Marcello Scala

List of Publications by Year
in descending order

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

Version: 2024-02-01

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	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
2	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
3	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10
4	The Pathophysiological Link Between Reelin and Autism: Overview and New Insights. Frontiers in Genetics, 2022, 13, 869002.	2.3	6
5	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
6	A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. Frontiers in Pediatrics, 2022, 10, 847549.	1.9	3
7	De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. Human Mutation, 2022, 43, 1299-1313.	2.5	6
8	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
9	Limits and pitfalls of indirect revascularization in moyamoya disease and syndrome. Neurosurgical Review, 2021, 44, 1877-1887.	2.4	17
10	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
11	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
12	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
13	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
14	Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. International Journal of Molecular Sciences, 2021, 22, 4471.	4.1	8
15	Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltageâ€gated sodium channel function. Epilepsia, 2021, 62, e82-e87.	5.1	9
16	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: A Single-Center Cohort Study. Cancers, 2021, 13, 1879.	3.7	21
17	Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401.	1.3	2
18	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

#	Article	IF	Citations
19	Italian cohort of Lafora disease: Clinical features, disease evolution, and genotype-phenotype correlations. Journal of the Neurological Sciences, 2021, 424, 117409.	0.6	11
20	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
21	RNF213 variant in a patient with Legius syndrome associated with moyamoya syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1669.	1.2	3
22	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
23	Symptomatic eating epilepsy: two novel pediatric patients and review of literature. Italian Journal of Pediatrics, 2021, 47, 137.	2.6	4
24	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.1	11
25	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
26	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
27	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
28	Hyperkinetic stereotyped movements in a boy with biallelic CNTNAP2 variants. Italian Journal of Pediatrics, 2021, 47, 208.	2.6	5
29	Pathophysiological Mechanisms in Neurodevelopmental Disorders Caused by Rac GTPases Dysregulation: Whatâ€™s behind Neuro-RACopathies. Cells, 2021, 10, 3395.	4.1	17
30	Diagnostic Approach to Macrocephaly in Children. Frontiers in Pediatrics, 2021, 9, 794069.	1.9	17
31	Radiation-Induced Moyamoya Syndrome in Children with Brain Tumors: Case Series and Literature Review. World Neurosurgery, 2020, 135, 118-129.	1.3	23
32	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
33	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
34	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
35	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
36	Sinus pericranii, skull defects, and structural brain anomalies in TRAF7 â€related disorder. Birth Defects Research, 2020, 112, 1085-1092.	1.5	5

#	Article	IF	Citations
37	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
38	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
39	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
40	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	2.8	22
41	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
42	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
43	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
44	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
45	CNNM2 homozygous mutations cause severe refractory hypomagnesemia, epileptic encephalopathy and brain malformations. European Journal of Medical Genetics, 2019, 62, 198-203.	1.3	28
46	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
47	Biallelic variants in CTU2 cause DREAMâ€PL syndrome and impair thiolation of tRNA wobble U34. Human Mutation, 2019, 40, 2108-2120.	2.5	25
48	Novel homozygous TSFM pathogenic variant associated with encephalocardiomyopathy with sensorineural hearing loss and peculiar neuroradiologic findings. Neurogenetics, 2019, 20, 165-172.	1.4	8
49	Abnormal circadian rhythm in patients with GRIN1-related developmental epileptic encephalopathy. European Journal of Paediatric Neurology, 2019, 23, 657-661.	1.6	4
50	Aggressive desmoid fibromatosis in Kabuki syndrome: Expanding the tumor spectrum. Pediatric Blood and Cancer, 2019, 66, e27831.	1.5	6
51	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
52	â€˜Distal 16p12.2 microdeletionâ€™ in a patient with autosomal recessive deafness-22. Journal of Genetics, 2019, 98, 1.	0.7	8
53	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
54	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

#	Article	IF	Citations
55	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
56	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
57	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
58	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
59	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
60	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
61	Precision medicine in early-onset epilepsy: the KCNQ2 paradigm. , 0, , .		1
62	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