Marcia G Ribeiro

List of Publications by Year
in descending order

39 papers	812 citations	566801 15 h-index	500791 28 g-index
42 all docs	42 docs citations	42 times ranked	1260 citing authors

#	Article	IF	Citations
1	Mucopolysaccharidosis I, II, and VI: brief review and guidelines for treatment. Genetics and Molecular Biology, 2010, 33, 589-604.	0.6	150
2	A clinical study of 77 patients with mucopolysaccharidosis type II. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 63-70.	0.7	112
3	Mucopolysaccharidoses in Brazil: What happens from birth to biochemical diagnosis?. American Journal of Medical Genetics, Part A, 2008, 146A, 1741-1747.	0.7	63
4	A novel nonsense mutation in KDM5C/JARID1C gene causing intellectual disability, short stature and speech delay. Neuroscience Letters, 2011, 498, 67-71.	1.0	57
5	Timed motor function tests capacity in healthy children. Archives of Disease in Childhood, 2016, 101, 147-151.	1.0	36
6	Novel mutations of the BSCL2 and AGPAT2 genes in 10 families with Berardinelliâ€“Seip congenital generalized lipodystrophy syndrome. Clinical Endocrinology, 2009, 71, 512-517.	1.2	35
7	Insulin resistance in adolescents with Down syndrome: a cross-sectional study. BMC Endocrine Disorders, 2005, 5, 6.	0.9	34
8	Single median maxillary central incisor: New data and mutation review. Birth Defects Research Part A: Clinical and Molecular Teratology, 2007, 79, 573-580.	1.6	32
9	Mental retardation in Duchenne muscular dystrophy. Jornal De Pediatria, 2012, 88, 6-16.	0.9	32
10	FindingFMR1mosaicism in Fragile X syndrome. Expert Review of Molecular Diagnostics, 2016, 16, 501-507.	1.5	29
11	Diploid/triploid mosaicism: Further delineation of the phenotype. American Journal of Medical Genetics Part A, 1994, 52, 399-401.	2.4	28
12	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.	1.4	24
13	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.	1.1	24
14	Understanding the Landscape of X-linked Variants Causing Intellectual Disability in Females Through Extreme X Chromosome Inactivation Skewing. Molecular Neurobiology, 2020, 57, 3671-3684.	1.9	21
15	Enzyme replacement therapy for Mucopolysaccharidosis Type I among patients followed within the MPS Brazil Network. Genetics and Molecular Biology, 2014, 37, 23-29.	0.6	19
16	Dental findings and oral health status in patients with mucopolysaccharidosis: a case series. Acta Odontologica Scandinavica, 2013, 71, 157-167.	0.9	18
17	High incidence of acquiring methicillin-resistant Staphylococcus aureus in Brazilian children with Atopic Dermatitis and associated risk factors. Journal of Microbiology, Immunology and Infection, 2020, 53, 724-730.	1.5	13
18	Betaineâ€“homocysteine methyltransferase 742G>A polymorphism and risk of down syndrome offspring in a Brazilian population. Molecular Biology Reports, 2013, 40, 4685-4689.	1.0	12

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19	Oral Aspects Identified in Atopic Dermatitis Patients: A Literature Review. Open Dentistry Journal, 2018, 12, 424-434.	0.2	9
20	Oral mycobiome identification in atopic dermatitis, leukemia, and HIV patients â€“ a systematic review. Journal of Oral Microbiology, 2020, 12, 1807179.	1.2	9
21	Profile of patients assessed for cochlear implants. Brazilian Journal of Otorhinolaryngology, 2014, 80, 305-310.	0.4	6
22	High-throughput nucleotide sequencing for bacteriome studies in oral squamous cell carcinoma: a systematic review. Oral and Maxillofacial Surgery, 2020, 24, 387-401.	0.6	6
23	Decompensated Chronic Heart Failure Reduces Plasma L-carnitine. Archives of Medical Research, 2018, 49, 278-281.	1.5	4
24	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
25	Clinical oral profile of pediatric patients with atopic dermatitis: A crossâ€sectional study. Oral Diseases, 2021, 27, 1834-1846.	1.5	3
26	Association between cytogenetic alteration and the audiometric profile of individuals with Turner syndrome. Brazilian Journal of Otorhinolaryngology, 2021, 87, 728-732.	0.4	3
27	Sanfilippo syndrome type B: Analysis of patients diagnosed by the <scp>MPS</scp> Brazil Network. American Journal of Medical Genetics, Part A, 2022, 188, 760-767.	0.7	3
28	BenefÃcios da inserÃ§Ã£o do fisioterapeuta sobre o perfil de prematuros de baixo risco internados em unidade de terapia intensiva. Fisioterapia E Pesquisa, 2019, 26, 51-57.	0.3	2
29	Xâ€linked hypophosphatemic rickets: Description of seven new variants in patients followed up in reference hospitals in Rio de Janeiro. Molecular Genetics & Genomic Medicine, 2022, , e1941.	0.6	2
30	Sangue de cordÃ£o umbilical para uso autÃ³logo ou grupo de pacientes especiais. Revista Brasileira De Hematologia E Hemoterapia, 0, 31, 36-44.	0.7	1
31	RepercussÃµes clÃnicas e radiolÃ³gicas do neurofibroma plexiforme na regiÃ£o pÃ©lvica. Radiologia Brasileira, 2014, 47, 326-328.	0.3	1
32	Frequency of GJB2 mutations in patients with nonsyndromic hearing loss from an ethnically characterized Brazilian population. Brazilian Journal of Otorhinolaryngology, 2019, 85, 92-98.	0.4	1
33	Can simple and low-cost motor function assessments help in the diagnostic suspicion of Duchenne muscular dystrophy?. Jornal De Pediatria, 2020, 96, 503-510.	0.9	1
34	Determination of Vitamin D Levels in Patients With Neurofibromatosis Type 1 in the Pediatric Age Group. BMC Clinical Pathology, 2020, 13, 2632010X2092893.	0.7	1
35	Moderately progressive Ullrich congenital muscular dystrophy. Jornal De Pediatria, 2011, 88, 93-6.	0.9	1
36	Alopecia Areata-Part III: Prognosis and Treatment. Skinmed, 2016, 14, 361-365.	0.0	1

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37	Can simple and low-cost motor function assessments help in the diagnostic suspicion of Duchenne muscular dystrophy?. Jornal De Pediatria (VersÃ£o Em PortuguÃªs), 2020, 96, 503-510.	0.2	0
38	PercepÃ§Ã£o de responsÃ¡veis sobre o envolvimento de crianÃ§as e adolescentes diabÃ©ticos em ocupaÃ§Ãµes durante o distanciamento social devido Ã pandemia por covid-19/ Carers perception on the involvement of diabetic children and adolescents in occupations during the social distancing due to the Covid-19 pandemic. Revista Interinstitucional Brasileira De Terapia Ocupacional, 2021, 5, 352-368.	0.1	0
39	Agreement Between Alberta Infant Motor Scale Assessment and Maternal Perception of Motor Development in Full-Term Infants. Developmental Neurorehabilitation, 2022, , 1-9.	0.5	0