Aglaia Vignoli

List of Publications by Year
in descending order

113 papers	2,800 citations	218677 26 h-index	254184 43 g-index
113 all docs	113 docs citations	113 times ranked	3547 citing authors

#	Article	IF	Citations
1	Malformations in Offspring of Women with Epilepsy: A Prospective Study. Epilepsia, 1999, 40, 1231-1236.	5.1	181
2	Chromosome 20 Ring: A Chromosomal Disorder Associated with a Particular Electroclinical Pattern. Epilepsia, 1998, 39, 942-951.	5.1	137
3	Frontal cognitive dysfunction in juvenile myoclonic epilepsy. Epilepsia, 2008, 49, 657-662.	5.1	125
4	Ring chromosome 20 syndrome: A link between epilepsy onset and neuropsychological impairment in three children. Epilepsia, 2009, 50, 2420-2427.	5.1	105
5	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. BMC Medical Genetics, 2010, 11, 146.	2.1	86
6	Autism spectrum disorder in tuberous sclerosis complex: searching for risk markers. Orphanet Journal of Rare Diseases, 2015, 10, 154.	2.7	74
7	Epilepsy in Rett syndrome: Clinical and genetic features. Epilepsy and Behavior, 2010, 19, 296-300.	1.7	68
8	The visual system in eyelid myoclonia with absences. Annals of Neurology, 2014, 76, 412-427.	5.3	68
9	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	10.2	67
10	Epilepsy in <scp>TSC</scp>: Certain etiology does not mean certain prognosis. Epilepsia, 2013, 54, 2134-2142.	5.1	66
11	Rett Syndrome: A Focus on Gut Microbiota. International Journal of Molecular Sciences, 2017, 18, 344.	4.1	63
12	Early diagnosis of tuberous sclerosis complex: a race against time. How to make the diagnosis before seizures?. Orphanet Journal of Rare Diseases, 2018, 13, 25.	2.7	60
13	Patients with epilepsy and patients with psychogenic non-epileptic seizures: Video-EEG, clinical and neuropsychological evaluation. Seizure: the Journal of the British Epilepsy Association, 2011, 20, 706-710.	2.0	56
14	Intrauterine growth in the offspring of epileptic women: a prospective multicenter study. Epilepsy Research, 1999, 36, 53-60.	1.6	55
15	Laryngeal motility alteration: A missing link between sleep apnea and vagus nerve stimulation for epilepsy. Epilepsia, 2016, 57, e24-7.	5.1	51
16	Clinical course and variability of non-Rasmussen, nonstroke motor and sensory epilepsia partialis continua: A European survey and analysis of 65 cases. Epilepsia, 2011, 52, 1168-1176.	5.1	50
17	Correlations between neurophysiological, behavioral, and cognitive function in Rett syndrome. Epilepsy and Behavior, 2010, 17, 489-496.	1.7	47
18	Epilepsy in Rett syndromeâ€"Lessons from the Rett networked database. Epilepsia, 2015, 56, 569-576.	5.1	47

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19	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	5.1	44
20	Ring chromosome 20. European Journal of Medical Genetics, 2012, 55, 381-387.	1.3	40
21	Focal epilepsies in adult patients attending two epilepsy centers: Classification of drugâ€resistance, assessment of risk factors, and usefulness of â€œnewâ€antiepileptic drugs. Epilepsia, 2012, 53, 733-740.	5.1	39
22	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. Journal of Clinical Immunology, 2015, 35, 168-181.	3.8	35
23	Current concepts on epilepsy management in tuberous sclerosis complex. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 299-308.	1.6	35
24	Electroclinical pattern in MECP2 duplication syndrome: Eight new reported cases and review of literature. Epilepsia, 2012, 53, 1146-1155.	5.1	34
25	An HLA-Gâ^—14bp insertion/deletion polymorphism associates with the development of autistic spectrum disorders. Brain, Behavior, and Immunity, 2015, 44, 207-212.	4.1	32
26	The hyperkinetic movement disorder of <scp>FOXG</scp>1â€related epilepticâ€“dyskinetic encephalopathy. Developmental Medicine and Child Neurology, 2016, 58, 93-97.	2.1	32
27	Vagus nerve stimulation in refractory epilepsy: New indications and outcome assessment. Epilepsy and Behavior, 2013, 28, 374-378.	1.7	31
28	Epilepsy in ring chromosome 20 syndrome. Epilepsy Research, 2016, 128, 83-93.	1.6	30
29	Pathogenic Variants in STXBP1 and in Genes for GABAa Receptor Subunities Cause Atypical Rett/Rett-like Phenotypes. International Journal of Molecular Sciences, 2019, 20, 3621.	4.1	29
30	Do patients with tuberous sclerosis complex have an increased risk for malignancies?. American Journal of Medical Genetics, Part A, 2016, 170, 1538-1544.	1.2	28
31	Medical care of adolescents and women with Rett syndrome: An Italian study. American Journal of Medical Genetics, Part A, 2012, 158A, 13-18.	1.2	26
32	Deep phenotyping of patients with Tuberous Sclerosis Complex and no mutation identified in TSC1 and TSC2. European Journal of Medical Genetics, 2018, 61, 403-410.	1.3	25
33	Rett Syndrome and Other Neurodevelopmental Disorders Share Common Changes in Gut Microbial Community: A Descriptive Review. International Journal of Molecular Sciences, 2019, 20, 4160.	4.1	25
34	Sleep-related hypermotor epilepsy (SHE): Contribution of known genes in 103 patients. Seizure: the Journal of the British Epilepsy Association, 2020, 74, 60-64.	2.0	25
35	Effectiveness and tolerability of antiepileptic drugs in 104 girls with Rett syndrome. Epilepsy and Behavior, 2017, 66, 27-33.	1.7	24
36	Healthcare transition from childhood to adulthood in Tuberous Sclerosis Complex. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2018, 178, 355-364.	1.6	24

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37	Epilepsy in adult patients with Down syndrome: a clinicalâ€video EEG study. Epileptic Disorders, 2011, 13, 125-132.	1.3	23
38	Hot water epilepsy and <scp>SYN</scp>1 variants. Epilepsia, 2018, 59, 2162-2163.	5.1	23
39	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	1.6	23
40	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.1	23
41	Low frequency mu-like activity characterizes cortical rhythms in epilepsy due to ring chromosome 20. Clinical Neurophysiology, 2014, 125, 239-249.	1.5	21
42	Lennoxâ€“Gastaut syndrome in adulthood: Long-term clinical follow-up of 38 patients and analysis of their recorded seizures. Epilepsy and Behavior, 2017, 77, 73-78.	1.7	21
43	Felbamate in refractory partial epilepsy. Epilepsy Research, 1999, 34, 43-48.	1.6	20
44	Another case of reversibility of visual-field defect induced by vigabatrin monotherapy: is young age a favorable factor?. Neurological Sciences, 2000, 21, 185-186.	1.9	20
45	Sudden unexpected death in epilepsy (SUDEP) disclosure in pediatric epilepsy: An Italian survey on â€œto tell or not to tellâ€. Epilepsy and Behavior, 2017, 67, 33-38.	1.7	20
46	Rett networked database: An integrated clinical and genetic network of rett syndrome databases. Human Mutation, 2012, 33, 1031-1036.	2.5	19
47	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.	3.3	19
48	Effects of chronic administration of valproic acid to epileptic patients on coagulation tests and primary hemostasis. Epilepsia, 2015, 56, e49-52.	5.1	19
49	Effects of Combined Transcranial Direct Current Stimulation with Cognitive Training in Girls with Rett Syndrome. Brain Sciences, 2020, 10, 276.	2.3	18
50	Worsening in seizure frequency and severity in relation to folic acid administration. European Journal of Neurology, 1998, 5, 301-303.	3.3	17
51	KMT2A: Umbrella Gene for Multiple Diseases. Genes, 2022, 13, 514.	2.4	17
52	Aggressive behavior and epilepsy: A multicenter study. Epilepsia, 2012, 53, e174-9.	5.1	16
53	Vaccination and Occurrence of Seizures in SCN1A Mutationâ€“Positive Patients: A Multicenter Italian Study. Pediatric Neurology, 2014, 50, 228-232.	2.1	16
54	A clinical and genetic study of 33 new cases with early-onset absence epilepsy. Epilepsy Research, 2011, 95, 221-226.	1.6	15

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55	Epilepsyâ€related brain networks in ring chromosome 20 syndrome: An EEGâ€<scp>fMRI</scp> study. Epilepsia, 2014, 55, 403-413.	5.1	15
56	Long-term outcome of epilepsy with onset in the first three years of life: Findings from a large cohort of patients. European Journal of Paediatric Neurology, 2016, 20, 566-572.	1.6	15
57	Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. Annals of Clinical and Translational Neurology, 2019, 6, 475-485.	3.7	15
58	Interstitial 6q microdeletion syndrome and epilepsy: A new patient and review of the literature. American Journal of Medical Genetics, Part A, 2013, 161, 2009-2015.	1.2	14
59	Clinical dissection of early onset absence epilepsy in children and prognostic implications. Epilepsia, 2013, 54, 1761-1770.	5.1	14
60	Dramatic relapse of seizures after everolimus withdrawal. European Journal of Paediatric Neurology, 2018, 22, 203-206.	1.6	14
61	Mapping the Effect of Interictal Epileptic Activity Density During Wakefulness on Brain Functioning in Focal Childhood Epilepsies With Centrotemporal Spikes. Frontiers in Neurology, 2019, 10, 1316.	2.4	14
62	Tuberous sclerosis complex (TSC), lymphangioleiomyomatosis, and COVID â€19: The experience of a TSC clinic in Italy. American Journal of Medical Genetics, Part A, 2020, 182, 2479-2485.	1.2	14
63	Ring Chromosome 20 Syndrome: Genetics, Clinical Characteristics, and Overlapping Phenotypes. Frontiers in Neurology, 2020, 11, 613035.	2.4	14
64	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	1.6	14
65	Evolution of stereotypies in adolescents and women with Rett syndrome. Movement Disorders, 2009, 24, 1379-1383.	3.9	13
66	Glioblastoma multiforme in a child with tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2015, 167, 2388-2393.	1.2	13
67	Antiepileptic drugs in Rett Syndrome. European Journal of Paediatric Neurology, 2015, 19, 446-452.	1.6	13
68	Antiepileptic drug use in Italian children over a decade. European Journal of Clinical Pharmacology, 2017, 73, 241-248.	1.9	13
69	Ictal signs in tuberous sclerosis complex: Clinical and video-EEG features in a large series of recorded seizures. Epilepsy and Behavior, 2018, 85, 14-20.	1.7	13
70	Ictal involvement of the nigrostriatal system in subtle seizures of ring chromosome 20 epilepsy. Epilepsia, 2012, 53, e156-60.	5.1	12
71	Early onset absence epilepsy with onset in the first year of life: A multicenter cohort study. Epilepsia, 2013, 54, 66-69.	5.1	12
72	The TAND checklist: a useful screening tool in children with tuberous sclerosis and neurofibromatosis type 1. Orphanet Journal of Rare Diseases, 2020, 15, 237.	2.7	12

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73	Missense variants in the Arg206 residue of HNRNPH2 : Further evidence of causality and expansion of the phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 823-828.	1.2	12
74	Seizure outcome after epilepsy surgery in tuberous sclerosis complex: Results and analysis of predictors from a multicenter study. Journal of the Neurological Sciences, 2021, 427, 117506.	0.6	12
75	Ictal EEG patterns in epilepsy with centro-temporal spikes. Brain and Development, 2011, 33, 301-309.	1.1	11
76	Emerging neuroimaging contribution to the diagnosis and management of the ring chromosome 20 syndrome. Epilepsy and Behavior, 2015, 45, 155-163.	1.7	11
77	Blood oxidative stress and metallothionein expression in Rett syndrome: Probing for markers. World Journal of Biological Psychiatry, 2016, 17, 198-209.	2.6	11
78	Sleep disturbances in Italian children and adolescents with epilepsy: A questionnaire study. Epilepsy and Behavior, 2020, 106, 107014.	1.7	11
79	Panayiotopoulos syndrome with convulsive status epilepticus at the onset: A long-term study. Seizure: the Journal of the British Epilepsy Association, 2014, 23, 728-731.	2.0	10
80	Extrastriate visual cortex in idiopathic occipital epilepsies: The contribution of retinotopic areas to spike generation. Epilepsia, 2016, 57, 896-906.	5.1	10
81	The role of laryngeal electromyography in vagus nerve stimulation-related vocal fold dysmotility. European Archives of Oto-Rhino-Laryngology, 2017, 274, 1585-1589.	1.6	10
82	In vivo magnetic resonance spectroscopy in the brain of Cdkl5 null mice reveals a metabolic profile indicative of mitochondrial dysfunctions. Journal of Neurochemistry, 2021, 157, 1253-1269.	3.9	10
83	7p22.1 microduplication syndrome: Clinical and molecular characterization of an adult case and review of the literature. European Journal of Medical Genetics, 2015, 58, 578-583.	1.3	9
84	Symptomatic and presumed symptomatic focal epilepsies in childhood: An observational, prospective multicentre study. Epilepsia, 2016, 57, 1808-1816.	5.1	9
85	Genetic and imaging features of cerebellar abnormalities in tuberous sclerosis complex: more insights into their pathogenesis. Developmental Medicine and Child Neurology, 2018, 60, 724-725.	2.1	9
86	Cerebellar lesions as potential predictors of neurobehavioural phenotype in tuberous sclerosis complex. Developmental Medicine and Child Neurology, 2019, 61, 1221-1228.	2.1	9
87	Is Gut Microbiota a Key Player in Epilepsy Onset? A Longitudinal Study in Drug-Naive Children. Frontiers in Cellular and Infection Microbiology, 2021, 11, 749509.	3.9	9
88	Sleep in ring chromosome 20 syndrome: a peculiar electroencephalographic pattern. Functional Neurology, 2013, 28, 47-53.	1.3	8
89	De novo ARHGEF9 missense variants associated with neurodevelopmental disorder in females: expanding the genotypic and phenotypic spectrum of ARHGEF9 disease in females. Neurogenetics, 2021, 22, 87-94.	1.4	7
90	Sleep and behavior in children and adolescents with tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2021, 185, 1421-1429.	1.2	7

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91	Chromatin Imbalance as the Vertex Between Fetal Valproate Syndrome and Chromatinopathies. Frontiers in Cell and Developmental Biology, 2021, 9, 654467.	3.7	7
92	Myoclonic epilepsy with photosensitivity in infants with Pallister-Killian Syndrome. European Journal of Paediatric Neurology, 2019, 23, 653-656.	1.6	6
93	Phenotypes in adult patients with Rett syndrome: results of a 13-year experience and insights into healthcare transition. Journal of Medical Genetics, 2022, 59, 39-45.	3.2	6
94	Epilepsy in adult patients with tuberous sclerosis complex. Acta Neurologica Scandinavica, 2021, 144, 29-40.	2.1	6
95	Rhinencephalon changes in tuberous sclerosis complex. Neuroradiology, 2018, 60, 813-820.	2.2	5
96	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546.	1.2	5
97	PIGWâ€related glycosylphosphatidylinositol deficiency: Description of a new patient and review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 1477-1482.	1.2	5
98	SLC35F1 as a candidate gene for neurodevelopmental disorders resembling Rett syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2238-2240.	1.2	5
99	Further delineation and long-term evolution of electroclinical phenotype in Mowat Wilson Syndrome. A longitudinal study in 40 individuals. Epilepsy and Behavior, 2021, 124, 108315.	1.7	5
100	Perinatal outcome and healthcare resource utilization in the first year of life after antiepileptic exposure during pregnancy. Epilepsy and Behavior, 2019, 92, 14-17.	1.7	4
101	Brivaracetam as add-on treatment in patients with post-stroke epilepsy: real-world data from the BRIVAracetam add-on First Italian netwoRk Study (BRIVAFIRST). Seizure: the Journal of the British Epilepsy Association, 2022, 97, 37-42.	2.0	4
102	Hot water epilepsy. Epileptic Disorders, 2014, 16, 96-100.	1.3	2
103	Effects of postpartum depression on the behaviour of children born to mothers with epilepsy. Seizure: the Journal of the British Epilepsy Association, 2019, 73, 31-38.	2.0	2
104	Sleep in Children With Pallister Killian Syndrome: A Prospective Clinical and Videopolysomnographic Study. Frontiers in Neurology, 2021, 12, 796828.	2.4	2
105	Dreaming experience as a useful diagnostic clue for syncopal episodes. European Journal of Neurology, 2011, 18, 1361-1363.	3.3	1
106	Cardiac asystoles misdiagnosed as epileptic seizures. BMJ Case Reports, 2015, 2015, bcr2014206969-bcr2014206969.	0.5	1
107	The contribution of microbiology to neuroscience: More complex than it seems?. Behavioral and Brain Sciences, 2019, 42, .	0.7	1
108	Chronic Hyponatriemia Associated With Rett Syndrome. Pediatric Neurology, 2014, 50, e1-e2.	2.1	0

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109	Non-epileptic myoclonic attacks in infancy: three cases. Epileptic Disorders, 2014, 16, 433-438.	1.3	0
110	Genetic Generalized Epilepsies. , 2019, , 461-473.		0
111	Ischemic stroke as presentation of DADA2: Case report and literature review. Journal of the Neurological Sciences, 2021, 429, 118886.	0.6	0
112	Late onset myoclonic epilepsy in Down syndrome and dementia. Clinical Management Issues, 2012, 6, 97-103.	0.3	0
113	Different circuitry dysfunction in drug-naive patients with juvenile myoclonic epilepsy and juvenile absence epilepsy. Epilepsy and Behavior, 2021, 125, 108443.	1.7	0