Romina Romaniello

List of Publications by Year
in descending order

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58	1,290	361413	414414
papers	citations	20 h-index	32 g-index

62	62	62	2380
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Remote cognitive training for children with congenital brain malformation or genetic syndrome: a scoping review. Journal of Intellectual Disabilities, 2023, 27, 808-841.	1.4	1
2	Cognitive predictors of Social processing in congenital atypical development. Journal of Autism and Developmental Disorders, 2023, 53, 3343-3355.	2.7	2
3	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
4	Virtual-Reality Performance-Based Assessment of Cognitive Functions in Adult Patients With Acquired Brain Injury: A Scoping Review. Neuropsychology Review, 2022, 32, 352-399.	4.9	9
5	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	3.6	12
6	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
7	Get Your Molar Tooth Right: Joubert Syndrome Misdiagnosis Unmasked by Whole-Exome Sequencing. Cerebellum, 2022, 21, 1144-1150.	2.5	7
8	Cerebellar Agenesis. , 2022, , 2113-2134.		0
9	SCN2A Pathogenic Variants and Epilepsy: Heterogeneous Clinical, Genetic and Diagnostic Features. Brain Sciences, 2022, 12, 18.	2.3	5
10	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	3.2	21
11	Defining the phenotypical spectrum associated with variants in TUBB2A. Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11
12	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
13	Novel SPTBN2 gene mutation and first intragenic deletion in early onset spinocerebellar ataxia type 5. Annals of Clinical and Translational Neurology, 2021, 8, 956-963.	3.7	12
14	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	1.6	7
15	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
16	The Fetus with Ganglionic Eminence Abnormality: Head Size and Extracranial Sonographic Findings Predict Genetic Diagnoses and Postnatal Outcomes. American Journal of Neuroradiology, 2021, 42, 1528-1534.	2.4	5
17	Social prediction in pediatric patients with congenital, non-progressive malformations of the cerebellum: From deficits in predicting movements to rehabilitation in virtual reality. Cortex, 2021, 144, 82-98.	2.4	8
18	Longâ€term followâ€up in a cohort of children with isolated corpus callosum agenesis at fetal MRI. Annals of Clinical and Translational Neurology, 2021, 8, 2280-2288.	3.7	4

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19	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
20	Characterizing White Matter Tract Organization in Polymicrogyria and Lissencephaly: A Multifiber Diffusion MRI Modeling and Tractography Study. American Journal of Neuroradiology, 2020, 41, 1495-1502.	2.4	9
21	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.1	26
22	Targeted next-generation sequencing identifies the disruption of the SHANK3 and RYR2 genes in a patient carrying a de novo t(1;22)(q43;q13.3) associated with signs of Phelan-McDermid syndrome. Molecular Cytogenetics, 2020, 13, 22.	0.9	4
23	Virtual Reality Social Prediction Improvement and Rehabilitation Intensive Training (VR-SPIRIT) for paediatric patients with congenital cerebellar diseases: study protocol of a randomised controlled trial. Trials, 2020, 21, 82.	1.6	16
24	Loss of ap4s1 in zebrafish leads to neurodevelopmental defects resembling spastic paraplegia 52. Annals of Clinical and Translational Neurology, 2020, 7, 584-589.	3.7	15
25	Cerebellar Agenesis. , 2020, , 1-23.		0
26	The spectrum of brainstem malformations associated to mutations of the tubulin genes family: MRI and DTI analysis. European Radiology, 2019, 29, 770-782.	4.5	22
27	Epilepsy in Tubulinopathy: Personal Series and Literature Review. Cells, 2019, 8, 669.	4.1	27
28	The phenotypic spectrum of WWOX-related disorders: 20 additional cases of WOREE syndrome and review of the literature. Genetics in Medicine, 2019, 21, 1308-1318.	2.4	48
29	Tubulin genes and malformations of cortical development. European Journal of Medical Genetics, 2018, 61, 744-754.	1.3	93
30	Feasibility of a home-based computerized cognitive training for pediatric patients with congenital or acquired brain damage: An explorative study. PLoS ONE, 2018, 13, e0199001.	2.5	22
31	Anterior Mesencephalic Cap Dysplasia: Novel Brain Stem Malformative Features Associated with Joubert Syndrome. American Journal of Neuroradiology, 2017, 38, 2385-2390.	2.4	15
32	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
33	Clinical Characterization, Genetics, and Long-Term Follow-up of a Large Cohort of Patients With Agenesis of the Corpus Callosum. Journal of Child Neurology, 2017, 32, 60-71.	1.4	34
34	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Porettiâ€“Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267.	2.8	43
35	Automatic localization of cerebral cortical malformations using fractal analysis. Physics in Medicine and Biology, 2016, 61, 6025-6040.	3.0	4
36	Aberrant supracallosal longitudinal bundle: MR features, pathogenesis and associated clinical phenotype. European Radiology, 2016, 26, 2587-2596.	4.5	7

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37	Learning to live without the cerebellum. NeuroReport, 2015, 26, 809-813.	1.2	17
38	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
39	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	3.8	30
40	A de-novo STXBP1 gene mutation in a patient showing the Rett syndrome phenotype. NeuroReport, 2015, 26, 254-257.	1.2	39
41	Neurophysiological and clinical findings on Nodding Syndrome in 21 South Sudanese children and a review of the literature. Seizure: the Journal of the British Epilepsy Association, 2015, 31, 64-71.	2.0	23
42	Cognitive rehabilitation in a child with Joubert Syndrome: Developmental trends and adaptive changes in a single case report. Research in Developmental Disabilities, 2015, 47, 375-384.	2.2	15
43	Mutations in Î±- and Î²-tubulin encoding genes: Implications in brain malformations. Brain and Development, 2015, 37, 273-280.	1.1	94
44	Epilepsy in Multigene Tubulin Family Mutations. Journal of Neurology & Neurophysiology, 2015, 06, .	0.1	1
45	A Novel Mutation in STXBP1 Gene in a Child With Epileptic Encephalopathy and an Atypical Electroclinical Pattern. Journal of Child Neurology, 2014, 29, 249-253.	1.4	18
46	Brain malformations and mutations in Î±â€•and Î²â€tubulin genes: a review of the literature and description of two new cases. Developmental Medicine and Child Neurology, 2014, 56, 354-360.	2.1	42
47	Novel SETX variants in a patient with ataxia, neuropathy, and oculomotor apraxia are associated with normal sensitivity to oxidative DNA damaging agents. Brain and Development, 2014, 36, 682-689.	1.1	8
48	Cerebroretinal Microangiopathy With Calcifications and Cysts Associated With CTC1 and NDP Mutations. Journal of Child Neurology, 2013, 28, 1702-1708.	1.4	11
49	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
50	Cerebellar Agenesis. , 2013, , 1855-1872.		4
51	A novel mutation in the Î²â€tubulin gene TUBB2B associated with complex malformation of cortical development and deficits in axonal guidance. Developmental Medicine and Child Neurology, 2012, 54, 765-769.	2.1	50
52	Electroclinical pattern in MECP2 duplication syndrome: Eight new reported cases and review of literature. Epilepsia, 2012, 53, 1146-1155.	5.1	34
53	Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. Orphanet Journal of Rare Diseases, 2012, 7, 4.	2.7	64
54	Novel spliceâ€site mutations and a large intragenic deletion in PLA2G6 associated with a severe and rapidly progressive form of infantile neuroaxonal dystrophy. Clinical Genetics, 2010, 78, 432-440.	2.0	30

#	Article	IF	Citations
55	A wide spectrum of clinical, neurophysiological and neuroradiological abnormalities in a family with a novel CACNA1A mutation. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 840-843.	1.9	51
56	Disturbi dell'attenzione ed epilessia: revisione della letteratura ed esperienza personale. Child Development & Disabilities, 2010, , 95-108.	0.0	0
57	Cryptogenic Epileptic Syndromes Related to SCN1A. Archives of Neurology, 2008, 65, 489.	4.5	43
58	Assigning single clinical features to their disease-locus in large deletions: the example of chromosome 1q23-25 deletion syndrome. , 0, , .		1