Hiroyuki Mishima

List of Publications by Year in Descending Order

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	Heterozygous missense variant of the proteasome subunit type 9 causes neonatal-onset autoinflammation and immunodeficiency. *Nature Communications*, **2021**, 12, 6819	17.4	4
42	Functionally validated SCN5A variants allow interpretation of pathogenicity and prediction of lethal events in Brugada syndrome. *European Heart Journal*, **2021**, 42, 2854-2863	9.5	4
41	Bile extracellular vesicles from end-stage liver disease patients show altered microRNA content. *Hepatology International*, **2021**, 15, 821-830	8.8	3
40	A unique missense variant in the E1A-binding protein P400 gene is implicated in schizophrenia by whole-exome sequencing and mutant mouse models. *Translational Psychiatry*, **2021**, 11, 132	8.6	
39	Multifaceted array-based keloidal gene expression profiling reveals specific MDFI upregulation in keloid lesions. *Clinical and Experimental Dermatology*, **2021**, 46, 1255-1261	1.8	0
38	Targeting Adaptive IRE1 Signaling and PLK2 in Multiple Myeloma: Possible Anti-Tumor Mechanisms of KIRA8 and Nilotinib. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	2
37	Genome analysis of myelodysplastic syndromes among atomic bomb survivors in Nagasaki. *Haematologica*, **2020**, 105, 358-365	6.6	3
36	Target Capture/Next-Generation Sequencing for Nonsyndromic Cleft Lip and Palate in the Japanese Population. *Cleft Palate-Craniofacial Journal*, **2020**, 57, 80-87	1.9	1
35	A homozygous splice site ROBO1 mutation in a patient with a novel syndrome with combined pituitary hormone deficiency. *Journal of Human Genetics*, **2019**, 64, 341-346	4.3	6
34	Evaluation of Face2Gene using facial images of patients with congenital dysmorphic syndromes recruited in Japan. *Journal of Human Genetics*, **2019**, 64, 789-794	4.3	26
33	Nonsense mutation in causes normal-pressure hydrocephalus with ciliary abnormalities. *Neurology*, **2019**, 92, e2364-e2374	6.5	34
32	Identification of a homozygous frameshift variant in RFLNA in a patient with a typical phenotype of spondylocarpotarsal synostosis syndrome. *Journal of Human Genetics*, **2019**, 64, 467-471	4.3	2
31	-related disorder in a patient with a novel frameshift variant (c.3925dup). *Human Genome Variation*, **2019**, 6, 54	1.8	1
30	Identification of a variant associated with early-onset diabetes in the intron of the insulin gene with exome sequencing. *Journal of Diabetes Investigation*, **2019**, 10, 947-950	3.9	6
29	Whole-exome sequencing and gene-based rare variant association tests suggest that PLA2G4E might be a risk gene for panic disorder. *Translational Psychiatry*, **2018**, 8, 41	8.6	11
28	Clonal dynamics in a case of acute monoblastic leukemia that later developed myeloproliferative neoplasm. *International Journal of Hematology*, **2018**, 108, 213-217	2.3	2
27	Detection of de novo single nucleotide variants in offspring of atomic-bomb survivors close to the hypocenter by whole-genome sequencing. *Journal of Human Genetics*, **2018**, 63, 357-363	4.3	12

26	Patients with SATB2-associated syndrome exhibiting multiple odontomas. *American Journal of Medical Genetics, Part A*, **2018**, 176, 2614-2622	2.5	7
25	Sick sinus syndrome with HCN4 mutations shows early onset and frequent association with atrial fibrillation and left ventricular noncompaction. *Heart Rhythm*, **2017**, 14, 717-724	6.7	27
24	Deep sequencing reveals variations in somatic cell mosaic mutations between monozygotic twins with discordant psychiatric disease. *Human Genome Variation*, **2017**, 4, 17032	1.8	15
23	Three brothers with a nonsense mutation in caused by parental germline mosaicism. *Human Genome Variation*, **2017**, 4, 17045	1.8	7
22	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. *Journal of Human Genetics*, **2016**, 61, 793-6	4.3	10
21	Pregnancy-associated microRNAs in plasma as potential molecular markers of ectopic pregnancy. *Fertility and Sterility*, **2015**, 103, 1202-8.e1	4.8	23
20	Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. *Clinical Immunology*, **2015**, 160, 255-60	9	22
19	Germline mutations causing familial lung cancer. *Journal of Human Genetics*, **2015**, 60, 597-603	4.3	16
18	Metabolic autopsy with next generation sequencing in sudden unexpected death in infancy: Postmortem diagnosis of fatty acid oxidation disorders. *Molecular Genetics and Metabolism Reports*, **2015**, 5, 26-32	1.8	10
17	Clinical applications of analysis of plasma circulating complete hydatidiform mole pregnancy-associated miRNAs in gestational trophoblastic neoplasia: a preliminary investigation. *Placenta*, **2014**, 35, 787-9	3.4	13
16	Circulating levels of maternal plasma cell-free pregnancy-associated placenta-specific microRNAs are associated with placental weight. *Placenta*, **2014**, 35, 848-51	3.4	11
15	Identification of endometrioid endometrial carcinoma-associated microRNAs in tissue and plasma. *Gynecologic Oncology*, **2014**, 132, 715-21	4.9	61
14	Single human papillomavirus 16 or 52 infection and later cytological findings in Japanese women with NILM or ASC-US. *Journal of Human Genetics*, **2014**, 59, 251-5	4.3	0
13	Predominantly placenta-expressed mRNAs in maternal plasma as predictive markers for twin-twin transfusion syndrome. *Prenatal Diagnosis*, **2014**, 34, 345-9	3.2	6
12	Genome-wide association study of HPV-associated cervical cancer in Japanese women. *Journal of Medical Virology*, **2014**, 86, 1153-8	19.7	21
11	Characterization of placenta-specific microRNAs in fetal growth restriction pregnancy. *Prenatal Diagnosis*, **2013**, 33, 214-22	3.2	102
10	Uniparental disomy analysis in trios using genome-wide SNP array and whole-genome sequencing data imply segmental uniparental isodisomy in general populations. *Gene*, **2013**, 512, 267-74	3.8	18
9	Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer. *Journal of Human Genetics*, **2013**, 58, 250-3	4.3	18

8	Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase IIo processing in transcription-coupled nucleotide-excision repair. *Nature Genetics*, **2012**, 44, 586-92	36.3	123
7	The Ruby UCSC API: accessing the UCSC genome database using Ruby. *BMC Bioinformatics*, **2012**, 13, 240	3.6	2
6	Biogem: an effective tool-based approach for scaling up open source software development in bioinformatics. *Bioinformatics*, **2012**, 28, 1035-7	7.2	22
5	Identification of novel schizophrenia loci by homozygosity mapping using DNA microarray analysis. *PLoS ONE*, **2011**, 6, e20589	3.7	19
4	Maternal uniparental isodisomy and heterodisomy on chromosome 6 encompassing a CUL7 gene mutation causing 3M syndrome. *Clinical Genetics*, **2011**, 80, 478-83	4	16
3	Agile parallel bioinformatics workflow management using Pwrake. *BMC Research Notes*, **2011**, 4, 331	2.3	17
2	Proteasome assembly defect due to a proteasome subunit beta type 8 (PSMB8) mutation causes the autoinflammatory disorder, Nakajo-Nishimura syndrome. *Proceedings of the National Academy of Sciences of the United States of America*, **2011**, 108, 14914-9	11.5	227
1	Application of the Linux cluster for exhaustive window haplotype analysis using the FBAT and Unphased programs. *BMC Bioinformatics*, **2008**, 9 Suppl 6, S10	3.6	5