Marcello Scala

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8231774/publications.pdf

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62	958	567281	580821
papers	citations	15 h-index	25 g-index

68	68	68	1572
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
2	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
3	Advances in genetic testing and optimization of clinical management in children and adults with epilepsy. Expert Review of Neurotherapeutics, 2020, 20, 251-269.	2.8	45
4	Three de novo DDX3X variants associated with distinctive brain developmental abnormalities and brain tumor in intellectually disabled females. European Journal of Human Genetics, 2019, 27, 1254-1259.	2.8	41
5	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	6.2	35
6	CNNM2 homozygous mutations cause severe refractory hypomagnesemia, epileptic encephalopathy and brain malformations. European Journal of Medical Genetics, 2019, 62, 198-203.	1.3	28
7	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
8	Biallelic variants in CTU2 cause DREAMâ€PL syndrome and impair thiolation of tRNA wobble U34. Human Mutation, 2019, 40, 2108-2120.	2.5	25
9	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
10	Radiation-Induced Moyamoya Syndrome in Children with Brain Tumors: Case Series and Literature Review. World Neurosurgery, 2020, 135, 118-129.	1.3	23
11	When and why is surgical revascularization indicated for the treatment of moyamoya syndrome in patients with RASopathies? A systematic review of the literature and a single institute experience. Child's Nervous System, 2018, 34, 1311-1323.	1.1	22
12	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	2.8	22
13	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
14	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
15	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: A Single-Center Cohort Study. Cancers, 2021, 13, 1879.	3.7	21
16	Limits and pitfalls of indirect revascularization in moyamoya disease and syndrome. Neurosurgical Review, 2021, 44, 1877-1887.	2.4	17
17	Targeted re-sequencing for early diagnosis of genetic causes of childhood epilepsy: the Italian experience from the â€˜beyond epilepsyâ€™ project. Italian Journal of Pediatrics, 2020, 46, 92.	2.6	17
18	Pathophysiological Mechanisms in Neurodevelopmental Disorders Caused by Rac GTPases Dysregulation: Whatâ€™s behind Neuro-RACopathies. Cells, 2021, 10, 3395.	4.1	17

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19	Diagnostic Approach to Macrocephaly in Children. Frontiers in Pediatrics, 2021, 9, 794069.	1.9	17
20	A novel pathogenic MYH3 mutation in a child with Sheldonâ€“Hall syndrome and vertebral fusions. American Journal of Medical Genetics, Part A, 2018, 176, 663-667.	1.2	16
21	Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Journal of Medical Genetics, 2021, 58, 495-504.	3.2	14
22	Familial ROBO1 deletion associated with ectopic posterior pituitary, duplication of the pituitary stalk and anterior pituitary hypoplasia. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 95-99.	0.9	11
23	Spatial coefficient of variation applied to arterial spin labeling MRI may contribute to predict surgical revascularization outcomes in pediatric moyamoya vasculopathy. Neuroradiology, 2020, 62, 1003-1015.	2.2	11
24	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
25	Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Journal of the Neurological Sciences, 2021, 424, 117409.	0.6	11
26	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.1	11
27	ZTTK syndrome: Clinical and molecular findings ofÂ15 cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3740-3753.	1.2	11
28	Homozygous missense WIPI2 variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Brain Communications, 2021, 3, fcab183.	3.3	10
29	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10
30	Novel CNS malformations and skeletal anomalies in a patient with Beaulieuâ€boycottâ€innes syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2835-2840.	1.2	9
31	Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltageâ€gated sodium channel function. Epilepsia, 2021, 62, e82-e87.	5.1	9
32	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
33	Atypical choroid plexus papilloma: spontaneous resolution of diffuse leptomeningeal contrast enhancement after primary tumor removal in 2 pediatric cases. Journal of Neurosurgery: Pediatrics, 2017, 20, 284-288.	1.3	8
34	Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. Neurogenetics, 2019, 20, 165-172.	1.4	8
35	â€˜Distal 16p12.2 microdeletionâ€™ in a patient with autosomal recessive deafness-22. Journal of Genetics, 2019, 98, 1.	0.7	8
36	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	2.8	8

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37	Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. International Journal of Molecular Sciences, 2021, 22, 4471.	4.1	8
38	Gain-of-function p.F28S variant inRAC3disrupts neuronal differentiation, migration and axonogenesis during cortical development, leading to neurodevelopmental disorder. Journal of Medical Genetics, 2023, 60, 223-232.	3.2	8
39	De novo ARHGEF9 missense variants associated with neurodevelopmental disorder in females: expanding the genotypic and phenotypic spectrum of ARHGEF9 disease in females. Neurogenetics, 2021, 22, 87-94.	1.4	7
40	Aggressive desmoid fibromatosis in Kabuki syndrome: Expanding the tumor spectrum. Pediatric Blood and Cancer, 2019, 66, e27831.	1.5	6
41	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
42	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
43	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	2.0	6
44	Exome survey of individuals affected by VATER / VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. American Journal of Medical Genetics, Part A, 2021, 185, 3784-3792.	1.2	6
45	The Pathophysiological Link Between Reelin and Autism: Overview and New Insights. Frontiers in Genetics, 2022, 13, 869002.	2.3	6
46	De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. Human Mutation, 2022, 43, 1299-1313.	2.5	6
47	Radiation-Induced Moyamoya Syndrome After Proton Therapy in Child with Clival Chordoma: Natural History and Surgical Treatment. World Neurosurgery, 2019, 123, 306-309.	1.3	5
48	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7 â€related disorder. Birth Defects Research, 2020, 112, 1085-1092.	1.5	5
49	Congenital posterior cervical spine malformation due to biallelic c.240â€4T>G RIPPLY2 variant: A discrete entity. American Journal of Medical Genetics, Part A, 2020, 182, 1466-1472.	1.2	5
50	Hyperkinetic stereotyped movements in a boy with biallelic CNTNAP2 variants. Italian Journal of Pediatrics, 2021, 47, 208.	2.6	5
51	Abnormal circadian rhythm in patients with GRIN1-related developmental epileptic encephalopathy. European Journal of Paediatric Neurology, 2019, 23, 657-661.	1.6	4
52	Symptomatic eating epilepsy: two novel pediatric patients and review of literature. Italian Journal of Pediatrics, 2021, 47, 137.	2.6	4
53	Structural brain anomalies in Cri-du-Chat syndrome: MRI findings in 14 patients and possible genotype-phenotype correlations. European Journal of Paediatric Neurology, 2020, 28, 110-119.	1.6	3
54	Novel likely disease-causing CLN5 variants identified in Pakistani patients with neuronal ceroid lipofuscinosis. Journal of the Neurological Sciences, 2020, 414, 116826.	0.6	3

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55	RNF213 variant in a patient with Legius syndrome associated with moyamoya syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1669.	1.2	3
56	A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. Frontiers in Pediatrics, 2022, 10, 847549.	1.9	3
57	Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401.	1.3	2
58	De novo POLR2A p.(Ile457Thr) variant associated with early-onset encephalopathy and cerebellar atrophy: expanding the phenotypic spectrum. Brain and Development, 2022, 44, 480-485.	1.1	2
59	Genotypeâ€“phenotype spectrum and correlations in <scp>Xiaâ€Gibbs</scp> syndrome: Report of five novel cases and literature review. Birth Defects Research, 0, , .	1.5	2
60	Pelizaeusâ€“Merzbacher Disease due to PLP1 Frameshift Mutation in a Female with Nonrandom Skewed X-Chromosome Inactivation. Neuropediatrics, 2019, 50, 268-270.	0.6	1
61	Precision medicine in early-onset epilepsy: the KCNQ2 paradigm. , 0, , .		1
62	Letter to the Editor Regarding â€œPrimary Aneurysmal Bone Cyst of the Thoracic Spine: A Pediatric Case Reportâ€. World Neurosurgery, 2020, 144, 322.	1.3	0