Monia Ginevrino

List of Publications by Year
in descending order

38 papers	853 citations	687363 13 h-index	552781 26 g-index
38 all docs	38 docs citations	38 times ranked	1922 citing authors

#	Article	IF	Citations
1	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	3.2	13
2	Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73.	2.5	9
3	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	3.2	21
4	Absence of FGFR3â€“TACC3 rearrangement in hematological malignancies with numerical chromosomal alteration. Hematology/ Oncology and Stem Cell Therapy, 2021, 14, 163-168.	0.9	3
5	Posterior Cortical Atrophy phenotype in a GBA N370S mutation carrier: a case report. BMC Neurology, 2021, 21, 17.	1.8	3
6	Genetic characterization of a cohort with familial parkinsonism and cognitive-behavioral syndrome: A Next Generation Sequencing study. Parkinsonism and Related Disorders, 2021, 84, 82-90.	2.2	2
7	A ZFHX4 mutation associated with a recognizable neuropsychological and facial phenotype. European Journal of Medical Genetics, 2021, 64, 104321.	1.3	4
8	A novel IRF2BPL truncating variant is associated with endolysosomal storage. Molecular Biology Reports, 2020, 47, 711-714.	2.3	16
9	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi006-A from a patient affected by an autosomal recessive form of long QT syndrome type 1. Stem Cell Research, 2020, 42, 101658.	0.7	4
10	Gamma-transcranial alternating current stimulation and theta-burst stimulation: inter-subject variability and the role of BDNF. Clinical Neurophysiology, 2020, 131, 2691-2699.	1.5	13
11	<scp>GBAâ€Related</scp> Parkinson's Disease: Dissection of Genotypeâ€“Phenotype Correlates in a Large Italian Cohort. Movement Disorders, 2020, 35, 2106-2111.	3.9	83
12	<scp>APP</scp>â€Related Corticobasal Syndrome: Expanding the List of Corticobasal Degeneration Look Alikes. Movement Disorders Clinical Practice, 2020, 7, 849-851.	1.5	6
13	Clinical and Molecular Characterization of a Novel Progranulin Deletion Associated with Different Phenotypes. Journal of Alzheimer's Disease, 2020, 76, 341-347.	2.6	5
14	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.1	26
15	Generation of two human induced pluripotent stem cell (hiPSC) lines from a long QT syndrome South African founder population. Stem Cell Research, 2019, 39, 101510.	0.7	3
16	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. Brain, 2019, 142, 2965-2978.	7.6	12
17	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi005-A from a patient carrying the KCNQ1-R190W mutation. Stem Cell Research, 2019, 37, 101437.	0.7	1
18	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi007-A from a Long QT Syndrome type 1 patient carrier of two common variants in the NOS1AP gene. Stem Cell Research, 2019, 36, 101416.	0.7	2

#	Article	IF	Citations
19	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi004-A from a carrier of the KCNQ1-R594Q mutation. Stem Cell Research, 2019, 37, 101431.	0.7	2
20	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi002-A from a patient affected by the Jervell and Lange-Nielsen syndrome and carrier of two compound heterozygous mutations on the KCNQ1 gene. Stem Cell Research, 2018, 29, 157-161.	0.7	3
21	Intrafamilial variability in a polish family harbouring a frameshift THAP1 mutation. Journal of the Neurological Sciences, 2018, 388, 158.	0.6	0
22	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	3.1	30
23	Generation of the human induced pluripotent stem cell (hiPSC) line PSMi003-A from a patient affected by an autosomal recessive form of Long QT Syndrome type 1. Stem Cell Research, 2018, 29, 170-173.	0.7	6
24	Between SCA5 and SCAR14: delineation of the SPTBN2 p.R480W-associated phenotype. European Journal of Human Genetics, 2018, 26, 928-929.	2.8	17
25	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	9.0	66
26	Progressive Supranuclear Palsyâ€“Like Phenotype in a GBA E326K Mutation Carrier. Movement Disorders Clinical Practice, 2017, 4, 444-446.	1.5	14
27	DYT2 screening in early-onset isolated dystonia. European Journal of Paediatric Neurology, 2017, 21, 269-271.	1.6	13
28	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13.	3.1	108
29	BDNF and LTP-/LTD-like plasticity of the primary motor cortex in Gilles de la Tourette syndrome. Experimental Brain Research, 2017, 235, 841-850.	1.5	12
30	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563.	6.2	45
31	Genetic Paradoxes in an Italian Family with <scp>PARK</scp>2 Multiexon Duplication. Movement Disorders Clinical Practice, 2017, 4, 889-892.	1.5	1
32	The multiple faces of TOR1A: different inheritance, different phenotype. Brain, 2017, 140, 2764-2767.	7.6	3
33	The <scp>C</scp>ontursi <scp>F</scp>amily 20 <scp>Y</scp>ears <scp>L</scp>ater: <scp>I</scp>ntrafamilial <scp>P</scp>henotypic <scp>V</scp>ariability of the <scp>SNCA</scp> p.<scp>A</scp>53T <scp>M</scp>utation. Movement Disorders, 2016, 31, 257-258.	3.9	86
34	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Porettiâ€“Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267.	2.8	43
35	Impulsive-compulsive behaviors in parkin -associated Parkinson disease. Neurology, 2016, 87, 1436-1441.	1.1	61
36	Very mild features of dysequilibrium syndrome associated with a novel VLDLR missense mutation. Neurogenetics, 2016, 17, 191-195.	1.4	9

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37	Phenotypic spectrum of alpha-synuclein mutations: New insights from patients and cellular models. Parkinsonism and Related Disorders, 2016, 22, S16-S20.	2.2	98
38	Brain Connectivity Changes in Autosomal Recessive Parkinson Disease: A Model for the Sporadic Form. PLoS ONE, 2016, 11, e0163980.	2.5	10