Francesca Moro

List of Publications by Year
in descending order

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30 papers	939 citations	471477 17 h-index	526264 27 g-index
30 all docs	30 docs citations	30 times ranked	1909 citing authors

#	Article	IF	Citations
1	Customized multigene panels in epilepsy: the best things come in small packages. Neurogenetics, 2020, 21, 1-18.	1.4	9
2	Autophagic vacuolar myopathy caused by a CLN3 mutation. A case report. Neuromuscular Disorders, 2019, 29, 67-69.	0.6	1
3	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. Orphanet Journal of Rare Diseases, 2018, 13, 170.	2.7	26
4	Diagnostic methods and emerging treatments for adult neuronal ceroid lipofuscinoses (Kufs disease). Expert Opinion on Orphan Drugs, 2017, 5, 487-501.	0.8	2
5	Phenotype and natural history of variant late infantile ceroidâ€lipofuscinosis 5. Developmental Medicine and Child Neurology, 2017, 59, 815-821.	2.1	31
6	Gain-of-function defects of astrocytic Kir4.1 channels in children with autism spectrum disorders and epilepsy. Scientific Reports, 2016, 6, 34325.	3.3	56
7	TMEM5-associated dystroglycanopathy presenting with CMD and mild limb-girdle muscle involvement. Neuromuscular Disorders, 2016, 26, 459-461.	0.6	15
8	Early infantile neuronal ceroid lipofuscinosis (CLN10 disease) associated with a novel mutation in CTSD. Journal of Neurology, 2016, 263, 1029-1032.	3.6	23
9	Targeted Gene Resequencing (Astrochip) to Explore the Tripartite Synapse in Autismâ€“Epilepsy Phenotype with Macrocephaly. NeuroMolecular Medicine, 2016, 18, 69-80.	3.4	19
10	Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease?. Cerebellum, 2014, 13, 79-88.	2.5	50
11	Clinical, ultrastructural, and molecular studies in a patient with Kufs disease. Neurological Sciences, 2014, 35, 605-607.	1.9	5
12	Genetically induced dysfunctions of Kir2.1 channels: implications for short QT3 syndrome and autismâ€“epilepsy phenotype. Human Molecular Genetics, 2014, 23, 4875-4886.	2.9	65
13	Novel DyneinDYNC1H1Neck and Motor Domain Mutations Link Distal Spinal Muscular Atrophy and Abnormal Cortical Development. Human Mutation, 2014, 35, 298-302.	2.5	77
14	Autism-epilepsy phenotype with macrocephaly suggests PTEN, but not GLIALCAM, genetic screening. BMC Medical Genetics, 2014, 15, 26.	2.1	55
15	Pseudo-dominant inheritance of a novel CTSF mutation associated with type B Kufs disease. Neurology, 2014, 83, 1769-1770.	1.1	24
16	Novel mutations in the fukutin gene in a boy with asymptomatic hyperCKemia. Neuromuscular Disorders, 2013, 23, 1010-1015.	0.6	5
17	TRPV4 mutations in children with congenital distal spinal muscular atrophy. Neurogenetics, 2012, 13, 195-203.	1.4	31
18	P3.2 Novel mutation of TRPV4 in congenital distal SMA with vocal cord paralysis. Neuromuscular Disorders, 2011, 21, 682.	0.6	0

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19	Autism with Seizures and Intellectual Disability: Possible Causative Role of Gain-of-function of the Inwardly-Rectifying K+ Channel Kir4.1. Neurobiology of Disease, 2011, 43, 239-247.	4.4	108
20	Arginine:glycine amidinotransferase (AGAT) deficiency in a newborn: Early treatment can prevent phenotypic expression of the disease. Journal of Pediatrics, 2006, 148, 828-830.	1.8	85
21	Stable formation of mutated p53 multimers in a Chinese hamster cell line causes defective p53 nuclear localization and abrogates its residual function. Journal of Cellular Biochemistry, 2006, 98, 1689-1700.	2.6	4
22	Abnormal Phonologic Processing in Familial Lateral Temporal Lobe Epilepsy Due to a New LGI1 Mutation. Epilepsia, 2005, 46, 118-123.	5.1	55
23	Generalized Epilepsy with Febrile Seizures Plus (GEFS+): Clinical Spectrum in Seven Italian Families Unrelated to SCN1A, SCN1B, and GABRG2 Gene Mutations. Epilepsia, 2004, 45, 149-158.	5.1	67
24	Nonsyndromic mental retardation and cryptogenic epilepsy in women withDoublecortin gene mutations. Annals of Neurology, 2003, 54, 30-37.	5.3	65
25	Defective nuclear localization of p53 protein in a Chinese hamster cell line is associated with the formation of stable cytoplasmic protein multimers in cells with gene amplification. Carcinogenesis, 2000, 21, 1631-1638.	2.8	8
26	Derivative Chromosome 17 in a Case of Burkitt Lymphoma with 8;14 Translocation. Cancer Genetics and Cytogenetics, 1999, 110, 1-6.	1.0	0
27	The Î² and Î³ Subunits of the Human Platelet-Activating Factor Acetyl Hydrolase Isoform Ib (PAFAH1B2 and) Tj ETQq1 1 0.784314 rgB	2.9	4
28	Heterogeneousp53 mutations in a Burkitt lymphoma from an AIDS patient with monoclonalc-myc andVDJ rearrangements. , 1997, 73, 816-821.		6
29	Study on aneuploidy and p53 mutations in astrocytonias. Cancer Genetics and Cytogenetics, 1996, 88, 95-102.	1.0	19
30	p53 Expression in normal versus transformed mammalian cells. Carcinogenesis, 1995, 16, 2435-2440.	2.8	24