Cecilia Jimenez-Mallebrera

List of Publications by Year
in descending order

70 papers	3,537 citations	159585 30 h-index	144013 57 g-index

75 all docs	75 docs citations	75 times ranked	3784 citing authors

#	Article	IF	Citations
1	Mutations in the human LARGE gene cause MDC1D, a novel form of congenital muscular dystrophy with severe mental retardation and abnormal glycosylation of Â-dystroglycan. Human Molecular Genetics, 2003, 12, 2853-2861.	2.9	389
2	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain, 2007, 130, 2725-2735.	7.6	385
3	Abnormalities in Î±-Dystroglycan Expression in MDC1C and LGMD2I Muscular Dystrophies. American Journal of Pathology, 2004, 164, 727-737.	3.8	154
4	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153
5	Fukutingene mutations in steroid-responsive limb girdle muscular dystrophy. Annals of Neurology, 2006, 60, 603-610.	5.3	140
6	Exon skipping mutations in collagen VI are common and are predictive for severity and inheritance. Human Mutation, 2008, 29, 809-822.	2.5	134
7	GDF-15 Is Elevated in Children with Mitochondrial Diseases and Is Induced by Mitochondrial Dysfunction. PLoS ONE, 2016, 11, e0148709.	2.5	133
8	Congenital muscular dystrophy: molecular and cellular aspects. Cellular and Molecular Life Sciences, 2005, 62, 809-823.	5.4	123
9	Congenital Myasthenic Syndromes in childhood: Diagnostic and management challenges. Journal of Neuroimmunology, 2008, 201-202, 6-12.	2.3	114
10	A Comparative Study of Î±â€Dystroglycan Glycosylation in Dystroglycanopathies Suggests that the Hypoglycosylation of Î±â€Dystroglycan Does Not Consistently Correlate with Clinical Severity. Brain Pathology, 2009, 19, 596-611.	4.1	107
11	Transcriptomic profiling of TK2 deficient human skeletal muscle suggests a role for the p53 signalling pathway and identifies growth and differentiation factor-15 as a potential novel biomarker for mitochondrial myopathies. BMC Genomics, 2014, 15, 91.	2.8	104
12	A comparative analysis of collagen VI production in muscle, skin and fibroblasts from 14 Ullrich congenital muscular dystrophy patients with dominant and recessive COL6A mutations. Neuromuscular Disorders, 2006, 16, 571-582.	0.6	97
13	Mild POMGnT1 Mutations Underlie a Novel Limb-Girdle Muscular Dystrophy Variant. Archives of Neurology, 2008, 65, 137-41.	4.5	73
14	Muscle magnetic resonance imaging in patients with congenital muscular dystrophy and Ullrich phenotype. Neuromuscular Disorders, 2003, 13, 554-558.	0.6	72
15	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
16	Deoxynucleoside Therapy for Thymidine Kinase 2â€“Deficient Myopathy. Annals of Neurology, 2019, 86, 293-303.	5.3	72
17	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	3.4	70
18	Congenital myopathies. Current Opinion in Neurology, 2008, 21, 569-575.	3.6	66

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19	Coenzyme Q10 deficiency in mitochondrial DNA depletion syndromes. Mitochondrion, 2013, 13, 337-341.	3.4	51
20	TRAPPC11 functions in autophagy by recruiting ATG2Bâ€WIPI4/WDR45 to preautophagosomal membranes. Traffic, 2019, 20, 325-345.	2.7	51
21	Absence of neuronal nitric oxide synthase (nNOS) as a pathological marker for the diagnosis of Becker muscular dystrophy with rod domain deletions. Neuropathology and Applied Neurobiology, 2004, 30, 540-545.	3.2	50
22	PGC-1Î± Induces Mitochondrial and Myokine Transcriptional Programs and Lipid Droplet and Glycogen Accumulation in Cultured Human Skeletal Muscle Cells. PLoS ONE, 2012, 7, e29985.	2.5	43
23	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
24	Interplay between DMD Point Mutations and Splicing Signals in Dystrophinopathy Phenotypes. PLoS ONE, 2013, 8, e59916.	2.5	42
25	Up71 and Up140, two novel transcripts of utrophin that are homologues of short forms of dystrophin. Human Molecular Genetics, 1999, 8, 1271-1278.	2.9	40
26	Prognostic value of X-chromosome inactivation in symptomatic female carriers of dystrophinopathy. Orphanet Journal of Rare Diseases, 2012, 7, 82.	2.7	40
27	Cyclosporine A treatment for Ullrich congenital muscular dystrophy: a cellular study of mitochondrial dysfunction and its rescue. Brain, 2009, 132, 147-155.	7.6	37
28	Prenatal diagnosis in laminin Î±2 chain (merosin)-deficient congenital muscular dystrophy: A collective experience of five international centers. Neuromuscular Disorders, 2005, 15, 588-594.	0.6	35
29	Profound skeletal muscle depletion of Î±-dystroglycan in Walker-Warburg syndrome. European Journal of Paediatric Neurology, 2003, 7, 129-137.	1.6	33
30	A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. JCI Insight, 2019, 4, .	5.0	33
31	KLHL40-related nemaline myopathy with a sustained, positive response to treatment with acetylcholinesterase inhibitors. Journal of Neurology, 2016, 263, 517-523.	3.6	30
32	Homozygous truncating mutation in prenatally expressed skeletal isoform of TTN gene results in arthrogryposis multiplex congenita and myopathy without cardiac involvement. Neuromuscular Disorders, 2017, 27, 188-192.	0.6	30
33	Prenatal diagnosis of Ullrich congenital muscular dystrophy using haplotype analysis and collagen VI immunocytochemistry. Prenatal Diagnosis, 2004, 24, 440-444.	2.3	29
34	Sub-cellular localisation of fukutin related protein in different cell lines and in the muscle of patients with MDC1C and LGMD2I. Neuromuscular Disorders, 2005, 15, 836-843.	0.6	29
35	A congenital myopathy with diaphragmatic weakness not linked to the SMARD1 locus. Neuromuscular Disorders, 2007, 17, 174-179.	0.6	29
36	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	2.7	29

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37	Digital PCR quantification of miR-30c and miR-181a as serum biomarkers for Duchenne muscular dystrophy. Neuromuscular Disorders, 2017, 27, 15-23.	0.6	27
38	Antenatal and Postnatal Brain Magnetic Resonance Imaging in Muscle-Eye-Brain Disease. Archives of Neurology, 2004, 61, 1301-6.	4.5	25
39	A homozygous COL6A2 intron mutation causes in-frame triple-helical deletion and nonsense-mediated mRNA decay in a patient with Ullrich congenital muscular dystrophy. Human Genetics, 2005, 117, 460-466.	3.8	24
40	Activation of osmolyte pathways in inflammatory myopathy and Duchenne muscular dystrophy points to osmoregulation as a contributing pathogenic mechanism. Laboratory Investigation, 2016, 96, 872-884.	3.7	24
41	Flow cytometry analysis: A quantitative method for collagen VI deficiency screening. Neuromuscular Disorders, 2012, 22, 139-148.	0.6	23
42	Transcriptome Analysis of Ullrich Congenital Muscular Dystrophy Fibroblasts Reveals a Disease Extracellular Matrix Signature and Key Molecular Regulators. PLoS ONE, 2015, 10, e0145107.	2.5	23
43	Gene Expression Profiling Identifies Molecular Pathways Associated with Collagen VI Deficiency and Provides Novel Therapeutic Targets. PLoS ONE, 2013, 8, e77430.	2.5	23
44	Muscle Fiber Atrophy and Regeneration Coexist in Collagen VI-Deficient Human Muscle: Role of Calpain-3 and Nuclear Factor-ÍºB Signaling. Journal of Neuropathology and Experimental Neurology, 2012, 71, 894-906.	1.7	22
45	Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. Scientific Reports, 2020, 10, 10111.	3.3	20
46	Longitudinal Study of Three microRNAs in Duchenne Muscular Dystrophy and Becker Muscular Dystrophy. Frontiers in Neurology, 2020, 11, 304.	2.4	17
47	Association between coenzyme Q10 and glucose transporter (GLUT1) deficiency. BMC Pediatrics, 2014, 14, 284.	1.7	15
48	Mutation loads in different tissues from six pathogenic mtDNA point mutations. Mitochondrion, 2015, 22, 17-22.	3.4	15
49	A study of short utrophin isoforms in mice deficient for full-length utrophin. Mammalian Genome, 2003, 14, 47-60.	2.2	14
50	Muscle Involvement in a Large Cohort of Pediatric Patients with Genetic Diagnosis of Mitochondrial Disease. Journal of Clinical Medicine, 2019, 8, 68.	2.4	14
51	Long-term survival in a child with severe encephalopathy, multiple respiratory chain deficiency and GFM1 mutations. Frontiers in Genetics, 2015, 6, 102.	2.3	13
52	Epilepsy in LAMA2â€related muscular dystrophy: An electroâ€clinicoâ€radiological characterization. Epilepsia, 2020, 61, 971-983.	5.1	12
53	The Phenotype and Genotype of Congenital Myopathies Based on a Large Pediatric Cohort. Pediatric Neurology, 2021, 115, 50-65.	2.1	11
54	Association of Initial Maximal Motor Ability With Long-term Functional Outcome in Patients With COL6-Related Dystrophies. Neurology, 2021, 96, e1413-e1424.	1.1	10

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55	Oxidative Phosphorylation Dysfunction Modifies the Cell Secretome. International Journal of Molecular Sciences, 2020, 21, 3374.	4.1	8
56	Glycogen branching enzyme deficiency in an infant with severe congenital hypotonia: an emerging diagnosis of muscle weakness in the perinatal period. Histopathology, 2009, 54, 765-768.	2.9	7
57	Differences in Adipose Tissue and Lean Mass Distribution in Patients with Collagen VI Related Myopathies Are Associated with Disease Severity and Physical Ability. Frontiers in Aging Neuroscience, 2017, 9, 268.	3.4	7
58	Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. Clinical Chemistry, 2021, 67, 1113-1121.	3.2	7
59	Recurrent rhabdomyolysis and exercise intolerance: A new phenotype of late-onset thymidine kinase 2 deficiency. Molecular Genetics and Metabolism Reports, 2021, 26, 100701.	1.1	6
60	Pediatric SMA patients with complex spinal anatomy: Implementation and evaluation of a decision-tree algorithm for administration of nusinersen. European Journal of Paediatric Neurology, 2021, 31, 92-101.	1.6	6
61	A Convolutional Neural Network for the automatic diagnosis of collagen VI-related muscular dystrophies. Applied Soft Computing Journal, 2019, 85, 105772.	7.2	5
62	CRISPR/Cas9-Mediated Allele-Specific Disruption of a Dominant COL6A1 Pathogenic Variant Improves Collagen VI Network in Patient Fibroblasts. International Journal of Molecular Sciences, 2022, 23, 4410.	4.1	5
63	The Capillary Morphogenesis Gene 2 Triggers the Intracellular Hallmarks of Collagen VI-Related Muscular Dystrophy. International Journal of Molecular Sciences, 2022, 23, 7651.	4.1	5
64	Personalized in vitro Extracellular Matrix Models of Collagen VI-Related Muscular Dystrophies. Frontiers in Bioengineering and Biotechnology, 2022, 10, 851825.	4.1	4
65	Early and long-term effect of the treatment with pyridostigmine in patients with GMPPB-related congenital myasthenic syndrome. Neuromuscular Disorders, 2020, 30, 719-726.	0.6	3
66	A case presenting with delayed motor milestones. Neuromuscular Disorders, 2005, 15, 817-818.	0.6	2
67	Biallelic mutations in Tenascin-X cause classical-like Ehlers-Danlos syndrome with slowly progressive muscular weakness. Neuromuscular Disorders, 2020, 30, 833-838.	0.6	2
68	Digital PCR quantification of miR-30c and miR-181a as serum biomarkers in Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, S159.	0.6	1
69	The importance of verifying the novelty of a finding and the value of combining results. Annals of Clinical and Translational Neurology, 0, , .	3.7	1
70	Response to letter from Bernardi. Brain, 2009, 132, e122-e122.	7.6	0