# 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 | 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 |
| 2 | Personalized in vitro Extracellular Matrix Models of Collagen VI-Related Muscular Dystrophies. Frontiers in Bioengineering and Biotechnology, 2022, 10, 851825. | 4.1 | 4 |
| 3 | 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 |
| 4 | The Phenotype and Genotype of Congenital Myopathies Based on a Large Pediatric Cohort. Pediatric Neurology, 2021, 115, 50-65. | 2.1 | 11 |
| 5 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. Clinical Chemistry, 2021, 67, 1113-1121. | 3.2 | 7 |
| 9 | Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308. | 3.6 | 43 |
| 10 | 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 |
| 11 | 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 |
| 12 | Oxidative Phosphorylation Dysfunction Modifies the Cell Secretome. International Journal of Molecular Sciences, 2020, 21, 3374. | 4.1 | 8 |
| 13 | Longitudinal Study of Three microRNAs in Duchenne Muscular Dystrophy and Becker Muscular Dystrophy. Frontiers in Neurology, 2020, 11, 304. | 2.4 | 17 |
| 14 | Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. Scientific Reports, 2020, 10, 10111. | 3.3 | 20 |
| 15 | Epilepsy in LAMA2â€related muscular dystrophy: An electroâ€clinicoâ€radiological characterization. Epilepsia, 2020, 61, 971-983. | 5.1 | 12 |
| 16 | A Convolutional Neural Network for the automatic diagnosis of collagen VI-related muscular dystrophies. Applied Soft Computing Journal, 2019, 85, 105772. | 7.2 | 5 |
| 17 | Deoxynucleoside Therapy for Thymidine Kinase 2â€“Deficient Myopathy. Annals of Neurology, 2019, 86, 293-303. | 5.3 | 72 |
| 18 | Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100. | 2.7 | 29 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | TRAPPC11 functions in autophagy by recruiting ATG2Bâ€WIPI4/WDR45 to preautophagosomal membranes. Traffic, 2019, 20, 325-345. | 2.7 | 51 |
| 20 | 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 |
| 21 | A recurrent COL6A1 pseudoexon insertion causes muscular dystrophy and is effectively targeted by splice-correction therapies. JCI Insight, 2019, 4, . | 5.0 | 33 |
| 22 | 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 |
| 23 | 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 |
| 24 | 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 |
| 25 | GDF-15 Is Elevated in Children with Mitochondrial Diseases and Is Induced by Mitochondrial Dysfunction. PLoS ONE, 2016, 11, e0148709. | 2.5 | 133 |
| 26 | Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58. | 3.4 | 70 |
| 27 | Digital PCR quantification of miR-30c and miR-181a as serum biomarkers in Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, S159. | 0.6 | 1 |
| 28 | 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 |
| 29 | KLHL40-related nemaline myopathy with a sustained, positive response to treatment with acetylcholinesterase inhibitors. Journal of Neurology, 2016, 263, 517-523. | 3.6 | 30 |
| 30 | 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 |
| 31 | 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 |
| 32 | Mutation loads in different tissues from six pathogenic mtDNA point mutations. Mitochondrion, 2015, 22, 17-22. | 3.4 | 15 |
| 33 | Association between coenzyme Q10 and glucose transporter (GLUT1) deficiency. BMC Pediatrics, 2014, 14, 284. | 1.7 | 15 |
| 34 | 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 |
| 35 | Coenzyme Q10 deficiency in mitochondrial DNA depletion syndromes. Mitochondrion, 2013, 13, 337-341. | 3.4 | 51 |
| 36 | Interplay between DMD Point Mutations and Splicing Signals in Dystrophinopathy Phenotypes. PLoS ONE, 2013, 8, e59916. | 2.5 | 42 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Gene Expression Profiling Identifies Molecular Pathways Associated with Collagen VI Deficiency and Provides Novel Therapeutic Targets. PLoS ONE, 2013, 8, e77430. | 2.5 | 23 |
| 38 | Muscle Fiber Atrophy and Regeneration Coexist in Collagen VI-Deficient Human Muscle: Role of Calpain-3 and Nuclear Factor-Î²B Signaling. Journal of Neuropathology and Experimental Neurology, 2012, 71, 894-906. | 1.7 | 22 |
| 39 | Flow cytometry analysis: A quantitative method for collagen VI deficiency screening. Neuromuscular Disorders, 2012, 22, 139-148. | 0.6 | 23 |
| 40 | Prognostic value of X-chromosome inactivation in symptomatic female carriers of dystrophinopathy. Orphanet Journal of Rare Diseases, 2012, 7, 82. | 2.7 | 40 |
| 41 | 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 |
| 42 | Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850. | 3.6 | 72 |
| 43 | Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172. | 6.2 | 153 |
| 44 | 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 |
| 45 | Response to letter from Bernardi. Brain, 2009, 132, e122-e122. | 7.6 | 0 |
| 46 | 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 |
| 47 | 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 |
| 48 | Exon skipping mutations in collagen VI are common and are predictive for severity and inheritance. Human Mutation, 2008, 29, 809-822. | 2.5 | 134 |
| 49 | Congenital Myasthenic Syndromes in childhood: Diagnostic and management challenges. Journal of Neuroimmunology, 2008, 201-202, 6-12. | 2.3 | 114 |
| 50 | Mild POMGnT1 Mutations Underlie a Novel Limb-Girdle Muscular Dystrophy Variant. Archives of Neurology, 2008, 65, 137-41. | 4.5 | 73 |
| 51 | Congenital myopathies. Current Opinion in Neurology, 2008, 21, 569-575. | 3.6 | 66 |
| 52 | Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain, 2007, 130, 2725-2735. | 7.6 | 385 |
| 53 | A congenital myopathy with diaphragmatic weakness not linked to the SMARD1 locus. Neuromuscular Disorders, 2007, 17, 174-179. | 0.6 | 29 |
| 54 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Fukutingene mutations in steroid-responsive limb girdle muscular dystrophy. Annals of Neurology, 2006, 60, 603-610. | 5.3 | 140 |
| 56 | Congenital muscular dystrophy: molecular and cellular aspects. Cellular and Molecular Life Sciences, 2005, 62, 809-823. | 5.4 | 123 |
| 57 | 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 |
| 58 | 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 |
| 59 | A case presenting with delayed motor milestones. Neuromuscular Disorders, 2005, 15, 817-818. | 0.6 | 2 |
| 60 | 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 |
| 61 | Antenatal and Postnatal Brain Magnetic Resonance Imaging in Muscle-Eye-Brain Disease. Archives of Neurology, 2004, 61, 1301-6. | 4.5 | 25 |
| 62 | 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 |
| 63 | Prenatal diagnosis of Ullrich congenital muscular dystrophy using haplotype analysis and collagen VI immunocytochemistry. Prenatal Diagnosis, 2004, 24, 440-444. | 2.3 | 29 |
| 64 | Abnormalities in Î±-Dystroglycan Expression in MDC1C and LGMD2I Muscular Dystrophies. American Journal of Pathology, 2004, 164, 727-737. | 3.8 | 154 |
| 65 | A study of short utrophin isoforms in mice deficient for full-length utrophin. Mammalian Genome, 2003, 14, 47-60. | 2.2 | 14 |
| 66 | Profound skeletal muscle depletion of Î±-dystroglycan in Walker-Warburg syndrome. European Journal of Paediatric Neurology, 2003, 7, 129-137. | 1.6 | 33 |
| 67 | Muscle magnetic resonance imaging in patients with congenital muscular dystrophy and Ullrich phenotype. Neuromuscular Disorders, 2003, 13, 554-558. | 0.6 | 72 |
| 68 | 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 |
| 69 | 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 |
| 70 | 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 |