# Maurizio Elia

List of Publications by Year
in descending order



| 233 | 9,037 | 41323 | 56687 |
|---|---|---|---|
| papers | citations | 49 h-index | 83 g-index |

| 241 | 241 | 241 | 10150 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | A genetic variant that disrupts MET transcription is associated with autism. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 16834-16839. | 3.3 | 389 |
| 2 | Summary of recommendations for the management of infantile seizures: Task <scp>F</scp>orce <scp>R</scp>eport for the <scp>ILAE C</scp>ommission of <scp>P</scp>ediatrics. Epilepsia, 2015, 56, 1185-1197. | 2.6 | 323 |
| 3 | Mutations in NHLRC1 cause progressive myoclonus epilepsy. Nature Genetics, 2003, 35, 125-127. | 9.4 | 294 |
| 4 | Audiogenic Seizures Susceptibility in Transgenic Mice with Fragile X Syndrome. Epilepsia, 2000, 41, 19-23. | 2.6 | 265 |
| 5 | Further delineation of the 15q13 microdeletion and duplication syndromes: a clinical spectrum varying from non-pathogenic to a severe outcome. Journal of Medical Genetics, 2009, 46, 511-523. | 1.5 | 250 |
| 6 | Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy. Neurology, 2003, 60, 1961-1967. | 1.5 | 241 |
| 7 | Epilepsy and EEG Findings in Males with Fragile X Syndrome. Epilepsia, 1999, 40, 1092-1099. | 2.6 | 230 |
| 8 | Altered calcium homeostasis in autism-spectrum disorders: evidence from biochemical and genetic studies of the mitochondrial aspartate/glutamate carrier AGC1. Molecular Psychiatry, 2010, 15, 38-52. | 4.1 | 184 |
| 9 | The mismatch negativity and the P3a components of the auditory event-related potentials in autistic low-functioning subjects. Clinical Neurophysiology, 2003, 114, 1671-1680. | 0.7 | 182 |
| 10 | Sleep in children with autistic spectrum disorder: A questionnaire and polysomnographic study. Sleep Medicine, 2007, 9, 64-70. | 0.8 | 169 |
| 11 | Sleep in subjects with autistic disorder: a neurophysiological and psychological study. Brain and Development, 2000, 22, 88-92. | 0.6 | 164 |
| 12 | Association of a Functional Deficit of the BK<sub>Ca</sub>Channel, a Synaptic Regulator of Neuronal Excitability, With Autism and Mental Retardation. American Journal of Psychiatry, 2006, 163, 1622-1629. | 4.0 | 158 |
| 13 | Methodology of photic stimulation revisited: Updated European algorithm for visual stimulation in the EEG laboratory. Epilepsia, 2012, 53, 16-24. | 2.6 | 155 |
| 14 | SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678. | 2.6 | 152 |
| 15 | Clinical, Morphological, and Biochemical Correlates of Head Circumference in Autism. Biological Psychiatry, 2007, 62, 1038-1047. | 0.7 | 131 |
| 16 | Sulphation deficit in â€œlow-functioningâ€•autistic children: a pilot study. Biological Psychiatry, 1999, 46, 420-424. | 0.7 | 123 |
| 17 | Paraoxonase gene variants are associated with autism in North America, but not in Italy: possible regional specificity in geneâ€“environment interactions. Molecular Psychiatry, 2005, 10, 1006-1016. | 4.1 | 115 |
| 18 | An open-label trial of levetiracetam in severe myoclonic epilepsy of infancy. Neurology, 2007, 69, 250-254. | 1.5 | 115 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Myoclonic status in nonprogressive encephalopathies: An update. Epilepsia, 2009, 50, 41-44. | 2.6 | 107 |
| 20 | Cyclic alternating pattern and spectral analysis of heart rate variability during normal sleep. Journal of Sleep Research, 2000, 9, 13-18. | 1.7 | 106 |
| 21 | Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932. | 5.8 | 105 |
| 22 | Sleep phenotypes of intellectual disability: A polysomnographic evaluation in subjects with Down syndrome and Fragile-X syndrome. Clinical Neurophysiology, 2008, 119, 1242-1247. | 0.7 | 97 |
| 23 | Sleep disturbances in Angelman syndrome: a questionnaire study. Brain and Development, 2004, 26, 233-240. | 0.6 | 96 |
| 24 | Association between the HOXA1 A218G polymorphism and increased head circumference in patients with autism. Biological Psychiatry, 2004, 55, 413-419. | 0.7 | 94 |
| 25 | Autism, epilepsy, and synaptopathies: a not rare association. Neurological Sciences, 2017, 38, 1353-1361. | 0.9 | 90 |
| 26 | Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096. | 2.6 | 89 |
| 27 | Principal pathogenetic components and biological endophenotypes in autism spectrum disorders. Autism Research, 2010, 3, 237-252. | 2.1 | 85 |
| 28 | CDKL5 mutations in boys with severe encephalopathy and early-onset intractable epilepsy. Neurology, 2008, 71, 997-999. | 1.5 | 84 |
| 29 | Evaluation of autism traits in Angelman syndrome: a resource to unfold autism genes. Neurogenetics, 2007, 8, 169-178. | 0.7 | 81 |
| 30 | Special Education Versus Inclusive Education: The Role of the TEACCH Program. Journal of Autism and Developmental Disorders, 2009, 39, 874-882. | 1.7 | 75 |
| 31 | Involvement of the PRKCB1 gene in autistic disorder: significant genetic association and reduced neocortical gene expression. Molecular Psychiatry, 2009, 14, 705-718. | 4.1 | 75 |
| 32 | Respiratory patterns during sleep in Down's syndrome: importance of central apnoeas. Journal of Sleep Research, 1997, 6, 134-141. | 1.7 | 71 |
| 33 | Sleep polygraphy in Angelman syndrome. Clinical Neurophysiology, 2004, 115, 938-945. | 0.7 | 71 |
| 34 | A novel KCNQ3 mutation in familial epilepsy with focal seizures and intellectual disability. Epilepsia, 2015, 56, e15-20. | 2.6 | 66 |
| 35 | Management of psychogenic nonâ€epileptic seizures: a multidisciplinary approach. European Journal of Neurology, 2019, 26, 205. | 1.7 | 64 |
| 36 | Autism: evidence of association with adenosine deaminase genetic polymorphism. Neurogenetics, 2001, 3, 111-113. | 0.7 | 62 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | A New Benign Adult Familial Myoclonic Epilepsy (BAFME) Pedigree Suggesting Linkage to Chromosome 2p11.1-q12.2. Epilepsia, 2004, 45, 190-192. | 2.6 | 62 |
| 38 | Heart rate variability during sleep in children with partial epilepsy. Journal of Sleep Research, 2002, 11, 153-160. | 1.7 | 61 |
| 39 | Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322. | 4.9 | 61 |
| 40 | Lacosamide in pediatric and adult patients: Comparison of efficacy and safety. Seizure: the Journal of the British Epilepsy Association, 2013, 22, 210-216. | 0.9 | 60 |
| 41 | Permutation entropy of scalp EEG: A tool to investigate epilepsies. Clinical Neurophysiology, 2014, 125, 13-20. | 0.7 | 59 |
| 42 | Cannabidiol and epilepsy: Rationale and therapeutic potential. Pharmacological Research, 2016, 107, 85-92. | 3.1 | 58 |
| 43 | Epilepsy and fragile X syndrome: A follow-up study. American Journal of Medical Genetics Part A, 1991, 38, 511-513. | 2.4 | 57 |
| 44 | Sleep breathing and periodic leg movement pattern in Angelman Syndrome: A polysomnographic study. Clinical Neurophysiology, 2005, 116, 2685-92. | 0.7 | 56 |
| 45 | Polysomnographic findings in Rett syndrome: a caseâ€“control study. Sleep and Breathing, 2013, 17, 93-98. | 0.9 | 56 |
| 46 | Neurofibromatosis type 1 and infantile spasms. Child's Nervous System, 2009, 25, 211-216. | 0.6 | 55 |
| 47 | Audiogenic seizure susceptibility is reduced in fragile X knockout mice after introduction of FMR1 transgenes. Experimental Neurology, 2007, 203, 233-240. | 2.0 | 54 |
| 48 | Myoclonic Absence-Like Seizures and Chromosome Abnormality Syndromes. Epilepsia, 1998, 39, 660-663. | 2.6 | 52 |
| 49 | The pharmacological management of Lennox-Gastaut syndrome and critical literature review. Seizure: the Journal of the British Epilepsy Association, 2018, 63, 17-25. | 0.9 | 52 |
| 50 | Clinical Correlates of Brain Morphometric Features of Subjects With Low-Functioning Autistic Disorder. Journal of Child Neurology, 2000, 15, 504-508. | 0.7 | 51 |
| 51 | Non-linear EEG measures during sleep: effects of the different sleep stages and cyclic alternating pattern. International Journal of Psychophysiology, 2002, 43, 273-286. | 0.5 | 51 |
| 52 | Case-control and family-based association studies of candidate genes in autistic disorder and its endophenotypes: TPH2 and GLO1. BMC Medical Genetics, 2007, 8, 11. | 2.1 | 51 |
| 53 | A functional polymorphism in the SCN1A gene does not influence antiepileptic drug responsiveness in Italian patients with focal epilepsy. Epilepsia, 2011, 52, e40-e44. | 2.6 | 50 |
| 54 | Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63. | 3.6 | 50 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | The time course of high-frequency bands (15â€“45 Hz) in all-night spectral analysis of sleep EEG. Clinical Neurophysiology, 2000, 111, 1258-1265. | 0.7 | 49 |
| 56 | Familial Cortical Tremor, Epilepsy, and Mental Retardation. Archives of Neurology, 1998, 55, 1569. | 4.9 | 48 |
| 57 | Familial Occurrence of Febrile Seizures and Epilepsy in Severe Myoclonic Epilepsy of Infancy (SMEI) Patients with SCN1A Mutations. Epilepsia, 2006, 47, 1629-1635. | 2.6 | 48 |
| 58 | Heart rate variability and apnea during sleep in Down's syndrome. Journal of Sleep Research, 1998, 7, 282-287. | 1.7 | 46 |
| 59 | Hypersensitivity to Aromatic Anticonvulsants: In Vivo and In Vitro Cross-Reactivity Studies. Current Pharmaceutical Design, 2006, 12, 3373-3381. | 0.9 | 46 |
| 60 | Advances in genetic testing and optimization of clinical management in children and adults with epilepsy. Expert Review of Neurotherapeutics, 2020, 20, 251-269. | 1.4 | 45 |
| 61 | 6q Terminal Deletion Syndrome Associated with a Distinctive EEG and Clinical Pattern: A Report of Five Cases. Epilepsia, 2006, 47, 830-838. | 2.6 | 44 |
| 62 | Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690. | 2.6 | 44 |
| 63 | Seizures and EEG pattern in the 22q13.3 deletion syndrome: Clinical report of six Italian cases. Seizure: the Journal of the British Epilepsy Association, 2014, 23, 774-779. | 0.9 | 42 |
| 64 | Lennoxâ€Gastaut syndrome with lateâ€onset and prominent reflex seizures in trisomy 21 patients. Epilepsia, 2009, 50, 1587-1595. | 2.6 | 40 |
| 65 | An atypical patient with Cowden syndrome and PTEN gene mutation presenting with cortical malformation and focal epilepsy. Brain and Development, 2012, 34, 873-876. | 0.6 | 40 |
| 66 | A nationwide survey of PMM2-CDG in Italy: high frequency of a mild neurological variant associated with the L32R mutation. Journal of Neurology, 2015, 262, 154-164. | 1.8 | 40 |
| 67 | Genetic and forensic implications in epilepsy and cardiac arrhythmias: a case series. International Journal of Legal Medicine, 2015, 129, 495-504. | 1.2 | 40 |
| 68 | Relationship between Delta, Sigma, Beta, and Gamma EEG bands at REM sleep onset and REM sleep end. Clinical Neurophysiology, 2001, 112, 2046-2052. | 0.7 | 39 |
| 69 | The association of rs4307059 and rs35678 markers with autism spectrum disorders is replicated in Italian families. Psychiatric Genetics, 2012, 22, 177-181. | 0.6 | 39 |
| 70 | Long-term Cognitive and Behavioral Therapies, Combined with Augmentative Communication, are Related to Uncinate Fasciculus Integrity in Autism. Journal of Autism and Developmental Disorders, 2012, 42, 585-592. | 1.7 | 39 |
| 71 | Proposal for a Multicenter Study on Epilepsy and Learning Disorders in Children. Epilepsia, 2001, 42, 10-12. | 2.6 | 38 |
| 72 | miRNAs Plasma Profiles in Vascular Dementia: Biomolecular Data and Biomedical Implications. Frontiers in Cellular Neuroscience, 2016, 10, 51. | 1.8 | 38 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Leigh syndrome and partial deficit of cytochrome c oxidase associated with epilepsia partialis continua. Brain and Development, 1996, 18, 207-211. | 0.6 | 37 |
| 74 | Different EEG frequency band synchronization during nocturnal frontal lobe seizures. Clinical Neurophysiology, 2004, 115, 1202-1211. | 0.7 | 35 |
| 75 | Risk factors for unprovoked epileptic seizures in multiple sclerosis: a systematic review and meta-analysis. Neurological Sciences, 2017, 38, 399-406. | 0.9 | 35 |
| 76 | An educational campaign about epilepsy among Italian primary school teachers. 2. The results of a focused training program. Epilepsy and Behavior, 2015, 42, 93-97. | 0.9 | 33 |
| 77 | Denaturing high-performance liquid chromatography of the myotubularin-related 2 gene (MTMR2) in unrelated patients with Charcot-Marie-Tooth disease suggests a low frequency of mutation in inherited neuropathy. Neurogenetics, 2001, 3, 107-109. | 0.7 | 32 |
| 78 | Low penetrance of autosomal dominant lateral temporal epilepsy in Italian families without <scp>LGI</scp>1 mutations. Epilepsia, 2013, 54, 1288-1297. | 2.6 | 32 |
| 79 | Unilateral Eye Blinking Arising From the Ictal Ipsilateral Occipital Area. Clinical EEG and Neuroscience, 2016, 47, 243-246. | 0.9 | 32 |
| 80 | Perampanel tolerability in children and adolescents with focal epilepsy: Effects on behavior and executive functions. Epilepsy and Behavior, 2020, 103, 106879. | 0.9 | 32 |
| 81 | Polysomnographic assessment of sleep disturbances in children with developmental disabilities and seizures. Neurological Sciences, 2010, 31, 575-583. | 0.9 | 31 |
| 82 | Seizures in klinefelterâ€™s syndrome. Pediatric Neurology, 1998, 19, 275-278. | 1.0 | 30 |
| 83 | Effects of repetitive transcranial magnetic stimulation in performing eyeâ€“hand integration tasks: Four preliminary studies with children showing low-functioning autism. Autism, 2014, 18, 638-650. | 2.4 | 30 |
| 84 | Mutation spectrum of NF1 gene in Italian patients with neurofibromatosis type 1 using Ion Torrent PGMâ„¢ platform. European Journal of Medical Genetics, 2017, 60, 93-99. | 0.7 | 30 |
| 85 | Ketogenic Diets in the Treatment of Epilepsy. Current Pharmaceutical Design, 2018, 23, 5691-5701. | 0.9 | 30 |
| 86 | Chaotic behavior of EEG slow-wave activity during sleep. Electroencephalography and Clinical Neurophysiology, 1996, 99, 539-543. | 0.3 | 29 |
| 87 | Enhanced APOE2 transmission rates in families with autistic probands. Psychiatric Genetics, 2004, 14, 73-82. | 0.6 | 29 |
| 88 | Homocysteine is a determinant of ApoA-I and both are associated with ankle brachial index, in an Ambulatory Elderly Population. Atherosclerosis, 2011, 214, 480-485. | 0.4 | 29 |
| 89 | Seizures in Chiari I Malformation: A Clinical and Electroencephalographic Study. Journal of Child Neurology, 1999, 14, 446-450. | 0.7 | 28 |
| 90 | Mild Generalized Epilepsy and Developmental Disorder Associated with Large Inv Dup(15). Epilepsia, 2002, 43, 1096-1100. | 2.6 | 27 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Nonlinear EEG analysis during sleep in premature and full-term newborns. Clinical Neurophysiology, 2003, 114, 1176-1180. | 0.7 | 27 |
| 92 | Expression of multidrug resistance type 1 gene (MDR1) P-glycoprotein in intractable epilepsy with different aetiologies: a double-labelling and electron microscopy study. Neurological Sciences, 2006, 27, 245-251. | 0.9 | 27 |
| 93 | Sleep alterations in children with refractory epileptic encephalopathies: A polysomnographic study. Epilepsy and Behavior, 2014, 35, 50-53. | 0.9 | 27 |
| 94 | Eyelid myoclonia with absences: Electroclinical features and prognostic factors. Epilepsia, 2019, 60, 1104-1113. | 2.6 | 27 |
| 95 | Scalp topographic mapping of middle-latency somatosensory evoked potentials in normal aging and dementia. Neurophysiologie Clinique, 1996, 26, 311-319. | 1.0 | 26 |
| 96 | Non-linear EEG analysis in children with epilepsy and electrical status epilepticus during slow-wave sleep (ESES). Clinical Neurophysiology, 2001, 112, 2274-2280. | 0.7 | 26 |
| 97 | HOXA1gene variants influence head growth rates in humans. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 388-390. | 1.1 | 26 |
| 98 | Autosomal dominant lateral temporal epilepsy: Absence of mutations in ADAM22 and Kv1 channel genes encoding LGI1-associated proteins. Epilepsy Research, 2008, 80, 1-8. | 0.8 | 26 |
| 99 | ALS dysphagia pathophysiology. Neurology, 2013, 80, 616-620. | 1.5 | 26 |
| 100 | Proposal of an Algorithm for Diagnosis and Treatment of Neonatal Seizures in Developing Countries. Epilepsia, 2007, 48, 1158-1164. | 2.6 | 25 |
| 101 | Electroclinical Features and Long-Term Outcome of Cryptogenic Epilepsy in Children with Down Syndrome. Journal of Pediatrics, 2013, 163, 1754-1758. | 0.9 | 25 |
| 102 | Epilepsy in the setting of full trisomy 18: A multicenter study on 18 affected children with and without structural brain abnormalities. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 288-295. | 0.7 | 24 |
| 103 | A de novo heterozygous mutation in KCNC2 gene implicated in severe developmental and epileptic encephalopathy. European Journal of Medical Genetics, 2020, 63, 103848. | 0.7 | 24 |
| 104 | Adjunctive Brivaracetam in Focal Epilepsy: Real-World Evidence from the BRIVAracetam add-on First Italian netwoRk STudy (BRIVAFIRST). CNS Drugs, 2021, 35, 1289-1301. | 2.7 | 24 |
| 105 | Homocysteine predicts increased NT-pro-BNP through impaired fatty acid oxidation. International Journal of Cardiology, 2013, 167, 768-775. | 0.8 | 23 |
| 106 | Validated outcome of treatment changes according to International League Against Epilepsy criteria in adults with drugâ€resistant focal epilepsy. Epilepsia, 2019, 60, 1114-1123. | 2.6 | 23 |
| 107 | Results From an Italian Expanded Access Program on Cannabidiol Treatment in Highly Refractory Dravet Syndrome and Lennoxâ€“Gastaut Syndrome. Frontiers in Neurology, 2021, 12, 673135. | 1.1 | 23 |
| 108 | Transient MRI abnormalities associated with partial status epilepticus: a case report. European Journal of Radiology, 2001, 38, 50-54. | 1.2 | 22 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | â€œPostural firstâ€•principle when balance is challenged in elderly people. International Journal of Neuroscience, 2014, 124, 558-566. | 0.8 | 22 |
| 110 | An educational campaign toward epilepsy among Italian primary school teachers. Epilepsy and Behavior, 2014, 32, 84-91. | 0.9 | 22 |
| 111 | BIT-mapped somatosensory evoked potentials in the fragile X syndrome. Neurophysiologie Clinique, 1994, 24, 413-426. | 1.0 | 21 |
| 112 | Topiramate potentiates the antiseizure activity of some anticonvulsants in DBA/2 mice. European Journal of Pharmacology, 2000, 388, 163-170. | 1.7 | 21 |
| 113 | Felbamate in therapy-resistant epilepsy: an Italian experience. Epilepsy Research, 1996, 25, 249-255. | 0.8 | 20 |
| 114 | No evidence of GABRG2 mutations in severe myoclonic epilepsy of infancy. Epilepsy Research, 2003, 53, 196-200. | 0.8 | 20 |
| 115 | Helicobacter pylori serologic status has no influence on the association between fucosyltransferase 2 polymorphism (FUT2 461 Gâ†'A) and vitamin B-12 in Europe and West Africa. American Journal of Clinical Nutrition, 2012, 95, 514-521. | 2.2 | 20 |
| 116 | The in cis T251I and P587L POLG1 base changes: Description of a new family and literature review. Neuromuscular Disorders, 2015, 25, 333-339. | 0.3 | 20 |
| 117 | Epilepsy and sleep disorders improve in adolescents and adults with Angelman syndrome: A multicenter study on 46 patients. Epilepsy and Behavior, 2017, 75, 225-229. | 0.9 | 20 |
| 118 | Age, sex and mental retardation related changes of brainstem auditory evoked potentials in Down's syndrome. Italian Journal of Neurological Sciences, 1995, 16, 377-383. | 0.1 | 19 |
| 119 | Eyelid myoclonia with absences in three subjects with mental retardation. Neurological Sciences, 2000, 21, 247-250. | 0.9 | 19 |
| 120 | Hypersensitivity to Lamotrigine and Nonaromatic Anticonvulsant Drugs: A Review. Current Pharmaceutical Design, 2008, 14, 2874-2882. | 0.9 | 19 |
| 121 | CDKL5 MUTATIONS IN BOYS WITH SEVERE ENCEPHALOPATHY AND EARLY-ONSET INTRACTABLE EPILEPSY. Neurology, 2009, 73, 77-78. | 1.5 | 19 |
| 122 | Earlyâ€•onset absence epilepsy: <scp>SLC</scp>2<scp>A</scp>1 gene analysis and treatment evolution. European Journal of Neurology, 2013, 20, 856-859. | 1.7 | 19 |
| 123 | A further family with epilepsy, dementia and yellow teeth: the KohlschÃ¼tter syndrome. Brain and Development, 1995, 17, 133-138. | 0.6 | 18 |
| 124 | Lack of association of HOXA1 and HOXB1 mutations and autism in Sicilian (Italian) patients. Molecular Psychiatry, 2003, 8, 716-717. | 4.1 | 18 |
| 125 | Evoked spikes and giant somatosensory evoked potentials in a patient with fragile-X syndrome. Italian Journal of Neurological Sciences, 1994, 15, 365-368. | 0.1 | 17 |
| 126 | Chromosome Abnormalities and Epilepsy. Epilepsia, 2001, 42, 24-27. | 2.6 | 17 |

| # | Article | IF | Citations |
|---|---|---|---|
| 127 | Analysis of LGI1 promoter sequence, PDYN and GABBR1 polymorphisms in sporadic and familial lateral temporal lobe epilepsy. Neuroscience Letters, 2008, 436, 23-26. | 1.0 | 17 |
| 128 | Epilepsy in ring 14 chromosome syndrome. Epilepsy and Behavior, 2012, 25, 585-592. | 0.9 | 17 |
| 129 | Sensory Profiles of Children with Autism Spectrum Disorder with and without Feeding Problems: A Comparative Study in Sicilian Subjects. Brain Sciences, 2020, 10, 336. | 1.1 | 17 |
| 130 | Seizures in Klinefelter's syndrome: A clinical and EEG study of five patients. Italian Journal of Neurological Sciences, 1995, 16, 231-238. | 0.1 | 16 |
| 131 | Polysomnographic abnormalities in patients with vascular cognitive impairment-no dementia. Sleep Medicine, 2013, 14, 1071-1075. | 0.8 | 16 |
| 132 | Correlation dimension of EEG slow-wave activity during sleep in children and young adults. Electroencephalography and Clinical Neurophysiology, 1998, 106, 424-428. | 0.3 | 15 |
| 133 | Trisomy 12p and epilepsy with myoclonic absences. Brain and Development, 1998, 20, 127-130. | 0.6 | 15 |
| 134 | Isolated monolateral neurosensory hearing loss as a rare sign of neuroborreliosis. Neurological Sciences, 2004, 25, 30-33. | 0.9 | 15 |
| 135 | A t(4;9)(q34;p22) Translocation Associated with Partial Epilepsy, Mental Retardation, and Dysmorphism. Epilepsia, 2005, 46, 1322-1324. | 2.6 | 15 |
| 136 | Self-injury in people with intellectual disability and epilepsy: A matched controlled study. Seizure: the Journal of the British Epilepsy Association, 2012, 21, 160-164. | 0.9 | 15 |
| 137 | Age-related changes of cortical excitability in subjects with sleep-enhanced centrotemporal spikes: a somatosensory evoked potential study. Clinical Neurophysiology, 2000, 111, 591-599. | 0.7 | 14 |
| 138 | Clinical dissection of early onset absence epilepsy in children and prognostic implications. Epilepsia, 2013, 54, 1761-1770. | 2.6 | 14 |
| 139 | Exome-Wide Association Study Identifies New Low-Frequency and Rare UGT1A1 Coding Variants and UGT1A6 Coding Variants Influencing Serum Bilirubin in Elderly Subjects. Medicine (United States), 2015, 94, e925. | 0.4 | 14 |
| 140 | Bit-mapped somatosensory evoked potentials in Down's syndrome individuals. Neurophysiologie Clinique, 1994, 24, 357-366. | 1.0 | 13 |
| 141 | Electroclinical findings in four patients with karyotype 47,XYY. Brain and Development, 2011, 33, 384-389. | 0.6 | 13 |
| 142 | Targeted re-sequencing in malformations of cortical development: genotype-phenotype correlations. Seizure: the Journal of the British Epilepsy Association, 2020, 80, 145-152. | 0.9 | 13 |
| 143 | Screening of subtelomeric rearrangements in autistic disorder: Identification of a partial trisomy of 13q34 in a patient bearing a 13q;21p translocation. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 584-590. | 1.1 | 12 |
| 144 | Biological Determinants of Postural Disorders in Elderly Women. International Journal of Neuroscience, 2012, 123, 24-30. | 0.8 | 12 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | Do neurologists agree in diagnosing drug resistance in adults with focal epilepsy?. Epilepsia, 2019, 60, 175-183. | 2.6 | 12 |
| 146 | Mutations in ACTL6B, coding for a subunit of the neuron-specific chromatin remodeling complex nBAF, cause early onset severe developmental and epileptic encephalopathy with brain hypomyelination and cerebellar atrophy. Human Genetics, 2019, 138, 187-198. | 1.8 | 12 |
| 147 | Cognitive, adaptive, and behavioral effects of adjunctive rufinamide in Lennoxâ€“Gastaut syndrome: A prospective observational clinical study. Epilepsy and Behavior, 2020, 112, 107445. | 0.9 | 12 |
| 148 | Severe encephalomyopathy in a patient with homoplasmic A5814G point mutation in mitochondrial tRNACys gene. Neuromuscular Disorders, 2007, 17, 258-261. | 0.3 | 11 |
| 149 | Posterior fossa abnormalities in hereditary spastic paraparesis with spastin mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 80, 440-443. | 0.9 | 11 |
| 150 | Electroclinical findings and long-term outcomes in epileptic patients with inv dup (15). Acta Neurologica Scandinavica, 2018, 137, 575-581. | 1.0 | 11 |
| 151 | Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Journal of the Neurological Sciences, 2021, 424, 117409. | 0.3 | 11 |
| 152 | Cutis verticis gyrata and chromosomal fragile sites. American Journal of Medical Genetics Part A, 1991, 38, 249-250. | 2.4 | 10 |
| 153 | Seizures and epileptiform EEG abnormalities in FRAXE syndrome. Clinical Neurophysiology, 2001, 112, 1954-1955. | 0.7 | 10 |
| 154 | Two-loci ADA haplotypes in autistic disorder. American Journal of Medical Genetics Part A, 2002, 108, 339-340. | 2.4 | 10 |
| 155 | Association study of autistic disorder and chromosome 16p. American Journal of Medical Genetics Part A, 2003, 119A, 242-246. | 2.4 | 10 |
| 156 | Analysis of the gastrinâ€releasing peptide receptor gene in Italian patients with autism spectrum disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 807-813. | 1.1 | 10 |
| 157 | Partial monosomy Xq(Xq23â†'qter) and trisomy 4p(4p15.33â†'pter) in a woman with intractable focal epilepsy, borderline intellectual functioning, and dysmorphic features. Brain and Development, 2008, 30, 425-429. | 0.6 | 10 |
| 158 | The management of epilepsy in clinical practice: Do the needs manifested by the patients correspond to the priorities of the caring physicians? Findings from the EPINEEDS Study. Epilepsy and Behavior, 2020, 102, 106641. | 0.9 | 10 |
| 159 | Tuberous sclerosis and down syndrome: A casual association?. Brain and Development, 1992, 14, 245-248. | 0.6 | 9 |
| 160 | Saethre-Chotzen syndrome: a clinical, EEG and neuroradiological study. Child's Nervous System, 1996, 12, 699-704. | 0.6 | 9 |
| 161 | A New Case of Trichothiodystrophy Associated with Autism, Seizures, and Mental Retardation. Pediatric Dermatology, 1997, 14, 125-128. | 0.5 | 9 |
| 162 | Acrofrontofacionasal dysostosis 1 in two sisters of Indian origin. American Journal of Medical Genetics, Part A, 2011, 155, 3125-3127. | 0.7 | 9 |

| # | Article | IF | Citations |
|---|---|---|---|
| 163 | Association of intronic variants of the KCNAB1 gene with lateral temporal epilepsy. Epilepsy Research, 2011, 94, 110-116. | 0.8 | 9 |
| 164 | Age- and height-dependent changes of amplitude and latency of somatosensory evoked potentials in children and young adults with Down's syndrome. Neurophysiologie Clinique, 1996, 26, 321-327. | 1.0 | 8 |
| 165 | A New Family With Periventricular Nodular Heterotopia and Peculiar Dysmorphic Features. Archives of Neurology, 1997, 54, 61. | 4.9 | 8 |
| 166 | Electroclinical features of epilepsy monosomy 1p36 syndrome and their implications. Acta Neurologica Scandinavica, 2018, 138, 523-530. | 1.0 | 8 |
| 167 | From Cannabis to Cannabidiol to Treat Epilepsy, Where Are We?. Current Pharmaceutical Design, 2017, 22, 6426-6433. | 0.9 | 8 |
| 168 | A case of epidermal nevus syndrome with carotid malformation. Italian Journal of Neurological Sciences, 1990, 11, 293-296. | 0.1 | 7 |
| 169 | Giant somatosensory evoked potentials and pathophysiology of hyperekplexia. Neurophysiological study of one patient. Neurophysiologie Clinique, 1994, 24, 318-324. | 1.0 | 7 |
| 170 | Reflex seizures in a patient with Angelman syndrome and trisomy 21. Neurological Sciences, 2016, 37, 1373-1374. | 0.9 | 7 |
| 171 | Epilepsy in â€œSunflower syndromeâ€‰ electroclinical features, therapeutic response, and long-term follow-up. Seizure: the Journal of the British Epilepsy Association, 2021, 93, 8-12. | 0.9 | 7 |
| 172 | Brainstem auditory evoked potentials in tuberous sclerosis. Italian Journal of Neurological Sciences, 1993, 14, 311-316. | 0.1 | 6 |
| 173 | Does antiepileptic therapy affect immune response?. Journal of Chemotherapy, 1998, 10, 184-186. | 0.7 | 6 |
| 174 | Heart rate variability and autonomic function during sleep in fragile X syndrome. , 1999, 83, 296-297. | | 6 |
| 175 | Somatosensory Evoked Potentials in Patients Affected by Unilateral Cerebrovascular Lesions With Onset During the Perinatal Period or Adulthood. Journal of Child Neurology, 2001, 16, 541-547. | 0.7 | 6 |
| 176 | Peripheral Markers of the Î³-Aminobutyric Acid (GABA)ergic System in Angelman's Syndrome. Journal of Child Neurology, 2003, 18, 21-25. | 0.7 | 6 |
| 177 | Suggestive evidence for association of D2S2188 marker (2q31.1) with autism in 143 Sicilian (Italian) TRIO families. Psychiatric Genetics, 2005, 15, 149-150. | 0.6 | 6 |
| 178 | Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. Neurological Sciences, 2006, 27, 425-431. | 0.9 | 6 |
| 179 | Schimmelpenning Syndrome: A Kind of Craniofacial Epidermal Nevus Associated with Cerebral and Ocular MR Imaging Abnormalities. American Journal of Neuroradiology, 2010, 31, E47-E48. | 1.2 | 6 |
| 180 | Analysis of RBFOX1 gene expression in lymphoblastoid cell lines of Italian discordant autism spectrum disorders sib-pairs. Molecular and Cellular Probes, 2014, 28, 242-245. | 0.9 | 6 |

| # | Article | IF | Citations |
|---|---|---|---|
| 181 | Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. Seizure: the Journal of the British Epilepsy Association, 2021, 88, 60-72. | 0.9 | 6 |
| 182 | Growth hormone deficit in autism. Journal of Autism and Developmental Disorders, 1993, 23, 421-422. | 1.7 | 5 |
| 183 | Novel deletion of the E3A ubiquitin protein ligase gene detected by multiplex ligation-dependent probe amplification in a patient with Angelman syndrome. Experimental and Molecular Medicine, 2010, 42, 842. | 3.2 | 5 |
| 184 | 7q11.23 microduplication syndrome: neurophysiological and neuroradiological insights into a rare chromosomal disorder. Journal of Intellectual Disability Research, 2018, 62, 359-370. | 1.2 | 5 |
| 185 | Determination of Perampanel in Dried Plasma Spots: Applicability to Therapeutic Drug Monitoring. Therapeutic Drug Monitoring, 2020, 42, 309-314. | 1.0 | 5 |
| 186 | Improvements in mealtime behaviors of children with special needs following a day-center-based behavioral intervention for feeding problems. Rivista Di Psichiatria, 2018, 53, 299-308. | 0.6 | 5 |
| 187 | Phosphatase and tensin homolog (PTEN) variants and epilepsy: A multicenter case series. Seizure: the Journal of the British Epilepsy Association, 2022, 100, 82-86. | 0.9 | 5 |
| 188 | An Italian consensus on the management of Lennox-Gastaut syndrome. Seizure: the Journal of the British Epilepsy Association, 2022, 101, 134-140. | 0.9 | 5 |
| 189 | Epileptic seizures and fra(X) syndrome. American Journal of Medical Genetics Part A, 1991, 38, 510-510. | 2.4 | 4 |
| 190 | Relevance of clinical context in the diagnosticâ€therapeutic approach to status epilepticus. Epilepsia, 2016, 57, 1527-1529. | 2.6 | 4 |
| 191 | First-aid management of tonic-clonic seizures among healthcare personnel: A survey by the Apulian section of the Italian League Against Epilepsy. Epilepsy and Behavior, 2018, 80, 321-325. | 0.9 | 4 |
| 192 | Impact of daytime routine modifications on people with severe intellectual disability amid COVIDâ€19 pandemic. Perspectives in Psychiatric Care, 2021, 57, 1536-1537. | 0.9 | 4 |
| 193 | Epilepsy, electroclinical features, and longâ€term outcomes in Pittâ€“Hopkins syndrome due to pathogenic variants in the TCF4 gene. European Journal of Neurology, 2022, 29, 19-25. | 1.7 | 4 |
| 194 | Modulation of the interictal epileptiform EEG activity during sleep: from oscillations to complex dynamics. Neurophysiologie Clinique, 1991, 21, 1-14. | 1.0 | 3 |
| 195 | Emotional facial paralysis in the Rett syndrome. Brain and Development, 1991, 13, 61-62. | 0.6 | 3 |
| 196 | Transient alternating hemichorea as presenting sign of progressive supranuclear palsy. Italian Journal of Neurological Sciences, 1991, 12, 99-101. | 0.1 | 3 |
| 197 | A CASE OF FG SYNDROME WITH GINGIVAL HYPERPLASIA AND KELOIDS. Pediatric Dermatology, 1995, 12, 387-388. | 0.5 | 3 |
| 198 | Brain Morphometry and Psychobehavioural Measures in Autistic Low-Functioning Subjects. The Neuroradiology Journal, 1997, 10, 431-436. | 0.1 | 3 |

| # | Article | IF | Citations |
|---|---|---|---|
| 199 | Associations between folate, vitamin B12, homocysteine and pathologies related to aging: the need to consider complex nutrient-nutrient and gene-nutrient interactions and the functional and socio-economic determinants in population-based studies. Clinical Chemistry and Laboratory Medicine, 2007, 45, 127-9. | 1.4 | 3 |
| 200 | Seizure aggravation caused by antiepileptic drugs in a patient with muscleâ€“eyeâ€“brain disease. Epilepsy and Behavior, 2010, 19, 666-668. | 0.9 | 3 |
| 201 | Connectivity measures suggest a sub-cortical generator of myoclonus in Angelman syndrome. Clinical Neurophysiology, 2019, 130, 2231-2237. | 0.7 | 3 |
| 202 | TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. Acta Neurologica Belgica, 2020, 120, 381-383. | 0.5 | 3 |
| 203 | A validation study of the clinical diagnosis of Dup15q syndrome: Which symptoms matter most?. Seizure: the Journal of the British Epilepsy Association, 2020, 74, 26-30. | 0.9 | 3 |
| 204 | The management of epilepsy in clinical practice: Do the timing and severity of the disease influence the priorities of patients and the caring physicians? Data from the EPINEEDS study. Epilepsy and Behavior, 2021, 114, 107201. | 0.9 | 3 |
| 205 | EEG Patterns in Patients with Praderâ€“Willi Syndrome. Brain Sciences, 2021, 11, 1045. | 1.1 | 3 |
| 206 | Scalp Topographic Distribution of Beta and Gamma Ratios During Sleep. Journal of Psychophysiology, 2002, 16, 107-113. | 0.3 | 3 |
| 207 | Metacognitive and emotional/motivational executive functions in individuals with autism spectrum disorder and attention deficit hyperactivity disorder: preliminary results. Rivista Di Psichiatria, 2016, 51, 104-9. | 0.6 | 3 |
| 208 | Letter to the Editor. Journal of Sleep Research, 1996, 5, 272-272. | 1.7 | 2 |
| 209 | Does a peculiar EEG pattern exist also for FRAXE mental retardation?. Clinical Neurophysiology, 2000, 111, 1632-1636. | 0.7 | 2 |
| 210 | Juvenile myoclonic epilepsy with generalised and focal electroencephalographic abnormalities: a case report with a molecular genetic study. Neurological Sciences, 2007, 28, 276-278. | 0.9 | 2 |
| 211 | Coexistence of mitochondrial and nuclear DNA mutations in a woman with mitochondrial encephalomyopathy and double cortex. Mitochondrion, 2010, 10, 548-554. | 1.6 | 2 |
| 212 | A case of savant syndrome in a child with autism spectrum disorder. International Journal on Disability and Human Development, 2015, 14, . | 0.2 | 2 |
| 213 | Are Mutations in the DHRS9 Gene Causally Linked to Epilepsy? A Case Report. Medicina (Lithuania), 2020, 56, 387. | 0.8 | 2 |
| 214 | The evolution of self-injurious behaviors in people with intellectual disability and epilepsy: A follow-up study. Seizure: the Journal of the British Epilepsy Association, 2020, 82, 99-104. | 0.9 | 2 |
| 215 | Somatosensory evoked potentials and hyplexia outcome. Neurophysiologie Clinique, 1995, 25, 28-30. | 1.0 | 1 |
| 216 | The 9-bp deletion in region V of mtDNA: a risk factor of hearing loss and encephalomyopathy in Caucasian populations?. Neurological Sciences, 2013, 34, 1223-1226. | 0.9 | 1 |

| # | Article | IF | Citations |
|---|---|---|---|
| 217 | Tuberous sclerosis underlying neonatal poliosis. Journal of the European Academy of Dermatology and Venereology, 2015, 29, 822-823. | 1.3 | 1 |
| 218 | Maternally derived 15q11.2â€q13.1 duplication in a child with Lennoxâ€“Gastautâ€type epilepsy and dysmorphic features: Clinicalâ€genetic characterization of the family and review of the literature. American Journal of Medical Genetics, Part A, 2017, 173, 556-560. | 0.7 | 1 |
| 219 | Early results from a combined low-intensive psychoeducational intervention for preschoolers with autism spectrum disorder. Disability and Rehabilitation, 2020, 42, 1275-1283. | 0.9 | 1 |
| 220 | Electroclinical Features of Epilepsy in Mucopolysaccharidosis III: Outcome Description in a Cohort of 15 Italian Patients. Frontiers in Neurology, 2021, 12, 705423. | 1.1 | 1 |
| 221 | Somatosensory Evoked Potentials in Patients Affected by Unilateral Cerebrovascular Lesions With Onset During the Perinatal Period or Adulthood. Journal of Child Neurology, 2001, 16, 541. | 0.7 | 1 |
| 222 | Exome sequencing in a child with neurodevelopmental disorder and epilepsy: Variant analysis of the <scp>AHNAK2</scp> gene. Molecular Genetics & Genomic Medicine, 0, , . | 0.6 | 1 |
| 223 | Congenital cerebellar ataxia, mental retardation, and atrophic retinal lesions in two brothers.. Journal of Neurology, Neurosurgery and Psychiatry, 1996, 61, 424-425. | 0.9 | 0 |
| 224 | Sleep Apnea in a patient with Becker Muscular Dystrophy. Italian Journal of Neurological Sciences, 1997, 18, 59-60. | 0.1 | 0 |
| 225 | Anomalie della migrazione neuronale e dell'organizzazione corticale nell'epilessia. The Neuroradiology Journal, 1998, 11, 443-452. | 0.1 | 0 |
| 226 | Facial and Skeletal Malformations, Mental Retardation, Aganglionosis, and Neurogenic Muscle Weakness: A Variant of Niikawa-Kuroki Syndrome or a New Syndrome?. Journal of Child Neurology, 2001, 16, 296-296. | 0.7 | 0 |
| 227 | M.P.1.05 The COII/tRNALys intergenic 9-bp deletion in mtDNA: A new possible cause of sensorineural hearing loss?. Neuromuscular Disorders, 2007, 17, 769. | 0.3 | 0 |
| 228 | M.P.1.11 Homoplasmic point mutations in mitochondrial tRNA genes in patients with encephalomyopathy. Neuromuscular Disorders, 2007, 17, 770-771. | 0.3 | 0 |
| 229 | Transient Brain Lesions in Neuro-BehÃ§et's Disease without Systemic Involvement. Neuroradiology Journal, 2012, 25, 319-324. | 0.6 | 0 |
| 230 | Copy Number Variants and Epilepsy: New Emerging Syndromes. , 2015, , 1-14. | | 0 |
| 231 | Chromosomal Abnormalities and Cortical Malformations. , 2019, , 547-585. | | 0 |
| 232 | Non-convulsive Status Epilepticus and Frontal Lobe Seizures in a Patient with a Chromosome Abnormality. , 2008, , 69-72. | | 0 |
| 233 | Letter to the Editor Regarding the Article â€œWhole-Exome Sequencing in NF1-Related West's Syndrome Leads to the Identification of KCNC2 as a Novel Candidate Gene for Epilepsyâ€. Neuropediatrics, 2021, 52, 153-153. | 0.3 | 0 |