# Rachel Honjo

List of Publications by Year
in descending order



| 42 | 543 | 687363 13 | 713466 21 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 44 | 44 | 44 | 1177 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | <scp>Cardiovascular findings in Williamsâ€“Beuren Syndrome</scp>: Experience of a single center with 127 cases. American Journal of Medical Genetics, Part A, 2022, 188, 676-682. | 1.2 | 4 |
| 2 | Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074. | 1.7 | 14 |
| 3 | The recurrent homozygous translation start site variant in CCDC134 in an individual with severe osteogenesis imperfecta of nonâ€Morrocan ancestry. American Journal of Medical Genetics, Part A, 2022, , . | 1.2 | 3 |
| 4 | Vertebral segmentation defects in a Brazilian cohort: Clinical and molecular analysis focused on spondylocostal dysostosis. Clinical Genetics, 2022, 101, 476-478. | 2.0 | 0 |
| 5 | Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918. | 2.5 | 8 |
| 6 | Back Cover, Volume 43, Issue 7. Human Mutation, 2022, 43, . | 2.5 | 0 |
| 7 | Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. Human Reproduction, 2021, 36, 506-518. | 0.9 | 16 |
| 8 | Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65. | 2.5 | 18 |
| 9 | Abnormal auditory event-related potentials in Williams syndrome. European Journal of Medical Genetics, 2021, 64, 104163. | 1.3 | 2 |
| 10 | Cerebellofaciodental syndrome in an adult patient: Expanding the phenotypic and natural history characteristics. American Journal of Medical Genetics, Part A, 2021, 185, 1561-1568. | 1.2 | 4 |
| 11 | Atypical, severe hypertrophic cardiomyopathy in a newborn presenting Noonan syndrome harboring a recurrent heterozygous <scp>MRAS</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 3099-3103. | 1.2 | 4 |
| 12 | Auditory hypersensitivity in Williams syndrome. International Journal of Pediatric Otorhinolaryngology, 2021, 146, 110740. | 1.0 | 3 |
| 13 | Twentyâ€year followâ€up of the facial phenotype of Brazilian patients with Sotos syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3916-3923. | 1.2 | 0 |
| 14 | Congenital limb deficiency: Genetic investigation of 44 individuals presenting mainly longitudinal defects in isolated or syndromic forms. Clinical Genetics, 2021, 100, 615-623. | 2.0 | 4 |
| 15 | Nationwide questionnaire data of 229 Williams-Beuren syndrome patients using WhatsApp tool. Arquivos De Neuro-Psiquiatria, 2021, 79, 950-956. | 0.8 | 1 |
| 16 | Expanding the role of SETD5 haploinsufficiency in neurodevelopment and neuroblastoma. Pediatric Blood and Cancer, 2020, 67, e28376. | 1.5 | 3 |
| 17 | Phenotypeâ€“genotype analysis of 242 individuals with <scp>RASopathies</scp>: 18â€year experience of a tertiary center in Brazil. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 896-911. | 1.6 | 10 |
| 18 | Enzyme replacement therapy interruption in patients with Mucopolysaccharidoses: Recommendations for distinct scenarios in Latin America. Molecular Genetics and Metabolism Reports, 2020, 23, 100572. | 1.1 | 10 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Mucopolysaccharidosis type VI: case report with first neonatal presentation with ascites fetalis and rapidly progressive cardiac manifestation. BMC Medical Genetics, 2020, 21, 37. | 2.1 | 6 |
| 20 | Genetic investigation of patients with tall stature. European Journal of Endocrinology, 2020, 182, 139-147. | 3.7 | 3 |
| 21 | Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. Journal of Human Genetics, 2019, 64, 885-890. | 2.3 | 11 |
| 22 | Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978. | 2.3 | 43 |
| 23 | Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta. American Journal of Human Genetics, 2019, 105, 836-843. | 6.2 | 36 |
| 24 | Genetic Disorders in Prenatal Onset Syndromic Short Stature Identified by Exome Sequencing. Journal of Pediatrics, 2019, 215, 192-198. | 1.8 | 36 |
| 25 | Primary immunodeficiency with chronic enteropathy and developmental delay in a boy arising from a novel homozygous RIPK1 variant. Journal of Human Genetics, 2019, 64, 955-960. | 2.3 | 28 |
| 26 | Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. American Journal of Human Genetics, 2019, 104, 925-935. | 6.2 | 92 |
| 27 | Growth and Clinical Characteristics of Children with Floating-Harbor Syndrome: Analysis of Current Original Data and a Review of the Literature. Hormone Research in Paediatrics, 2019, 92, 115-123. | 1.8 | 7 |
| 28 | Cri du Chat syndrome: Characteristics of 73 Brazilian patients. Journal of Intellectual Disability Research, 2018, 62, 467-473. | 2.0 | 16 |
| 29 | Richieriâ€Costaâ€Pereira syndrome: Expanding its phenotypic and genotypic spectrum. Clinical Genetics, 2018, 93, 800-811. | 2.0 | 15 |
| 30 | Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. Hormone Research in Paediatrics, 2018, 89, 13-21. | 1.8 | 29 |
| 31 | Diagnosis and management of systemic hypertension due to renovascular and aortic stenosis in patients with Williams-Beuren syndrome. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2018, 64, 723-728. | 0.7 | 8 |
| 32 | Large deletion in PIGL: a common mutational mechanism in CHIME syndrome?. Genetics and Molecular Biology, 2018, 41, 85-91. | 1.3 | 6 |
| 33 | A Multicentric Brazilian Investigative Study of Copy Number Variations in Patients with Congenital Anomalies and Intellectual Disability. Scientific Reports, 2018, 8, 13382. | 3.3 | 1 |
| 34 | Cognitive and behavioral profile of Williams Syndrome toddlers. CoDAS, 2018, 30, e20170188. | 0.7 | 5 |
| 35 | Natural history of 39 patients with Achondroplasia. Clinics, 2018, 73, e324. | 1.5 | 20 |
| 36 | Short Communication Impact of early enzyme-replacement therapy for mucopolysaccharidosis VI: results of a long-term follow-up of Brazilian siblings. Genetics and Molecular Research, 2016, 15, . | 0.2 | 21 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | StÃ¼ve-Wiedemann Syndrome: Update on Clinical and Genetic Aspects. Molecular Syndromology, 2016, 7, 12-18. | 0.8 | 18 |
| 39 | Menkes disease: importance of diagnosis with molecular analysis in the neonatal period. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2015, 61, 407-410. | 0.7 | 5 |
| 40 | Williams-Beuren Syndrome: A Clinical Study of 55 Brazilian Patients and the Diagnostic Use of MLPA. BioMed Research International, 2015, 2015, 1-6. | 1.9 | 13 |
| 41 | Multicentric study on the diagnosis of Fabry's disease using angiokeratoma biopsy registries. International Journal of Dermatology, 2015, 54, e241-4. | 1.0 | 2 |
| 42 | Lipoid proteinosis: Rare case confirmed by ECM1 mutation detection. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2314-2315. | 1.0 | 3 |