

Hiroyuki Mishima

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

43
papers

937
citations

16
h-index

30
g-index

46
ext. papers

1,172
ext. citations

5.6
avg, IF

3.27
L-index

#	Paper	IF	Citations
43	Proteasome assembly defect due to a proteasome subunit beta type 8 (PSMB8) mutation causes the autoinflammatory disorder, Nakajo-Nishimura syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011 , 108, 14914-9	11.5	227
42	Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase II processing in transcription-coupled nucleotide-excision repair. <i>Nature Genetics</i> , 2012 , 44, 586-92	36.3	123
41	Characterization of placenta-specific microRNAs in fetal growth restriction pregnancy. <i>Prenatal Diagnosis</i> , 2013 , 33, 214-22	3.2	102
40	Identification of endometrioid endometrial carcinoma-associated microRNAs in tissue and plasma. <i>Gynecologic Oncology</i> , 2014 , 132, 715-21	4.9	61
39	Nonsense mutation in causes normal-pressure hydrocephalus with ciliary abnormalities. <i>Neurology</i> , 2019 , 92, e2364-e2374	6.5	34
38	Sick sinus syndrome with HCN4 mutations shows early onset and frequent association with atrial fibrillation and left ventricular noncompaction. <i>Heart Rhythm</i> , 2017 , 14, 717-724	6.7	27
37	Evaluation of Face2Gene using facial images of patients with congenital dysmorphic syndromes recruited in Japan. <i>Journal of Human Genetics</i> , 2019 , 64, 789-794	4.3	26
36	Pregnancy-associated microRNAs in plasma as potential molecular markers of ectopic pregnancy. <i>Fertility and Sterility</i> , 2015 , 103, 1202-8.e1	4.8	23
35	Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. <i>Clinical Immunology</i> , 2015 , 160, 255-60	9	22
34	Biogem: an effective tool-based approach for scaling up open source software development in bioinformatics. <i>Bioinformatics</i> , 2012 , 28, 1035-7	7.2	22
33	Genome-wide association study of HPV-associated cervical cancer in Japanese women. <i>Journal of Medical Virology</i> , 2014 , 86, 1153-8	19.7	21
32	Identification of novel schizophrenia loci by homozygosity mapping using DNA microarray analysis. <i>PLoS ONE</i> , 2011 , 6, e20589	3.7	19
31	Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply segmental uniparental isodisomy in general populations. <i>Gene</i> , 2013 , 512, 267-74	3.8	18
30	Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer. <i>Journal of Human Genetics</i> , 2013 , 58, 250-3	4.3	18
29	Agile parallel bioinformatics workflow management using Pwrake. <i>BMC Research Notes</i> , 2011 , 4, 331	2.3	17
28	Germline mutations causing familial lung cancer. <i>Journal of Human Genetics</i> , 2015 , 60, 597-603	4.3	16
27	Maternal uniparental isodisomy and heterodisomy on chromosome 6 encompassing a CUL7 gene mutation causing 3M syndrome. <i>Clinical Genetics</i> , 2011 , 80, 478-83	4	16

26	Deep sequencing reveals variations in somatic cell mosaic mutations between monozygotic twins with discordant psychiatric disease. <i>Human Genome Variation</i> , 2017 , 4, 17032	1.8	15
25	Clinical applications of analysis of plasma circulating complete hydatidiform mole pregnancy-associated miRNAs in gestational trophoblastic neoplasia: a preliminary investigation. <i>Placenta</i> , 2014 , 35, 787-9	3.4	13
24	Detection of de novo single nucleotide variants in offspring of atomic-bomb survivors close to the hypocenter by whole-genome sequencing. <i>Journal of Human Genetics</i> , 2018 , 63, 357-363	4.3	12
23	Whole-exome sequencing and gene-based rare variant association tests suggest that PLA2G4E might be a risk gene for panic disorder. <i>Translational Psychiatry</i> , 2018 , 8, 41	8.6	11
22	Circulating levels of maternal plasma cell-free pregnancy-associated placenta-specific microRNAs are associated with placental weight. <i>Placenta</i> , 2014 , 35, 848-51	3.4	11
21	A significant association between rs8067378 at 17q12 and invasive cervical cancer originally identified by a genome-wide association study in Han Chinese is replicated in a Japanese population. <i>Journal of Human Genetics</i> , 2016 , 61, 793-6	4.3	10
20	Metabolic autopsy with next generation sequencing in sudden unexpected death in infancy: Postmortem diagnosis of fatty acid oxidation disorders. <i>Molecular Genetics and Metabolism Reports</i> , 2015 , 5, 26-32	1.8	10
19	Three brothers with a nonsense mutation in caused by parental germline mosaicism. <i>Human Genome Variation</i> , 2017 , 4, 17045	1.8	7
18	Patients with SATB2-associated syndrome exhibiting multiple odontomas. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2614-2622	2.5	7
17	A homozygous splice site ROBO1 mutation in a patient with a novel syndrome with combined pituitary hormone deficiency. <i>Journal of Human Genetics</i> , 2019 , 64, 341-346	4.3	6
16	Predominantly placenta-expressed mRNAs in maternal plasma as predictive markers for twin-twin transfusion syndrome. <i>Prenatal Diagnosis</i> , 2014 , 34, 345-9	3.2	6
15	Identification of a variant associated with early-onset diabetes in the intron of the insulin gene with exome sequencing. <i>Journal of Diabetes Investigation</i> , 2019 , 10, 947-950	3.9	6
14	Application of the Linux cluster for exhaustive window haplotype analysis using the FBAT and Unphased programs. <i>BMC Bioinformatics</i> , 2008 , 9 Suppl 6, S10	3.6	5
13	Heterozygous missense variant of the proteasome subunit β type 9 causes neonatal-onset autoinflammation and immunodeficiency. <i>Nature Communications</i> , 2021 , 12, 6819	17.4	4
12	Functionally validated SCN5A variants allow interpretation of pathogenicity and prediction of lethal events in Brugada syndrome. <i>European Heart Journal</i> , 2021 , 42, 2854-2863	9.5	4
11	Bile extracellular vesicles from end-stage liver disease patients show altered microRNA content. <i>Hepatology International</i> , 2021 , 15, 821-830	8.8	3
10	Genome analysis of myelodysplastic syndromes among atomic bomb survivors in Nagasaki. <i>Haematologica</i> , 2020 , 105, 358-365	6.6	3
9	Identification of a homozygous frameshift variant in RFLNA in a patient with a typical phenotype of spondylocarpotarsal synostosis syndrome. <i>Journal of Human Genetics</i> , 2019 , 64, 467-471	4.3	2

8	Clonal dynamics in a case of acute monoblastic leukemia that later developed myeloproliferative neoplasm. <i>International Journal of Hematology</i> , 2018 , 108, 213-217	2.3	2
7	The Ruby UCSC API: accessing the UCSC genome database using Ruby. <i>BMC Bioinformatics</i> , 2012 , 13, 240	3.6	2
6	Targeting Adaptive IRE1 β Signaling and PLK2 in Multiple Myeloma: Possible Anti-Tumor Mechanisms of KIRA8 and Nilotinib. <i>International Journal of Molecular Sciences</i> , 2020 , 21, -related disorder in a patient with a novel frameshift variant (c.3925dup). <i>Human Genome Variation</i> , 2019 , 6, 54	6.3	2
5	Target Capture/Next-Generation Sequencing for Nonsyndromic Cleft Lip and Palate in the Japanese Population. <i>Cleft Palate-Craniofacial Journal</i> , 2020 , 57, 80-87	1.8	1
4	Single human papillomavirus 16 or 52 infection and later cytological findings in Japanese women with NILM or ASC-US. <i>Journal of Human Genetics</i> , 2014 , 59, 251-5	1.9	1
3	Multifaceted array-based keloidal gene expression profiling reveals specific MDFI upregulation in keloid lesions. <i>Clinical and Experimental Dermatology</i> , 2021 , 46, 1255-1261	4.3	0
2	A unique missense variant in the E1A-binding protein P400 gene is implicated in schizophrenia by whole-exome sequencing and mutant mouse models. <i>Translational Psychiatry</i> , 2021 , 11, 132	1.8	0
1		8.6	