Shinya Yamamoto

List of Publications by Year
in descending order

101 papers	5,892 citations	81434 41 h-index	104191 69 g-index
124 all docs	124 docs citations	124 times ranked	9593 citing authors

#	Article	IF	Citations
1	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders. , 2022, , 390-404.		0
2	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. Brain, 2022, 145, 1684-1697.	3.7	5
3	Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling. Science Advances, 2022, 8, eabl5613.	4.7	12
4	Axillary Lymph Node Swelling Mimicking Breast Cancer Metastasis After COVID-19 Vaccination: A Japanese Case Report and Literature Review. In Vivo, 2022, 36, 1041-1046.	0.6	3
5	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	2.9	24
6	ModelMatcher: A scientistâ€centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research. Human Mutation, 2022, , .	1.1	5
7	Fly Cell Atlas: A single-nucleus transcriptomic atlas of the adult fruit fly. Science, 2022, 375, eabk2432.	6.0	295
8	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
9	Role of Drosophila in Human Disease Research 2.0. International Journal of Molecular Sciences, 2022, 23, 4203.	1.8	4
10	Timing and Duration of Axillary Lymph Node Swelling After COVID-19 Vaccination: Japanese Case Report and Literature Review. In Vivo, 2022, 36, 1333-1336.	0.6	2
11	Functional Studies of Genetic Variants Associated with Human Diseases in Notch Signaling-Related Genes Using Drosophila. Methods in Molecular Biology, 2022, , 235-276.	0.4	1
12	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
13	Drosophila as a Model for Infectious Diseases. International Journal of Molecular Sciences, 2021, 22, 2724.	1.8	35
14	Rare deleterious de novo missense variants in Rnf2/Ring2 are associated with a neurodevelopmental disorder with unique clinical features. Human Molecular Genetics, 2021, 30, 1283-1292.	1.4	17
15	Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. Orphanet Journal of Rare Diseases, 2021, 16, 206.	1.2	53
16	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	1.1	13
17	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	2.6	23
18	COVID-19 Screening of Breast Cancer Patients During Treatment: A Single Center Experience in Japan. Cancer Diagnosis & Prognosis, 2021, 1, 423-425.	0.3	0

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19	TM2D genes regulate Notch signaling and neuronal function in Drosophila. PLoS Genetics, 2021, 17, e1009962.	1.5	5
20	Erdheim-Chester Disease. Internal Medicine, 2020, 59, 309-310.	0.3	1
21	Maternal almondex, a neurogenic gene, is required for proper subcellular Notch distribution in early Drosophila embryogenesis. Development Growth and Differentiation, 2020, 62, 80-93.	0.6	5
22	A Genetic Screen for Genes That Impact Peroxisomes in Drosophila Identifies Candidate Genes for Human Disease. G3: Genes, Genomes, Genetics, 2020, 10, 69-77.	0.8	6
23	BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. American Journal of Human Genetics, 2020, 107, 1096-1112.	2.6	32
24	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. Human Molecular Genetics, 2020, 29, 1568-1579.	1.4	29
25	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. American Journal of Human Genetics, 2020, 106, 717-725.	2.6	23
26	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	3.8	71
27	Post-Developmental Roles of Notch Signaling in the Nervous System. Biomolecules, 2020, 10, 985.	1.8	16
28	Making sense out of missense mutations: Mechanistic dissection of Notch receptors through structureâ€function studies in Drosophila. Development Growth and Differentiation, 2020, 62, 15-34.	0.6	14
29	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	60
30	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	2.6	43
31	Using MARRVEL v1.2 for Bioinformatics Analysis of Human Genes and Variant Pathogenicity. Current Protocols in Bioinformatics, 2019, 67, e85.	25.8	14
32	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43
33	In Vivo Functional Study of Disease-associated Rare Human Variants Using Drosophila. Journal of Visualized Experiments, 2019, , .	0.2	34
34	Navigating MARRVEL, a Web-Based Tool that Integrates Human Genomics and Model Organism Genetics Information. Journal of Visualized Experiments, 2019, , .	0.2	20
35	The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases. Human Molecular Genetics, 2019, 28, R207-R214.	1.4	72
36	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	1.1	19

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37	Acute infectious purpura fulminans with Enterobacter aerogenes post-neurosurgery. IDCases, 2019, 15, e00514.	0.4	0
38	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	0.6	8
39	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic spliceâ€site and Charcotâ€Marieâ€Tooth phenotype with early onset symptoms. Molecular Genetics & Genomic Medicine, 2019, 7, e00676.	0.6	18
40	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
41	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	2.6	59
42	An efficient CRISPR-based strategy to insert small and large fragments of DNA using short homology arms. ELife, 2019, 8, .	2.8	105
43	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	2.6	59
44	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	1.1	34
45	Phenotypic heterogeneity of ZMPSTE24 deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1175-1179.	0.7	11
46	A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network. Journal of Pediatrics, 2018, 196, 291-297.e2.	0.9	15
47	Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?. Journal of Genetic Counseling, 2018, 27, 935-946.	0.9	49
48	Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network. Journal of Genetic Counseling, 2018, 27, 1087-1101.	0.9	12
49	Pleiotropic neuropathological and biochemical alterations associated with Myo5a mutation in a rat Model. Brain Research, 2018, 1679, 155-170.	1.1	14
50	Mild encephalitis/encephalopathy with a reversible splenial lesion due to Plasmodium falciparum malaria: a case report. Tropical Medicine and Health, 2018, 46, 37.	1.0	6
51	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	13.9	261
52	Rapid and Integrative Discovery of Retina Regulatory Molecules. Cell Reports, 2018, 24, 2506-2519.	2.9	28
53	Characteristics of undiagnosed diseases network applicants: implications for referring providers. BMC Health Services Research, 2018, 18, 652.	0.9	23
54	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. American Journal of Human Genetics, 2018, 103, 154-162.	2.6	56

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55	A gene-specific T2A-GAL4 library for Drosophila. ELife, 2018, 7, .	2.8	203
56	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
57	Integration of Drosophila and Human Genetics to Understand Notch Signaling Related Diseases. Advances in Experimental Medicine and Biology, 2018, 1066, 141-185.	0.8	35
58	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	1.4	54
59	Unraveling Novel Mechanisms of Neurodegeneration Through a Large-Scale Forward Genetic Screen in Drosophila. Frontiers in Genetics, 2018, 9, 700.	1.1	31
60	Unweaving the role of nuclear Lamins in neural circuit integrity. Cell Stress, 2018, 2, 219-224.	1.4	3
61	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	2.6	142
62	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	2.6	35
63	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	2.6	181
64	Neutral Competition forDrosophilaFollicle and Cyst Stem Cell Niches Requires Vesicle Trafficking Genes. Genetics, 2017, 206, 1417-1428.	1.2	14
65	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	2.6	96
66	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	3.8	95
67	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
68	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	1.2	165
69	A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network. Orphanet Journal of Rare Diseases, 2017, 12, 71.	1.2	53
70	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	1.5	80
71	Loss of Frataxin induces iron toxicity, sphingolipid synthesis, and Pdk1/Mef2 activation, leading to neurodegeneration. ELife, 2016, 5, .	2.8	74
72	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	2.6	68

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73	WAC Regulates mTOR Activity by Acting as an Adaptor for the TTT and Pontin/Reptin Complexes. Developmental Cell, 2016, 36, 139-151.	3.1	47
74	Ubr3, a Novel Modulator of Hh Signaling Affects the Degradation of Costal-2 and Kif7 through Poly-ubiquitination. PLoS Genetics, 2016, 12, e1006054.	1.5	17
75	Rare Functional Variant in TM2D3 is Associated with Late-Onset Alzheimer's Disease. PLoS Genetics, 2016, 12, e1006327.	1.5	47
76	Glial Lipid Droplets and ROS Induced by Mitochondrial Defects Promote Neurodegeneration. Cell, 2015, 160, 177-190.	13.5	617
77	Fruit Flies in Biomedical Research. Genetics, 2015, 199, 639-653.	1.2	149
78	A Voltage-Gated Calcium Channel Regulates Lysosomal Fusion with Endosomes and Autophagosomes and Is Required for Neuronal Homeostasis. PLoS Biology, 2015, 13, e1002103.	2.6	85
79	Morganâ€™s Legacy: Fruit Flies and the Functional Annotation of Conserved Genes. Cell, 2015, 163, 12-14.	13.5	79
80	Impaired Mitochondrial Energy Production Causes Light-Induced Photoreceptor Degeneration Independent of Oxidative Stress. PLoS Biology, 2015, 13, e1002197.	2.6	48
81	The Retromer Complex Is Required for Rhodopsin Recycling and Its Loss Leads to Photoreceptor Degeneration. PLoS Biology, 2014, 12, e1001847.	2.6	75
82	Drosophila Tempura, a Novel Protein Prenyltransferase Î± Subunit, Regulates Notch Signaling Via Rab1 and Rab11. PLoS Biology, 2014, 12, e1001777.	2.6	45
83	Shared mechanisms between Drosophila peripheral nervous system development and human neurodegenerative diseases. Current Opinion in Neurobiology, 2014, 27, 158-164.	2.0	25
84	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	13.5	322
85	Introduction to Notch Signaling. Methods in Molecular Biology, 2014, 1187, 1-14.	0.4	78
86	Large-scale identification of chemically induced mutations in Drosophila melanogaster. Genome Research, 2014, 24, 1707-1718.	2.4	67
87	Dopamine Dynamics and Signaling in <i>Drosophila</i>: An Overview of Genes, Drugs and Behavioral Paradigms. Experimental Animals, 2014, 63, 107-119.	0.7	124
88	Mitochondrial fusion but not fission regulates larval growth and synaptic development through steroid hormone production. ELife, 2014, 3, .	2.8	109
89	dEHBP1 regulates Scabrous secretion during Notch mediated lateral inhibition. Journal of Cell Science, 2013, 126, 3686-96.	1.2	10
90	The C8ORF38 homologue Sicily is a cytosolic chaperone for a mitochondrial complex I subunit. Journal of Cell Biology, 2013, 200, 807-820.	2.3	56

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91	Protein Phosphatase 1ÃŸ Limits Ring Canal Constriction during Drosophila Germline Cyst Formation. PLoS ONE, 2013, 8, e70502.	1.1	27
92	Crag Is a GEF for Rab11 Required for Rhodopsin Trafficking and Maintenance of Adult Photoreceptor Cells. PLoS Biology, 2012, 10, e1001438.	2.6	93
93	dEHBP1 controls exocytosis and recycling of Delta during asymmetric divisions. Journal of Cell Biology, 2012, 196, 65-83.	2.3	35
94	A Mutation in EGF Repeat-8 of Notch Discriminates Between Serrate/Jagged and Delta Family Ligands. Science, 2012, 338, 1229-1232.	6.0	92
95	Endocytosis and Intracellular Trafficking of Notch and Its Ligands. Current Topics in Developmental Biology, 2010, 92, 165-200.	1.0	113
96	Sequoia regulates cell fate decisions in the external sensory organs of adult Drosophila. EMBO Reports, 2009, 10, 636-641.	2.0	13
97	Up-Regulation of NOD1 and NOD2 through TLR4 and TNF-.ALPHA. in LPS-treated Murine Macrophages. Journal of Veterinary Medical Science, 2006, 68, 471-478.	0.3	76
98	Intestinal Gene Expression in TNBS Treated Mice Using GeneChip and Subtractive cDNA Analysis: Implications for Crohn's Disease. Biological and Pharmaceutical Bulletin, 2005, 28, 2046-2053.	0.6	10
99	Regulation of embryo outgrowth by a morphogenic factor, epimorphin, in the mouse. Molecular Reproduction and Development, 2005, 70, 455-463.	1.0	14
100	Effects of Progranulin on Blastocyst Hatching and Subsequent Adhesion and Outgrowth in the Mouse1. Biology of Reproduction, 2005, 73, 434-442.	1.2	46
101	Use of DNA Array to Screen Blastocyst Genes Potentially Involved in the Process of Murine Implantation. Journal of Reproduction and Development, 2003, 49, 473-484.	0.5	8