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	Mutations in Î±- and Î²-tubulin encoding genes: Implications in brain malformations. Brain and Development, 2015, 37, 273-280.	1.1	94
2	Tubulin genes and malformations of cortical development. European Journal of Medical Genetics, 2018, 61, 744-754.	1.3	93
3	Delineation and Diagnostic Criteria of Oral-Facial-Digital Syndrome Type VI. Orphanet Journal of Rare Diseases, 2012, 7, 4.	2.7	64
4	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
5	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. ELife, 2015, 4, e06602.	6.0	64
6	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
7	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
8	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
9	Cryptogenic Epileptic Syndromes Related to SCN1A. Archives of Neurology, 2008, 65, 489.	4.5	43
10	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
11	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
12	A de-novo STXBP1 gene mutation in a patient showing the Rett syndrome phenotype. NeuroReport, 2015, 26, 254-257.	1.2	39
13	Tubulin-related cerebellar dysplasia: definition of a distinct pattern of cerebellar malformation. European Radiology, 2017, 27, 5080-5092.	4.5	36
14	Electroclinical pattern in MECP2 duplication syndrome: Eight new reported cases and review of literature. Epilepsia, 2012, 53, 1146-1155.	5.1	34
15	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
16	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
17	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. Human Genetics, 2015, 134, 123-126.	3.8	30
18	Epilepsy in Tubulinopathy: Personal Series and Literature Review. Cells, 2019, 8, 669.	4.1	27

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19	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.1	26
20	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
21	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
22	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
23	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	3.2	21
24	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
25	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
26	Learning to live without the cerebellum. NeuroReport, 2015, 26, 809-813.	1.2	17
27	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
28	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
29	Anterior Mesencephalic Cap Dysplasia: Novel Brain Stem Malformative Features Associated with Joubert Syndrome. American Journal of Neuroradiology, 2017, 38, 2385-2390.	2.4	15
30	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
31	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
32	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
33	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
34	Monoallelic KIF1A-related disorders: a multicenter cross sectional study and systematic literature review. Journal of Neurology, 2022, 269, 437-450.	3.6	12
35	Cerebroretinal Microangiopathy With Calcifications and Cysts Associated With CTC1 and NDP Mutations. Journal of Child Neurology, 2013, 28, 1702-1708.	1.4	11
36	Defining the phenotypical spectrum associated with variants in TUBB2A. Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11

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37	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
38	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
39	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
40	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
41	Aberrant supracallosal longitudinal bundle: MR features, pathogenesis and associated clinical phenotype. European Radiology, 2016, 26, 2587-2596.	4.5	7
42	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	1.6	7
43	Get Your Molar Tooth Right: Joubert Syndrome Misdiagnosis Unmasked by Whole-Exome Sequencing. Cerebellum, 2022, 21, 1144-1150.	2.5	7
44	Basal Ganglia Dysmorphism in Patients With Aicardi Syndrome. Neurology, 2021, 96, e1319-e1333.	1.1	6
45	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
46	SCN2A Pathogenic Variants and Epilepsy: Heterogeneous Clinical, Genetic and Diagnostic Features. Brain Sciences, 2022, 12, 18.	2.3	5
47	Automatic localization of cerebral cortical malformations using fractal analysis. Physics in Medicine and Biology, 2016, 61, 6025-6040.	3.0	4
48	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
49	Cerebellar Agenesis. , 2013, , 1855-1872.		4
50	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
51	Cognitive predictors of Social processing in congenital atypical development. Journal of Autism and Developmental Disorders, 2023, 53, 3343-3355.	2.7	2
52	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
53	Epilepsy in Multigene Tubulin Family Mutations. Journal of Neurology & Neurophysiology, 2015, 06, .	0.1	1
54	Assigning single clinical features to their disease-locus in large deletions: the example of chromosome 1q23-25 deletion syndrome. , 0, , .		1

#	Article	IF	Citations
55	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
56	Disturbi dell'attenzione ed epilessia: revisione della letteratura ed esperienza personale. Child Development & Disabilities, 2010, , 95-108.	0.0	0
57	Cerebellar Agenesis. , 2020, , 1-23.		0
58	Cerebellar Agenesis. , 2022, , 2113-2134.		0