

Carlotta Spagnoli

List of Publications by Year in descending order

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77
papers

1,036
citations

516561

16
h-index

526166

27
g-index

79
all docs

79
docs citations

79
times ranked

1417
citing authors

#	ARTICLE	IF	CITATIONS
1	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. <i>Journal of Neurology</i> , 2022, 269, 437-450.	1.8	12
2	Infantile-Onset Spinocerebellar Ataxia Type 5 (SCA5) with Optic Atrophy and Peripheral Neuropathy. <i>Cerebellum</i> , 2021, 20, 481-483.	1.4	3
3	Early-onset Dopamine Transporter Deficiency Syndrome: Long-term Follow-up. <i>Canadian Journal of Neurological Sciences</i> , 2021, 48, 285-286.	0.3	5
4	Acute symptomatic neonatal seizures, brain injury, and long-term outcome: The role of neuroprotective strategies. <i>Expert Review of Neurotherapeutics</i> , 2021, 21, 189-203.	1.4	6
5	Seizures in the neonate: A review of etiologies and outcomes. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 85, 48-56.	0.9	39
6	Genetic Neonatal-Onset Epilepsies and Developmental/Epileptic Encephalopathies with Movement Disorders: A Systematic Review. <i>International Journal of Molecular Sciences</i> , 2021, 22, 4202.	1.8	10
7	Rett Syndrome Spectrum in Monogenic Developmental-Epileptic Encephalopathies and Epilepsies: A Review. <i>Genes</i> , 2021, 12, 1157.	1.0	11
8	Further delineation of PIGB-related early infantile epileptic encephalopathy. <i>European Journal of Medical Genetics</i> , 2021, 64, 104268.	0.7	1
9	Pharmacological Treatment of Severe Breathing Abnormalities in a Case of <i>HNRNPU</i> Epileptic Encephalopathy. <i>Molecular Syndromology</i> , 2021, 12, 101-105.	0.3	4
10	A Novel De Novo KIF21A Variant in a Patient With Congenital Fibrosis of the Extraocular Muscles With a Syndromic CFEOM Phenotype. <i>Journal of Neuro-Ophthalmology</i> , 2021, 41, e85-e88.	0.4	5
11	Beneficial effects of the ketogenic diet on drug-resistant epileptic encephalopathy associated with a <i>de novo</i> NBEA pathogenic variant. <i>Epileptic Disorders</i> , 2021, 23, 739-743.	0.7	3
12	SPG6 (NIPA1 variant): A report of a case with early-onset complex hereditary spastic paraplegia and brief literature review. <i>Journal of Clinical Neuroscience</i> , 2021, 94, 281-285.	0.8	11
13	<i>IRF2BPL</i> gene variants: One new case. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 255-256.	0.7	7
14	Neonatal seizures in preterm infants: A systematic review of mortality risk and neurological outcomes from studies in the 2000's. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 75, 7-17.	0.9	17
15	Linking acute symptomatic neonatal seizures, brain injury and outcome in preterm infants. <i>Epilepsy and Behavior</i> , 2020, 112, 107406.	0.9	7
16	Long-term follow-up in infantile-onset SCAR18: A case report. <i>Journal of Clinical Neuroscience</i> , 2020, 77, 232-234.	0.8	2
17	Paroxysmal movement disorder with response to carbamazepine in a patient with RHOBTB2 developmental and epileptic encephalopathy. <i>Parkinsonism and Related Disorders</i> , 2020, 76, 54-55.	1.1	10
18	A highly unusual case of osmotic demyelination syndrome and extrapontine myelinolysis in a 3-month-old infant with Bartter syndrome. <i>Journal of International Medical Research</i> , 2020, 48, 030006052096649.	0.4	1

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19	Severe intellectual disability, absence of language, epilepsy, microcephaly and progressive cerebellar atrophy related to the recurrent de novo variant p.(P139L) of the <i>CAMK2B</i> gene: A case report and brief review. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2675-2679.	0.7	12
20	EEG Monitoring of the Epileptic Newborn. <i>Current Neurology and Neuroscience Reports</i> , 2020, 20, 6.	2.0	12
21	New biallelic GBA2 variant in a patient with SPG46. <i>Clinical Neurology and Neurosurgery</i> , 2020, 191, 105676.	0.6	8
22	Paediatric-onset hereditary spastic paraplegias: a retrospective cohort study. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 1068-1074.	1.1	12
23	Neuropsychological and behavioral disorders as presentation symptoms in two brothers with early-infantile Niemann-Pick type C. <i>Acta Biomedica</i> , 2020, 91, e2020075.	0.2	0
24	Co-occurrence of an HSPG2 Missense Variant and Functional Polymorphisms in Atypical Schwartz-Jampel Syndrome Type 1 with Obesity: A Case Report. <i>Journal of Pediatric Neurology</i> , 2019, 17, 149-152.	0.0	1
25	Outcome in preterm infants with seizures. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2019, 162, 401-414.	1.0	6
26	Autosomal recessive axonal neuropathy caused by HINT1 mutation: New association of a psychiatric disorder to the neurologic phenotype. <i>Neuromuscular Disorders</i> , 2019, 29, 979.	0.3	14
27	Biallelic SZT2 mutation with early onset of focal status epilepticus: Useful diagnostic clues other than epilepsy, intellectual disability and macrocephaly. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 69, 296-297.	0.9	8
28	Prolonged survival in a patient with a novel pyrroline-5-carboxylase reductase 2 genetic variant. <i>European Journal of Neurology</i> , 2019, 26, e45-e46.	1.7	3
29	The independent role of neonatal seizures in epilepsy and other long-term neurological outcomes. <i>Developmental Medicine and Child Neurology</i> , 2019, 61, 624-624.	1.1	0
30	Long-term follow-up until early adulthood in autosomal dominant, complex SPG30 with a novel KIF1A variant: a case report. <i>Italian Journal of Pediatrics</i> , 2019, 45, 155.	1.0	10
31	Early infantile SCN1A epileptic encephalopathy: Expanding the genotype-phenotype correlations. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2019, 65, 62-64.	0.9	12
32	Restless Legs Syndrome in NKX2-1-related chorea: An expansion of the disease spectrum. <i>Brain and Development</i> , 2019, 41, 250-256.	0.6	6
33	On CALFAN syndrome: report of a patient with a novel variant in SCYL1 gene and recurrent respiratory failure. <i>Genetics in Medicine</i> , 2019, 21, 1663-1664.	1.1	13
34	Diagnosis and Management of Acute Seizures in Neonates. , 2019, , 111-129.		1
35	Genetic diagnosis in neonatal-onset epilepsies: Back to the future. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 354-357.	0.7	14
36	Brain MRI abnormalities resembling Unidentified Bright Objects in a patient with Phelan-McDermid syndrome. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 568-569.	0.7	1

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37	Corticosteroid treatment in Sydenham's chorea. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 327-331.	0.7	12
38	Acute symptomatic neonatal seizures in preterm neonates: etiologies and treatments. <i>Seminars in Fetal and Neonatal Medicine</i> , 2018, 23, 191-196.	1.1	11
39	KCNQ2 encephalopathy: A case due to a de novo deletion. <i>Brain and Development</i> , 2018, 40, 65-68.	0.6	18
40	RNASEH2B Pathogenic Gene Variant in Uncomplicated Hereditary Spastic Paraplegia: Report of a New Patient. <i>Neuropediatrics</i> , 2018, 49, 419-419.	0.3	6
41	Symptomatic seizures in preterm newborns: a review on clinical features and prognosis. <i>Italian Journal of Pediatrics</i> , 2018, 44, 115.	1.0	27
42	<i>HCN1</i> mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. <i>Brain</i> , 2018, 141, 3160-3178.	3.7	96
43	Ocular flutter, generalized myoclonus, and ataxia associated with anti-GM1, GD1a, and GD1b antibodies in a 6-year-old child. <i>Neurological Sciences</i> , 2018, 39, 1801-1803.	0.9	1
44	Peripheral neuropathy and gastroenterologic disorders: an overview on an underrecognized association. <i>Acta Biomedica</i> , 2018, 89, 22-32.	0.2	5
45	Seizures in preterm newborns. <i>Minerva Pediatrics</i> , 2018, 70, 105-107.	0.2	0
46	Long-term follow-up in spastic paraplegia due to SPG56/CYP2U1: age-dependency rather than genetic variability?. <i>Journal of Neurology</i> , 2017, 264, 586-588.	1.8	9
47	Monitoring infants by automatic video processing: A unified approach to motion analysis. <i>Computers in Biology and Medicine</i> , 2017, 80, 158-165.	3.9	43
48	Minimal holoprosencephaly in a 14q deletion syndrome patient. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3216-3220.	0.7	2
49	Infantile neuroaxonal dystrophy and PLA2G6-associated neurodegeneration: An update for the diagnosis. <i>Brain and Development</i> , 2017, 39, 93-100.	0.6	28
50	Hereditary neuropathy with liability to pressure palsy (HNPP): report of a family with a new point mutation in PMP22 gene. <i>Italian Journal of Pediatrics</i> , 2017, 43, 97.	1.0	12
51	Reflex Myoclonic Epilepsy of Infancy: Seizures Induced by Tactile Stimulation. <i>Journal of Pediatrics</i> , 2016, 173, 250-253.e4.	0.9	3
52	Monitoring of newborns at high risk for brain injury. <i>Italian Journal of Pediatrics</i> , 2016, 42, 48.	1.0	20
53	Hereditary neuropathy with liability to pressure palsies in childhood: Case series and literature update. <i>Neuromuscular Disorders</i> , 2016, 26, 394.	0.3	5
54	Optimizing Detection Rate and Characterization of Subtle Paroxysmal Neonatal Abnormal Facial Movements with Multi-Camera Video-Electroencephalogram Recordings. <i>Neuropediatrics</i> , 2016, 47, 169-174.	0.3	9

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55	Phenobarbital for Neonatal Seizures: Response Rate and Predictors of Refractoriness. <i>Neuropediatrics</i> , 2016, 47, 318-326.	0.3	25
56	A Case of Alpers's "Huttenlocher Syndrome Due to a New POLG1 Mutation with Rapid Onset of Partial Status Epilepticus: Serial Neuroradiological and Neurophysiological Evaluation. <i>Journal of Pediatric Neurology</i> , 2016, 14, 112-118.	0.0	1
57	CMV-associated axonal sensory-motor Guillain-Barré syndrome in a child: Case report and review of the literature. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 168-175.	0.7	12
58	Epileptic and non-epileptic paroxysmal motor phenomena in newborns. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2016, 29, 3652-3659.	0.7	25
59	Neonatal Seizures: A Review of Outcomes and Outcome Predictors. <i>Neuropediatrics</i> , 2016, 47, 012-019.	0.3	40
60	Neonatal seizures in preterm newborns: A predictive model for outcome. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 243-251.	0.7	60
61	Expanding phenotype of PRRT2 gene mutations: A new case with epilepsy and benign myoclonus of early infancy. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 454-456.	0.7	21
62	Steroids efficacy in the acute management of seizure clusters in one case of PCDH19 female epilepsy. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2015, 32, 45-46.	0.9	8
63	Neonatal status epilepticus: Differences between preterm and term newborns. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 314-319.	0.7	35
64	A Painful Stiff Neck following an Ear, Nose, and Throat Surgical Procedure: Case Report. <i>Neuropediatrics</i> , 2015, 46, 069-071.	0.3	5
65	Symptomatic Neonatal Seizures Followed by Febrile Status Epilepticus. <i>Journal of Child Neurology</i> , 2015, 30, 615-618.	0.7	9
66	Children's Headache: Drawings in the Diagnostic Work Up. <i>Neuropediatrics</i> , 2015, 46, 261-268.	0.3	15
67	Epileptic spasms and early-onset photosensitive epilepsy in Patau syndrome: An EEG study. <i>Brain and Development</i> , 2015, 37, 704-713.	0.6	10
68	Epilepsy after neonatal seizures: Literature review. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 6-14.	0.7	75
69	Retrospective Study of the Investigations of Children Presenting with Lower Limbs Spasticity in a Single Institution. <i>Neuropediatrics</i> , 2014, 45, 109-116.	0.3	0
70	Real-time automated detection of clonic seizures in newborns. <i>Clinical Neurophysiology</i> , 2014, 125, 1533-1540.	0.7	26
71	Brown-Vialetto-van Laere syndrome: A riboflavin responsive neuronopathy of infancy with singular features. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 231-234.	0.7	18
72	Neonatal Forearm Compartment Syndrome: Look for Cerebral Stroke. <i>Journal of Pediatrics</i> , 2014, 164, 427-427.e1.	0.9	6

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73	Tapia's syndrome secondary to laterocervical localization of diffuse large cell lymphoma. Brain and Development, 2014, 36, 548-550.	0.6	7
74	A wire-free, non-invasive, low-cost video processing-based approach to neonatal apnoea detection. , 2014, , .		13
75	Neonatal seizures therapy: we are still looking for the efficacious drug. Italian Journal of Pediatrics, 2013, 39, 37.	1.0	5
76	Unusual Trigeminal Autonomic Pain Heraldng Hemichorea due to Zoster Sine Herpete Vasculopathy. Pediatric Neurology, 2013, 49, 205-208.	1.0	4
77	Brown's-Vialto's-Van Laere syndrome and Fazio's-Londe disease " treatable motor neuron diseases of childhood. Developmental Medicine and Child Neurology, 2012, 54, 292-293.	1.1	20