# Enrico Bertini

List of Publications by Year
in descending order

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|---|---|---|---|
| 727 papers | 42,990 citations | 2215 99 h-index | 5255 165 g-index |
| 759 all docs | 759 docs citations | 759 times ranked | 36015 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409. | 3.2 | 13 |
| 2 | PIGQ-Related Glycophosphatidylinositol Deficiency Associated with Nonprogressive Congenital Ataxia. Cerebellum, 2022, 21, 525-530. | 2.5 | 2 |
| 3 | Personalized profiles of antioxidant signaling pathway in patients with tuberculosis. Journal of Microbiology, Immunology and Infection, 2022, 55, 405-412. | 3.1 | 3 |
| 4 | SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894. | 3.2 | 19 |
| 5 | Long-Term Efficacy of T3 Analogue Triac in Children and Adults With MCT8 Deficiency: A Real-Life Retrospective Cohort Study. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e1136-e1147. | 3.6 | 15 |
| 6 | Revised upper limb module in type II and III spinal muscular atrophy: 24-month changes. Neuromuscular Disorders, 2022, 32, 36-42. | 0.6 | 13 |
| 7 | Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73. | 2.5 | 9 |
| 8 | Ataluren delays loss of ambulation and respiratory decline in nonsense mutation Duchenne muscular dystrophy patients. Journal of Comparative Effectiveness Research, 2022, 11, 139-155. | 1.4 | 29 |
| 9 | A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis. Neurological Sciences, 2022, 43, 2849-2852. | 1.9 | 0 |
| 10 | Movement disorders in MCT8 deficiency/Allan-Herndon-Dudley Syndrome. Molecular Genetics and Metabolism, 2022, 135, 109-113. | 1.1 | 17 |
| 11 | Toward the inÂvitro understanding of iPSC nucleoskeletal and cytoskeletal biology, and their relevance for organoid development. , 2022, , 137-150. | | 0 |
| 12 | Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial. Lancet Neurology, The, 2022, 21, 42-52. | 10.2 | 89 |
| 13 | Body mass index in type 2 spinal muscular atrophy: a longitudinal study. European Journal of Pediatrics, 2022, , 1. | 2.7 | 2 |
| 14 | Cerebellar Agenesis and Bilateral Polimicrogyria Associated with Rare Variants of CUB and Sushi Multiple Domains 1 Gene (CSMD1): A Longitudinal Neuropsychological and Neuroradiological Case Study. International Journal of Environmental Research and Public Health, 2022, 19, 1224. | 2.6 | 0 |
| 15 | Neurological and Neuroimaging Features of CYB5R3-Related Recessive Hereditary Methemoglobinemia Type II. Brain Sciences, 2022, 12, 182. | 2.3 | 4 |
| 16 | Therapy Trial Design in Vanishing White Matter. Neurology: Genetics, 2022, 8, e657. | 1.9 | 12 |
| 17 | Upper Body Physical Rehabilitation for Children with Ataxia through IMU-Based Exergame. Journal of Clinical Medicine, 2022, 11, 1065. | 2.4 | 7 |
| 18 | Nusinersen efficacy data for 24â€month in type 2 and 3 spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2022, 9, 404-409. | 3.7 | 22 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Modeling PCDH19-CE: From 2D Stem Cell Model to 3D Brain Organoids. International Journal of Molecular Sciences, 2022, 23, 3506. | 4.1 | 1 |
| 20 | Novel SEPSECS Pathogenic Variants Featuring Unusual Phenotype of Complex Movement Disorder With Thin Corpus Callosum. Neurology: Genetics, 2022, 8, e661. | 1.9 | 0 |
| 21 | Clinical-Genetic Features Influencing Disability in Spastic Paraplegia Type 4. Neurology: Genetics, 2022, 8, e664. | 1.9 | 9 |
| 22 | Clinical and Genetic Aspects of Phelanâ€“McDermid Syndrome: An Interdisciplinary Approach to Management. Genes, 2022, 13, 504. | 2.4 | 9 |
| 23 | A novel homozygous variant in <scp>COX5A</scp> causes an attenuated phenotype with failure to thrive, lactic acidosis, hypoglycemia, and short stature. Clinical Genetics, 2022, 102, 56-60. | 2.0 | 3 |
| 24 | Response to: Phenotypic heterogeneity of Leigh syndrome due to NDUFA12 variants is multicausal. Human Mutation, 2022, 43, 99-100. | 2.5 | 0 |
| 25 | Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study. Acta Neuropathologica Communications, 2022, 10, 54. | 5.2 | 3 |
| 26 | Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894. | 3.6 | 2 |
| 27 | â€œAtypicalâ€•Krabbe disease in two siblings harboring biallelic GALC mutations including a deep intronic variant. European Journal of Human Genetics, 2022, , . | 2.8 | 4 |
| 28 | Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390. | 7.6 | 3 |
| 29 | Superior Cerebellar Atrophy: An Imaging Clue to Diagnose ITPR1-Related Disorders. International Journal of Molecular Sciences, 2022, 23, 6723. | 4.1 | 4 |
| 30 | New Insights into the Neurodegeneration Mechanisms Underlying Riboflavin Transporter Deficiency (RTD): Involvement of Energy Dysmetabolism and Cytoskeletal Derangement. Biomedicines, 2022, 10, 1329. | 3.2 | 5 |
| 31 | Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483. | 3.2 | 21 |
| 32 | Sometimes they come back: New and old spinal muscular atrophy adults in the era of nusinersen. European Journal of Neurology, 2021, 28, 602-608. | 3.3 | 9 |
| 33 | Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154. | 1.6 | 13 |
| 34 | Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. Neurology, 2021, 96, e587-e599. | 1.1 | 36 |
| 35 | Novel ACTA1 mutation causes late-presenting nemaline myopathy with unusual dark cores. Neuromuscular Disorders, 2021, 31, 139-148. | 0.6 | 4 |
| 36 | Clinical and radiological profile of patients with spinal muscular atrophy type 4. European Journal of Neurology, 2021, 28, 609-619. | 3.3 | 23 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Friedreich ataxia in COVID-19 time: current impact and future possibilities. Cerebellum and Ataxias, 2021, 8, 4. | 1.9 | 8 |
| 38 | Type I SMA â€œnew natural historyâ€• longâ€term data in nusinersenâ€treated patients. Annals of Clinical and Translational Neurology, 2021, 8, 548-557. | 3.7 | 35 |
| 39 | The Spinal Muscular Atrophy Health Index: Italian validation of a disease-specific outcome measure. Neuromuscular Disorders, 2021, 31, 409-418. | 0.6 | 7 |
| 40 | Nuclear Factor Erythroid 2-Related Factor 2 Activation Might Mitigate Clinical Symptoms in Friedreichâ€™s Ataxia: Clues of an â€œOut-Brain Originâ€•of the Disease From a Family Study. Frontiers in Neuroscience, 2021, 15, 638810. | 2.8 | 5 |
| 41 | Altered cytoskeletal arrangement in induced pluripotent stem cells and motor neurons from patients with riboflavin transporter deficiency. DMM Disease Models and Mechanisms, 2021, 14, . | 2.4 | 5 |
| 42 | Mitochondrial Dynamics: Molecular Mechanisms, Related Primary Mitochondrial Disorders and Therapeutic Approaches. Genes, 2021, 12, 247. | 2.4 | 25 |
| 43 | Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710. | 2.5 | 12 |
| 44 | CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69. | 1.6 | 7 |
| 45 | Predictive fat mass equations for spinal muscular atrophy type I children: Development and internal validation. Clinical Nutrition, 2021, 40, 1578-1587. | 5.0 | 3 |
| 46 | Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. Disability and Rehabilitation, 2021, , 1-8. | 1.8 | 1 |
| 47 | Biâ€allelic KARS1 pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. Human Mutation, 2021, 42, 745-761. | 2.5 | 7 |
| 48 | Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063. | 2.4 | 8 |
| 49 | Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. Brain, 2021, 144, 3020-3035. | 7.6 | 11 |
| 50 | Dissecting the Role of PCDH19 in Clustering Epilepsy by Exploiting Patient-Specific Models of Neurogenesis. Journal of Clinical Medicine, 2021, 10, 2754. | 2.4 | 13 |
| 51 | Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1622-1634. | 3.7 | 27 |
| 52 | Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582. | 0.6 | 94 |
| 53 | The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. Frontiers in Neurology, 2021, 12, 677551. | 2.4 | 15 |
| 54 | North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. PLoS ONE, 2021, 16, e0253882. | 2.5 | 6 |

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|---|---|---|---|
| 55 | The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488. | 0.6 | 0 |
| 56 | Circadian Genes as Exploratory Biomarkers in DMD: Results From Both the mdx Mouse Model and Patients. Frontiers in Physiology, 2021, 12, 678974. | 2.8 | 1 |
| 57 | Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602. | 0.6 | 29 |
| 58 | Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359. | 2.4 | 8 |
| 59 | Different trajectories in upper limb and gross motor function in spinal muscular atrophy. Muscle and Nerve, 2021, 64, 552-559. | 2.2 | 18 |
| 60 | Ageâ€related sensory neuropathy in patients with spinal muscular atrophy type 1. Muscle and Nerve, 2021, 64, 599-603. | 2.2 | 3 |
| 61 | Growth patterns in children with spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2021, 16, 375. | 2.7 | 19 |
| 62 | SMA-miRs (miR-181a-5p, -324-5p, and -451a) are overexpressed in spinal muscular atrophy skeletal muscle and serum samples. ELife, 2021, 10, . | 6.0 | 13 |
| 63 | Broadening the spectrum phenotype of TBCE-related neuron neurodegeneration. Brain and Development, 2021, 43, 939-944. | 1.1 | 0 |
| 64 | Expanding the Clinical and Mutational Spectrum of the PLP1-Related Hypomyelination of Early Myelinated Structures (HEMS). Brain Sciences, 2021, 11, 93. | 2.3 | 3 |
| 65 | Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases. Orphanet Journal of Rare Diseases, 2021, 16, 413. | 2.7 | 16 |
| 66 | Myocardial and Arrhythmic Spectrum of Neuromuscular Disorders in Children. Biomolecules, 2021, 11, 1578. | 4.0 | 5 |
| 67 | Artificial Intelligence for Dysarthria Assessment in Children With Ataxia: A Hierarchical Approach. IEEE Access, 2021, 9, 166720-166735. | 4.2 | 4 |
| 68 | Induced Pluripotent Stem Cells (iPSCs) and Gene Therapy: A New Era for the Treatment of Neurological Diseases. International Journal of Molecular Sciences, 2021, 22, 13674. | 4.1 | 13 |
| 69 | Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202. | 0.7 | 15 |
| 70 | Speech and Language Disorders in Friedreich Ataxia: Highlights on Phenomenology, Assessment, and Therapy. Cerebellum, 2020, 19, 126-130. | 2.5 | 12 |
| 71 | Hereditary spastic paraplegia is a novel phenotype for germline de novo ATP1A1 mutation. Clinical Genetics, 2020, 97, 521-526. | 2.0 | 14 |
| 72 | 244th ENMC international workshop: Newborn screening in spinal muscular atrophy May 10â€“12, 2019, Hoofdorp, The Netherlands. Neuromuscular Disorders, 2020, 30, 93-103. | 0.6 | 55 |

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|---|---|---|---|
| 73 | Systemic Activation of Nrf2 Pathway in Parkinson's Disease. Movement Disorders, 2020, 35, 180-184. | 3.9 | 66 |
| 74 | Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152. | 3.7 | 26 |
| 75 | RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93. | 3.7 | 18 |
| 76 | Development of SaraHome: A novel, well-accepted, technology-based assessment tool for patients with ataxia. Computer Methods and Programs in Biomedicine, 2020, 188, 105257. | 4.7 | 21 |
| 77 | Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96. | 1.4 | 14 |
| 78 | SMA â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S123-S124. | 0.6 | 6 |
| 79 | <scp>PPP1R21</scp>â€related syndromic intellectual disability: Report of an adult patient and review. American Journal of Medical Genetics, Part A, 2020, 182, 3014-3022. | 1.2 | 8 |
| 80 | Long-Term Outcome of LVAD in Duchenne Population with End Stage Cardiomyopathy. Journal of Heart and Lung Transplantation, 2020, 39, S219-S220. | 0.6 | 0 |
| 81 | Microtubule Dysfunction: A Common Feature of Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 7354. | 4.1 | 63 |
| 82 | A Recurrent Pathogenic Variant of INPP5K Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. Frontiers in Genetics, 2020, 11, 565868. | 2.3 | 8 |
| 83 | Spatio-temporal parameters of ataxia gait dataset obtained with the Kinect. Data in Brief, 2020, 32, 106307. | 1.0 | 10 |
| 84 | Antioxidant Amelioration of Riboflavin Transporter Deficiency in Motoneurons Derived from Patient-Specific Induced Pluripotent Stem Cells. International Journal of Molecular Sciences, 2020, 21, 7402. | 4.1 | 8 |
| 85 | Validation of low-cost system for gait assessment in children with ataxia. Computer Methods and Programs in Biomedicine, 2020, 196, 105705. | 4.7 | 17 |
| 86 | Aicardiâ€“GoutiÃ¨res Syndrome Type 2: A Report on Two Cases with Different Phenotypes Caused by RNASEH2B Gene Mutations. Journal of Pediatric Neurology, 2020, 18, 206-209. | 0.2 | 0 |
| 87 | Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605. | 11.4 | 50 |
| 88 | Response to â€œAutosomal recessive axonal neuropathy caused by HINT1 mutation: New association of a psychiatric disorder to the neurological phenotypeâ€• Neuromuscular Disorders, 2020, 30, 265-266. | 0.6 | 4 |
| 89 | Ferroptosis in Friedreichâ€™s Ataxia: A Metal-Induced Neurodegenerative Disease. Biomolecules, 2020, 10, 1551. | 4.0 | 21 |
| 90 | Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. Genetics in Medicine, 2020, 22, 1851-1862. | 2.4 | 30 |

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|---|---|---|---|
| 91 | Cardiovascular Involvement in Pediatric Laminopathies. Report of Six Patients and Literature Revision. Frontiers in Pediatrics, 2020, 8, 374. | 1.9 | 9 |
| 92 | Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. Neuromuscular Disorders, 2020, 30, 756-764. | 0.6 | 25 |
| 93 | Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2020, 9, 973-984. | 1.4 | 41 |
| 94 | Mitochondrial and Peroxisomal Alterations Contribute to Energy Dysmetabolism in Riboflavin Transporter Deficiency. Oxidative Medicine and Cellular Longevity, 2020, 2020, 1-19. | 4.0 | 13 |
| 95 | Clinical Variability in Spinal Muscular Atrophy Type <scp>III</scp>. Annals of Neurology, 2020, 88, 1109-1117. | 5.3 | 34 |
| 96 | The clinical, histologic, and genotypic spectrum of SEPN1 -related myopathy. Neurology, 2020, 95, e1512-e1527. | 1.1 | 44 |
| 97 | Mitochondrial Abnormalities in Induced Pluripotent Stem Cells-Derived Motor Neurons from Patients with Riboflavin Transporter Deficiency. Antioxidants, 2020, 9, 1252. | 5.1 | 11 |
| 98 | Movement disorders in ADAR1 disease: Insights from a comprehensive cohort. Parkinsonism and Related Disorders, 2020, 79, 100-104. | 2.2 | 6 |
| 99 | Long-term follow-up of patients with type 2 and non-ambulant type 3 spinal muscular atrophy (SMA) treated with olesoxime in the OLEOS trial. Neuromuscular Disorders, 2020, 30, 959-969. | 0.6 | 15 |
| 100 | Oxidative Stress in DNA Repeat Expansion Disorders: A Focus on NRF2 Signaling Involvement. Biomolecules, 2020, 10, 702. | 4.0 | 17 |
| 101 | Randomized phase 2 trial and open-label extension of domagrozumab in Duchenne muscular dystrophy. Neuromuscular Disorders, 2020, 30, 492-502. | 0.6 | 40 |
| 102 | Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801. | 1.1 | 26 |
| 103 | Predictive energy equations for spinal muscular atrophy type I children. American Journal of Clinical Nutrition, 2020, 111, 983-996. | 4.7 | 8 |
| 104 | The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131. | 2.3 | 49 |
| 105 | Diagnostic journey in Spinal Muscular Atrophy: Is it still an odyssey?. PLoS ONE, 2020, 15, e0230677. | 2.5 | 38 |
| 106 | Co-occurrence of mutations in KIF7 and KIAA0556 in Joubert syndrome with ocular coloboma, pituitary malformation and growth hormone deficiency: a case report and literature review. BMC Pediatrics, 2020, 20, 120. | 1.7 | 12 |
| 107 | Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605. | 2.3 | 9 |
| 108 | The NRF2 Signaling Network Defines Clinical Biomarkers and Therapeutic Opportunity in Friedreichâ€™s Ataxia. International Journal of Molecular Sciences, 2020, 21, 916. | 4.1 | 27 |

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|---|---|---|---|
| 109 | Respiratory Needs in Patients with Type 1 Spinal Muscular Atrophy TreatedÂwith Nusinersen. Journal of Pediatrics, 2020, 219, 223-228.e4. | 1.8 | 51 |
| 110 | TUBB Variants Underlying Different Phenotypes Result in Altered Vesicle Trafficking and Microtubule Dynamics. International Journal of Molecular Sciences, 2020, 21, 1385. | 4.1 | 20 |
| 111 | Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798. | 3.7 | 36 |
| 112 | A wearable video-oculography based evaluation of saccades and respective clinical correlates in patients with early onset ataxia. Journal of Neuroscience Methods, 2020, 338, 108697. | 2.5 | 3 |
| 113 | Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263. | 5.3 | 52 |
| 114 | A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. Neurobiology of Disease, 2020, 141, 104880. | 4.4 | 29 |
| 115 | Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251. | | 1 |
| 116 | HDAC inhibitors tune miRNAs in extracellular vesicles of dystrophic muscleâ€resident mesenchymal cells. EMBO Reports, 2020, 21, e50863. | 4.5 | 45 |
| 117 | Clinical and Genetic Overview of Paroxysmal Movement Disorders and Episodic Ataxias. International Journal of Molecular Sciences, 2020, 21, 3603. | 4.1 | 36 |
| 118 | A clinical diagnostic algorithm for early onset cerebellar ataxia. European Journal of Paediatric Neurology, 2019, 23, 692-706. | 1.6 | 37 |
| 119 | Nrf2 Induction Re-establishes a Proper Neuronal Differentiation Program in Friedreichâ€™s Ataxia Neural Stem Cells. Frontiers in Cellular Neuroscience, 2019, 13, 356. | 3.7 | 36 |
| 120 | Effectiveness and safety of the tri-iodothyronine analogue Triac in children and adults with MCT8 deficiency: an international, single-arm, open-label, phase 2 trial. Lancet Diabetes and Endocrinology,the, 2019, 7, 695-706. | 11.4 | 77 |
| 121 | Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. Journal of Neurology, 2019, 266, 2657-2664. | 3.6 | 19 |
| 122 | Observations from a nationwide vigilance program in medical care for spinal muscular atrophy patients in Chile. Arquivos De Neuro-Psiquiatria, 2019, 77, 470-477. | 0.8 | 6 |
| 123 | Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism and Related Disorders, 2019, 68, 8-16. | 2.2 | 25 |
| 124 | â€˜Amish Nemaline Myopathyâ€™ in 2 Italian siblings harbouring a novel homozygous mutation in Troponin-I gene. Neuromuscular Disorders, 2019, 29, 766-770. | 0.6 | 13 |
| 125 | Evaluation of gait in Duchenne Muscular Dystrophy: Relation of 3D gait analysis to clinical assessment. Neuromuscular Disorders, 2019, 29, 920-929. | 0.6 | 14 |
| 126 | Targeting NRF2 for the Treatment of Friedreichâ€™s Ataxia: A Comparison among Drugs. International Journal of Molecular Sciences, 2019, 20, 5211. | 4.1 | 45 |

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|---|---|---|---|
| 127 | Development of an academic disease registry for spinal muscular atrophy. Neuromuscular Disorders, 2019, 29, 794-799. | 0.6 | 29 |
| 128 | Longitudinal natural history in young boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2019, 29, 857-862. | 0.6 | 23 |
| 129 | Corticospinal tract damage in HHH syndrome: a metabolic cause of hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2019, 14, 208. | 2.7 | 12 |
| 130 | Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508. | 6.2 | 48 |
| 131 | Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscular Disorders, 2019, 29, 842-856. | 0.6 | 401 |
| 132 | Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130. | 3.2 | 42 |
| 133 | Heart rate reduction strategy using ivabradine in end-stage Duchenne cardiomyopathy. International Journal of Cardiology, 2019, 280, 99-103. | 1.7 | 17 |
| 134 | Clinical, radiological, and genetic characteristics of 16 patients with ACO2 gene defects: Delineation of an emerging neurometabolic syndrome. Journal of Inherited Metabolic Disease, 2019, 42, 264-275. | 3.6 | 18 |
| 135 | Nusinersen in type 1 spinal muscular atrophy: Twelveâ€month realâ€world data. Annals of Neurology, 2019, 86, 443-451. | 5.3 | 83 |
| 136 | Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. PLoS ONE, 2019, 14, e0218683. | 2.5 | 47 |
| 137 | Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750. | 2.4 | 29 |
| 138 | Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. Clinical Genetics, 2019, 96, 169-175. | 2.0 | 27 |
| 139 | Response to Jardim and colleagues regarding comments on â€˜Natural history of a cohort of <scp>ABCD</scp>1 variant female carriersâ€™. European Journal of Neurology, 2019, 26, e77. | 3.3 | 0 |
| 140 | Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748. | 2.5 | 31 |
| 141 | Mitochondrial Neurodegenerative Disorders II: Ataxia, Dystonia and Leukodystrophies. , 2019, , 241-256. | | 1 |
| 142 | Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. PLoS ONE, 2019, 14, e0214250. | 2.5 | 59 |
| 143 | An unusual case of late-infantile onset Krabbe disease with selective bilateralÂcorticospinal tract involvement, peripheral demyelinating neuropathy, and mild phenotype. Acta Neurologica Belgica, 2019, 119, 619-620. | 1.1 | 1 |
| 144 | Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. Cerebellum, 2019, 18, 422-432. | 2.5 | 10 |

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|---|---|---|---|
| 145 | Clinical-genetic features and peculiar muscle histopathology in infantileDNM1L-related mitochondrial epileptic encephalopathy. Human Mutation, 2019, 40, 601-618. | 2.5 | 31 |
| 146 | Cardiac and Neuromuscular Features of Patients WithLMNA-Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458. | 3.9 | 33 |
| 147 | One-year outcome of coenzyme Q10 supplementation in ADCK3 ataxia (ARCA2). Cerebellum and Ataxias, 2019, 6, 15. | 1.9 | 15 |
| 148 | Diagnostic Yield of a Targeted Next-Generation Sequencing Gene Panel for Pediatric-Onset Movement Disorders: A 3-Year Cohort Study. Frontiers in Genetics, 2019, 10, 1026. | 2.3 | 33 |
| 149 | Primary muscle involvement in a 15â€yearâ€old girl with the recurrent homozygous c.362dupC variant in FKBP14. American Journal of Medical Genetics, Part A, 2019, 179, 317-321. | 1.2 | 3 |
| 150 | A novel KCTD17 mutation is associated with childhood early-onset hyperkinetic movement disorder. Parkinsonism and Related Disorders, 2019, 61, 4-6. | 2.2 | 22 |
| 151 | Italian recommendations for diagnosis and management of congenital myasthenic syndromes. Neurological Sciences, 2019, 40, 457-468. | 1.9 | 24 |
| 152 | APOPT 1/ COA 8 assists COX assembly and is oppositely regulated by UPS and ROS. EMBO Molecular Medicine, 2019, 11, . | 6.9 | 19 |
| 153 | SLC2A1 mutations are a rare cause of pediatric-onset hereditary spastic paraplegia. European Journal of Paediatric Neurology, 2019, 23, 329-332. | 1.6 | 11 |
| 154 | Phenomenology and clinical course of movement disorder in GNAO1 variants: Results from an analytical review. Parkinsonism and Related Disorders, 2019, 61, 19-25. | 2.2 | 64 |
| 155 | Natural history of a cohort of <scp>ABCD</scp>1 variant female carriers. European Journal of Neurology, 2019, 26, 326-332. | 3.3 | 19 |
| 156 | Neonatal Hypotonia. , 2019, , 223-233. | | 0 |
| 157 | SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125. | 8.2 | 65 |
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