Stefano D'Arrigo

List of Publications by Year in descending order

95 papers	4,224 citations	147786 31 h-index	123420 61 g-index
97 all docs	97 docs citations	97 times ranked	7187 citing authors

#	Article	IF	Citations
1	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	3.2	13
2	SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	3.2	19
3	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	3.2	21
4	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
5	Milder presentation of TELO2-related syndrome in two sisters homozygous for the p.Arg609His pathogenic variant. European Journal of Medical Genetics, 2021, 64, 104116.	1.3	5
6	Clinical, Cognitive and Behavioural Assessment in Children with Cerebellar Disorder. Applied Sciences (Switzerland), 2021, 11, 544.	2.5	1
7	A Clinical-Based Diagnostic Approach to Cerebellar Atrophy in Children. Applied Sciences (Switzerland), 2021, 11, 2333.	2.5	0
8	A Missense De Novo Variant in the CASK-interactor KIRREL3 Gene Leading to Neurodevelopmental Disorder with Mild Cerebellar Hypoplasia. Neuropediatrics, 2021, 52, 484-488.	0.6	3
9	Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. Disability and Rehabilitation, 2021, , 1-8.	1.8	1
10	Electroclinical features of MEF2C haploinsufficiency-related epilepsy: A multicenter European study. Seizure: the Journal of the British Epilepsy Association, 2021, 88, 60-72.	2.0	6
11	Severe epilepsy in CNTNAP2-related Pitt-Hopkins-like syndrome successfully treated with stiripentol. Seizure: the Journal of the British Epilepsy Association, 2021, 88, 143-145.	2.0	4
12	CGH Findings in Children with Complex and Essential Autistic Spectrum Disorder. Journal of Autism and Developmental Disorders, 2021, , 1.	2.7	2
13	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.7	15
14	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
15	A Case of Severe Early-Onset Neuropathy Caused by a Compound Heterozygous Deletion of the PMP22 Gene: Clinical and Neurographic Aspects. Neuropediatrics, 2020, 51, 173-177.	0.6	3
16	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	2.4	31
17	Neurological phenotype of <scp>Potockiâ€“Lupski</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2317-2324.	1.2	7
18	Identification of an Identical de Novo SCAMP5 Missense Variant in Four Unrelated Patients With Seizures and Severe Neurodevelopmental Delay. Frontiers in Pharmacology, 2020, 11, 599191.	3.5	2

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19	Abnormal cerebellar foliation in EBF3 mutation. Neurology, 2020, 94, 933-935.	1.1	6
20	Chromosomal Microarray Analysis Has a Poor Diagnostic Yield in Children with Developmental Delay/Intellectual Disability When Concurrent Cerebellar Anomalies Are Present. Cerebellum, 2020, 19, 629-635.	2.5	3
21	Severe Phenotype in a Patient With Homozygous 15q21.2 Microdeletion Involving BCL2L10, GNB5, and MYO5C Genes, Resembling Infantile Developmental Disorder With Cardiac Arrhythmias (IDDCA). Frontiers in Genetics, 2020, 11, 399.	2.3	5
22	Postural Control in Children with Cerebellar Ataxia. Applied Sciences (Switzerland), 2020, 10, 1606.	2.5	20
23	Consolidating the Role of TDP2 Mutations in Recessive Spinocerebellar Ataxia Associated with Pediatric Onset Drug Resistant Epilepsy and Intellectual Disability (SCAR23). Cerebellum, 2019, 18, 972-975.	2.5	12
24	Flunarizine and Aspirin for Transient Hemiparesis in Sturgeâ€“Weber Syndrome. Neuropediatrics, 2019, 50, 406-407.	0.6	1
25	Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	2.4	29
26	Spontaneous MRI improvement and absence of cerebral calcification in Aicardi-GoutiÃ¨res syndrome: Diagnostic and disease-monitoring implications. Molecular Genetics and Metabolism, 2019, 126, 489-494.	1.1	10
27	EED and EZH2 constitutive variants: A study to expand the Cohenâ€Gibson syndrome phenotype and contrast it with Weaver syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 588-594.	1.2	24
28	Clinical spectrum of PTEN mutation in pediatric patients. A bicenter experience. European Journal of Medical Genetics, 2019, 62, 103596.	1.3	39
29	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic POLR1C pathogenic variants. Neurology: Genetics, 2019, 5, e369.	1.9	38
30	Comprehensive molecular screening strategy of <scp>OCLN</scp> in bandâ€like calcification with simplified gyration and polymicrogyria. Clinical Genetics, 2018, 93, 228-234.	2.0	9
31	Between SCA5 and SCAR14: delineation of the SPTBN2 p.R480W-associated phenotype. European Journal of Human Genetics, 2018, 26, 928-929.	2.8	17
32	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	2.7	17
33	The noncoding RNA AK127244 in 2p16.3 locus: A new susceptibility region for neuropsychiatric disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 557-562.	1.7	4
34	Cognitive aspects: sequencing, behavior, and executive functions. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 154, 167-180.	1.8	8
35	ZC4H2 deletions can cause severe phenotype in female carriers. American Journal of Medical Genetics, Part A, 2017, 173, 1358-1363.	1.2	23
36	De novo p.T362R mutation in MORC2 causes early onset cerebellar ataxia, axonal polyneuropathy and nocturnal hypoventilation. Brain, 2017, 140, e34-e34.	7.6	17

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37	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563.	6.2	45
38	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
39	Epilepsy is a possible feature in Williamsâ€Beuren syndrome patients harboring typical deletions of the 7q11.23 critical region. American Journal of Medical Genetics, Part A, 2016, 170, 148-155.	1.2	29
40	Cognitive, adaptive, and behavioral features in Joubert syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3115-3124.	1.2	22
41	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	3.2	55
42	Chromosome 17q21.31 duplication syndrome: Description of a new familiar case and further delineation of the clinical spectrum. European Journal of Paediatric Neurology, 2016, 20, 183-187.	1.6	10
43	The Diagnostic Yield of Array Comparative Genomic Hybridization Is High Regardless of Severity of Intellectual Disability/Developmental Delay in Children. Journal of Child Neurology, 2016, 31, 691-699.	1.4	37
44	Children with rare diseases: do they really have an increased risk of developing epilepsy?. Italian Journal of Pediatrics, 2015, 41, .	2.6	0
45	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
46	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	1.2	447
47	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	3.8	30
48	Electroencephalographic (EEG) Photoparoxysmal Responses Under 5 Years of Age. Journal of Child Neurology, 2015, 30, 1824-1830.	1.4	12
49	Novel Mutations in TSEN54 in Pontocerebellar Hypoplasia Type 2. Journal of Child Neurology, 2014, 29, 520-525.	1.4	15
50	Consensus Paper: The Cerebellum's Role in Movement and Cognition. Cerebellum, 2014, 13, 151-177.	2.5	815
51	Little folks, little myelin, and little teeth. Neurology, 2014, 83, 1884-1885.	1.1	0
52	Seizures and EEG features in 74 patients with geneticâ€dysmorphic syndromes. American Journal of Medical Genetics, Part A, 2014, 164, 3154-3161.	1.2	16
53	Supratentorial and pontine <scp>MRI</scp> abnormalities characterize recessive spastic ataxia of <scp>C</scp>harlevoixâ€<scp>S</scp>aguenay. A comprehensive study of an <scp>I</scp>talian series. European Journal of Neurology, 2013, 20, 138-146.	3.3	57
54	Dandy-Walker malformation and Wisconsin syndrome: novel cases add further insight into the genotype-phenotype correlations of 3q23q25 deletions. Orphanet Journal of Rare Diseases, 2013, 8, 75.	2.7	19

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55	5p13 microduplication syndrome: A new case and better clinical definition of the syndrome. European Journal of Medical Genetics, 2013, 56, 54-58.	1.3	14
56	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
57	Novel POMGNT1 point mutations and intragenic rearrangements associated with muscle-eye-brain disease. Journal of the Neurological Sciences, 2012, 318, 45-50.	0.6	20
58	Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. Orphanet Journal of Rare Diseases, 2012, 7, 4.	2.7	64
59	Duplications of FOXG1 in 14q12 are associated with developmental epilepsy, mental retardation, and severe speech impairment. European Journal of Human Genetics, 2011, 19, 102-107.	2.8	104
60	Iatrogenic diabetes mellitus during ACTH therapy in an infant with West syndrome. Acta Diabetologica, 2011, 48, 345-347.	2.5	6
61	Pontine tegmental cap dysplasia: developmental and cognitive outcome in three adolescent patients. Orphanet Journal of Rare Diseases, 2011, 6, 36.	2.7	44
62	The â€œEye-of-the-Tigerâ€•Sign may be Absent in the Early Stages of Classic Pantothenate Kinase Associated Neurodegeneration. Neuropediatrics, 2011, 42, 159-162.	0.6	34
63	Molecular Mechanisms Generating and Stabilizing Terminal 22q13 Deletions in 44 Subjects with Phelan/McDermid Syndrome. PLoS Genetics, 2011, 7, e1002173.	3.5	172
64	Recessive Mutations in the Gene Encoding the Tight Junction Protein Occludin Cause Band-like Calcification with Simplified Gyration and Polymicrogyria. American Journal of Human Genetics, 2010, 87, 354-364.	6.2	123
65	Identification of previously unreported mutations in CHRNA1, CHRNE and RAPSN genes in three unrelated Italian patients with congenital myasthenic syndromes. Journal of Neurology, 2010, 257, 1119-1123.	3.6	11
66	The syndrome of perisylvian polymicrogyria with congenital arthrogryposis. Brain and Development, 2010, 32, 550-555.	1.1	14
67	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
68	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
69	Bandâ€like intracranial calcification with simplified gyration and polymicrogyria: A distinct â€œpseudoâ€TORCHâ€•phenotype. American Journal of Medical Genetics, Part A, 2008, 146A, 3173-3180.	1.2	46
70	Verbal dichotic listening performance and its relationship with EEG features in benign childhood epilepsy with centrotemporal spikes. Epilepsy Research, 2008, 79, 31-38.	1.6	39
71	Aicardiâ€Goutiã¨res syndrome: description of a late onset case. Developmental Medicine and Child Neurology, 2008, 50, 631-634.	2.1	35
72	RPGRIP1L mutations are mainly associated with the cerebelloâ€renal phenotype of Joubert syndromeâ€related disorders. Clinical Genetics, 2008, 74, 164-170.	2.0	64

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73	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
74	Ataxia With Oculomotor Apraxia Type 1 (AOA1): Clinical and Neuropsychological Features in 2 New Patients and Differential Diagnosis. Journal of Child Neurology, 2008, 23, 895-900.	1.4	22
75	The Effectiveness of Hospitalization in the Treatment of Paediatric Idiopathic Headache Patients. Psychopathology, 2007, 40, 1-7.	1.5	14
76	Craniodigital Syndrome of Scott: Clinical and Neuroradiological Features of a New Case. Journal of Child Neurology, 2007, 22, 883-886.	1.4	1
77	Verbal and Gestural Communication in Children With Bilateral Perisylvian Polymicrogyria. Journal of Child Neurology, 2007, 22, 1090-1098.	1.4	12
78	Intellectual and language findings and their relationship to EEG characteristics in benign childhood epilepsy with centrotemporal spikes. Epilepsy and Behavior, 2007, 10, 278-285.	1.7	113
79	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	6.2	137
80	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
81	Cognitive and Behavioural Effects of Migraine in Childhood and Adolescence. Cephalalgia, 2006, 26, 596-603.	3.9	46
82	De Novo Duplication of Chromosome 13(q32-q34) in a Child With Developmental Delay. Journal of Child Neurology, 2006, 21, 1084-1085.	1.4	1
83	Unilateral frontal lobe epilepsy affects executive functions in children. Neurological Sciences, 2005, 26, 263-270.	1.9	53
84	The natural history of Aicardi-Goutieres syndrome: Follow-up of 11 Italian patients. Neurology, 2005, 64, 1621-1624.	1.1	42
85	Progressive Encephalopathy with Edema, Hypsarrhythmia, and Optic Nerve Atrophy (PEHO)-Like Syndrome: What Diagnostic Characteristics Are Defining?. Journal of Child Neurology, 2005, 20, 454-456.	1.4	10
86	Diagnostic Approach to Cerebellar Disease in Children. Journal of Child Neurology, 2005, 20, 859-866.	1.4	22
87	Another Patient With MECP2 Mutation Without Classic Rett Syndrome Phenotype. Pediatric Neurology, 2005, 32, 355-357.	2.1	20
88	Unusual neurophysiological features in Cockayne's syndrome: a report of two cases as a contribution to diagnosis and classification. Brain and Development, 2004, 26, 273-280.	1.1	13
89	Are Vascular Disorders More Prevalent in the Relatives of Children and Adolescents with Migraine?. Cephalalgia, 2003, 23, 887-891.	3.9	12
90	La sindrome di Aicardi-GoutiÃ¨res. The Neuroradiology Journal, 2003, 16, 511-514.	0.1	0

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91	Aicardi-GoutiÃ¨res syndrome: a description of 21 new cases and a comparison with the literature. European Journal of Paediatric Neurology, 2002, 6, A9-A22.	1.6	50
92	Oligoyric microcephaly in a child with Williams syndrome. , 2002, 117A, 169-171.		6
93	Papillitis as an onset sign of Leber's hereditary optic neuropathy: a case report. Brain and Development, 2001, 23, 125-127.	1.1	0
94	Personality Traits in Childhood and Adolescent Headache. Cephalalgia, 2001, 21, 53-60.	3.9	39
95	A case of 3-methylglutaconic aciduria misdiagnosed as cerebral palsy. Pediatric Neurology, 2000, 23, 442-444.	2.1	7