MÃ¡rcia Pimentel

List of Publications by Year
in descending order

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32 papers	557 citations	687363 13 h-index	677142 22 g-index
32 all docs	32 docs citations	32 times ranked	1150 citing authors

#	Article	IF	Citations
1	Copy-Number Gains of HUWE1 Due to Replication- and Recombination-Based Rearrangements. American Journal of Human Genetics, 2012, 91, 252-264.	6.2	71
2	KDM5C mutational screening among males with intellectual disability suggestive of X-Linked inheritance and review of the literature. European Journal of Medical Genetics, 2014, 57, 138-144.	1.3	51
3	p.Q223R leptin receptor polymorphism associated with obesity in Brazilian multiethnic subjects. American Journal of Human Biology, 2006, 18, 448-453.	1.6	42
4	A study of LRRK2 mutations and Parkinson's disease in Brazil. Neuroscience Letters, 2008, 433, 17-21.	2.1	35
5	Glucocerebrosidase N370S and L444P mutations as risk factors for Parkinsonâ€™s disease in Brazilian patients. Parkinsonism and Related Disorders, 2012, 18, 688-689.	2.2	29
6	FindingFMR1mosaicism in Fragile X syndrome. Expert Review of Molecular Diagnostics, 2016, 16, 501-507.	3.1	29
7	Circulating Endocannabinoids and the Polymorphism 385C>A in Fatty Acid Amide Hydrolase (FAAH) Gene May Identify the Obesity Phenotype Related to Cardiometabolic Risk: A Study Conducted in a Brazilian Population of Complex Interethnic Admixture. PLoS ONE, 2015, 10, e0142728.	2.5	27
8	Mutational analysis of GIGYF2, ATP13A2 and GBA genes in Brazilian patients with early-onset Parkinson's disease. Neuroscience Letters, 2010, 485, 121-124.	2.1	26
9	Using global team science to identify genetic parkinson's disease worldwide. Annals of Neurology, 2019, 86, 153-157.	5.3	26
10	A novel in-frame deletion affecting the BAR domain of OPHN1 in a family with intellectual disability and hippocampal alterations. European Journal of Human Genetics, 2014, 22, 644-651.	2.8	24
11	Novel microduplications at Xp11.22 including HUWE1: clinical and molecular insights into these genomic rearrangements associated with intellectual disability. Journal of Human Genetics, 2015, 60, 207-211.	2.3	24
12	Understanding the Landscape of X-linked Variants Causing Intellectual Disability in Females Through Extreme X Chromosome Inactivation Skewing. Molecular Neurobiology, 2020, 57, 3671-3684.	4.0	21
13	rs3851179 Polymorphism at 5â€² to the PICALM Gene is Associated with Alzheimer and Parkinson Diseases in Brazilian Population. NeuroMolecular Medicine, 2017, 19, 293-299.	3.4	18
14	High Frequency of Nonrecurrent MECP2 Duplications Among Brazilian Males with Mental Retardation. Journal of Molecular Neuroscience, 2010, 41, 105-109.	2.3	14
15	Genetic Analysis ofPARK2andPINK1Genes in Brazilian Patients with Early-Onset Parkinson's Disease. Disease Markers, 2013, 35, 181-185.	1.3	14
16	Low significance of MECP2 mutations as a cause of mental retardation in Brazilian males. Brain and Development, 2007, 29, 293-297.	1.1	13
17	Network Profiling of Brain-Expressed X-Chromosomal MicroRNA Genes Implicates Shared Key MicroRNAs in Intellectual Disability. Journal of Molecular Neuroscience, 2019, 67, 295-304.	2.3	12
18	Association of LRRK2 and GBA mutations in a Brazilian family with Parkinson's disease. Parkinsonism and Related Disorders, 2015, 21, 825-826.	2.2	11

#	Article	IF	Citations
19	ARX mutation c.428â€“451dup (24bp) in a Brazilian family with X-linked mental retardation. European Journal of Medical Genetics, 2006, 49, 269-275.	1.3	9
20	A MECP2 missense mutation within the MBD domain in a Brazilian male with autistic disorder. Brain and Development, 2011, 33, 807-809.	1.1	9
21	Exon dosage variations in Brazilian patients with Parkinson's disease: analysis of SNCA, PARKIN, PINK1 and DJ-1 genes. Disease Markers, 2012, 32, 173-8.	1.3	8
22	Mutational screening of ARX gene in Brazilian males with mental retardation of unknown etiology. Journal of Human Genetics, 2006, 51, 737-740.	2.3	7
23	Molecular and clinical insights into complex genomic rearrangements related to MECP2 duplication syndrome. European Journal of Medical Genetics, 2021, 64, 104367.	1.3	7
24	Lack of FMR3 expression in a male with non-syndromic mental retardation and a microdeletion immediately distal to FRAXE CCG repeat. Neuroscience Letters, 2006, 397, 245-248.	2.1	6
25	Influence of low frequency PSEN1 variants on familial Alzheimerâ€™s disease risk in Brazil. Neuroscience Letters, 2017, 653, 341-345.	2.1	6
26	The A140V mutation in the MECP2 gene is not a common etiological factor among Brazilian mentally retarded males. Neuroscience Letters, 2005, 379, 13-16.	2.1	5
27	A MECP2 mutation in a highly conserved aminoacid causing mental retardation in a male. Brain and Development, 2009, 31, 176-178.	1.1	4
28	Concurrence of fragile X and Klinefelter syndromes: report of a new case of paternal nondisjunction. Annales De GÃ©nÃ©tique, 2003, 46, 53-55.	0.4	3
29	De novo balanced translocation (2;10)(q24;q22) associated with mental retardation. Annales De GÃ©nÃ©tique, 2003, 46, 471-473.	0.4	3
30	Chromosome 6q deletion: Report of a new case and review of the literature. Genetics and Molecular Biology, 1998, 21, 145-149.	1.3	2
31	Strict network analysis of evolutionary conserved and brain-expressed genes reveals new putative candidates implicated in Intellectual Disability and in Global Development Delay. World Journal of Biological Psychiatry, 2021, 22, 1-11.	2.6	1
32	Geleophysic dysplasia: Report on two sibs. Genetics and Molecular Biology, 1998, 21, 159-162.	1.3	0