# 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 | 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 |
| 2 | Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1. | 2.5 | 5 |
| 3 | Synaptopathies in Developmental and Epileptic Encephalopathies: A Focus on Pre-synaptic Dysfunction. Frontiers in Neurology, 2022, 13, 826211. | 2.4 | 12 |
| 4 | 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 |
| 5 | De novo GRIN2A variants associated with epilepsy and autism and literature review. Epilepsy and Behavior, 2022, 129, 108604. | 1.7 | 9 |
| 6 | 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 |
| 7 | 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 |
| 8 | Paroxysmal limb dystonias associated with GABBR2 pathogenic variant: A case-based literature review. Brain and Development, 2022, , . | 1.1 | 2 |
| 9 | Mutations inTAF8cause a neurodegenerative disorder. Brain, 2022, 145, 3022-3034. | 7.6 | 3 |
| 10 | 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 |
| 11 | 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 |
| 12 | Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41. | 5.1 | 11 |
| 13 | 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 |
| 14 | 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 |
| 15 | Allelic and phenotypic heterogeneity in Junctophillin-3 related neurodevelopmental and movement disorders. European Journal of Human Genetics, 2021, 29, 1027-1031. | 2.8 | 5 |
| 16 | Tay-Sachs Disease: Two Novel Rare HEXA Mutations from Pakistan and Morocco. Klinische Padiatrie, 2021, 233, 226-230. | 0.6 | 4 |
| 17 | A paradigmatic autistic phenotype associated with loss of PCDH11Y and NLGN4Y genes. BMC Medical Genomics, 2021, 14, 98. | 1.5 | 4 |
| 18 | Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. International Journal of Molecular Sciences, 2021, 22, 4471. | 4.1 | 8 |

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|---|---|---|---|
| 19 | De novo mutation in SLC25A22 gene: expansion of the clinical and electroencephalographic phenotype. Journal of Neurogenetics, 2021, 35, 67-73. | 1.4 | 3 |
| 20 | Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401. | 1.3 | 2 |
| 21 | 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 |
| 22 | 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 |
| 23 | Prominent and Regressive Brain Developmental Disorders Associated with Nance-Horan Syndrome. Brain Sciences, 2021, 11, 1150. | 2.3 | 10 |
| 24 | 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 |
| 25 | Epileptic Phenotypes Associated With SNAREs and Related Synaptic Vesicle Exocytosis Machinery. Frontiers in Neurology, 2021, 12, 806506. | 2.4 | 9 |
| 26 | Diagnostic Approach to Macrocephaly in Children. Frontiers in Pediatrics, 2021, 9, 794069. | 1.9 | 17 |
| 27 | 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 |
| 28 | New Trends and Most Promising Therapeutic Strategies for Epilepsy Treatment. Frontiers in Neurology, 2021, 12, 753753. | 2.4 | 23 |
| 29 | 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 |
| 30 | Identification of common genetic markers of paroxysmal neurological disorders using a network analysis approach. Neurological Sciences, 2020, 41, 851-857. | 1.9 | 3 |
| 31 | 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 |
| 32 | Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397. | 7.6 | 28 |
| 33 | 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 |
| 34 | MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399. | 1.9 | 13 |
| 35 | 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 |
| 36 | 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 |

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|---|---|---|---|
| 37 | Novel variants underlying autosomal recessive intellectual disability in Pakistani consanguineous families. BMC Medical Genetics, 2020, 21, 59. | 2.1 | 3 |
| 38 | Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644. | 2.8 | 22 |
| 39 | Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197. | 1.6 | 14 |
| 40 | 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 |
| 41 | Genetic epilepsies and the Kv super-family. European Journal of Paediatric Neurology, 2020, 24, 5-6. | 1.6 | 1 |
| 42 | RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31. | 7.6 | 6 |
| 43 | Genotype-phenotype correlations in patients with de novo KCNQ2 pathogenic variants. Neurology: Genetics, 2020, 6, e528. | 1.9 | 24 |
| 44 | Age-dependent epileptic encephalopathy associated with an unusual co-occurrence of ZEB2 and SCN1A variants. , 2020, 22, 111-115. | | 2 |
| 45 | Benign familial infantile epilepsy associated with KCNQ3 mutation: a rare occurrence or an underestimated event?. Epileptic Disorders, 2020, 22, 807-810. | 1.3 | 6 |
| 46 | Kleineâ€Łevin syndrome is associated with LMOD3 variants. Journal of Sleep Research, 2019, 28, e12718. | 3.2 | 12 |
| 47 | AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094. | 12.8 | 150 |
| 48 | 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 |
| 49 | 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 |
| 50 | Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964. | 7.6 | 43 |
| 51 | 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 |
| 52 | 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 |
| 53 | Further supporting evidence for REEP1 phenotypic and allelic heterogeneity. Neurology: Genetics, 2019, 5, e379. | 1.9 | 3 |
| 54 | Pioneers and Emerging Pediatric Neurologists and Epileptologists in the World. Journal of Pediatric Epilepsy, 2019, 08, 001-001. | 0.2 | 0 |

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|---|---|---|---|
| 55 | 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 |
| 56 | A homozygous ATAD1 mutation impairs postsynaptic AMPA receptor trafficking and causes a lethal encephalopathy. Brain, 2018, 141, 651-661. | 7.6 | 52 |
| 57 | A homozygous lossâ€ofâ€function mutation in PDE2A associated to earlyâ€onset hereditary chorea. Movement Disorders, 2018, 33, 482-488. | 3.9 | 52 |
| 58 | PRRT2 Regulates Synaptic Fusion by Directly Modulating SNARE Complex Assembly. Cell Reports, 2018, 22, 820-831. | 6.4 | 67 |
| 59 | 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 |
| 60 | Pediatric stroke: current diagnostic and management challenges. Quantitative Imaging in Medicine and Surgery, 2018, 8, 984-991. | 2.0 | 10 |
| 61 | 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 |
| 62 | A Review of Copy Number Variants in Inherited Neuropathies. Current Genomics, 2018, 19, 412-419. | 1.6 | 19 |
| 63 | PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. Movement Disorders, 2018, 33, 1961-1965. | 3.9 | 38 |
| 64 | Reply: ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain, 2018, 141, e50-e50. | 7.6 | 1 |
| 65 | 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 |
| 66 | 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 |
| 67 | Homozygous mutations in <scp>VAMP</scp>1 cause a presynaptic congenital myasthenic syndrome. Annals of Neurology, 2017, 81, 597-603. | 5.3 | 48 |
| 68 | Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain, 2017, 140, 2820-2837. | 7.6 | 64 |
| 69 | Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429. | 3.8 | 122 |
| 70 | The phenotypic and molecular spectrum of PEHO syndrome and PEHO-like disorders. Brain, 2017, 140, e49-e49. | 7.6 | 33 |
| 71 | PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952. | 7.6 | 62 |
| 72 | 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 |

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|---|---|---|---|
| 73 | PKD or Not PKD: That is the question. Annals of Neurology, 2016, 80, 167-168. | 5.3 | 3 |
| 74 | Delineation of the movement disorders associated with FOXG1 mutations. Neurology, 2016, 86, 1794-1800. | 1.1 | 55 |
| 75 | 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 |
| 76 | 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 |