

Mirella Vinci

List of Publications by Year in descending order

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Version: 2024-02-01

26
papers

772
citations

1040018

9
h-index

713444

21
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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. <i>Nature Genetics</i> , 2017, 49, 515-526.	21.4	443
2	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. <i>Nature Neuroscience</i> , 2017, 20, 1043-1051.	14.8	152
3	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. <i>European Journal of Human Genetics</i> , 2018, 26, 54-63.	2.8	32
4	A de novo heterozygous mutation in KCNC2 gene implicated in severe developmental and epileptic encephalopathy. <i>European Journal of Medical Genetics</i> , 2020, 63, 103848.	1.3	24
5	The epilepsy phenotypic spectrum associated with a recurrent <i>CUX2</i> variant. <i>Annals of Neurology</i> , 2018, 83, 926-934.	5.3	20
6	Definition of minimal duplicated region encompassing the <i>XIAP</i> and <i>STAC2</i> genes in the Xq25 microduplication syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1923-1930.	1.2	15
7	Biallelic intragenic duplication in ADGRB3 (BAI3) gene associated with intellectual disability, cerebellar atrophy, and behavioral disorder. <i>European Journal of Human Genetics</i> , 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. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1381-1385.	1.2	14
9	Exon deletions of the phenylalanine hydroxylase gene in Italian hyperphenylalaninemics. <i>Experimental and Molecular Medicine</i> , 2010, 42, 81.	7.7	13
10	Interpreting Genetic Variants: Hints from a Family Cluster of Parkinson's Disease. <i>Journal of Parkinson's Disease</i> , 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. <i>Neural Plasticity</i> , 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. <i>Experimental and Molecular Medicine</i> , 2010, 42, 842.	7.7	5
13	Familial 1q22 microduplication associated with psychiatric disorders, intellectual disability and late-onset autoimmune inflammatory response. <i>Molecular Cytogenetics</i> , 2014, 7, 90.	0.9	5
14	Expression of Phosphodiesterase 4B cAMP-specific Gene in Subjects With Cryptorchidism and Down's Syndrome. <i>Journal of Clinical Laboratory Analysis</i> , 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. <i>Journal of Genetics</i> , 2018, 97, 1469-1472.	0.7	3
16	GRIN2A: involvement in movement disorders and intellectual disability without seizures. <i>Neurological Sciences</i> , 2019, 40, 2405-2406.	1.9	3
17	Are Mutations in the DHRS9 Gene Causally Linked to Epilepsy? A Case Report. <i>Medicina (Lithuania)</i> , 2020, 56, 387.	2.0	2
18	Novel compound heterozygous mutation in NPC1 gene cause Niemann-Pick disease type C with juvenile onset. <i>Journal of Genetics</i> , 2020, 99, 1.	0.7	2

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