

Stefano D'Arrigo

List of Publications by Year in descending order

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95
papers

4,224
citations

147786

31
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123420

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97
docs citations

97
times ranked

7187
citing authors

#	ARTICLE	IF	CITATIONS
1	Consensus Paper: The Cerebellum's Role in Movement and Cognition. <i>Cerebellum</i> , 2014, 13, 151-177.	2.5	815
2	Characterization of human disease phenotypes associated with mutations in <i>TREX1</i> , <i>RNASEH2A</i> , <i>RNASEH2B</i> , <i>RNASEH2C</i> , <i>SAMHD1</i> , <i>ADAR</i> , and <i>IFIH1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 296-312.	1.2	447
3	Clinical and Molecular Phenotype of Aicardi-Goutières Syndrome. <i>American Journal of Human Genetics</i> , 2007, 81, 713-725.	6.2	375
4	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. <i>PLoS Genetics</i> , 2011, 7, e1002173.	3.5	172
5	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndrome-Related Disorders. <i>American Journal of Human Genetics</i> , 2007, 81, 104-113.	6.2	137
6	Recessive Mutations in the Gene Encoding the Tight Junction Protein Occludin Cause Band-like Calcification with Simplified Gyration and Polymicrogyria. <i>American Journal of Human Genetics</i> , 2010, 87, 354-364.	6.2	123
7	Intellectual and language findings and their relationship to EEG characteristics in benign childhood epilepsy with centrotemporal spikes. <i>Epilepsy and Behavior</i> , 2007, 10, 278-285.	1.7	113
8	Duplications of <i>FOXP1</i> in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. <i>European Journal of Human Genetics</i> , 2011, 19, 102-107.	2.8	104
9	<i>MKS3/TMEM67</i> mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. <i>Human Mutation</i> , 2009, 30, E432-E442.	2.5	96
10	Novel <i>TMEM67</i> mutations and genotype-phenotype correlates in meckelin-related ciliopathies. <i>Human Mutation</i> , 2010, 31, n/a-n/a.	2.5	77
11	Healthcare recommendations for Joubert syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 229-249.	1.2	66
12	<i>RPGRIPL1</i> mutations are mainly associated with the cerebello-renal phenotype of Joubert syndrome-related disorders. <i>Clinical Genetics</i> , 2008, 74, 164-170.	2.0	64
13	Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 4.	2.7	64
14	Phenotypic spectrum and prevalence of <i>INPP5E</i> mutations in Joubert Syndrome and related disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 1074-1078.	2.8	64
15	Functional genome-wide siRNA screen identifies <i>KIAA0586</i> as mutated in Joubert syndrome. <i>ELife</i> , 2015, 4, e06602.	6.0	64
16	Supratentorial and pontine <i>MRI</i> abnormalities characterize recessive spastic ataxia of <i>Charlevoix-Saguenay</i> . A comprehensive study of an Italian series. <i>European Journal of Neurology</i> , 2013, 20, 138-146.	3.3	57
17	Mutations in <i>CEP120</i> cause Joubert syndrome as well as complex ciliopathy phenotypes. <i>Journal of Medical Genetics</i> , 2016, 53, 608-615.	3.2	55
18	Unilateral frontal lobe epilepsy affects executive functions in children. <i>Neurological Sciences</i> , 2005, 26, 263-270.	1.9	53

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19	Aicardi-Goutières syndrome: a description of 21 new cases and a comparison with the literature. <i>European Journal of Paediatric Neurology</i> , 2002, 6, A9-A22.	1.6	50
20	Cognitive and Behavioural Effects of Migraine in Childhood and Adolescence. <i>Cephalalgia</i> , 2006, 26, 596-603.	3.9	46
21	Band-like intracranial calcification with simplified gyration and polymicrogyria: A distinct â€œpseudo-TORCHâ€ phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 3173-3180.	1.2	46
22	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. <i>American Journal of Human Genetics</i> , 2017, 101, 552-563.	6.2	45
23	Pontine tegmental cap dysplasia: developmental and cognitive outcome in three adolescent patients. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 36.	2.7	44
24	The natural history of Aicardi-Goutieres syndrome: Follow-up of 11 Italian patients. <i>Neurology</i> , 2005, 64, 1621-1624.	1.1	42
25	Personality Traits in Childhood and Adolescent Headache. <i>Cephalalgia</i> , 2001, 21, 53-60.	3.9	39
26	Verbal dichotic listening performance and its relationship with EEG features in benign childhood epilepsy with centrotemporal spikes. <i>Epilepsy Research</i> , 2008, 79, 31-38.	1.6	39
27	Clinical spectrum of PTEN mutation in pediatric patients. A bicenter experience. <i>European Journal of Medical Genetics</i> , 2019, 62, 103596.	1.3	39
28	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic <i>POLR1C</i> pathogenic variants. <i>Neurology: Genetics</i> , 2019, 5, e369.	1.9	38
29	The Diagnostic Yield of Array Comparative Genomic Hybridization Is High Regardless of Severity of Intellectual Disability/Developmental Delay in Children. <i>Journal of Child Neurology</i> , 2016, 31, 691-699.	1.4	37
30	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. <i>European Radiology</i> , 2017, 27, 5080-5092.	4.5	36
31	Aicardi-Goutières syndrome: description of a late onset case. <i>Developmental Medicine and Child Neurology</i> , 2008, 50, 631-634.	2.1	35
32	The â€œEye-of-the-Tigerâ€ Sign may be Absent in the Early Stages of Classic Pantothenate Kinase Associated Neurodegeneration. <i>Neuropediatrics</i> , 2011, 42, 159-162.	0.6	34
33	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaides-Baraitser syndrome. <i>Genetics in Medicine</i> , 2020, 22, 1838-1850.	2.4	31
34	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. <i>Human Genetics</i> , 2015, 134, 123-126.	3.8	30
35	Epilepsy is a possible feature in Williams-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
36	Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. <i>Journal of Clinical Medicine</i> , 2019, 8, 750.	2.4	29

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37	<i>EED</i> and <i>EZH2</i> constitutive variants: A study to expand the Cohen-Gibson syndrome phenotype and contrast it with Weaver syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 588-594.	1.2	24
38	<i>ZC4H2</i> deletions can cause severe phenotype in female carriers. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1358-1363.	1.2	23
39	Diagnostic Approach to Cerebellar Disease in Children. <i>Journal of Child Neurology</i> , 2005, 20, 859-866.	1.4	22
40	Ataxia With Oculomotor Apraxia Type 1 (AOA1): Clinical and Neuropsychological Features in 2 New Patients and Differential Diagnosis. <i>Journal of Child Neurology</i> , 2008, 23, 895-900.	1.4	22
41	Cognitive, adaptive, and behavioral features in Joubert syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3115-3124.	1.2	22
42	Heterozygous <i>KIF1A</i> variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. <i>Journal of Medical Genetics</i> , 2021, 58, 475-483.	3.2	21
43	Another Patient With MECP2 Mutation Without Classic Rett Syndrome Phenotype. <i>Pediatric Neurology</i> , 2005, 32, 355-357.	2.1	20
44	Novel POMGNT1 point mutations and intragenic rearrangements associated with muscle-eye-brain disease. <i>Journal of the Neurological Sciences</i> , 2012, 318, 45-50.	0.6	20
45	Postural Control in Children with Cerebellar Ataxia. <i>Applied Sciences (Switzerland)</i> , 2020, 10, 1606.	2.5	20
46	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 75.	2.7	19
47	<i>SUFU</i> haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. <i>Journal of Medical Genetics</i> , 2022, 59, 888-894.	3.2	19
48	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. <i>Brain</i> , 2017, 140, e34-e34.	7.6	17
49	Between SCA5 and SCAR14: delineation of the SPTBN2 p.R480W-associated phenotype. <i>European Journal of Human Genetics</i> , 2018, 26, 928-929.	2.8	17
50	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 135.	2.7	17
51	Seizures and EEG features in 74 patients with genetic dysmorphic syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 3154-3161.	1.2	16
52	Novel Mutations in <i>TSEN54</i> in Pontocerebellar Hypoplasia Type 2. <i>Journal of Child Neurology</i> , 2014, 29, 520-525.	1.4	15
53	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. <i>Nephrology Dialysis Transplantation</i> , 2020, 35, 1195-1202.	0.7	15
54	The Effectiveness of Hospitalization in the Treatment of Paediatric Idiopathic Headache Patients. <i>Psychopathology</i> , 2007, 40, 1-7.	1.5	14

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55	The syndrome of perisylvian polymicrogyria with congenital arthrogryposis. <i>Brain and Development</i> , 2010, 32, 550-555.	1.1	14
56	5p13 microduplication syndrome: A new case and better clinical definition of the syndrome. <i>European Journal of Medical Genetics</i> , 2013, 56, 54-58.	1.3	14
57	Unusual neurophysiological features in Cockayne's syndrome: a report of two cases as a contribution to diagnosis and classification. <i>Brain and Development</i> , 2004, 26, 273-280.	1.1	13
58	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. <i>Journal of Medical Genetics</i> , 2022, 59, 399-409.	3.2	13
59	Are Vascular Disorders More Prevalent in the Relatives of Children and Adolescents with Migraine?. <i>Cephalalgia</i> , 2003, 23, 887-891.	3.9	12
60	Verbal and Gestural Communication in Children With Bilateral Perisylvian Polymicrogyria. <i>Journal of Child Neurology</i> , 2007, 22, 1090-1098.	1.4	12
61	Electroencephalographic (EEG) Photoparoxysmal Responses Under 5 Years of Age. <i>Journal of Child Neurology</i> , 2015, 30, 1824-1830.	1.4	12
62	Consolidating the Role of TDP2 Mutations in Recessive Spinocerebellar Ataxia Associated with Pediatric Onset Drug Resistant Epilepsy and Intellectual Disability (SCAR23). <i>Cerebellum</i> , 2019, 18, 972-975.	2.5	12
63	Identification of previously unreported mutations in CHRNA1, CHRNE and RAPSN genes in three unrelated Italian patients with congenital myasthenic syndromes. <i>Journal of Neurology</i> , 2010, 257, 1119-1123.	3.6	11
64	Progressive Encephalopathy with Edema, Hypsarrhythmia, and Optic Nerve Atrophy (PEHO)-Like Syndrome: What Diagnostic Characteristics Are Defining?. <i>Journal of Child Neurology</i> , 2005, 20, 454-456.	1.4	10
65	Chromosome 17q21.31 duplication syndrome: Description of a new familiar case and further delineation of the clinical spectrum. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 183-187.	1.6	10
66	Spontaneous MRI improvement and absence of cerebral calcification in Aicardi-GoutiÃ“res syndrome: Diagnostic and disease-monitoring implications. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 489-494.	1.1	10
67	Comprehensive molecular screening strategy of <i><sc>OCLN</sc></i> in bandâ€“like calcification with simplified gyration and polymicrogyria. <i>Clinical Genetics</i> , 2018, 93, 228-234.	2.0	9
68	Cognitive aspects: sequencing, behavior, and executive functions. <i>Handbook of Clinical Neurology</i> / Edited By PJ Vinken and G W Bruyn, 2018, 154, 167-180.	1.8	8
69	A case of 3-methylglutaconic aciduria misdiagnosed as cerebral palsy. <i>Pediatric Neurology</i> , 2000, 23, 442-444.	2.1	7
70	Neurological phenotype of <sc>Potockiâ€“Lupski</sc> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2317-2324.	1.2	7
71	Oligoeyric microcephaly in a child with Williams syndrome. , 2002, 117A, 169-171.		6
72	Iatrogenic diabetes mellitus during ACTH therapy in an infant with West syndrome. <i>Acta Diabetologica</i> , 2011, 48, 345-347.	2.5	6

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73	Abnormal cerebellar foliation in EBF3 mutation. <i>Neurology</i> , 2020, 94, 933-935.	1.1	6
74	Basal Ganglia Dismorphism in Patients With Aicardi Syndrome. <i>Neurology</i> , 2021, 96, e1319-e1333.	1.1	6
75	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 88, 60-72.	2.0	6
76	Severe Phenotype in a Patient With Homozygous 15q21.2 Microdeletion Involving BCL2L10, GNB5, and MYO5C Genes, Resembling Infantile Developmental Disorder With Cardiac Arrhythmias (IDDCA). <i>Frontiers in Genetics</i> , 2020, 11, 399.	2.3	5
77	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. <i>European Journal of Medical Genetics</i> , 2021, 64, 104116.	1.3	5
78	The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 557-562.	1.7	4
79	Severe epilepsy in CNTNAP2-related Pitt-Hopkins-like syndrome successfully treated with stiripentol. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 88, 143-145.	2.0	4
80	A Case of Severe Early-Onset Neuropathy Caused by a Compound Heterozygous Deletion of the PMP22 Gene: Clinical and Neurographic Aspects. <i>Neuropediatrics</i> , 2020, 51, 173-177.	0.6	3
81	Chromosomal Microarray Analysis Has a Poor Diagnostic Yield in Children with Developmental Delay/Intellectual Disability When Concurrent Cerebellar Anomalies Are Present. <i>Cerebellum</i> , 2020, 19, 629-635.	2.5	3
82	A Missense De Novo Variant in the CASK-interactor KIRREL3 Gene Leading to Neurodevelopmental Disorder with Mild Cerebellar Hypoplasia. <i>Neuropediatrics</i> , 2021, 52, 484-488.	0.6	3
83	Identification of an Identical de Novo SCAMP5 Missense Variant in Four Unrelated Patients With Seizures and Severe Neurodevelopmental Delay. <i>Frontiers in Pharmacology</i> , 2020, 11, 599191.	3.5	2
84	CGH Findings in Children with Complex and Essential Autistic Spectrum Disorder. <i>Journal of Autism and Developmental Disorders</i> , 2021, , 1.	2.7	2
85	De Novo Duplication of Chromosome 13(q32-q34) in a Child With Developmental Delay. <i>Journal of Child Neurology</i> , 2006, 21, 1084-1085.	1.4	1
86	Craniodigital Syndrome of Scott: Clinical and Neuroradiological Features of a New Case. <i>Journal of Child Neurology</i> , 2007, 22, 883-886.	1.4	1
87	Flunarizine and Aspirin for Transient Hemiparesis in Sturge-Weber Syndrome. <i>Neuropediatrics</i> , 2019, 50, 406-407.	0.6	1
88	Clinical, Cognitive and Behavioural Assessment in Children with Cerebellar Disorder. <i>Applied Sciences (Switzerland)</i> , 2021, 11, 544.	2.5	1
89	Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. <i>Disability and Rehabilitation</i> , 2021, , 1-8.	1.8	1
90	Papillitis as an onset sign of Leber's hereditary optic neuropathy: a case report. <i>Brain and Development</i> , 2001, 23, 125-127.	1.1	0

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91	La sindrome di Aicardi-GoutiÃ¨res. The Neuroradiology Journal, 2003, 16, 511-514.	0.1	0
92	G.P.2.07 Alpha-dystroglycanopathy in an Italian patient due to large intragenic and single nucleotide deletions in the POMGnT1 gene. Neuromuscular Disorders, 2008, 18, 737.	0.6	0
93	Little folks, little myelin, and little teeth. Neurology, 2014, 83, 1884-1885.	1.1	0
94	Children with rare diseases: do they really have an increased risk of developing epilepsy?. Italian Journal of Pediatrics, 2015, 41, .	2.6	0
95	A Clinical-Based Diagnostic Approach to Cerebellar Atrophy in Children. Applied Sciences (Switzerland), 2021, 11, 2333.	2.5	0