

Salvatore Savasta

List of Publications by Year in descending order

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Version: 2024-02-01

87
papers

1,680
citations

304743

22
h-index

345221

36
g-index

91
all docs

91
docs citations

91
times ranked

2435
citing authors

#	ARTICLE	IF	CITATIONS
1	Infantile Encephalopathy and Defective Mitochondrial DNA Translation in Patients with Mutations of Mitochondrial Elongation Factors EFG1 and EFTu. American Journal of Human Genetics, 2007, 80, 44-58.	6.2	172
2	Phenotype and genotype of 87 patients with Mowatâ€Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
3	<i>CDKL5</i> geneâ€related epileptic encephalopathy: electroclinical findings in the first year of life. Developmental Medicine and Child Neurology, 2011, 53, 354-360.	2.1	64
4	Improving molecular diagnosis in epilepsy by a dedicated high-throughput sequencing platform. European Journal of Human Genetics, 2015, 23, 354-362.	2.8	64
5	Lacosamide in pediatric and adult patients: Comparison of efficacy and safety. Seizure: the Journal of the British Epilepsy Association, 2013, 22, 210-216.	2.0	60
6	Challenges and management of neurological and psychiatric manifestations in SARS-CoV-2 (COVID-19) patients. Neurological Sciences, 2020, 41, 2353-2366.	1.9	60
7	Benign convulsions associated with mild gastroenteritis: A multicenter clinical study. Epilepsy Research, 2011, 93, 107-114.	1.6	57
8	Neuroimaging findings in Mowatâ€Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
9	Neurofibromatosis type 1 with external genitalia involvement. Journal of Pediatric Surgery, 2008, 43, 1998-2003.	1.6	44
10	Epilepsy in Mowatâ€Wilson syndrome: Delineation of the electroclinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 273-284.	1.2	42
11	Pediatric idiopathic intracranial hypertension and the underlying endocrine-metabolic dysfunction: a pilot study. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 107-115.	0.9	41
12	Relapse risk factors in antiâ€methylâ€caspate receptor encephalitis. Developmental Medicine and Child Neurology, 2019, 61, 1101-1107.	2.1	40
13	Lower Airway Microbiota. Frontiers in Pediatrics, 2019, 7, 393.	1.9	38
14	Total Hemi-overgrowth in Pigmentary Mosaicism of the (Hypomelanosis of) Ito Type. Medicine (United Tj ETQq 0 0 rgBT /Overlock 10	1.0	33
15	Italian Consensus Conference on Epilepsy and Pregnancy, Labor and Puerperium. Epilepsia, 2009, 50, 7-23.	5.1	31
16	Seizures and epilepsy in Sotos syndrome: Analysis of 19 Caucasian patients with longâ€term followâ€up. Epilepsia, 2012, 53, e102-5.	5.1	30
17	Clinical guidelines in pediatric headache: evaluation of quality using the AGREE II instrument. Journal of Headache and Pain, 2014, 15, 57.	6.0	30
18	Long-term follow-up in children with benign convulsions associated with gastroenteritis. European Journal of Paediatric Neurology, 2014, 18, 572-577.	1.6	30

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19	Epilepsy is a possible feature in Williamsâ€Beuren syndrome patients harboring typical deletions of the 7q11.23 critical region. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 148-155.	1.2	29
20	Melkerssonâ€Rosenthal Syndrome in Childhood: Report of Three Paediatric Cases and a Review of the Literature. <i>International Journal of Environmental Research and Public Health</i> , 2019, 16, 1289.	2.6	28
21	Acute ataxia in paediatric emergency departments: a multicentre Italian study. <i>Archives of Disease in Childhood</i> , 2019, 104, 768-774.	1.9	27
22	Ehlersâ€Danlos syndrome and neurological features: a review. <i>Child's Nervous System</i> , 2011, 27, 365-371.	1.1	25
23	Electroclinical Features and Long-Term Outcome of Cryptogenic Epilepsy in Children with Down Syndrome. <i>Journal of Pediatrics</i> , 2013, 163, 1754-1758.	1.8	25
24	Gastrointestinal phenotype of ATR-X syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1172-1176.	1.2	24
25	Seizures in fetal alcohol spectrum disorders: Evaluation of clinical, electroencephalographic, and neuroradiologic features in a pediatric case series. <i>Epilepsia</i> , 2014, 55, e60-6.	5.1	24
26	Epilepsy in the setting of full trisomy 18: A multicenter study on 18 affected children with and without structural brain abnormalities. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 288-295.	1.6	24
27	Clinical characteristics of headache in Italian adolescents aged 11â€16 years: a cross-sectional questionnaire school-based study. <i>Italian Journal of Pediatrics</i> , 2018, 44, 44.	2.6	24
28	Epilepsy in patients with Cornelia de Lange syndrome: A clinical series. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2013, 22, 356-359.	2.0	23
29	Understanding Childhood Neuroimmune Diseases of the Central Nervous System. <i>Frontiers in Pediatrics</i> , 2019, 7, 511.	1.9	23
30	Innovative therapies for malignant brain tumors: the road to a tailored cure. <i>Acta Biomedica</i> , 2020, 91, 5-17.	0.3	21
31	Mitochondrial DNA Deletion in a Child With Mitochondrial Encephalomyopathy, Growth Hormone Deficiency, and Hypoparathyroidism. <i>Journal of Child Neurology</i> , 2006, 21, 983-985.	1.4	20
32	Pai syndrome: a further report of a case with bifid nose, lipoma, and agenesis of the corpus callosum. <i>Child's Nervous System</i> , 2008, 24, 773-776.	1.1	20
33	CHARGEâ€like presentation, craniosynostosis and mild Mowatâ€Wilson Syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2557-2566.	1.2	20
34	HLA haplotype segregation and ultrastructural study in familial immotile-cilia syndrome. <i>Human Genetics</i> , 1992, 89, 270-4.	3.8	19
35	Nucleotide variation in Sabin type 3 poliovirus from an Albanian infant with agammaglobulinemia and vaccine associated poliomyelitis. <i>BMC Infectious Diseases</i> , 2016, 16, 277.	2.9	19
36	Coexistence of childhood absence epilepsy and benign epilepsy with centrotemporal spikes: A case series. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 570-575.	1.6	19

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37	The motilin gene: subregional localisation, tissue expression, DNA polymorphisms and exclusion as a candidate gene for the HLA-associated immotile cilia syndrome. <i>Human Genetics</i> , 1994, 94, 671-4.	3.8	15
38	Targeting the medulloblastoma: a molecular-based approach. <i>Acta Biomedica</i> , 2020, 91, 79-100.	0.3	14
39	FISH characterization of a supernumerary r(1)(:cen?q22::q22?sq21::) chromosome associated with multiple anomalies and bilateral cataracts. <i>American Journal of Medical Genetics Part A</i> , 2001, 104, 157-164.	2.4	13
40	Seizures and EEG patterns in Pallisterâ€“Killian syndrome: 13Â“New Italian patients. <i>European Journal of Paediatric Neurology</i> , 2012, 16, 636-641.	1.6	13
41	Wolfâ€“Hirschhorn syndrome-associated chromosome changes are not mediated by olfactory receptor gene clusters nor by inversion polymorphism on 4p16. <i>Human Genetics</i> , 2007, 122, 423-430.	3.8	12
42	Measurement of nitric oxide and assessment of airway diseases in children: an update. <i>Minerva Pediatrica</i> , 2019, 71, 524-532.	2.7	12
43	Adoptive immunotherapies in neuro-oncology: classification, recent advances, and translational challenges. <i>Acta Biomedica</i> , 2020, 91, 18-31.	0.3	12
44	Potential roads for reaching the summit: an overview on target therapies for high-grade gliomas. <i>Acta Biomedica</i> , 2020, 91, 61-78.	0.3	12
45	Muscle phosphofructokinase deficiency in a myopathic child with severe mental retardation and aplasia of cerebellar vermis. <i>Child's Nervous System</i> , 1992, 8, 237-241.	1.1	10
46	Long-term outcome of epilepsy in Kabuki syndrome. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2011, 20, 650-654.	2.0	10
47	Kikuchi-Fujimoto Disease Complicated by Peripheral Neuropathy. <i>Pediatric Neurology</i> , 2012, 46, 319-321.	2.1	10
48	Long-term outcome of epilepsy in patients with Praderâ€“Willi syndrome. <i>Journal of Neurology</i> , 2015, 262, 116-123.	3.6	10
49	X-Linked Hypohidrotic Ectodermal Dysplasia: New Features and a Novel <i>EDA</i> Gene Mutation. <i>Cytogenetic and Genome Research</i> , 2017, 152, 111-116.	1.1	10
50	The impact of stem cells in neuro-oncology: applications, evidence, limitations and challenges. <i>Acta Biomedica</i> , 2020, 91, 51-60.	0.3	10
51	Agenesis of Internal Carotid Artery and Hypopituitarism: Case Report and Review of Literature. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2012, 97, 3414-3420.	3.6	9
52	Stickler syndrome associated with epilepsy: report of three cases. <i>European Journal of Pediatrics</i> , 2015, 174, 697-701.	2.7	9
53	Gene therapies for high-grade gliomas: from the bench to the bedside. <i>Acta Biomedica</i> , 2020, 91, 32-50.	0.3	9
54	Subependymal Periventricular Heterotopias in a Patient With Ehlers-Danlos Syndrome: A New Case. <i>Journal of Child Neurology</i> , 2007, 22, 317-320.	1.4	8

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55	Gelastic seizures not associated with hypothalamic hamartoma: A long-term follow-up study. <i>Epilepsy and Behavior</i> , 2020, 103, 106578.	1.7	8
56	Advanced pharmacological therapies for neurofibromatosis type 1-related tumors. <i>Acta Biomedica</i> , 2020, 91, 101-114.	0.3	7
57	Holoprosencephaly with neurogenic hypernatremia: a new case. <i>Child's Nervous System</i> , 2007, 24, 139-142.	1.1	6
58	Paediatric clinically isolated syndromes: report of seven cases, differential diagnosis and literature review. <i>Child's Nervous System</i> , 2016, 32, 69-77.	1.1	6
59	Gradenigo's syndrome with abscess of the petrous apex in pediatric patients: what is the best treatment?. <i>Child's Nervous System</i> , 2019, 35, 2265-2272.	1.1	6
60	Ehlers-Danlos Syndromes. , 2008, , 887-906.		6
61	Infantile Mitochondria Encephalomyopathies: Report on 4 Cases. <i>European Neurology</i> , 1993, 33, 54-61.	1.4	5
62	Poly-epiphyseal overgrowth: description of a previously unreported skeletal dysplasia. <i>Pediatric Radiology</i> , 2007, 37, 1025-1030.	2.0	5
63	Long-term prognosis of patients with Ehlers-Danlos syndrome and epilepsy. <i>Epilepsia</i> , 2014, 55, 1213-1219.	5.1	5
64	Gelastic epilepsy without hypothalamic hamartoma: Three additional cases. <i>Epilepsy and Behavior</i> , 2014, 37, 87-90.	1.7	5
65	Efficacy of rituximab as third-line therapy in combined central and peripheral demyelination. <i>Neurology: Clinical Practice</i> , 2017, 7, 534-537.	1.6	5
66	Further delineation and long-term evolution of electroclinical phenotype in Mowat Wilson Syndrome. A longitudinal study in 40 individuals. <i>Epilepsy and Behavior</i> , 2021, 124, 108315.	1.7	5
67	A Rare Cause of Recurrent Spontaneous Pneumothorax. <i>Clinical Pediatrics</i> , 2011, 50, 456-458.	0.8	4
68	Sleep in Mowat-Wilson Syndrome: a clinical and video-polysomnographic study. <i>Sleep Medicine</i> , 2019, 61, 44-51.	1.6	4
69	Pai syndrome: a review. <i>Child's Nervous System</i> , 2020, 36, 2635-2640.	1.1	4
70	Wolf-Hirschhorn syndrome: diagnosis using hand radiograph performed for bone age. <i>Pediatric Radiology</i> , 2010, 40, 1580-1580.	2.0	3
71	Intramedullary spinal cord cavernous malformations presenting with unexplained chest pain: case report and review of the literature. <i>Child's Nervous System</i> , 2013, 29, 323-328.	1.1	3
72	Unilateral periventricular heterotopia and epilepsy in a girl with Ehlers-Danlos syndrome. <i>Epilepsy & Behavior Case Reports</i> , 2015, 4, 27-29.	1.5	3

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73	Clinical variability in children with dolichoarteriopathies of the internal carotid artery. <i>Child's Nervous System</i> , 2020, 36, 621-628.	1.1	3
74	Absence of lingual frenulum in children with Ehlers-Danlos Syndrome: a retrospective study of forty cases and literature review of a twenty years long debate. <i>Minerva Pediatrics</i> , 2021, 73, .	0.4	3
75	Absence of lingual frenulum in children with Ehlers-Danlos Syndrome: a retrospective study of forty cases and literature review of a twenty years long debate. <i>Minerva Pediatrics</i> , 2021, 73, 230-235.	0.4	3
76	Microcephaly associated with Legg-CalvÃ©-Perthes disease in two siblings. <i>Neurological Sciences</i> , 2012, 33, 1401-1405.	1.9	2
77	Headache and Diplopia after Rapid Maxillary Expansion: A Clue to Underdiagnosed Pseudotumor Cerebri Syndrome?. <i>Journal of Pediatric Neurology</i> , 2015, 13, 031-034.	0.2	2
78	Hypomelanosis of Ito. <i>Journal of Pediatric Neurology</i> , 2018, 16, 265-275.	0.2	2
79	Transient symptomatic zinc deficiency in a breastâ€fed African infant: case report and literature review. <i>International Journal of Dermatology</i> , 2019, 58, 963-965.	1.0	2
80	Headache in progressive facial hemiatrophy (Parry-Romberg syndrome): A paradigmatic case and systematic review of the literature. <i>Cephalalgia</i> , 2022, 42, 409-425.	3.9	2
81	Postural analysis in a pediatric cohort of patients with Ehlers-Danlos Syndrome: a pilot study. <i>Minerva Pediatrica</i> , 2020, 72, 73-78.	2.7	2
82	Management of Pediatric Pseudotumor Cerebri Syndrome. <i>Journal of Pediatric Neurology</i> , 2015, 13, 058-061.	0.2	1
83	Tinnitus and Pediatric Pseudotumor Cerebri Syndrome. <i>Journal of Pediatric Neurology</i> , 2015, 13, 038-041.	0.2	1
84	Challenges and management of neurological and psychiatric manifestations in SARS-CoV-2 (COVID-19) patients. , 2020, 41, 2353.		1
85	A Novel 4q32.3 Deletion in a Boy: Additional Signs and the Role of MARCH1. <i>Journal of Pediatric Genetics</i> , 2021, 10, 259-265.	0.7	1
86	Hemorrhagic Shock and Encephalopathy: Diagnostic Criteria. <i>JAMA Pediatrics</i> , 1992, 146, 279.	3.0	0
87	Octreotide: A Therapeutic Option for Pediatric Pseudotumor Cerebri Syndrome?. <i>Journal of Pediatric Neurology</i> , 2015, 13, 046-048.	0.2	0