Maurizio Elia

List of Publications by Year
in descending order

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233 papers	9,037 citations	47409 49 h-index	64407 83 g-index

241 all docs	241 docs citations	241 times ranked	11034 citing authors

#	Article	IF	Citations
1	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
2	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
3	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
4	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
5	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
6	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
7	Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Journal of the Neurological Sciences, 2021, 424, 117409.	0.3	11
8	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
9	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
10	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
11	EEG Patterns in Patients with Praderâ€“Willi Syndrome. Brain Sciences, 2021, 11, 1045.	1.1	3
12	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
13	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
14	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
15	TBC1D24 gene mRNA expression in a boy with early infantile epileptic encephalopathy-16. Acta Neurologica Belgica, 2020, 120, 381-383.	0.5	3
16	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
17	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
18	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

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19	Determination of Perampanel in Dried Plasma Spots: Applicability to Therapeutic Drug Monitoring. Therapeutic Drug Monitoring, 2020, 42, 309-314.	1.0	5
20	Are Mutations in the DHRS9 Gene Causally Linked to Epilepsy? A Case Report. Medicina (Lithuania), 2020, 56, 387.	0.8	2
21	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
22	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
23	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
24	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
25	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
26	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
27	Perampanel tolerability in children and adolescents with focal epilepsy: Effects on behavior and executive functions. Epilepsy and Behavior, 2020, 103, 106879.	0.9	32
28	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
29	Chromosomal Abnormalities and Cortical Malformations. , 2019, , 547-585.		0
30	Eyelid myoclonia with absences: Electroclinical features and prognostic factors. Epilepsia, 2019, 60, 1104-1113.	2.6	27
31	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
32	Connectivity measures suggest a sub-cortical generator of myoclonus in Angelman syndrome. Clinical Neurophysiology, 2019, 130, 2231-2237.	0.7	3
33	Do neurologists agree in diagnosing drug resistance in adults with focal epilepsy?. Epilepsia, 2019, 60, 175-183.	2.6	12
34	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
35	Management of psychogenic nonâ€epileptic seizures: a multidisciplinary approach. European Journal of Neurology, 2019, 26, 205.	1.7	64
36	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

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37	Electroclinical findings and long-term outcomes in epileptic patients with inv dup (15). Acta Neurologica Scandinavica, 2018, 137, 575-581.	1.0	11
38	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
39	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
40	Electroclinical features of epilepsy monosomy 1p36 syndrome and their implications. Acta Neurologica Scandinavica, 2018, 138, 523-530.	1.0	8
41	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
42	Ketogenic Diets in the Treatment of Epilepsy. Current Pharmaceutical Design, 2018, 23, 5691-5701.	0.9	30
43	Risk factors for unprovoked epileptic seizures in multiple sclerosis: a systematic review and meta-analysis. Neurological Sciences, 2017, 38, 399-406.	0.9	35
44	Autism, epilepsy, and synaptopathies: a not rare association. Neurological Sciences, 2017, 38, 1353-1361.	0.9	90
45	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
46	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
47	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
48	From Cannabis to Cannabidiol to Treat Epilepsy, Where Are We?. Current Pharmaceutical Design, 2017, 22, 6426-6433.	0.9	8
49	Unilateral Eye Blinking Arising From the Ictal Ipsilateral Occipital Area. Clinical EEG and Neuroscience, 2016, 47, 243-246.	0.9	32
50	miRNAs Plasma Profiles in Vascular Dementia: Biomolecular Data and Biomedical Implications. Frontiers in Cellular Neuroscience, 2016, 10, 51.	1.8	38
51	Relevance of clinical context in the diagnosticâ€therapeutic approach to status epilepticus. Epilepsia, 2016, 57, 1527-1529.	2.6	4
52	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
53	Reflex seizures in a patient with Angelman syndrome and trisomy 21. Neurological Sciences, 2016, 37, 1373-1374.	0.9	7
54	Cannabidiol and epilepsy: Rationale and therapeutic potential. Pharmacological Research, 2016, 107, 85-92.	3.1	58

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55	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
56	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
57	A novel KCNQ3 mutation in familial epilepsy with focal seizures and intellectual disability. Epilepsia, 2015, 56, e15-20.	2.6	66
58	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
59	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
60	Tuberous sclerosis underlying neonatal poliosis. Journal of the European Academy of Dermatology and Venereology, 2015, 29, 822-823.	1.3	1
61	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
62	A case of savant syndrome in a child with autism spectrum disorder. International Journal on Disability and Human Development, 2015, 14, .	0.2	2
63	Copy Number Variants and Epilepsy: New Emerging Syndromes. , 2015, , 1-14.		0
64	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
65	Genetic and forensic implications in epilepsy and cardiac arrhythmias: a case series. International Journal of Legal Medicine, 2015, 129, 495-504.	1.2	40
66	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
67	â€œPostural firstâ€•principle when balance is challenged in elderly people. International Journal of Neuroscience, 2014, 124, 558-566.	0.8	22
68	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
69	Permutation entropy of scalp EEG: A tool to investigate epilepsies. Clinical Neurophysiology, 2014, 125, 13-20.	0.7	59
70	An educational campaign toward epilepsy among Italian primary school teachers. Epilepsy and Behavior, 2014, 32, 84-91.	0.9	22
71	Sleep alterations in children with refractory epileptic encephalopathies: A polysomnographic study. Epilepsy and Behavior, 2014, 35, 50-53.	0.9	27
72	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

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73	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
74	Polysomnographic findings in Rett syndrome: a caseâ€“control study. Sleep and Breathing, 2013, 17, 93-98.	0.9	56
75	Polysomnographic abnormalities in patients with vascular cognitive impairment-no dementia. Sleep Medicine, 2013, 14, 1071-1075.	0.8	16
76	Homocysteine predicts increased NT-pro-BNP through impaired fatty acid oxidation. International Journal of Cardiology, 2013, 167, 768-775.	0.8	23
77	Electroclinical Features and Long-Term Outcome of Cryptogenic Epilepsy in Children with Down Syndrome. Journal of Pediatrics, 2013, 163, 1754-1758.	0.9	25
78	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
79	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
80	ALS dysphagia pathophysiology. Neurology, 2013, 80, 616-620.	1.5	26
81	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
82	Clinical dissection of early onset absence epilepsy in children and prognostic implications. Epilepsia, 2013, 54, 1761-1770.	2.6	14
83	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
84	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.9	61
85	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
86	Transient Brain Lesions in Neuro-BehÃ§et's Disease without Systemic Involvement. Neuroradiology Journal, 2012, 25, 319-324.	0.6	0
87	Epilepsy in ring 14 chromosome syndrome. Epilepsy and Behavior, 2012, 25, 585-592.	0.9	17
88	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
89	Biological Determinants of Postural Disorders in Elderly Women. International Journal of Neuroscience, 2012, 123, 24-30.	0.8	12
90	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

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91	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
92	Methodology of photic stimulation revisited: Updated European algorithm for visual stimulation in the EEG laboratory. Epilepsia, 2012, 53, 16-24.	2.6	155
93	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
94	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
95	Electroclinical findings in four patients with karyotype 47,XYY. Brain and Development, 2011, 33, 384-389.	0.6	13
96	Acrofrontofacionasal dysostosis 1 in two sisters of Indian origin. American Journal of Medical Genetics, Part A, 2011, 155, 3125-3127.	0.7	9
97	Association of intronic variants of the KCNAB1 gene with lateral temporal epilepsy. Epilepsy Research, 2011, 94, 110-116.	0.8	9
98	Principal pathogenetic components and biological endophenotypes in autism spectrum disorders. Autism Research, 2010, 3, 237-252.	2.1	85
99	Polysomnographic assessment of sleep disturbances in children with developmental disabilities and seizures. Neurological Sciences, 2010, 31, 575-583.	0.9	31
100	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
101	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
102	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
103	Coexistence of mitochondrial and nuclear DNA mutations in a woman with mitochondrial encephalomyopathy and double cortex. Mitochondrion, 2010, 10, 548-554.	1.6	2
104	Seizure aggravation caused by antiepileptic drugs in a patient with muscleâ€“eyeâ€“brain disease. Epilepsy and Behavior, 2010, 19, 666-668.	0.9	3
105	Neurofibromatosis type 1 and infantile spasms. Child's Nervous System, 2009, 25, 211-216.	0.6	55
106	Special Education Versus Inclusive Education: The Role of the TEACCH Program. Journal of Autism and Developmental Disorders, 2009, 39, 874-882.	1.7	75
107	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
108	Lennoxâ€Gastaut syndrome with lateâ€onset and prominent reflex seizures in trisomy 21 patients. Epilepsia, 2009, 50, 1587-1595.	2.6	40

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109	SCN1A duplications and deletions detected in Dravet syndrome: Implications for molecular diagnosis. Epilepsia, 2009, 50, 1670-1678.	2.6	152
110	Myoclonic status in nonprogressive encephalopathies: An update. Epilepsia, 2009, 50, 41-44.	2.6	107
111	CDKL5 MUTATIONS IN BOYS WITH SEVERE ENCEPHALOPATHY AND EARLY-ONSET INTRACTABLE EPILEPSY. Neurology, 2009, 73, 77-78.	1.5	19
112	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
113	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
114	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
115	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
116	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
117	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
118	Posterior fossa abnormalities in hereditary spastic paraparesis with spastin mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 80, 440-443.	0.9	11
119	CDKL5 mutations in boys with severe encephalopathy and early-onset intractable epilepsy. Neurology, 2008, 71, 997-999.	1.5	84
120	Hypersensitivity to Lamotrigine and Nonaromatic Anticonvulsant Drugs: A Review. Current Pharmaceutical Design, 2008, 14, 2874-2882.	0.9	19
121	Non-convulsive Status Epilepticus and Frontal Lobe Seizures in a Patient with a Chromosome Abnormality. , 2008, , 69-72.		0
122	An open-label trial of levetiracetam in severe myoclonic epilepsy of infancy. Neurology, 2007, 69, 250-254.	1.5	115
123	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
124	Sleep in children with autistic spectrum disorder: A questionnaire and polysomnographic study. Sleep Medicine, 2007, 9, 64-70.	0.8	169
125	Severe encephalomyopathy in a patient with homoplasmic A5814G point mutation in mitochondrial tRNACys gene. Neuromuscular Disorders, 2007, 17, 258-261.	0.3	11
126	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

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127	M.P.1.11 Homoplasmic point mutations in mitochondrial tRNA genes in patients with encephalomyopathy. Neuromuscular Disorders, 2007, 17, 770-771.	0.3	0
128	Audiogenic seizure susceptibility is reduced in fragile X knockout mice after introduction of FMR1 transgenes. Experimental Neurology, 2007, 203, 233-240.	2.0	54
129	Clinical, Morphological, and Biochemical Correlates of Head Circumference in Autism. Biological Psychiatry, 2007, 62, 1038-1047.	0.7	131
130	HOXA1gene variants influence head growth rates in humans. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 388-390.	1.1	26
131	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
132	Proposal of an Algorithm for Diagnosis and Treatment of Neonatal Seizures in Developing Countries. Epilepsia, 2007, 48, 1158-1164.	2.6	25
133	Brain MRI Findings in Severe Myoclonic Epilepsy in Infancy and Genotype?Phenotype Correlations. Epilepsia, 2007, 48, 1092-1096.	2.6	89
134	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	2.6	44
135	Evaluation of autism traits in Angelman syndrome: a resource to unfold autism genes. Neurogenetics, 2007, 8, 169-178.	0.7	81
136	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
137	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
138	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
139	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
140	Nail aplasia, microcephaly, severe mental retardation and MRI abnormalities: report of two unrelated cases. Neurological Sciences, 2006, 27, 425-431.	0.9	6
141	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
142	Association of a Functional Deficit of the BK_{Ca}Channel, a Synaptic Regulator of Neuronal Excitability, With Autism and Mental Retardation. American Journal of Psychiatry, 2006, 163, 1622-1629.	4.0	158
143	Hypersensitivity to Aromatic Anticonvulsants: In Vivo and In Vitro Cross-Reactivity Studies. Current Pharmaceutical Design, 2006, 12, 3373-3381.	0.9	46
144	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

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145	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
146	A t(4;9)(q34;p22) Translocation Associated with Partial Epilepsy, Mental Retardation, and Dysmorphism. Epilepsia, 2005, 46, 1322-1324.	2.6	15
147	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
148	Sleep breathing and periodic leg movement pattern in Angelman Syndrome: A polysomnographic study. Clinical Neurophysiology, 2005, 116, 2685-92.	0.7	56
149	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
150	Sleep disturbances in Angelman syndrome: a questionnaire study. Brain and Development, 2004, 26, 233-240.	0.6	96
151	Isolated monolateral neurosensory hearing loss as a rare sign of neuroborreliosis. Neurological Sciences, 2004, 25, 30-33.	0.9	15
152	Association between the HOXA1 A218G polymorphism and increased head circumference in patients with autism. Biological Psychiatry, 2004, 55, 413-419.	0.7	94
153	Sleep polygraphy in Angelman syndrome. Clinical Neurophysiology, 2004, 115, 938-945.	0.7	71
154	Different EEG frequency band synchronization during nocturnal frontal lobe seizures. Clinical Neurophysiology, 2004, 115, 1202-1211.	0.7	35
155	Enhanced APOE2 transmission rates in families with autistic probands. Psychiatric Genetics, 2004, 14, 73-82.	0.6	29
156	Association study of autistic disorder and chromosome 16p. American Journal of Medical Genetics Part A, 2003, 119A, 242-246.	2.4	10
157	No evidence of GABRG2 mutations in severe myoclonic epilepsy of infancy. Epilepsy Research, 2003, 53, 196-200.	0.8	20
158	Lack of association of HOXA1 and HOXB1 mutations and autism in Sicilian (Italian) patients. Molecular Psychiatry, 2003, 8, 716-717.	4.1	18
159	Mutations in NHLRC1 cause progressive myoclonus epilepsy. Nature Genetics, 2003, 35, 125-127.	9.4	294
160	Nonlinear EEG analysis during sleep in premature and full-term newborns. Clinical Neurophysiology, 2003, 114, 1176-1180.	0.7	27
161	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
162	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

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163	Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy. Neurology, 2003, 60, 1961-1967.	1.5	241
164	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
165	Two-loci ADA haplotypes in autistic disorder. American Journal of Medical Genetics Part A, 2002, 108, 339-340.	2.4	10
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