Caio Robledo D'Angioli Costa Quaio

List of Publications by Year
in descending order

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30	487	840776 11	713466 21
papers	citations	h-index	g-index

33	33	33	1319
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Vertebral segmentation defects in a Brazilian cohort: Clinical and molecular analysis focused on spondylocostal dysostosis. Clinical Genetics, 2022, 101, 476-478.	2.0	0
2	Frequency of carriers for rare metabolic diseases in a Brazilian cohort of 320 patients. Molecular Biology Reports, 2022, 49, 3911-3918.	2.3	2
3	Parental segregation study reveals rare benign and likely benign variants in a Brazilian cohort of rare diseases. Scientific Reports, 2022, 12, 7764.	3.3	1
4	Exome sequencing and targeted gene panels: a simulated comparison of diagnostic yield using data from 158 patients with rare diseases. Genetics and Molecular Biology, 2021, 44, 20210061.	1.3	4
5	Does SCA45 Cause Very Late-Onset Pure Cerebellar Ataxia?. Neurology: Genetics, 2021, 7, e581.	1.9	3
6	Frequency of carriers for rare recessive Mendelian diseases in a Brazilian cohort of 320 patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 364-372.	1.6	6
7	Diagnostic power and clinical impact of exome sequencing in a cohort of 500 patients with rare diseases. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 955-964.	1.6	22
8	Gene expression profile suggesting immunological dysregulation in two Brazilian Bloom's syndrome cases. Molecular Genetics & Genomic Medicine, 2020, 8, e1133.	1.2	7
9	Biochemical profile in an infant with neonatal hemochromatosis shows evidence of impairment of mitochondrial long-chain fatty acid oxidation. Clinical and Molecular Hepatology, 2019, 25, 86-91.	8.9	0
10	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
11	Development and validation of a variant detection workflow for BRCA1 and BRCA2 genes and its clinical application based on the Ion Torrent technology. Human Genomics, 2017, 11, 14.	2.9	11
12	Clinical description of 41 Brazilian patients with oculo-auriculo-vertebral dysplasia. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2016, 62, 202-206.	0.7	12
13	Multicentric study on the diagnosis of Fabry's disease using angiokeratoma biopsy registries. International Journal of Dermatology, 2015, 54, e241-4.	1.0	2
14	Lipoid proteinosis: Rare case confirmed by ECM1 mutation detection. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2314-2315.	1.0	3
15	Mucopolysaccharidosis type IVA: Evidence of primary and secondary central nervous system involvement. American Journal of Medical Genetics, Part A, 2014, 164, 1162-1169.	1.2	19
16	New insights in mucopolysaccharidosis type VI: Neurological perspective. Brain and Development, 2014, 36, 585-592.	1.1	20
17	Case Report Johanson-Blizzard syndrome: a report of gender-discordant twins with a novel UBR1 mutation. Genetics and Molecular Research, 2014, 13, 4159-4164.	0.2	1
18	Discrepant outcomes in two Brazilian patients with Bloom syndrome and Wilmsâ€™ tumor: two case reports. Journal of Medical Case Reports, 2013, 7, 284.	0.8	9

#	Article	IF	Citations
19	A DominantABCC8-Related Hyperinsulinism: Familial Case ReportMoreiraet al.ABCC8-Related Hyperinsulinism. Fetal and Pediatric Pathology, 2013, 32, 384-386.	0.7	1
20	Vertical transmission of a frontonasal phenotype caused by a novel ALX4 mutation. American Journal of Medical Genetics, Part A, 2013, 161, 600-604.	1.2	24
21	Tegumentary manifestations of Noonan and Noonan-related syndromes. Clinics, 2013, 68, 1079-1083.	1.5	12
22	The first cardiac transplant experience in a patient with mucopolysaccharidosis. Cardiovascular Pathology, 2012, 21, 358-360.	1.6	6
23	Role of SNAP29, LZTR1 and P2RXL1 genes on immune regulation in a patient with atypical 0.5Mb deletion in 22q11.2 region. Clinical Immunology, 2012, 145, 55-58.	3.2	6
24	A clinical follow-up of 35 Brazilian patients with Prader-Willi Syndrome. Clinics, 2012, 67, 917-921.	1.5	4
25	Autoimmune disease and multiple autoantibodies in 42 patients with RASopathies. American Journal of Medical Genetics, Part A, 2012, 158A, 1077-1082.	1.2	73
26	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	6.2	157
27	A Possible Role of Different PTPN Genes in Immune Regulation. Scandinavian Journal of Immunology, 2012, 75, 540-541.	2.7	2
28	Manchas hipomelanocÃticas associadas Ã epilepsia: alerta ao diagnÃ³stico de esclerose tuberosa. , 2012, 91, 117.	0.1	0
29	Report of a Large Brazilian Family With a Very Attenuated Form of Hunter Syndrome (MPS II). JIMD Reports, 2011, 4, 125-128.	1.5	15
30	Estudo comparativo entre a sÃndrome antifosfolÃpide primÃ¡ria e a secundÃ¡ria: caracterÃsticas clÃnico-laboratoriais em 149 pacientes. Revista Brasileira De Reumatologia, 2008, 48, .	0.8	0