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	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
2	KMT2A: Umbrella Gene for Multiple Diseases. Genes, 2022, 13, 514.	2.4	17
3	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
4	Natural History Study of STXBP1-Developmental and Epileptic Encephalopathy Into Adulthood. Neurology, 2022, 99, .	1.1	23
5	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
6	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
7	Epilepsy in adult patients with tuberous sclerosis complex. Acta Neurologica Scandinavica, 2021, 144, 29-40.	2.1	6
8	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
9	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
10	Chromatin Imbalance as the Vertex Between Fetal Valproate Syndrome and Chromatinopathies. Frontiers in Cell and Developmental Biology, 2021, 9, 654467.	3.7	7
11	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
12	Ischemic stroke as presentation of DADA2: Case report and literature review. Journal of the Neurological Sciences, 2021, 429, 118886.	0.6	0
13	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
14	Different circuitry dysfunction in drug-naive patients with juvenile myoclonic epilepsy and juvenile absence epilepsy. Epilepsy and Behavior, 2021, 125, 108443.	1.7	0
15	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
16	Sleep in Children With Pallister Killian Syndrome: A Prospective Clinical and Videopolysomnographic Study. Frontiers in Neurology, 2021, 12, 796828.	2.4	2
17	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
18	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

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19	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
20	Ring Chromosome 20 Syndrome: Genetics, Clinical Characteristics, and Overlapping Phenotypes. Frontiers in Neurology, 2020, 11, 613035.	2.4	14
21	Effects of Combined Transcranial Direct Current Stimulation with Cognitive Training in Girls with Rett Syndrome. Brain Sciences, 2020, 10, 276.	2.3	18
22	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
23	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	1.6	14
24	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
25	Sleep disturbances in Italian children and adolescents with epilepsy: A questionnaire study. Epilepsy and Behavior, 2020, 106, 107014.	1.7	11
26	Cerebellar lesions as potential predictors of neurobehavioural phenotype in tuberous sclerosis complex. Developmental Medicine and Child Neurology, 2019, 61, 1221-1228.	2.1	9
27	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
28	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
29	Myoclonic epilepsy with photosensitivity in infants with Pallister-Killian Syndrome. European Journal of Paediatric Neurology, 2019, 23, 653-656.	1.6	6
30	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
31	Genetic Generalized Epilepsies. , 2019, , 461-473.		0
32	Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9.	1.6	23
33	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
34	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
35	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
36	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

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37	The contribution of microbiology to neuroscience: More complex than it seems?. Behavioral and Brain Sciences, 2019, 42, .	0.7	1
38	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
39	Dramatic relapse of seizures after everolimus withdrawal. European Journal of Paediatric Neurology, 2018, 22, 203-206.	1.6	14
40	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
41	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
42	Hot water epilepsy and <scp>SYN</scp>1 variants. Epilepsia, 2018, 59, 2162-2163.	5.1	23
43	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
44	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
45	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
46	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
47	Rhinencephalon changes in tuberous sclerosis complex. Neuroradiology, 2018, 60, 813-820.	2.2	5
48	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
49	Antiepileptic drug use in Italian children over a decade. European Journal of Clinical Pharmacology, 2017, 73, 241-248.	1.9	13
50	Effectiveness and tolerability of antiepileptic drugs in 104 girls with Rett syndrome. Epilepsy and Behavior, 2017, 66, 27-33.	1.7	24
51	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
52	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
53	Rett Syndrome: A Focus on Gut Microbiota. International Journal of Molecular Sciences, 2017, 18, 344.	4.1	63
54	Extrastriate visual cortex in idiopathic occipital epilepsies: The contribution of retinotopic areas to spike generation. Epilepsia, 2016, 57, 896-906.	5.1	10

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55	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
56	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
57	Laryngeal motility alteration: A missing link between sleep apnea and vagus nerve stimulation for epilepsy. Epilepsia, 2016, 57, e24-7.	5.1	51
58	Symptomatic and presumed symptomatic focal epilepsies in childhood: An observational, prospective multicentre study. Epilepsia, 2016, 57, 1808-1816.	5.1	9
59	Epilepsy in ring chromosome 20 syndrome. Epilepsy Research, 2016, 128, 83-93.	1.6	30
60	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
61	Blood oxidative stress and metallothionein expression in Rett syndrome: Probing for markers. World Journal of Biological Psychiatry, 2016, 17, 198-209.	2.6	11
62	Autism spectrum disorder in tuberous sclerosis complex: searching for risk markers. Orphanet Journal of Rare Diseases, 2015, 10, 154.	2.7	74
63	Glioblastoma multiforme in a child with tuberous sclerosis complex. American Journal of Medical Genetics, Part A, 2015, 167, 2388-2393.	1.2	13
64	Cardiac asystoles misdiagnosed as epileptic seizures. BMJ Case Reports, 2015, 2015, bcr2014206969-bcr2014206969.	0.5	1
65	Antiepileptic drugs in Rett Syndrome. European Journal of Paediatric Neurology, 2015, 19, 446-452.	1.6	13
66	Emerging neuroimaging contribution to the diagnosis and management of the ring chromosome 20 syndrome. Epilepsy and Behavior, 2015, 45, 155-163.	1.7	11
67	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. Journal of Clinical Immunology, 2015, 35, 168-181.	3.8	35
68	Effects of chronic administration of valproic acid to epileptic patients on coagulation tests and primary hemostasis. Epilepsia, 2015, 56, e49-52.	5.1	19
69	Epilepsy in Rett syndromeâ€”Lessons from the Rett networked database. Epilepsia, 2015, 56, 569-576.	5.1	47
70	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
71	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
72	Hot water epilepsy. Epileptic Disorders, 2014, 16, 96-100.	1.3	2

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73	The visual system in eyelid myoclonia with absences. Annals of Neurology, 2014, 76, 412-427.	5.3	68
74	Epilepsyâ€related brain networks in ring chromosome 20 syndrome: An EEGâ€<scp>fMRI</scp> study. Epilepsia, 2014, 55, 403-413.	5.1	15
75	Vaccination and Occurrence of Seizures in SCN1A Mutationâ€"Positive Patients: A Multicenter Italian Study. Pediatric Neurology, 2014, 50, 228-232.	2.1	16
76	Low frequency mu-like activity characterizes cortical rhythms in epilepsy due to ring chromosome 20. Clinical Neurophysiology, 2014, 125, 239-249.	1.5	21
77	Chronic Hyponatriemia Associated With Rett Syndrome. Pediatric Neurology, 2014, 50, e1-e2.	2.1	0
78	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
79	Non-epileptic myoclonic attacks in infancy: three cases. Epileptic Disorders, 2014, 16, 433-438.	1.3	0
80	Vagus nerve stimulation in refractory epilepsy: New indications and outcome assessment. Epilepsy and Behavior, 2013, 28, 374-378.	1.7	31
81	Epilepsy in <scp>TSC</scp>: Certain etiology does not mean certain prognosis. Epilepsia, 2013, 54, 2134-2142.	5.1	66
82	Early onset absence epilepsy with onset in the first year of life: A multicenter cohort study. Epilepsia, 2013, 54, 66-69.	5.1	12
83	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
84	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
85	Clinical dissection of early onset absence epilepsy in children and prognostic implications. Epilepsia, 2013, 54, 1761-1770.	5.1	14
86	Sleep in ring chromosome 20 syndrome: a peculiar electroencephalographic pattern. Functional Neurology, 2013, 28, 47-53.	1.3	8
87	Ictal involvement of the nigrostriatal system in subtle seizures of ring chromosome 20 epilepsy. Epilepsia, 2012, 53, e156-60.	5.1	12
88	Aggressive behavior and epilepsy: A multicenter study. Epilepsia, 2012, 53, e174-9.	5.1	16
89	Ring chromosome 20. European Journal of Medical Genetics, 2012, 55, 381-387.	1.3	40
90	Rett networked database: An integrated clinical and genetic network of rett syndrome databases. Human Mutation, 2012, 33, 1031-1036.	2.5	19

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91	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
92	Electroclinical pattern in MECP2 duplication syndrome: Eight new reported cases and review of literature. Epilepsia, 2012, 53, 1146-1155.	5.1	34
93	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
94	Late onset myoclonic epilepsy in Down syndrome and dementia. Clinical Management Issues, 2012, 6, 97-103.	0.3	0
95	Epilepsy in adult patients with Down syndrome: a clinicalâ€video EEG study. Epileptic Disorders, 2011, 13, 125-132.	1.3	23
96	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
97	Dreaming experience as a useful diagnostic clue for syncopal episodes. European Journal of Neurology, 2011, 18, 1361-1363.	3.3	1
98	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
99	Ictal EEG patterns in epilepsy with centro-temporal spikes. Brain and Development, 2011, 33, 301-309.	1.1	11
100	A clinical and genetic study of 33 new cases with early-onset absence epilepsy. Epilepsy Research, 2011, 95, 221-226.	1.6	15
101	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. BMC Medical Genetics, 2010, 11, 146.	2.1	86
102	Correlations between neurophysiological, behavioral, and cognitive function in Rett syndrome. Epilepsy and Behavior, 2010, 17, 489-496.	1.7	47
103	Epilepsy in Rett syndrome: Clinical and genetic features. Epilepsy and Behavior, 2010, 19, 296-300.	1.7	68
104	Evolution of stereotypies in adolescents and women with Rett syndrome. Movement Disorders, 2009, 24, 1379-1383.	3.9	13
105	Ring chromosome 20 syndrome: A link between epilepsy onset and neuropsychological impairment in three children. Epilepsia, 2009, 50, 2420-2427.	5.1	105
106	Frontal cognitive dysfunction in juvenile myoclonic epilepsy. Epilepsia, 2008, 49, 657-662.	5.1	125
107	Mutational Analysis of EFHC1 Gene in Italian Families with Juvenile Myoclonic Epilepsy. Epilepsia, 2007, 48, 1686-1690.	5.1	44
108	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

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109	Felbamate in refractory partial epilepsy. Epilepsy Research, 1999, 34, 43-48.	1.6	20
110	Intrauterine growth in the offspring of epileptic women: a prospective multicenter study. Epilepsy Research, 1999, 36, 53-60.	1.6	55
111	Malformations in Offspring of Women with Epilepsy: A Prospective Study. Epilepsia, 1999, 40, 1231-1236.	5.1	181
112	Chromosome 20 Ring: A Chromosomal Disorder Associated with a Particular Electroclinical Pattern. Epilepsia, 1998, 39, 942-951.	5.1	137
113	Worsening in seizure frequency and severity in relation to folic acid administration. European Journal of Neurology, 1998, 5, 301-303.	3.3	17