Vincenzo Salpietro

List of Publications by Year
in descending order

76	1,703	361413	377865
papers	citations	20 h-index	34 g-index

78	78	78	3193
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	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	122
3	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
4	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
5	PRRT2 Regulates Synaptic Fusion by Directly Modulating SNARE Complex Assembly. Cell Reports, 2018, 22, 820-831.	6.4	67
6	Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837.	7.6	64
7	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
8	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
9	Delineation of the movement disorders associated with FOXG1 mutations. Neurology, 2016, 86, 1794-1800.	1.1	55
10	A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661.	7.6	52
11	A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488.	3.9	52
12	Pontocerebellar hypoplasia type 2D and optic nerve atrophy further expand the spectrum associated with selenoprotein biosynthesis deficiency. European Journal of Paediatric Neurology, 2016, 20, 483-488.	1.6	49
13	Homozygous mutations in <scp>VAMP</scp>1 cause a presynaptic congenital myasthenic syndrome. Annals of Neurology, 2017, 81, 597-603.	5.3	48
14	A loss-of-function homozygous mutation in DDX59 implicates a conserved DEAD-box RNA helicase in nervous system development and function. Human Mutation, 2018, 39, 187-192.	2.5	44
15	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
16	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. Movement Disorders, 2018, 33, 1961-1965.	3.9	38
17	The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. Brain, 2017, 140, e49-e49.	7.6	33
18	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32

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19	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
20	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
21	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	6.2	24
22	Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528.	1.9	24
23	New Trends and Most Promising Therapeutic Strategies for Epilepsy Treatment. Frontiers in Neurology, 2021, 12, 753753.	2.4	23
24	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	2.8	22
25	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
26	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
27	A Review of Copy Number Variants in Inherited Neuropathies. Current Genomics, 2018, 19, 412-419.	1.6	19
28	Genotype-phenotype correlations and expansion of the molecular spectrum of AP4M1-related hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2017, 12, 172.	2.7	17
29	Diagnostic Approach to Macrocephaly in Children. Frontiers in Pediatrics, 2021, 9, 794069.	1.9	17
30	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	1.6	14
31	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	1.9	13
32	Targeted re-sequencing in malformations of cortical development: genotype-phenotype correlations. Seizure: the Journal of the British Epilepsy Association, 2020, 80, 145-152.	2.0	13
33	A novel SLC1A4 homozygous mutation causing congenital microcephaly, epileptic encephalopathy and spastic tetraparesis: a video-EEG and tractography â€“ case study. Journal of Neurogenetics, 2018, 32, 316-321.	1.4	12
34	Kleineâ€Łevin syndrome is associated with LMOD3 variants. Journal of Sleep Research, 2019, 28, e12718.	3.2	12
35	Rare novel CYP2U1 and ZFYVE26 variants identified in two Pakistani families with spastic paraplegia. Journal of the Neurological Sciences, 2020, 411, 116669.	0.6	12
36	Synaptopathies in Developmental and Epileptic Encephalopathies: A Focus on Pre-synaptic Dysfunction. Frontiers in Neurology, 2022, 13, 826211.	2.4	12

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37	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
38	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
39	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
40	Pediatric stroke: current diagnostic and management challenges. Quantitative Imaging in Medicine and Surgery, 2018, 8, 984-991.	2.0	10
41	Paroxysmal Movement Disorder and Epilepsy Caused by a De Novo Truncating Mutation in KAT6A. Journal of Pediatric Genetics, 2018, 07, 114-116.	0.7	10
42	Complex Neurological Phenotype Associated with a De Novo DHDDS Mutation in a Boy with Intellectual Disability, Refractory Epilepsy, and Movement Disorder. Journal of Pediatric Genetics, 2021, 10, 236-238.	0.7	10
43	Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. Brain Sciences, 2021, 11, 1150.	2.3	10
44	Epileptic Phenotypes Associated With SNAREs and Related Synaptic Vesicle Exocytosis Machinery. Frontiers in Neurology, 2021, 12, 806506.	2.4	9
45	De novo GRIN2A variants associated with epilepsy and autism and literature review. Epilepsy and Behavior, 2022, 129, 108604.	1.7	9
46	Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. International Journal of Molecular Sciences, 2021, 22, 4471.	4.1	8
47	Electroclinical history of a five-year-old girl with GRIN1-related early-onset epileptic encephalopathy: a video-case study. Epileptic Disorders, 2018, 20, 423-427.	1.3	7
48	Genotype-Phenotype Correlations in Charcot-Marie-Tooth Disease Due to MTMR2 Mutations and Implications in Membrane Trafficking. Frontiers in Neuroscience, 2019, 13, 974.	2.8	7
49	Biotin-Thiamine Responsive Encephalopathy: Report of an Egyptian Family with a Novel SLC19A3 Mutation and Review of the Literature. Journal of Pediatric Genetics, 2019, 08, 100-108.	0.7	7
50	Electroclinical features and outcome of ANKRD11-related KBG syndrome: A novel report and literature review. Seizure: the Journal of the British Epilepsy Association, 2021, 85, 151-154.	2.0	7
51	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
52	Neuromuscular and Neuroendocrinological Features Associated With ZC4H2-Related Arthrogryposis Multiplex Congenita in a Sicilian Family: A Case Report. Frontiers in Neurology, 2021, 12, 704747.	2.4	6
53	Benign familial infantile epilepsy associated with KCNQ3 mutation: a rare occurrence or an underestimated event?. Epileptic Disorders, 2020, 22, 807-810.	1.3	6
54	Broad neurodevelopmental features and cortical anomalies associated with a novel de novo KMT2A variant in Wiedemannâ^'Steiner syndrome. European Journal of Medical Genetics, 2021, 64, 104133.	1.3	5

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55	Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. European Journal of Human Genetics, 2021, 29, 1027-1031.	2.8	5
56	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	2.5	5
57	Commonalities and distinctions between two neurodevelopmental disorder subtypes associated with <scp>SCN2A</scp> and <scp>SCN8A</scp> variants and literature review. Molecular Genetics & Genomic Medicine, 2022, 10, e1911.	1.2	5
58	Tay-Sachs Disease: Two Novel Rare HEXA Mutations from Pakistan and Morocco. Klinische Padiatrie, 2021, 233, 226-230.	0.6	4
59	A paradigmatic autistic phenotype associated with loss of PCDH11Y and NLGN4Y genes. BMC Medical Genomics, 2021, 14, 98.	1.5	4
60	PKD or Not PKD: That is the question. Annals of Neurology, 2016, 80, 167-168.	5.3	3
61	Further supporting evidence for REEP1 phenotypic and allelic heterogeneity. Neurology: Genetics, 2019, 5, e379.	1.9	3
62	A rare PANK2 deletion in the first north African patient affected with pantothenate kinase associated neurodegeneration. Journal of the Neurological Sciences, 2020, 410, 116639.	0.6	3
63	Identification of common genetic markers of paroxysmal neurological disorders using a network analysis approach. Neurological Sciences, 2020, 41, 851-857.	1.9	3
64	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. Journal of Medical Genetics, 2021, 58, 815-831.	3.2	3
65	Novel variants underlying autosomal recessive intellectual disability in Pakistani consanguineous families. BMC Medical Genetics, 2020, 21, 59.	2.1	3
66	De novo mutation in SLC25A22 gene: expansion of the clinical and electroencephalographic phenotype. Journal of Neurogenetics, 2021, 35, 67-73.	1.4	3
67	Mutations inTAF8cause a neurodegenerative disorder. Brain, 2022, 145, 3022-3034.	7.6	3
68	Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401.	1.3	2
69	Age-dependent epileptic encephalopathy associated with an unusual co-occurrence of ZEB2 and SCN1A variants. , 2020, 22, 111-115.		2
70	A complex epileptic and dysmorphic phenotype associated with a novel frameshift KDM5B variant and deletion of SCN gene cluster. Seizure: the Journal of the British Epilepsy Association, 2022, 97, 20-22.	2.0	2
71	Paroxysmal limb dystonias associated with GABBR2 pathogenic variant: A case-based literature review. Brain and Development, 2022, , .	1.1	2
72	Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50.	7.6	1

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73	Genetic epilepsies and the Kv super-family. European Journal of Paediatric Neurology, 2020, 24, 5-6.	1.6	1
74	Recurrent missense variant in the nuclear export signal of FMR1 associated with FXS-like phenotype including intellectual disability, ASD, facial abnormalities. European Journal of Medical Genetics, 2022, 65, 104441.	1.3	1
75	KCNQ2-Related Neonatal Epilepsy Treated With Vitamin B6: A Report of Two Cases and Literature Review. Frontiers in Neurology, 2022, 13, 826225.	2.4	1
76	Pioneers and Emerging Pediatric Neurologists and Epileptologists in the World. Journal of Pediatric Epilepsy, 2019, 08, 001-001.	0.2	0