Mirella Vinci

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8629979/publications.pdf

Version: 2024-02-01

26 papers	772 citations	1040056 9 h-index	713466 21 g-index
26 all docs	26 docs citations	26 times ranked	2346 citing authors

#	Article	IF	Citations
1	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
2	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
3	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
4	A de novo heterozygous mutation in KCNC2 gene implicated in severe developmental and epileptic encephalopathy. European Journal of Medical Genetics, 2020, 63, 103848.	1.3	24
5	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
6	Definition of minimal duplicated region encompassing the XIAP and STAG2 genes in the Xq25 microduplication syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1923-1930.	1.2	15
7	Biallelic intragenic duplication in ADGRB3 (BAI3) gene associated with intellectual disability, cerebellar atrophy, and behavioral disorder. European Journal of Human Genetics, 2019, 27, 594-602.	2.8	15
8	Intragenic ILRAPL1 deletion in a male patient with intellectual disability, mild dysmorphic signs, deafness, and behavioral problems. American Journal of Medical Genetics, Part A, 2013, 161, 1381-1385.	1.2	14
9	Exon deletions of the phenylalanine hydroxylase gene in Italian hyperphenylalaninemics. Experimental and Molecular Medicine, 2010, 42, 81.	7.7	13
10	Interpreting Genetic Variants: Hints from a Family Cluster of Parkinsonâ€™s Disease. Journal of Parkinson's Disease, 2019, 9, 203-206.	2.8	11
11	A Customized Next-Generation Sequencing-Based Panel to Identify Novel Genetic Variants in Dementing Disorders: A Pilot Study. Neural Plasticity, 2020, 2020, 1-10.	2.2	6
12	Novel deletion of the E3A ubiquitin protein ligase gene detected by multiplex ligation-dependent probe amplification in a patient with Angelman syndrome. Experimental and Molecular Medicine, 2010, 42, 842.	7.7	5
13	Familial 1q22 microduplication associated with psychiatric disorders, intellectual disability and late-onset autoimmune inflammatory response. Molecular Cytogenetics, 2014, 7, 90.	0.9	5
14	Expression of Phosphodiesterase 4B cAMPâ€Specific Gene in Subjects With Cryptorchidism and Down's Syndrome. Journal of Clinical Laboratory Analysis, 2016, 30, 196-199.	2.1	3
15	Novel c.C2254T (p.Q752*) mutation in ZFYVE26 (SPG15) gene in a patient with hereditary spastic paraparesis. Journal of Genetics, 2018, 97, 1469-1472.	0.7	3
16	GRIN2A: involvement in movement disorders and intellectual disability without seizures. Neurological Sciences, 2019, 40, 2405-2406.	1.9	3
17	Are Mutations in the DHRS9 Gene Causally Linked to Epilepsy? A Case Report. Medicina (Lithuania), 2020, 56, 387.	2.0	2
18	Novel compound heterozygous mutation in NPC1 gene cause Niemannâ€“Pick disease type C with juvenile onset. Journal of Genetics, 2020, 99, 1.	0.7	2

#	Article	IF	Citations
19	Comparative multiplex dosage analysis in spinocerebellar ataxia type 2 patients. Genetics and Molecular Research, 2013, 12, 1176-1181.	0.2	1
20	Identification of novel mutations in L1CAM gene by a DHPLC-based assay. Genes and Genomics, 2016, 38, 1159-1164.	1.4	1
21	Novel SPINK5 variants in a patient with Netherton syndrome and intellectual disability. The diagnostic value of trichoscopy. Giornale Italiano Di Dermatologia E Venereologia, 2020, 155, 239-240.	0.8	1
22	Exome sequencing in a child with neurodevelopmental disorder and epilepsy: Variant analysis of the <scp>AHNAK2</scp> gene. Molecular Genetics & Genomic Medicine, 0, , .	1.2	1
23	LDOC1 expression in fibroblasts of patients with Down syndrome. Open Life Sciences, 2015, 10, .	1.4	0
24	Pigmented porokeratosis with dermal deposits of amyloid: the different chromatic features. Giornale Italiano Di Dermatologia E Venereologia, 2020, 155, 240-241.	0.8	0
25	Letter to the Editor Regarding the Article â€œWhole-Exome Sequencing in NF1-Related West's Syndrome Leads to the Identification of KCNC2 as a Novel Candidate Gene for Epilepsyâ€. Neuropediatrics, 2021, 52, 153-153.	0.6	0
26	Novel c.C2254T (p.Q752*) mutation in (SPG15) gene in a patient with hereditary spastic paraparesis. Journal of Genetics, 2018, 97, 1469-1472.	0.7	0