCOL6Loci. Neuropediatrics, 2004, 35, 103-112.	0.3	35
52	Laminopathies: Involvement of structural nuclear proteins in the pathogenesis of an increasing number of human diseases. Journal of Cellular Physiology, 2005, 203, 319-327.	2.0	34
53	Laminopathies: A chromatin affair. Advances in Enzyme Regulation, 2006, 46, 33-49.	2.9	34
54	Interleukinâ€6 neutralization ameliorates symptoms in prematurely aged mice. Aging Cell, 2021, 20, e13285.	3.0	34

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55	Decreased expression of laminin Î²1 in chromosome 21-linked Bethlem myopathy. Neuromuscular Disorders, 1999, 9, 326-329.	0.3	33
56	Hyper-CK-emia as the sole manifestation of myotonic dystrophy type 2. Muscle and Nerve, 2005, 31, 764-767.	1.0	33
57	Vascular Smooth Muscle Emilin-1 Is a Regulator of Arteriolar Myogenic Response and Blood Pressure. Arteriosclerosis, Thrombosis, and Vascular Biology, 2012, 32, 2178-2184.	1.1	33
58	Collagen VIâ€“NG2 axis in human tendon fibroblasts under conditions mimicking injury response. Matrix Biology, 2016, 55, 90-105.	1.5	33
59	Novel COL6A1 splicing mutation in a family affected by mild Bethlem myopathy. Muscle and Nerve, 2002, 25, 513-519.	1.0	31
60	Emerin expression at the early stages of myogenic differentiation. Differentiation, 2000, 66, 208-217.	1.0	30
61	Melanocytesâ€”A novel tool to study mitochondrial dysfunction in Duchenne muscular dystrophy. Journal of Cellular Physiology, 2013, 228, 1323-1331.	2.0	30
62	166th ENMC International Workshop on Collagen type VI-related Myopathies, 22â€“24 May 2009, Naarden, The Netherlands. Neuromuscular Disorders, 2010, 20, 346-354.	0.3	29
63	Improving clinical trial design for Duchenne muscular dystrophy. BMC Neurology, 2015, 15, 153.	0.8	29
64	Altered expression of the MCSP/NG2 chondroitin sulfate proteoglycan in collagen VI deficiency. Molecular and Cellular Neurosciences, 2005, 30, 408-417.	1.0	27
65	Identification of a deep intronic mutation in the COL6A2 gene by a novel custom oligonucleotide CGH array designed to explore allelic and genetic heterogeneity in collagen VI-related myopathies. BMC Medical Genetics, 2010, 11, 44.	2.1	27
66	Defective collagen VI Î±6 chain expression in the skeletal muscle of patients with collagen VI-related myopathies. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1604-1612.	1.8	27
67	Hereditary motor and sensory neuropathy Lom type in an Italian Gypsy family. Neuromuscular Disorders, 1998, 8, 182-185.	0.3	25
68	Antisense-Induced Messenger Depletion Corrects a COL6A2 Dominant Mutation in Ullrich Myopathy. Human Gene Therapy, 2012, 23, 1313-1318.	1.4	25
69	Elevated TGF Î²2 serum levels in Emery-Dreifuss Muscular Dystrophy: Implications for myocyte and tenocyte differentiation and fibrogenic processes. Nucleus, 2018, 9, 337-349.	0.6	25
70	Ullrich myopathy phenotype with secondary ColVI defect identified by confocal imaging and electron microscopy analysis. Neuromuscular Disorders, 2007, 17, 587-596.	0.3	24
71	Functional domains of the nucleus: implications for Emeryâ€“Dreifuss muscular dystrophy. Neuromuscular Disorders, 2002, 12, 815-823.	0.3	22
72	Muscle Fiber Atrophy and Regeneration Coexist in Collagen VI-Deficient Human Muscle: Role of Calpain-3 and Nuclear Factor-ÎºB Signaling. Journal of Neuropathology and Experimental Neurology, 2012, 71, 894-906.	0.9	22

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73	Ultrastructural defects of collagen VI filaments in an Ullrich syndrome patient with loss of the Î±3(VI) N10-N7 domains. Journal of Cellular Physiology, 2006, 206, 160-166.	2.0	21
74	Collagen VI myopathies: From the animal model to the clinical trial. Advances in Enzyme Regulation, 2009, 49, 197-211.	2.9	21
75	Biodistribution and Molecular Studies on Orally Administered Nanoparticle-AON Complexes Encapsulated with Alginate Aiming at Inducing Dystrophin Rescue inmdxMice. BioMed Research International, 2013, 2013, 1-13.	0.9	21
76	Immunocytochemical evaluation of protein kinase C translocation to the inner nuclear matrix in 3T3 mouse fibroblasts after IGF-I treatment. Histochemistry, 1995, 103, 447-457.	1.9	19
77	Persistent Dystrophin Protein Restoration 90 Days after a Course of Intraperitoneally Administered Naked 2â€²OMePS AON and ZM2 NP-AON Complexes in mdx Mice. Journal of Biomedicine and Biotechnology, 2012, 2012, 1-8.	3.0	19
78	Integrin binding site within the gC1q domain orchestrates EMILIN-1-induced lymphangiogenesis. Matrix Biology, 2019, 81, 34-49.	1.5	19
79	Treatment with a triazole inhibitor of the mitochondrial permeability transition pore fully corrects the pathology of sapje zebrafish lacking dystrophin. Pharmacological Research, 2021, 165, 105421.	3.1	19
80	Interleukin-1Î± induces variations of the intranuclear amount of phosphatidylinositol 4,5-bisphosphate and phospholipase C Î²1 in human osteosarcoma Saos-2 cells. The Histochemical Journal, 1996, 28, 495-504.	0.6	18
81	AggresomeÃ¢â‚¬â€œAutophagy Involvement in a Sarcopenic Patient with Rigid Spine Syndrome and a p.C150R Mutation in FHL1 Gene. Frontiers in Aging Neuroscience, 2014, 6, 215.	1.7	18
82	Localization of dystrophin COOH-terminal domain by the fracture-label technique.. Journal of Cell Biology, 1992, 118, 1401-1409.	2.3	17
83	Combined use of malachite green fixation and PLA2-gold complex technique to localize phospholipids in areas of early calcification of rat epiphyseal cartilage and bone. Bone, 1996, 18, 559-565.	1.4	17
84	Hepatitis C virus infection and myositis: a polymerase chain reaction study. Acta Neuropathologica, 2000, 99, 271-276.	3.9	17
85	Critical evaluation of the use of cell cultures for inclusion in clinical trials of patients affected by collagen VI myopathies. Journal of Cellular Physiology, 2012, 227, 2927-2935.	2.0	16
86	Deep RNA profiling identified clock and molecular clock genes as pathophysiological signatures in collagen VI myopathy. Journal of Cell Science, 2016, 129, 1671-84.	1.2	16
87	The epg5 knockout zebrafish line: a model to study Vici syndrome. Autophagy, 2019, 15, 1438-1454.	4.3	16
88	The myotonic dystrophy type 2 (DM2) gene product zinc finger protein 9 (ZNF9) is associated with sarcomeres and normally localized in DM2 patients' muscles. Neuropathology and Applied Neurobiology, 2010, 36, 275-284.	1.8	15
89	Emeryâ€“dreifuss muscular dystrophy, nuclear cell signaling and chromatin remodeling. Advances in Enzyme Regulation, 2002, 42, 1-18.	2.9	13
90	Cultured muscle cells display defects of mitochondrial myopathy ameliorated by anti-oxidants. Brain, 2007, 130, 2715-2724.	3.7	13

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91	Effect of Mechanical Strain on the Collagen VI Pericellular Matrix in Anterior Cruciate Ligament Fibroblasts. Journal of Cellular Physiology, 2014, 229, 878-886.	2.0	13
92	A combined ultrastructural approach to the study of nuclear matrix thermal stabilization. Histochemistry, 1992, 98, 121-129.	1.9	12
93	Intracellular detection of laminin Î±2 chain in skin by electron microscopy immunocytochemistry: Comparison between normal and laminin Î±2 chain deficient subjects. Neuromuscular Disorders, 1997, 7, 91-98.	0.3	12
94	Emerin presence in platelets. Acta Neuropathologica, 2000, 100, 291-298.	3.9	12
95	Unusual Laminin Î±2 Processing in Myoblasts from a Patient with a Novel Variant of Congenital Muscular Dystrophy. Biochemical and Biophysical Research Communications, 2000, 277, 639-642.	1.0	12
96	Melanocytes from Patients Affected by Ullrich Congenital Muscular Dystrophy and Bethlem Myopathy have Dysfunctional Mitochondria That Can be Rescued with Cyclophilin Inhibitors. Frontiers in Aging Neuroscience, 2014, 6, 324.	1.7	12
97	Tendon Extracellular Matrix Remodeling and Defective Cell Polarization in the Presence of Collagen VI Mutations. Cells, 2020, 9, 409.	1.8	12
98	Ambra1 deficiency impairs mitophagy in skeletal muscle. Journal of Cachexia, Sarcopenia and Muscle, 2022, 13, 2211-2224.	2.9	12
99	Immunofluorescence study of a muscle biopsy from a 1-year-old patient with Walker-Warburg syndrome. Acta Neuropathologica, 1998, 96, 651-654.	3.9	10
100	Macrophages: A minimally invasive tool for monitoring collagen VI myopathies. Muscle and Nerve, 2011, 44, 80-84.	1.0	10
101	Tendon Extracellular Matrix Alterations in Ullrich Congenital Muscular Dystrophy. Frontiers in Aging Neuroscience, 2016, 8, 131.	1.7	10
102	Localization of the laminin Î±2 chain in normal human skeletal muscle and peripheral nerve: an ultrastructural immunolabeling study. Acta Neuropathologica, 1997, 93, 166-172.	3.9	9
103	Characterization of a rare case of Ullrich congenital muscular dystrophy due to truncating mutations within the COL6A1 gene C-Terminal domain: a case report. BMC Medical Genetics, 2013, 14, 59.	2.1	9
104	Congenital myopathy with hanging big toe due to homozygous myopalladin (MYPN) mutation. Skeletal Muscle, 2019, 9, 14.	1.9	9
105	Ultrastructural changes in muscle cells of patients with collagen VI-related myopathies. Muscles, Ligaments and Tendons Journal, 2013, 3, 281-6.	0.1	9
106	On the pathogenesis of collagen VI muscular dystrophies--Comment on article of Hicks et al.. Brain, 2009, 132, e121-e121.	3.7	8
107	Histochemical, ultrastructural and biochemical study of muscle mitochondria in Leber's hereditary optic atrophy. Journal of Inherited Metabolic Disease, 1988, 11, 193-197.	1.7	5
108	Multidrug-resistance (MDR) phenotype of human osteosarcoma cells evaluated by quantitative morphological and electron microscopy analyses. Biology of the Cell, 1995, 84, 195-204.	0.7	5

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109	At the nucleus of the problem: nuclear proteins and disease. Advances in Enzyme Regulation, 2003, 43, 411-443.	2.9	5
110	Influence of specimen preparation on the identification of phospholipids by the phospholipase A2-gold method in mineralizing cartilage and bone. Histochemistry and Cell Biology, 1996, 105, 283-296.	0.8	4
111	Homozygous Recessive Versican Missense Variation Is Associated With Early Teeth Loss in a Pakistani Family. Frontiers in Genetics, 2018, 9, 723.	1.1	4
112	epg5 knockout leads to the impairment of reproductive success and courtship behaviour in a zebrafish model of autophagy-related diseases. Biomedical Journal, 2022, 45, 377-386.	1.4	4
113	Urine-Derived Stem Cells Express 571 Neuromuscular Disorders Causing Genes, Making Them a Potential in vitro Model for Rare Genetic Diseases. Frontiers in Physiology, 2021, 12, 716471.	1.3	4
114	Transfer of HIV-1 to Human Tonsillar Stromal Cells Following Cocultivation with Infected Lymphocytes. AIDS Research and Human Retroviruses, 1994, 10, 675-682.	0.5	3
115	Detecting Collagen VI in Bethlem Myopathy. Journal of Biological Chemistry, 2015, 290, 8011.	1.6	3
116	Protein aggregates and autophagy involvement in a family with a mutation in Z-band alternatively spliced PDZ-motif protein. Neuromuscular Disorders, 2021, 31, 44-51.	0.3	3
117	Early Morphological Changes of the Rectus Femoris Muscle and Deep Fascia in Ullrich Congenital Muscular Dystrophy. International Journal of Environmental Research and Public Health, 2022, 19, 1252.	1.2	3
118	Cytoplasmic and nuclear localization sites of phosphatidylinositol 3-kinase in human osteosarcoma sensitive and multidrug-resistant Saos-2 cells. Histochemistry and Cell Biology, 1996, 106, 457-464.	0.8	2
119	EM.P.5.04 Genetic ablation of cyclophilin D rescues mitochondrial defects and prevents muscle apoptosis in collagen VI myopathic mice. Neuromuscular Disorders, 2009, 19, 631.	0.3	1
120	Cytoskeletal and extracellular matrix alterations in limb girdle muscular dystrophy 2I muscle fibers. Neurology India, 2012, 60, 510.	0.2	1
121	T.P.22 Nanoparticles as delivery systems for antisense oligoribonucleotides: Biodistribution studies and definition of the release kinetic in treated mdx mice. Neuromuscular Disorders, 2012, 22, 859.	0.3	1
122	Morphometric and biochemical study of muscle mitochondria in adult chronic progressive external ophthalmoplegia. Journal of Inherited Metabolic Disease, 1988, 11, 198-201.	1.7	0
123	P.P.7 01 Confocal imaging and electron microscopy analysis to identify secondary collagen VI defects. Neuromuscular Disorders, 2006, 16, 713-714.	0.3	0
124	P.P.7 02 Effect on collagen VI extra-cellular assembly of COL6A1 and COL6A2 C-terminal mutations in Ullrich congenital muscular dystrophy. Neuromuscular Disorders, 2006, 16, 714.	0.3	0
125	G.P.3.02 In vivo biodistribution of non-viral systems for oligoribonucleotides delivery. Neuromuscular Disorders, 2007, 17, 782.	0.3	0
126	G.P.5.08 PCNA staining pattern is altered in Emeryâ€“Dreifuss fibroblasts. Neuromuscular Disorders, 2007, 17, 799-800.	0.3	0

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127	G.P.12.02 How much dystrophin to avoid muscular dystrophy?. Neuromuscular Disorders, 2007, 17, 839-840.	0.3	0
128	C.P.2.05 Molecular analysis of COL6 genes in patients with Bethlem myopathy and Ullrich congenital muscular dystrophy. Neuromuscular Disorders, 2007, 17, 844-845.	0.3	0
129	C.P.2.07 Autosomal recessive myosclerosis myopathy is a collagen type VI disorder. Neuromuscular Disorders, 2007, 17, 845.	0.3	0
130	G.P.1.04 Design of a novel array-CGH to explore allelic and genetic heterogeneity in COLVI related myopathies. Neuromuscular Disorders, 2008, 18, 731-732.	0.3	0
131	T.P.2.06 Modulation of small mutations in dystrophin â€œskippableâ€•exons: In vitro studies to identify the optimal PS-AONs. Neuromuscular Disorders, 2008, 18, 757-758.	0.3	0
132	T.P.2.07 The systemic administration of a low dose of 2OMePS-AON combined with novel cationic polymethylmethacrylate nanoparticles induces the rescue of dystrophin expression in the mdx murine model. Neuromuscular Disorders, 2008, 18, 758.	0.3	0
133	M.P.1.01 Pilot trial with cyclosporin A in patients with collagen VI myopathies. Neuromuscular Disorders, 2009, 19, 546.	0.3	0
134	T.P.1.01 Pre-trial antisense screening of myogenic cells from boys with Duchenne muscular dystrophy and genomic and transcriptomic biomarkers discovery for treatment monitoring. Neuromuscular Disorders, 2009, 19, 576-577.	0.3	0
135	EM.I.2 Toward a mitochondrial therapy of collagen VI muscular dystrophies. Neuromuscular Disorders, 2009, 19, 598.	0.3	0
136	EM.P.4.03 Extensive sequencing of COL6A genes in a cohort of 65 patients with collagen type VI related myopathies. Focus on splicing mutations causing Ullrich congenital muscular dystrophy. Neuromuscular Disorders, 2009, 19, 607.	0.3	0
137	EM.P.4.07 Autosomal recessive Bethlem myopathy. Neuromuscular Disorders, 2009, 19, 608-609.	0.3	0
138	EM.P.4.09 Immunofluorescence and morphological alterations of capillary wall in skeletal muscle of two myosclerosis myopathy patients. Neuromuscular Disorders, 2009, 19, 609.	0.3	0
139	EM.P.5.01 Gene expression and proteome profiles in Col6a1â^'/â^' mice, a model of Ullrich congenital muscular dystrophy (UCMD). Neuromuscular Disorders, 2009, 19, 630.	0.3	0
140	EM.P.5.02 Role of mitochondria in the pathogenesis of muscular dystrophies. Neuromuscular Disorders, 2009, 19, 630.	0.3	0
141	EM.P.5.03 The cyclophilin inhibitor Debio 025 normalizes mitochondrial function, muscle apoptosis and ultrastructural defects in Col6a1<mml:math xmlns:mml="http://www.w3.org/1998/Math/MathML" altimg="si1.gif" overflow="scroll"><mml:mrow><mml:mtext>-</mml:mtext><mml:mo>/</mml:mo><mml:mtext>-</mml:mtext></mml:mrow></mml:math> myopathic mice. Neuromuscular Disorders, 2009, 19, 630.	0.3	0
142	EM.P.5.06 Collagen VI alpha5 chain exhibits a restricted localization at junctions in human skeletal muscle and skin. Neuromuscular Disorders, 2009, 19, 631.	0.3	0
143	EM.P.5.07 Abnormal elastin deposits and altered organization of elastic fibers in collagen VI- related disorders. Neuromuscular Disorders, 2009, 19, 631-632.	0.3	0
144	EM.P.5.08 Novel collagen VI alpha chains distribution in murine skeletal muscle: Possible implications for neuromuscular disorders. Neuromuscular Disorders, 2009, 19, 632.	0.3	0

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145	G.P.15.04 Collagen VI deficiency in skin fibroblasts from progeroid laminopathies. Neuromuscular Disorders, 2009, 19, 648.	0.3	0
146	P1.09 Clinical features in collagen VI deficiency. Neuromuscular Disorders, 2010, 20, 602.	0.3	0
147	P1.13 Pathological spectrum of collagen VI related myopathies: Does the pathology tell us something about the disease?. Neuromuscular Disorders, 2010, 20, 603.	0.3	0
148	P3.03 Nanoparticles are effective vehicles for systemic delivery of 2â€²OMePS antisense oligonucleotides in exon skipping-mediated dystrophin restoration. Neuromuscular Disorders, 2010, 20, 641.	0.3	0
149	P4.50 Mitochondrial therapy with Cyclosporine A in patients with Ullrich Congenital Muscular Dystrophy. Neuromuscular Disorders, 2011, 21, 719.	0.3	0
150	O.17 Autophagy thwarts collagen VI muscular dystrophies. Neuromuscular Disorders, 2011, 21, 749.	0.3	0
151	P1.28 Dystrophin mediates melanocytes attachment to dermal-epidermal junction in human skin. Neuromuscular Disorders, 2011, 21, 649-650.	0.3	0
152	P2.3 Monoamine oxidase inhibitors reduce mitochondrial ROS accumulation and dysfunction in patients with collagen VI myopathies. Neuromuscular Disorders, 2011, 21, 661.	0.3	0
153	P2.7 Collagen VI alpha5 and alpha6 chains expression in human muscle. Neuromuscular Disorders, 2011, 21, 662-663.	0.3	0
154	O.14 Biocompatible nanoparticles as slow-release delivery system of 2â€²OMePS AON administered both intraperitoneally and orally in the mdx mice: dystrophin rescue and nanoparticles biodistribution. Neuromuscular Disorders, 2011, 21, 704.	0.3	0
155	Transcriptomics analysis in collagen VI myopathy: Role of circadian genes using novel fluidic card tools. Neuromuscular Disorders, 2016, 26, S90-S91.	0.3	0
156	A mitochondrial therapy for Duchenne muscular dystrophy. Biochimica Et Biophysica Acta - Bioenergetics, 2018, 1859, e112.	0.5	0
157	Mitochondrial Pathogenesis of Myopathies Due to Collagen VI Mutations. , 2007, , 133-144.		0