# Caio Robledo D'Angioli Costa Quaio

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/8556884/publications.pdf

Version: 2024-02-01

| 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 | 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 |
| 2 | Autoimmune disease and multiple autoantibodies in 42 patients with RASopathies. American Journal of Medical Genetics, Part A, 2012, 158A, 1077-1082. | 1.2 | 73 |
| 3 | 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 |
| 4 | 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 |
| 5 | 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 |
| 6 | New insights in mucopolysaccharidosis type VI: Neurological perspective. Brain and Development, 2014, 36, 585-592. | 1.1 | 20 |
| 7 | 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 |
| 8 | 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 |
| 9 | Tegumentary manifestations of Noonan and Noonan-related syndromes. Clinics, 2013, 68, 1079-1083. | 1.5 | 12 |
| 10 | 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 |
| 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 | 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 |
| 13 | Gene expression profile suggesting immunological dysregulation in two Brazilian Bloom's syndrome cases. Molecular Genetics & Genomic Medicine, 2020, 8, e1133. | 1.2 | 7 |
| 14 | The first cardiac transplant experience in a patient with mucopolysaccharidosis. Cardiovascular Pathology, 2012, 21, 358-360. | 1.6 | 6 |
| 15 | 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 |
| 16 | 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 |
| 17 | A clinical follow-up of 35 Brazilian patients with Prader-Willi Syndrome. Clinics, 2012, 67, 917-921. | 1.5 | 4 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Lipoid proteinosis: Rare case confirmed by ECM1 mutation detection. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2314-2315. | 1.0 | 3 |
| 20 | Does SCA45 Cause Very Late-Onset Pure Cerebellar Ataxia?. Neurology: Genetics, 2021, 7, e581. | 1.9 | 3 |
| 21 | A Possible Role of Different PTPN Genes in Immune Regulation. Scandinavian Journal of Immunology, 2012, 75, 540-541. | 2.7 | 2 |
| 22 | Multicentric study on the diagnosis of Fabry's disease using angiokeratoma biopsy registries. International Journal of Dermatology, 2015, 54, e241-4. | 1.0 | 2 |
| 23 | Frequency of carriers for rare metabolic diseases in a Brazilian cohort of 320 patients. Molecular Biology Reports, 2022, 49, 3911-3918. | 2.3 | 2 |
| 24 | A DominantABCC8-Related Hyperinsulinism: Familial Case ReportMoreiraet al.ABCC8-Related Hyperinsulinism. Fetal and Pediatric Pathology, 2013, 32, 384-386. | 0.7 | 1 |
| 25 | 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 |
| 26 | 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 |
| 27 | 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 |
| 28 | Manchas hipomelanocÃ¡ticas associadas Ã  epilepsia: alerta ao diagnÃ³stico de esclerose tuberosa. , 2012, 91, 117. | 0.1 | 0 |
| 29 | 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 |
| 30 | Vertebral segmentation defects in a Brazilian cohort: Clinical and molecular analysis focused on spondylocostal dysostosis. Clinical Genetics, 2022, 101, 476-478. | 2.0 | 0 |