Gen Tamiya

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2680693/publications.pdf

Version: 2024-02-01

120 papers	5,675 citations	126907 33 h-index	95266 68 g-index
124 all docs	124 docs citations	124 times ranked	7568 citing authors

#	Article	IF	Citations
1	A patient with early-onset SMAX3 and a novel variant of ATP7A. Brain and Development, 2022, 44, 63-67.	1.1	1
2	Maternal Baseline Characteristics and Perinatal Outcomes: The Tohoku Medical Megabank Project Birth and Three-Generation Cohort Study. Journal of Epidemiology, 2022, 32, 69-79.	2.4	13
3	Heterozygous calcyclin-binding protein/Siah1-interacting protein (CACYBP/SIP) gene pathogenic variant linked to a dominant family with paucity of interlobular bile duct. Journal of Human Genetics, 2022, , .	2.3	0
4	Two Siblings with Cerebellar Ataxia, Mental Retardation, and Disequilibrium Syndrome 4 and a Novel Variant of <i>ATP8A2</i>. Tohoku Journal of Experimental Medicine, 2022, 256, 321-326.	1.2	6
5	The longest reported sibling survivors of a severe form of congenital myasthenic syndrome with the <scp> ALG14 </scp> pathogenic variant. American Journal of Medical Genetics, Part A, 2022, 188, 1293-1298.	1.2	2
6	Development of a prognostic prediction support system for cervical intraepithelial neoplasia using artificial intelligence-based diagnosis. Journal of Gynecologic Oncology, 2022, 33, .	2.2	3
7	Study Profile of the Tohoku Medical Megabank Community-Based Cohort Study. Journal of Epidemiology, 2021, 31, 65-76.	2.4	81
8	Artificial intelligence powered statistical genetics in biobanks. Journal of Human Genetics, 2021, 66, 61-65.	2.3	16
9	Facial UV photo imaging for skin pigmentation assessment using conditional generative adversarial networks. Scientific Reports, 2021, 11, 1213.	3.3	9
10	Estimation of the carrier frequencies and proportions of potential patients by detecting causative gene variants associated with autosomal recessive bone dysplasia using a whole-genome reference panel of Japanese individuals. Human Genome Variation, 2021, 8, 2.	0.7	3
11	Genome-wide meta-analysis identifies 127 open-angle glaucoma loci with consistent effect across ancestries. Nature Communications, 2021, 12, 1258.	12.8	196
12	Body mass index and colorectal cancer risk: A Mendelian randomization study. Cancer Science, 2021, 112, 1579-1588.	3.9	25
13	ALOX12 mutation in a family with dominantly inherited bleeding diathesis. Journal of Human Genetics, 2021, 66, 753-759.	2.3	5
14	Ethnic and trans-ethnic genome-wide association studies identify new loci influencing Japanese Alzheimerâ€™s disease risk. Translational Psychiatry, 2021, 11, 151.	4.8	34
15	GWAS Identified IL4R and the Major Histocompatibility Complex Region as the Associated Loci of Total Serum IgE Levels in 9,260 Japanese Individuals. Journal of Investigative Dermatology, 2021, 141, 2749-2752.	0.7	4
16	Japonica Array NEO with increased genome-wide coverage and abundant disease risk SNPs. Journal of Biochemistry, 2021, 170, 399-410.	1.7	17
17	Identification and Validation of Combination Plasma Biomarker of Afamin, Fibronectin and Sex Hormone-Binding Globulin to Predict Pre-eclampsia. Biological and Pharmaceutical Bulletin, 2021, 44, 804-815.	1.4	10
18	Smooth-threshold multivariate genetic prediction incorporating geneâ€“environment interactions. G3: Genes, Genomes, Genetics, 2021, 11, .	1.8	0

#	Article	IF	Citations
19	A cross-population atlas of genetic associations for 220 human phenotypes. Nature Genetics, 2021, 53, 1415-1424.	21.4	560
20	Construction and integration of three de novo Japanese human genome assemblies toward a population-specific reference. Nature Communications, 2021, 12, 226.	12.8	31
21	jMorp updates in 2020: large enhancement of multi-omics data resources on the general Japanese population. Nucleic Acids Research, 2021, 49, D536-D544.	14.5	107
22	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	27.8	353
23	dbTMM: an integrated database of large-scale cohort, genome and clinical data for the Tohoku Medical Megabank Project. Human Genome Variation, 2021, 8, 44.	0.7	7
24	Cohort Profile: Tohoku Medical Megabank Project Birth and Three-Generation Cohort Study (TMM) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 2020, 49, 18-19m.	1.9	107
25	Endogenization and excision of human herpesvirus 6 in human genomes. PLoS Genetics, 2020, 16, e1008915.	3.5	22
26	Clustering by phenotype and genome-wide association study in autism. Translational Psychiatry, 2020, 10, 290.	4.8	29
27	Machine learning for effectively avoiding overfitting is a crucial strategy for the genetic prediction of polygenic psychiatric phenotypes. Translational Psychiatry, 2020, 10, 294.	4.8	11
28	Machine learning to reveal hidden risk combinations for the trajectory of posttraumatic stress disorder symptoms. Scientific Reports, 2020, 10, 21726.	3.3	3
29	Identification of critical genetic variants associated with metabolic phenotypes of the Japanese population. Communications Biology, 2020, 3, 662.	4.4	16
30	Improved metabolomic data-based prediction of depressive symptoms using nonlinear machine learning with feature selection. Translational Psychiatry, 2020, 10, 157.	4.8	24
31	Genome-wide association study identifies new loci for albuminuria in the Japanese population. Clinical and Experimental Nephrology, 2020, 24, 1-9.	1.6	9
32	A genotype imputation method for de-identified haplotype reference information by using recurrent neural network. PLoS Computational Biology, 2020, 16, e1008207.	3.2	11
33	Design and Progress of Oral Health Examinations in the Tohoku Medical Megabank Project. Tohoku Journal of Experimental Medicine, 2020, 251, 97-115.	1.2	3
34	3.5KJPNv2: an allele frequency panel of 3552 Japanese individuals including the X chromosome. Human Genome Variation, 2019, 6, 28.	0.7	115
35	Quick assessment for systematic test statistic inflation/deflation due to null model misspecifications in genome-wide environment interaction studies. PLoS ONE, 2019, 14, e0219825.	2.5	5
36	GWAS with principal component analysis identifies a gene comprehensively controlling rice architecture. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 21262-21267.	7.1	122

#	Article	IF	Citations
37	InÂVivo Expression of NUP93 and Its Alteration by NUP93 Mutations Causing Focal Segmental Glomerulosclerosis. Kidney International Reports, 2019, 4, 1312-1322.	0.8	10
38	Growth impairment in individuals with citrin deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 501-508.	3.6	14
39	Susceptibility Loci for Tanning Ability in the JapaneseÂPopulation Identified by aÂGenome-WideÂAssociation Study from the TohokuÂMedical Megabank Project Cohort Study. Journal of Investigative Dermatology, 2019, 139, 1605-1608.e13.	0.7	14
40	Estimating carrier frequencies of newborn screening disorders using a whole-genome reference panel of 3552 Japanese individuals. Human Genetics, 2019, 138, 389-409.	3.8	7
41	Outlier detection for questionnaire data in biobanks. International Journal of Epidemiology, 2019, 48, 1305-1315.	1.9	9
42	The prevalence of GALM mutations that cause galactosemia: A database of functionally evaluated variants. Molecular Genetics and Metabolism, 2019, 126, 362-367.	1.1	19
43	Respiratory resistance among adults in a population-based cohort study in Northern Japan. Respiratory Investigation, 2019, 57, 274-281.	1.8	3
44	Automated acquisition of explainable knowledge from unannotated histopathology images. Nature Communications, 2019, 10, 5642.	12.8	92
45	Genome analyses for the Tohoku Medical Megabank Project towards establishment of personalized healthcare. Journal of Biochemistry, 2019, 165, 139-158.	1.7	33
46	Establishment of Integrated Biobank for Precision Medicine and Personalized Healthcare: The Tohoku Medical Megabank Project. JMA Journal, 2019, 2, 113-122.	0.8	21
47	Omics research project on prospective cohort studies from the Tohoku Medical Megabank Project. Genes To Cells, 2018, 23, 406-417.	1.2	38
48	Evaluation of reported pathogenic variants and their frequencies in a Japanese population based on a whole-genome reference panel of 2049 individuals. Journal of Human Genetics, 2018, 63, 213-230.	2.3	35
49	Potential identification of vitamin B6 responsiveness in autism spectrum disorder utilizing phenotype variables and machine learning methods. Scientific Reports, 2018, 8, 14840.	3.3	18
50	Female Japanese quail visually differentiate testosterone-dependent male attractiveness for mating preferences. Scientific Reports, 2018, 8, 10012.	3.3	7
51	Regional genetic differences among Japanese populations and performance of genotype imputation using whole-genome reference panel of the Tohoku Medical Megabank Project. BMC Genomics, 2018, 19, 551.	2.8	14
52	Ethylene-gibberellin signaling underlies adaptation of rice to periodic flooding. Science, 2018, 361, 181-186.	12.6	188
53	Detecting genetic association through shortest paths in a bidirected graph. Genetic Epidemiology, 2017, 41, 481-497.	1.3	5
54	Security controls in an integrated Biobank to protect privacy in data sharing: rationale and study design. BMC Medical Informatics and Decision Making, 2017, 17, 100.	3.0	30

#	Article	IF	Citations
55	Genome-wide meta-analysis in Japanese populations identifies novel variants at the TMC6â€“TMC8 and SIX3â€“SIX2 loci associated with HbA1c. Scientific Reports, 2017, 7, 16147.	3.3	28
56	Smooth-Threshold Multivariate Genetic Prediction with Unbiased Model Selection. Genetic Epidemiology, 2016, 40, 233-243.	1.3	10
57	Immunohistopathological analysis of immature melanocytes from hair follicles of patients with Rhododenol-induced leukoderma. Journal of Dermatological Science, 2016, 84, e177.	1.9	0
58	Whole-exome sequencing confirmation of multiple MC1R variants associated with extensive freckles and red hair: Analysis of a Mongolian family. Journal of Dermatological Science, 2016, 84, 216-219.	1.9	1
59	The Tohoku Medical Megabank Project: Design and Mission. Journal of Epidemiology, 2016, 26, 493-511.	2.4	236
60	Analysis of the genes responsible for steroid-resistant nephrotic syndrome and/or focal segmental glomerulosclerosis in Japanese patients by whole-exome sequencing analysis. Journal of Human Genetics, 2016, 61, 137-141.	2.3	21
61	Immunohistopathological analysis of frizzled-4-positive immature melanocytes from hair follicles of patients with Rhododenol-induced leukoderma. Journal of Dermatological Science, 2015, 80, 156-158.	1.9	8
62	Waardenburg syndrome type <scp>IIE</scp> in a Japanese patient caused by a novel missense mutation in the <scp>SOX</scp>10 gene. Journal of Dermatology, 2015, 42, 1211-1212.	1.2	6
63	Positive selection with diversity in oculocutaneous albinisms type 2 gene (OCA2) among Japanese. Pigment Cell and Melanoma Research, 2015, 28, 233-235.	3.3	2
64	Association of neonatal hyperbilirubinemia in breast-fed infants with UGT1A1 or SLCOs polymorphisms. Journal of Human Genetics, 2015, 60, 35-40.	2.3	15
65	Whole-exome sequencing confirmation of a novel heterozygous mutation in RUNX1 in a pregnant woman with platelet disorder. Platelets, 2015, 26, 364-369.	2.3	6
66	Divergence of East Asians and Europeans Estimated Using Male- and Female-Specific Genetic Markers. Genome Biology and Evolution, 2014, 6, 466-473.	2.5	8
67	A Pollen Coatâ€“Inducible Autoinhibited Ca2+-ATPase Expressed in Stigmatic Papilla Cells Is Required for Compatible Pollination in the Brassicaceae. Plant Cell, 2014, 26, 636-649.	6.6	83
68	Variants in melanogenesisâ€related genes associate with skin cancer risk among <scp>J</scp>apanese populations. Journal of Dermatology, 2014, 41, 296-302.	1.2	13
69	A Mutation of COX6A1 Causes a Recessive Axonal or Mixed Form of Charcot-Marie-Tooth Disease. American Journal of Human Genetics, 2014, 95, 294-300.	6.2	65
70	Gamete Attachment Requires GEX2 for Successful Fertilization in Arabidopsis. Current Biology, 2014, 24, 170-175.	3.9	108
71	Case of a <scp>M</scp>ongolian child with extensive <scp>M</scp>ongolian spots in mucopolysaccharidosis type VI: Identification of a novel mutation in the arylsulfatase <scp>B</scp> gene. Journal of Dermatology, 2013, 40, 758-759.	1.2	0
72	Association of melanogenesis genes with skin color variation among Japanese females. Journal of Dermatological Science, 2013, 69, 167-172.	1.9	29

#	Article	IF	Citations
73	Comparison of caregiver strain in Parkinson's disease between Yamagata, Japan, and Maryland, The United States. Parkinsonism and Related Disorders, 2013, 19, 628-633.	2.2	11
74	A founder haplotype of APOE-Sendai mutation associated with lipoprotein glomerulopathy. Journal of Human Genetics, 2013, 58, 254-258.	2.3	15
75	Generation of a Monoclonal Antibody Specifically Reacting with Neuron-specific TATA-Box Binding Protein-Associated Factor 1 (N-TAF1). Antibodies, 2013, 2, 1-8.	2.5	0
76	The confounding effect of cryptic relatedness for environmental risks of systolic blood pressure on cohort studies. Molecular Genetics & Genomic Medicine, 2013, 1, 45-53.	1.2	4
77	Association of breast-fed neonatal hyperbilirubinemia with UGT1A1 polymorphisms: 211G>A (G71R) mutation becomes a risk factor under inadequate feeding. Journal of Human Genetics, 2013, 58, 7-10.	2.3	39
78	Sustained expression of a neuron-specific isoform of the Taf1 gene in development stages and aging in mice. Biochemical and Biophysical Research Communications, 2012, 425, 273-277.	2.1	20
79	Ultrahigh-dimensional variable selection method for whole-genome gene-gene interaction analysis. BMC Bioinformatics, 2012, 13, 72.	2.6	33
80	Epistatic interaction between Toll-like receptor 3 (TLR3) and prostaglandin E receptor 3 (PTGER3) genes. Journal of Allergy and Clinical Immunology, 2012, 129, 1413-1416.e11.	2.9	28
81	Impaired Glucose Metabolism Slows Executive Function Independent of Cerebral Ischemic Lesions in Japanese Elderly: The Takahata Study. Internal Medicine, 2011, 50, 1671-1678.	0.7	8
82	Polymorphism of proinflammatory cytokine genes and albuminuria in the Japanese general population: the Takahata study. Nephrology Dialysis Transplantation, 2011, 26, 3902-3907.	0.7	3
83	Identification ofMICAas a Susceptibility Gene for PulmonaryMycobacterium aviumComplex Infection. Journal of Infectious Diseases, 2009, 199, 1707-1715.	4.0	23
84	Transcriptional dysregulation: a cause of dystonia?. Lancet Neurology, The, 2009, 8, 416-418.	10.2	11
85	Hepatic AdipoR2 signaling plays a protective role against progression of nonalcoholic steatohepatitis in mice. Hepatology, 2008, 48, 458-473.	7.3	109
86	Genetic Analysis of Craniofacial Traits in the Medaka. Genetics, 2007, 177, 2379-2388.	2.9	31
87	Reduced Neuron-Specific Expression of the TAF1 Gene Is Associated with X-Linked Dystonia-Parkinsonism. American Journal of Human Genetics, 2007, 80, 393-406.	6.2	239
88	TAF1 as the Most Plausible Disease Gene for XDP/DYT3. American Journal of Human Genetics, 2007, 81, 417-418.	6.2	9
89	Refinement of a locus for autosomal dominant hereditary motor and sensory neuropathy with proximal dominancy (HMSN-P) and genetic heterogeneity. Journal of Human Genetics, 2007, 52, 907-914.	2.3	21
90	Genomewide Association Analysis of Human Narcolepsy and a New Resistance Gene. American Journal of Human Genetics, 2006, 79, 252-263.	6.2	64

#	Article	IF	Citations
91	AICAR, an AMPK Activator, Has Protective Effects on Alcoholâ€Induced Fatty Liver in Rats. Alcoholism: Clinical and Experimental Research, 2005, 29, 240S-5S.	2.4	98
92	Functional anatomy of the basal ganglia in Xâ€linked recessive dystoniaâ€parkinsonism. Annals of Neurology, 2005, 58, 7-17.	5.3	166
93	Identification and characterization of novel variants of the thioredoxin reductase 3 new transcript 1 TXNRD3NT1. Mammalian Genome, 2005, 16, 41-49.	2.2	7
94	Gene expression profiling of Japanese psoriatic skin reveals an increased activity in molecular stress and immune response signals. Journal of Molecular Medicine, 2005, 83, 964-975.	3.9	62
95	Whole genome association study of rheumatoid arthritis using 27â€...039 microsatellites. Human Molecular Genetics, 2005, 14, 2305-2321.	2.9	122
96	Genetic features of Mongolian ethnic groups revealed by Y-chromosomal analysis. Gene, 2005, 346, 63-70.	2.2	43
97	Genetic features of Khoton Mongolians revealed by SNP analysis of the X chromosome. Gene, 2005, 357, 95-102.	2.2	3
98	Genetic linkage map of medaka with polymerase chain reaction length polymorphisms. Gene, 2005, 363, 24-31.	2.2	31
99	Novel algorithm for automated genotyping of microsatellites. Nucleic Acids Research, 2004, 32, 6069-6077.	14.5	12
100	Leptin deficiency enhances sensitivity of rats to alcoholic steatohepatitis through suppression of metallothionein. American Journal of Physiology - Renal Physiology, 2004, 287, G1078-G1085.	3.4	26
101	Integrative Annotation of 21,037 Human Genes Validated by Full-Length cDNA Clones. PLoS Biology, 2004, 2, e162.	5.6	290
102	hRDH-E2 gene polymorphisms, variable transcriptional start sites, and psoriasis. Mammalian Genome, 2004, 15, 668-675.	2.2	5
103	Pioglitazone prevents alcohol-induced fatty liver in rats through up-regulation of c-Met. Gastroenterology, 2004, 126, 873-885.	1.3	123
104	Identification, expression analysis and polymorphism of a novel RLTPR gene encoding a RGD motif, tropomodulin domain and proline/leucine-rich regions. Gene, 2004, 343, 291-304.	2.2	30
105	Natural Selection and Population History in the Human Angiotensinogen Gene (AGT): 736 Complete AGT Sequences in Chromosomes from Around the World. American Journal of Human Genetics, 2004, 74, 898-916.	6.2	122
106	Identification of NAD+-dependent isocitrate dehydrogenase 3 Î³-like (IDH3GL) gene and its genetic polymorphisms. Gene, 2003, 323, 141-148.	2.2	2
107	Identification of IÎºBL as the Second Major Histocompatibility Complexâ€“Linked Susceptibility Locus for Rheumatoid Arthritis. American Journal of Human Genetics, 2003, 72, 303-312.	6.2	125
108	SLURP-2, a novel member of the human Ly-6 superfamily that is up-regulated in psoriasis vulgarisâ˜†â˜†Sequence data from this article have been deposited with the DDBJ/EMBL/GenBank Data Libraries under Accession No. AB081838.. Genomics, 2003, 81, 26-33.	2.9	80

#	Article	IF	Citations
109	Association of a Determinant on Mouse Chromosome 18 with Experimental Severe Plasmodium berghei Malaria. Infection and Immunity, 2002, 70, 512-516.	2.2	30
110	Identification of the hRDH-E2 gene, a novel member of the SDR family, and its increased expression in psoriatic lesion. Biochemical and Biophysical Research Communications, 2002, 297, 1171-1180.	2.1	15
111	Identification of novel candidate genes in the diffuse panbronchiolitis critical region of the class I human MHC. Immunogenetics, 2002, 54, 301-309.	2.4	30
112	Genomic Organization, Chromosomal Localization, and the Complete 22 kb DNA Sequence of the Human GCMa/GCM1, a Placenta- Specific Transcription Factor Gene. Biochemical and Biophysical Research Communications, 2000, 278, 134-139.	2.1	14
113	Gene structure and promoter for Crad2 encoding mouse cis-retinol/3Î±-hydroxysterol short-chain dehydrogenase isozyme. Gene, 2000, 251, 175-186.	2.2	14
114	Fine Localization of a Major Disease-Susceptibility Locus for Diffuse Panbronchiolitis. American Journal of Human Genetics, 2000, 66, 501-507.	6.2	72
115	Genome sequencing analysis of the 1.8 Mb entire human MHC class I region. Immunological Reviews, 1999, 167, 193-199.	6.0	65
116	The Critical Region for BehÃ§et Disease in the Human Major Histocompatibility Complex Is Reduced to a 46-kb Segment Centromeric of HLA-B, by Association Analysis Using Refined Microsatellite Mapping. American Journal of Human Genetics, 1999, 64, 1406-1410.	6.2	104
117	Microsatellite polymorphism within the MICB gene among japanese patients with behÃ§etâ€™s disease. Human Immunology, 1998, 59, 500-502.	2.4	32
118	Nucleotide Sequencing Analysis of the 146-Kilobase Segment around theIκBLandMICAGenes at the Centromeric End of the HLA Class I Region. Genomics, 1998, 47, 372-382.	2.9	83
119	Physical Mapping 220 kb Centromeric of the Human MHC and DNA Sequence Analysis of the 43-kb Segment Including theRING1, HKE6,andHKE4Genes. Genomics, 1997, 42, 422-435.	2.9	26
120	An embryological study of ventralization of dorsal structures in the tail of medaka (Oryzias latipes) Da mutants. Development Growth and Differentiation, 1997, 39, 531-538.	1.5	14