

# Rachel Honjo

## List of Publications by Year in descending order

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Version: 2024-02-01

42  
papers

543  
citations

687363

13  
h-index

713466

21  
g-index

44  
all docs

44  
docs citations

44  
times ranked

1177  
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	2.3	43
3	Autosomal-Recessive Mutations in MESD Cause Osteogenesis Imperfecta. American Journal of Human Genetics, 2019, 105, 836-843.	6.2	36
4	Genetic Disorders in Prenatal Onset Syndromic Short Stature Identified by Exome Sequencing. Journal of Pediatrics, 2019, 215, 192-198.	1.8	36
5	Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. Hormone Research in Paediatrics, 2018, 89, 13-21.	1.8	29
6	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
7	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
8	Natural history of 39 patients with Achondroplasia. Clinics, 2018, 73, e324.	1.5	20
9	StÅ½ve-Wiedemann Syndrome: Update on Clinical and Genetic Aspects. Molecular Syndromology, 2016, 7, 12-18.	0.8	18
10	Efficient detection of copy number variations using exome data: Batch and sex based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
11	Cri du Chat syndrome: Characteristics of 73 Brazilian patients. Journal of Intellectual Disability Research, 2018, 62, 467-473.	2.0	16
12	Insights from the genetic characterization of central precocious puberty associated with multiple anomalies. Human Reproduction, 2021, 36, 506-518.	0.9	16
13	Richieri-Costa-Pereira syndrome: Expanding its phenotypic and genotypic spectrum. Clinical Genetics, 2018, 93, 800-811.	2.0	15
14	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
15	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
16	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
17	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
18	Phenotype-genotype analysis of 242 individuals with RASopathies: 18-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

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19	Enzyme replacement therapy interruption in patients with Mucopolysaccharidoses: Recommendations for distinct scenarios in Latin America. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 23, 100572.	1.1	10
20	Diagnosis and management of systemic hypertension due to renovascular and aortic stenosis in patients with Williams-Beuren syndrome. <i>Revista Da Associação Médica Brasileira</i> , 2018, 64, 723-728.	0.7	8
21	Phenotypic and mutational spectrum of <i>ROR2</i> -related Robinow syndrome. <i>Human Mutation</i> , 2022, 43, 900-918.	2.5	8
22	Growth and Clinical Characteristics of Children with Floating-Harbor Syndrome: Analysis of Current Original Data and a Review of the Literature. <i>Hormone Research in Paediatrics</i> , 2019, 92, 115-123.	1.8	7
23	Large deletion in PIGL: a common mutational mechanism in CHIME syndrome?. <i>Genetics and Molecular Biology</i> , 2018, 41, 85-91.	1.3	6
24	Mucopolysaccharidosis type VI: case report with first neonatal presentation with ascites fetalis and rapidly progressive cardiac manifestation. <i>BMC Medical Genetics</i> , 2020, 21, 37.	2.1	6
25	Menkes disease: importance of diagnosis with molecular analysis in the neonatal period. <i>Revista Da Associação Médica Brasileira</i> , 2015, 61, 407-410.	0.7	5
26	Cognitive and behavioral profile of Williams Syndrome toddlers. <i>CoDAS</i> , 2018, 30, e20170188.	0.7	5
27	Cerebellofaciodental syndrome in an adult patient: Expanding the phenotypic and natural history characteristics. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1561-1568.	1.2	4
28	Atypical, severe hypertrophic cardiomyopathy in a newborn presenting Noonan syndrome harboring a recurrent heterozygous <i>MRAS</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3099-3103.	1.2	4
29	Congenital limb deficiency: Genetic investigation of 44 individuals presenting mainly longitudinal defects in isolated or syndromic forms. <i>Clinical Genetics</i> , 2021, 100, 615-623.	2.0	4
30	Cardiovascular findings in Williams-Beuren Syndrome: Experience of a single center with 127 cases. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 676-682.	1.2	4
31	Lipoid proteinosis: Rare case confirmed by ECM1 mutation detection. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2014, 78, 2314-2315.	1.0	3
32	Expanding the role of <i>SETD5</i> haploinsufficiency in neurodevelopment and neuroblastoma. <i>Pediatric Blood and Cancer</i> , 2020, 67, e28376.	1.5	3
33	Auditory hypersensitivity in Williams syndrome. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2021, 146, 110740.	1.0	3
34	Genetic investigation of patients with tall stature. <i>European Journal of Endocrinology</i> , 2020, 182, 139-147.	3.7	3
35	The recurrent homozygous translation start site variant in <i>CCDC134</i> in an individual with severe osteogenesis imperfecta of non-Moroccan ancestry. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	1.2	3
36	Multicentric study on the diagnosis of Fabry's disease using angiokeratoma biopsy registries. <i>International Journal of Dermatology</i> , 2015, 54, e241-4.	1.0	2

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37	Abnormal auditory event-related potentials in Williams syndrome. <i>European Journal of Medical Genetics</i> , 2021, 64, 104163.	1.3	2
38	A Multicentric Brazilian Investigative Study of Copy Number Variations in Patients with Congenital Anomalies and Intellectual Disability. <i>Scientific Reports</i> , 2018, 8, 13382.	3.3	1
39	Nationwide questionnaire data of 229 Williams-Beuren syndrome patients using WhatsApp tool. <i>Arquivos De Neuro-Psiquiatria</i> , 2021, 79, 950-956.	0.8	1
40	Twenty-year follow-up of the facial phenotype of Brazilian patients with Sotos syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3916-3923.	1.2	0
41	Vertebral segmentation defects in a Brazilian cohort: Clinical and molecular analysis focused on spondylocostal dysostosis. <i>Clinical Genetics</i> , 2022, 101, 476-478.	2.0	0
42	Back Cover, Volume 43, Issue 7. <i>Human Mutation</i> , 2022, 43, .	2.5	0