Shinya Yamamoto

List of Publications by Year in Descending Order

101 papers	3,481 citations	35 h-index	56 g-index
124 ext. papers	4,870 ext. citations	9.5 avg, IF	5.12 L-index

#	Paper	IF	Citations
101	Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling.. *Science Advances*, **2022**, 8, eabl5613	14.3	1
100	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders **2022**, 390-404		
99	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	2.3	0
98	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases.. *Cell Reports*, **2022**, 38, 110517	10.6	2
97	ModelMatcher: A scientist-centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research.. *Human Mutation*, **2022**,	4.7	2
96	Fly Cell Atlas: A single-nucleus transcriptomic atlas of the adult fruit fly.. *Science*, **2022**, 375, eabk2432	33.3	23
95	Timing and Duration of Axillary Lymph Node Swelling After COVID-19 Vaccination: Japanese Case Report and Literature Review.. *In Vivo*, **2022**, 36, 1333-1336	2.3	0
94	Functional Studies of Genetic Variants Associated with Human Diseases in Notch Signaling-Related Genes Using Drosophila. *Methods in Molecular Biology*, **2022**, 235-276	1.4	0
93	COVID-19 Screening of Breast Cancer Patients During Treatment: A Single Center Experience in Japan.. *Cancer Diagnosis & Prognosis*, **2021**, 1, 423-425		
92	De novo FZR1 loss-of-function variants cause developmental and epileptic encephalopathies. *Brain*, **2021**,	11.2	2
91	as a Model for Infectious Diseases. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	6
90	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	5.6	4
89	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	4.2	9
88	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. *Genetics in Medicine*, **2021**, 23, 1889-1900	8.1	5
87	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. *Genetics in Medicine*, **2021**, 23, 259-271	8.1	6
86	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	11	4
85	TM2D genes regulate Notch signaling and neuronal function in Drosophila.. *PLoS Genetics*, **2021**, 17, e1009962	6	2

84	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. *Human Molecular Genetics*, **2020**, 29, 1568-1579	5.6	11
83	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	11	10
82	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. *Neuron*, **2020**, 106, 589-606.e6	13.9	32
81	Post-Developmental Roles of Notch Signaling in the Nervous System. *Biomolecules*, **2020**, 10,	5.9	6
80	Making sense out of missense mutations: Mechanistic dissection of Notch receptors through structure-function studies in Drosophila. *Development Growth and Differentiation*, **2020**, 62, 15-34	3	6
79	Maternal almondex, a neurogenic gene, is required for proper subcellular Notch distribution in early Drosophila embryogenesis. *Development Growth and Differentiation*, **2020**, 62, 80-93	3	3
78	A Genetic Screen for Genes That Impact Peroxisomes in Identifies Candidate Genes for Human Disease. *G3: Genes, Genomes, Genetics*, **2020**, 10, 69-77	3.2	4
77	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	11	9
76	Erdheim-Chester Disease. *Internal Medicine*, **2020**, 59, 309-310	1.1	0
75	Navigating MARRVEL, a Web-Based Tool that Integrates Human Genomics and Model Organism Genetics Information. *Journal of Visualized Experiments*, **2019**,	1.6	10
74	The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases. *Human Molecular Genetics*, **2019**, 28, R207-R214	5.6	33
73	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. *Human Mutation*, **2019**, 40, 1115-1126	4.7	7
72	Acute infectious purpura fulminans with post-neurosurgery. *IDCases*, **2019**, 15, e00514	2	
71	IgG4-related disease: Association with a rare gene variant expressed in cytotoxic T cells. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e686	2.3	6
70	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic splice-site and Charcot-Marie-Tooth phenotype with early onset symptoms. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00676	2.3	11
69	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. *Genetics in Medicine*, **2019**, 21, 161-172	8.1	36
68	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	11	19
67	Using MARRVEL v1.2 for Bioinformatics Analysis of Human Genes and Variant Pathogenicity. *Current Protocols in Bioinformatics*, **2019**, 67, e85	24.2	6

66	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature Communications*, **2019**, 10, 4679	17.4	21
65	In Vivo Functional Study of Disease-associated Rare Human Variants Using Drosophila. *Journal of Visualized Experiments*, **2019**,	1.6	19
64	An efficient CRISPR-based strategy to insert small and large fragments of DNA using short homology arms. *ELife*, **2019**, 8,	8.9	41
63	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
62	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	11	27
61	Unraveling Novel Mechanisms of Neurodegeneration Through a Large-Scale Forward Genetic Screen in. *Frontiers in Genetics*, **2018**, 9, 700	4.5	20
60	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. *American Journal of Human Genetics*, **2018**, 102, 494-504	11	44
59	Genotype-phenotype correlations in individuals with pathogenic RERE variants. *Human Mutation*, **2018**, 39, 666-675	4.7	19
58	Phenotypic heterogeneity of ZMPSTE24 deficiency. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1175-1179	2.5	10
57	A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network. *Journal of Pediatrics*, **2018**, 196, 291-297.e2	3.6	12
56	Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?. *Journal of Genetic Counseling*, **2018**, 27, 935-946	2.5	30
55	Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network. *Journal of Genetic Counseling*, **2018**, 27, 1087-1101	2.5	7
54	IRF2BPL Is Associated with Neurological Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 245-260	11	39
53	Integration of Drosophila and Human Genetics to Understand Notch Signaling Related Diseases. *Advances in Experimental Medicine and Biology*, **2018**, 1066, 141-185	3.6	24
52	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. *Human Molecular Genetics*, **2018**, 27, 2454-2465	5.6	35
51	Unweaving the role of nuclear Lamins in neural circuit integrity. *Cell Stress*, **2018**, 2, 219-224	5.5	1
50	Pleiotropic neuropathological and biochemical alterations associated with Myo5a mutation in a rat Model. *Brain Research*, **2018**, 1679, 155-170	3.7	8
49	Mild encephalitis/encephalopathy with a reversible splenial lesion due to malaria: a case report. *Tropical Medicine and Health*, **2018**, 46, 37	3.4	4

48	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. *New England Journal of Medicine*, **2018**, 379, 2131-2139	59.2	129
47	Rapid and Integrative Discovery of Retina Regulatory Molecules. *Cell Reports*, **2018**, 24, 2506-2519	10.6	8
46	Characteristics of undiagnosed diseases network applicants: implications for referring providers. *BMC Health Services Research*, **2018**, 18, 652	2.9	11
45	De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features. *American Journal of Human Genetics*, **2018**, 103, 154-162	11	25
44	A gene-specific library for. *ELife*, **2018**, 7,	8.9	85
43	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. *American Journal of Human Genetics*, **2017**, 100, 185-192	11	102
42	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	11	23
41	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	11	104
40	Neutral Competition for Follicle and Cyst Stem Cell Niches Requires Vesicle Trafficking Genes. *Genetics*, **2017**, 206, 1417-1428	4	7
39	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. *American Journal of Human Genetics*, **2017**, 100, 128-137	11	65
38	Loss of Nardilysin, a Mitochondrial Co-chaperone for Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. *Neuron*, **2017**, 93, 115-131	13.9	65
37	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. *American Journal of Human Genetics*, **2017**, 101, 768-788	11	81
36	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. *Genetics*, **2017**, 207, 9-27	4	99
35	A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network. *Orphanet Journal of Rare Diseases*, **2017**, 12, 71	4.2	28
34	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. *PLoS Genetics*, **2017**, 13, e1006905	6	42
33	WAC Regulates mTOR Activity by Acting as an Adaptor for the TTT and Pontin/Reptin Complexes. *Developmental Cell*, **2016**, 36, 139-51	10.2	35
32	Ubr3, a Novel Modulator of Hh Signaling Affects the Degradation of Costal-2 and Kif7 through Poly-ubiquitination. *PLoS Genetics*, **2016**, 12, e1006054	6	13
31	Rare Functional Variant in TM2D3 is Associated with Late-Onset Alzheimer's Disease. *PLoS Genetics*, **2016**, 12, e1006327	6	38

30	Loss of Frataxin induces iron toxicity, sphingolipid synthesis, and Pdk1/Mef2 activation, leading to neurodegeneration. *ELife*, **2016**, 5,	8.9	49
29	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. *American Journal of Human Genetics*, **2016**, 99, 991-999	11	42
28	Fruit flies in biomedical research. *Genetics*, **2015**, 199, 639-53	4	108
27	A voltage-gated calcium channel regulates lysosomal fusion with endosomes and autophagosomes and is required for neuronal homeostasis. *PLoS Biology*, **2015**, 13, e1002103	9.7	71
26	Morgan\'s legacy: fruit flies and the functional annotation of conserved genes. *Cell*, **2015**, 163, 12-4	56.2	61
25	Glial lipid droplets and ROS induced by mitochondrial defects promote neurodegeneration. *Cell*, **2015**, 160, 177-90	56.2	395
24	Impaired Mitochondrial Energy Production Causes Light-Induced Photoreceptor Degeneration Independent of Oxidative Stress. *PLoS Biology*, **2015**, 13, e1002197	9.7	36
23	Shared mechanisms between Drosophila peripheral nervous system development and human neurodegenerative diseases. *Current Opinion in Neurobiology*, **2014**, 27, 158-64	7.6	17
22	A drosophila genetic resource of mutants to study mechanisms underlying human genetic diseases. *Cell*, **2014**, 159, 200-214	56.2	239
21	Introduction to Notch signaling. *Methods in Molecular Biology*, **2014**, 1187, 1-14	1.4	66
20	Large-scale identification of chemically induced mutations in Drosophila melanogaster. *Genome Research*, **2014**, 24, 1707-18	9.7	48
19	Dopamine dynamics and signaling in Drosophila: an overview of genes, drugs and behavioral paradigms. *Experimental Animals*, **2014**, 63, 107-19	1.8	79
18	The retromer complex is required for rhodopsin recycling and its loss leads to photoreceptor degeneration. *PLoS Biology*, **2014**, 12, e1001847	9.7	52
17	Drosophila Tempura, a novel protein prenyltransferase □subunit, regulates notch signaling via Rab1 and Rab11. *PLoS Biology*, **2014**, 12, e1001777	9.7	37
16	Mitochondrial fusion but not fission regulates larval growth and synaptic development through steroid hormone production. *ELife*, **2014**, 3,	8.9	73
15	Drosophila EHBP1 regulates Scabrous secretion during Notch-mediated lateral inhibition. *Journal of Cell Science*, **2013**, 126, 3686-96	5.3	8
14	The C8ORF38 homologue Sicily is a cytosolic chaperone for a mitochondrial complex I subunit. *Journal of Cell Biology*, **2013**, 200, 807-20	7.3	47
13	Protein phosphatase 1□limits ring canal constriction during Drosophila germline cyst formation. *PLoS ONE*, **2013**, 8, e70502	3.7	21

List of Publications

12	A mutation in EGF repeat-8 of Notch discriminates between Serrate/Jagged and Delta family ligands. *Science*, **2012**, 338, 1229-32	33.3	75
11	Crag is a GEF for Rab11 required for rhodopsin trafficking and maintenance of adult photoreceptor cells. *PLoS Biology*, **2012**, 10, e1001438	9.7	72
10	dEHBP1 controls exocytosis and recycling of Delta during asymmetric divisions. *Journal of Cell Biology*, **2012**, 196, 65-83	7.3	26
9	Endocytosis and intracellular trafficking of Notch and its ligands. *Current Topics in Developmental Biology*, **2010**, 92, 165-200	5.3	86
8	Sequoia regulates cell fate decisions in the external sensory organs of adult Drosophila. *EMBO Reports*, **2009**, 10, 636-41	6.5	12
7	Up-regulation of NOD1 and NOD2 through TLR4 and TNF-alpha in LPS-treated murine macrophages. *Journal of Veterinary Medical Science*, **2006**, 68, 471-8	1.1	70
6	Effects of progranulin on blastocyst hatching and subsequent adhesion and outgrowth in the mouse. *Biology of Reproduction*, **2005**, 73, 434-42	3.9	40
5	Intestinal gene expression in TNBS treated mice using genechip and subtractive cDNA analysis: implications for Crohn\'s disease. *Biological and Pharmaceutical Bulletin*, **2005**, 28, 2046-53	2.3	8
4	Regulation of embryo outgrowth by a morphogenic factor, epimorphin, in the mouse. *Molecular Reproduction and Development*, **2005**, 70, 455-63	2.6	14
3	Use of DNA array to screen blastocyst genes potentially involved in the process of murine implantation. *Journal of Reproduction and Development*, **2003**, 49, 473-84	2.1	8
2	Loss-of-function in IRF2BPL is associated with neurological phenotypes		2
1	Drosophila functional screening of de novo variants in autism uncovers deleterious variants and facilitates discovery of rare neurodevelopmental diseases		2