Hiroyuki Mishima

List of Publications by Year
in descending order

45 papers	1,337 citations	393982 19 h-index	360668 35 g-index
46 all docs	46 docs citations	46 times ranked	2951 citing authors

#	Article	IF	Citations
1	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, 14914-14919.	3.3	288
2	Mutations in UVSSA cause UV-sensitive syndrome and impair RNA polymerase IIo processing in transcription-coupled nucleotide-excision repair. Nature Genetics, 2012, 44, 586-592.	9.4	162
3	Characterization of placentaâ€specific microRNAs in fetal growth restriction pregnancy. Prenatal Diagnosis, 2013, 33, 214-222.	1.1	135
4	Identification of endometrioid endometrial carcinoma-associated microRNAs in tissue and plasma. Gynecologic Oncology, 2014, 132, 715-721.	0.6	74
5	Nonsense mutation in CFAP43 causes normal-pressure hydrocephalus with ciliary abnormalities. Neurology, 2019, 92, e2364-e2374.	1.5	65
6	Evaluation of Face2Gene using facial images of patients with congenital dysmorphic syndromes recruited in Japan. Journal of Human Genetics, 2019, 64, 789-794.	1.1	51
7	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.	0.3	43
8	Functionally validated SCN5A variants allow interpretation of pathogenicity and prediction of lethal events in Brugada syndrome. European Heart Journal, 2021, 42, 2854-2863.	1.0	37
9	Novel compound heterozygous DNA ligase IV mutations in an adolescent with a slowly-progressing radiosensitive-severe combined immunodeficiency. Clinical Immunology, 2015, 160, 255-260.	1.4	29
10	Biogem: an effective tool-based approach for scaling up open source software development in bioinformatics. Bioinformatics, 2012, 28, 1035-1037.	1.8	27
11	Genomeâ€wide association study of HPVâ€associated cervical cancer in Japanese women. Journal of Medical Virology, 2014, 86, 1153-1158.	2.5	27
12	Pregnancy-associated microRNAs inÂplasma as potential molecular markers of ectopic pregnancy. Fertility and Sterility, 2015, 103, 1202-1208.e1.	0.5	27
13	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, 267-274.	1.0	26
14	Deep sequencing reveals variations in somatic cell mosaic mutations between monozygotic twins with discordant psychiatric disease. Human Genome Variation, 2017, 4, 17032.	0.4	22
15	Maternal uniparental isodisomy and heterodisomy on chromosome 6 encompassing a CUL7 gene mutation causing 3M syndrome. Clinical Genetics, 2011, 80, 478-483.	1.0	21
16	Copy number variation of the antimicrobial-gene, defensin beta 4, is associated with susceptibility to cervical cancer. Journal of Human Genetics, 2013, 58, 250-253.	1.1	21
17	Identification of Novel Schizophrenia Loci by Homozygosity Mapping Using DNA Microarray Analysis. PLoS ONE, 2011, 6, e20589.	1.1	20
18	Agile parallel bioinformatics workflow management using Pwrake. BMC Research Notes, 2011, 4, 331.	0.6	20

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19	Germline mutations causing familial lung cancer. Journal of Human Genetics, 2015, 60, 597-603.	1.1	20
20	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.	1.1	20
21	Heterozygous missense variant of the proteasome subunit Î²-type 9 causes neonatal-onset autoinflammation and immunodeficiency. Nature Communications, 2021, 12, 6819.	5.8	20
22	Circulating levels of maternal plasma cell-free pregnancy-associated placenta-specific microRNAs are associated with placental weight. Placenta, 2014, 35, 848-851.	0.7	16
23	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.	2.4	16
24	Clinical applications of analysis of plasma circulating complete hydatidiform mole pregnancy-associated miRNAs in gestational trophoblastic neoplasia: A preliminary investigation. Placenta, 2014, 35, 787-789.	0.7	14
25	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.	0.4	13
26	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, 793-796.	1.1	13
27	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.	1.1	13
28	Patients with SATB2â€associated syndrome exhibiting multiple odontomas. American Journal of Medical Genetics, Part A, 2018, 176, 2614-2622.	0.7	12
29	Three brothers with a nonsense mutation in KAT6A caused by parental germline mosaicism. Human Genome Variation, 2017, 4, 17045.	0.4	10
30	Targeting Adaptive IRE1Î± Signaling and PLK2 in Multiple Myeloma: Possible Anti-Tumor Mechanisms of KIRA8 and Nilotinib. International Journal of Molecular Sciences, 2020, 21, 6314.	1.8	9
31	Itpr1 regulates the formation of anterior eye segment tissues derived from neural crest cells. Development (Cambridge), 2021, 148, .	1.2	9
32	Identification of a variant associated with earlyâ€onset diabetes in the intron of the insulin gene with exome sequencing. Journal of Diabetes Investigation, 2019, 10, 947-950.	1.1	8
33	Predominantly placentaâ€expressed mRNAs in maternal plasma as predictive markers for twinâ€“twin transfusion syndrome. Prenatal Diagnosis, 2014, 34, 345-349.	1.1	7
34	Bile extracellular vesicles from end-stage liver disease patients show altered microRNA content. Hepatology International, 2021, 15, 821-830.	1.9	7
35	Application of the Linux cluster for exhaustive window haplotype analysis using the FBAT and Unphased programs. BMC Bioinformatics, 2008, 9, S10.	1.2	6
36	Target Capture/Next-Generation Sequencing for Nonsyndromic Cleft Lip and Palate in the Japanese Population. Cleft Palate-Craniofacial Journal, 2020, 57, 80-87.	0.5	6

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37	Genome analysis of myelodysplastic syndromes among atomic bomb survivors in Nagasaki. Haematologica, 2020, 105, 358-365.	1.7	5
38	BCS1L mutations produce Fanconi syndrome with developmental disability. Journal of Human Genetics, 2022, 67, 143-148.	1.1	5
39	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.	1.1	4
40	The Ruby UCSC API: accessing the UCSC genome database using Ruby. BMC Bioinformatics, 2012, 13, 240.	1.2	3
41	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, 251-255.	1.1	2
42	Clonal dynamics in a case of acute monoblastic leukemia that later developed myeloproliferative neoplasm. International Journal of Hematology, 2018, 108, 213-217.	0.7	2
43	KAT6B-related disorder in a patient with a novel frameshift variant (c.3925dup). Human Genome Variation, 2019, 6, 54.	0.4	1
44	Multifaceted arrayâ€based keloidal gene expression profiling reveals specific MDFI upregulation in keloid lesions. Clinical and Experimental Dermatology, 2021, 46, 1255-1261.	0.6	1
45	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.	2.4	0