SHINYA YAMAMOTO, D.V.M., PH.D.

ASSOCIATE PROFESSOR, DEPARTMENT OF MOLECULAR & HUMAN GENETICS
GRADUATE PROGRAMS IN GENETICS & GENOMICS, NEUROSCIENCE, AND DDMT (DEVELOPMENT, DISEASE MODELS & THERAPEUTICS), BAYLOR COLLEGE OF MEDICINE

INVESTIGATOR, JAN AND DAN DUNCAN NEUROLOGICAL RESEARCH INSTITUTE, TEXAS CHILDREN'S HOSPITAL

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EDUCATION

1999 - 2005 *Bachelor in Veterinary Medicine (B.S.)*
The University of Tokyo, Tokyo, Japan

2005 *Doctor of Veterinary Medicine (D.V.M.)*
Ministry of Agriculture, Forestry & Fisheries, Tokyo, Japan

2005 - 2012 *Doctor of Philosophy in Developmental Biology (Ph.D.)*
Baylor College of Medicine (BCM), Houston, Texas, U.S.A.
Thesis Title: "Novel Insights into the Notch Signaling Pathway through Forward Genetic Approaches: *In vivo* Analyses of *Notch* and *EHBP1* in *Drosophila*"
Advisor: Dr. Hugo J. Bellen

POSTGRADUATE TRAINING

2012 - 2013 *Postdoctoral Fellow*
Department of Molecular & Human Genetics, BCM, Houston, Texas, U.S.A.
Project Title: "Integration of *Drosophila* Phenotypic Screening and Human Whole-Exome Sequencing Datasets to Identify and Study Novel Human Disease Genes"
Advisors: Dr. Hugo J. Bellen [In close collaboration with the Baylor-Hopkins Center for Mendelian Genomics (led by Dr. James R. Lupski) and Human Genome Sequencing Center (led by Dr. Richard Gibbs) at BCM]

ACADEMIC APPOINTMENTS

2013 – 2016 *NRI Fellow (Independent Research Position)*, Jan and Dan Duncan Neurological Research Institute (NRI), Texas Children's Hospital (TCH)

2014 – 2016 *Assistant Professor (non-tenure track)*, Department of Molecular & Human Genetics (primary), Graduate Program in Developmental Biology, BCM

2017 – 2024 *Assistant Professor (tenure track)*, Department of Molecular & Human Genetics (primary), Department of Neuroscience (secondary), Graduate Programs in Genetics & Genomics, Developmental Biology, Neuroscience and DDMT, BCM

2017 – present *Investigator*, NRI, TCH

2024 – present *Associate Professor (tenured)*, Department of Molecular & Human Genetics (primary), Department of Neuroscience (secondary), Graduate Programs in Genetics & Genomics, Developmental Biology, Neuroscience and DDMT, BCM

HONORS AND AWARDS

2005 "Dean's Award for Excellence", Graduate School of Biomedical Sciences (GSBS), BCM

2010 "Mavis P. Kelsey Student Speaker Award", 22nd Annual Graduate Studies Symposium, BCM

2011 "Milton Gregory Poster Award, First place", 23rd Annual Graduate Studies Symposium, BCM

2013 "Best Student Paper Award", Department of Molecular & Human Genetics, BCM

2018 "Nancy Chang, Ph.D. Award for Research Excellence", BCM

2020 "Rolanette & Berdon Lawrence Family Achievement Award in Genetics", BCM

2021 "Best Course Award for Graduate Program in Genetics & Genomics", a recognition for co-directing the *Method and Logic* Course in AY2021, GSBS, BCM

2022 "Outstanding Lecturer Award for Graduate Program in Genetics & Genomics", GSBS, BCM

2022 "Marc Dresden Excellence in Education Award", GSBS, BCM

2023 "Young Alumnus Award", Alumni Executive Committee, BCM

2024 Nominee for "2024 RARE Champion in Research Award", Global Genes

"Johannes Ludwig Janson Award", Alumni Association, University of Tokyo Veterinary Medical School

2025 "Genetics Society of America Early Career Medal", Genetics Society of America (GSA)

2025 "2025 UJA Science Outreach Award", United Japanese researchers Around the world (UJA)

2025 "Kenneth Scott Outstanding Mentorship Award", Department of Molecular & Human Genetics, BCM

2025 "Best Course Award for Graduate Program in Genetics & Genomics", a recognition for co-directing a Ph.D.-level Course in AY2024-2025, GSBS, BCM

2025 "2025 NISTEP Selection", National Institute of Science and Technology Policy (NISTEP, Japan)

TEACHING RELATED ACTIVITIES

GRADUATE PROGRAM LEADERSHIP

2020-present Member, Program Executive Committee. Genetics & Genomics Ph.D. Program, BCM.
2021-2023 Co-Director, Curriculum Committee, Genetics & Genomics Ph.D. Program, BCM.
2021-present Member, Curriculum Committee, GSBS, BCM.
2022-present Associate Director, Genetics & Genomics Ph.D. Program, BCM.
2023-present Member, Student Services Committee, GSBS, BCM.
2024-present Director, Curriculum Committee, Genetics & Genomics Ph.D. Program, BCM.

COURSE DIRECTOR/CO-DIRECTOR (GRADUATE EDUCATION LEVEL)

2020-present *Model Systems in Developmental Biology and Disease,* Ph.D. level course, BCM.
Co-Director with Dr. Ross Poché.
2021-present *Genetics & Genomics Journal Club*, Ph.D. level course, BCM.
Co-Director with Dr. Hamed Jafar-Nejad.

2021-present *Methods and Logic*, Ph.D. level course, Ph.D. level course, BCM.
Co-Director with Dr. Ross Poché.

LECTURES & DISCUSSION FACILITATION (GRADUATE EDUCATION LEVEL)

2007-2018 *Classical Developmental Biology*, Ph.D. level course. Three lectures. BCM
2014-2018 *Developmental Biology Journal Club*. Ph.D. student course. One-two sessions. BCM
2016-2023 *Neural Development*, Ph.D. level course. Three lectures. BCM
2016-present *Responsible Conduct of Research (Ethics) training*, Ph.D. level course. One lecture. BCM
2016-present *Neuro Lab II,* Ph.D. level course. One lecture and one lab. BCM
2017-2022 *Anatomy of the Nervous System,* Ph.D. level course. One lecture and one lab. BCM
2017 *Genetics Ethics Session*. M.D. level course. One discussion session. BCM
2017-present *Genetics Journal Club*. Ph.D. level course. Three terms/tear. BCM
2017-present *Responsible Conduct of Research (Ethics) training*, Postdoc level course. One lecture. BCM
2017-present *Methods and Logic*, Ph.D. level course. Four lectures & discussion sessions/year. BCM
2019 *TBMM Bench to Bedside-Neurodegeneration Journal Club*. Ph.D. level course. One discussion session, BCM
2019-2020 *Powerful Presentations*, Ph.D. level course. Three lectures + 1:1 student meetings. BCM
2019-2023, 2025 *Concepts in Genetics & Genomics*, Ph.D. level course. One lecture. MD Anderson Cancer Center
2020-present *Model Systems in Developmental Biology and Disease.* Ph.D. level course. Four lectures. BCM
2020 CNS Diseases (BIOH 441), B.S. level course. One lecture, University of Montana
2021-2024 *Animal Models of Human Disease*, Ph.D. level course. One lecture. BCM
2022 *CTR-CAQ Lecture Series*, Clinical Translational Research Certificate of Added Qualification, Ph.D. level course. One lecture. BCM
2025-present *Introduction to Genetics & Genomics Research (MEGNT 553)*, M.D. level course. One lecture. BCM

LECTURES (POST-GRADUATE EDUCATION LEVEL)

2023-2024 *Evaluation of Undiagnosed and Rare Condition*, CME Credit Course (ABMS/ACGME), One lecture. Harvard Medical School https://cmecatalog.hms.harvard.edu/evaluation-undiagnosed-and-rare-conditions
2025 *Diagnóstico genético de las enfermedades raras (Genetic diagnosis of rare diseases)*, Training Course for Health Professionals, One lecture. Red de Expertos en Salud de Enfermedades Raras (RIBERSER, a.k.a Iberoamerican Health Expert Network for Rare Diseases) https://intercoonecta.aecid.es/programaci%C3%B3n-de-actividades/diagn-stico-gen-tico-de-las-enfermedades-raras
2026 *Evaluation of Undiagnosed and Rare Condition*, Training Course for Healthcare Providers One lecture. Undiagnosed Diseases Network https://undiagnosed.hms.harvard.edu/events/course/

TEACHING ASSISTANT

2003 *Animal Resource Sciences*, DVM student Course, The University of Tokyo
2006 *Genetics A*, PhD student course, BCM

EDITORIAL & OTHER SCIENTIFIC/ACADEMIC ACTIVITIES

Peer-reviewed Manuscripts for: *Acta Neuropathologica Communications*; *American Journal of Human Genetics (AJHG); Antioxidants; Biochimica et Biophysica Acta (BBA) Molecular Cell Research; Bioinformatics; Cell; Cell*

& Bioscience; Cell Death & Differentiation; Cell Reports; Cell Stress; Clinical and Translational Medicine; Current Biology; Circulation; Development; Developmental Biology; Developmental Cell; Disease Models and Mechanisms; eBioMedicine; eLife; European Journal of Medical Genetics (EJMG); Experinmental Cell Research; Experinmental Neurology; Frontiers in Cell and Developmental Biology; Frontiers in Immunology; Genes, Genomes, Genetics (G3); Genetics; Genetics in Medicine; Genetics in Medicine Open; Genes & Development; Genome Biology; Genome Research, Heliyon; Human Molecular Genetics, Human Mutation; International Journal of Molecular Sciences (IJMS), iScience; Journal of Alzheimer's Disease; Journal of Cell Biology (JCB); Journal of Cell Science (JCS); Journal of Neurodevelopmental Disorders; Journal of Neuroscience; Journal of Visualized Experiments (JoVE); Metallomics; Mitochondrion; Molecular Psychiatry, Mutation Research; Nature Cell Biology; Nucleic Acid Research, PLoS Biology; PLoS Genetics; PLoS One; Proceedings of the National Academy of Sciences (PNAS); *Rare*; *Review Commons; Science Signaling; Scientific Reports*; and *Traffic*.

Peer-reviewed Grants for: *Agency for Medical Research and Development* (Japan, 2019); *Biotechnology and Biological Sciences Research Council* [United Kingdom (UK), 2016, 2019]; *Cerebral Palsy Alliance Research Foundation* (Australia, 2019); CURE Epilepsy (USA, 2026); *Czech Science Foundation* (Czech Republic, 2020); *Department of Defense(DOD)- Congressionally Directed Medical Research Programs (CDMRP) Rare Cancer Research Program (RCRP) Concept-Thoracic Cancers (CON-TC) Peer Review Panel* (USA, 2025), *European Research Counsel* (European Commission, 2022); *Deutsche Forschungsgemeinschaft* (Germany, 2026); *Gulf Coast Consortia* (USA, 2019); *Human Frontier Science Program* (International, 2023 & 2024), *Israel Science Foundation* (Israel, 2021); *Medical Research Council* (UK, 2017, 2022); *National Institutes of Health(NIH)- Cellular and Molecular Neurodegeneration (CMND) Study Section* (USA, 2021 & 2023); *NIH-Genetics of Health and Disease (GHD) Study Section* (USA, 2024, 2025); *Swiss National Science Foundation* (Switzerland, 2022, 2023, 2024); *UK Research and Innovation* (UK, 2024), and *United States-Israel Binational Science Foundation* (USA & Israel, 2024).

Associate Faculty Member, Faculty of 1000 (F1000) (2011-2019)

Chair, Gordon Research Seminar "*Notch Signaling in Development, Regeneration and Disease*", Gordon Research Conferences (2014)"

Editor of the book "*Notch Signaling- Methods and Protocols*", Editors: Hugo J. Bellen and Shinya Yamamoto, Humana Press/Springer (2014)

SFARI Investigator, Simons Foundation Autism Research Initiative (2015-2018)

Co-Director, Model Organisms Screening Center *Drosophila* Core, Undiagnosed Diseases Network (UDN) (2015-present)

Guest Editor of the "*Notch Signaling* (in Japanese)" issue of *Jikken Igaku* (*Experimental Medicine*), Editors: Shinya Yamamoto and Mitsuru Morimoto, Yodosha (Japan) (2016)

Platform Judge for "*29th Annual BCM Graduate Student Research Symposium Speaker Award*" (2017)

Member, Seminar Committee, Department of Neuroscience, BCM (2017-2023)

Member, Functional Study Working Group, Undiagnosed Diseases Network International (2018-present)

Scientific Member, Undiagnosed Diseases Network International (2019-present)

Workshop Chair, Annual *Drosophila* Research Conference, Genetics Society of America (2019)

Chair, Functional Study Working Group, Undiagnosed Diseases Network International (2020-present)

Workshop Chair, The Allied Genetics Conference (TAGC2020), Genetics Society of America (2020)

Workshop Chair, The 43rd Annual Meeting of the Molecular Biology Society of Japan, (2020)

Member, GSA Award Committee, Genetics Society of America (2020-2023)

Guest Editor of the special issue "Role of *Drosophila* in Human Disease Research" and "Role of *Drosophila* in Human Disease Research 2.0", *International Journal of Molecular Sciences* (2020-2021)

Guest Editor of the commentary article collection "Impact of COVID-19 on scientific research and graduate education in the USA and Europe (in Japanese)", *Jikken Igaku* (*Experimental Medicine*), Yodosha (Japan) (2021)

Editorial Board Member, *Rare*, Elsevier (the Netherlands) (2023-present)

Guest Editor of the article collection "Maximizing your research training abroad (in Japanese)", *Jikken Igaku* (*Experimental Medicine*), Yodosha (Japan) (2023)

Co-Chair, Executive Committee and Steering Committee, UDN (2023-2025)

Editorial Supporter, *Jikken Igaku* (*Experimental Medicine*), Yodosha (Japan) (2024-2025)

Co-Chair, UDN Environmental Triggers Workshop, NIH (NINDS & NHEIS) (2024)

Lead author and editor of the book "A practical guide to conducting research abroad (in Japanese)", Authors: Shinya Yamamoto and Daisuke Nakada, Yodosha (Japan) (2024)

Editor, *Disease Models & Mechanisms*, Company of Biologists (UK) (2025-present)

Member, Awards Committee, BCM Alumi Association (2025-present)

Member, Faculty Committee on Awards, BCM (2025-present)

Member, Proposal Review Committee, Department of Molecular & Human Genetics, BCM (2025-present)

FUNDING

CURRENT SUPPORT

U54 OD030165 (ORIP) Heaney/Lee/Milosavljevic/**Yamamoto** (PD/PI) 9/15/2020-8/31/2030
The major goal of this project is to establish a BCM Center for Precision Medicine Models, which will support local, national, and international programs and individual researchers in the development of precision animal models that end the diagnostic odyssey of patients with undiagnosed, rare, and Mendelian diseases and serve as resources for pre-clinical studies investigating personalized medicine approaches to their care.
Role: MPI

RF1 AG071557 (NIA) **Yamamoto** (PD/PI) 5/1/2021-4/30/2026
The major goal of this proposal is to study the function of TM2D3 and its related genes in the context of Alzheimer's disease pathogenesis in Notch signaling.
Role: PI

R01 HG011795 (NHGRI) Bellen/Wangler (PD/PI) 7/13/2021-4/30/2026
The major goal of this project is to facilitate the implementation of genomic medicine in a medically underserved population by combining the latest clinical diagnostics, bioinformatics and *Drosophila* genetics tools in a clinical setting.
Role: Co-I

#2023-32824 (CHAN ZUCKERBERG INITIATIVE) Gibbs/King/Bellen (PD/PI) 08/01/2023 - 07/31/2027
The major goal of this subcontract is to provide diagnosis and study mechanisms of rare undiagnosed pediatric disorders at Texas Children's Hospital.
Role: Co-I

#2023-332162 (CHAN ZUCKERBERG INITIATIVE) **Yamamoto** (PD/PI) 11/01/2023 - 10/31/2025
The major goal of this grant is to further develop MARRVEL, AI-MARRVEL and ModelMatcher to facilitate diagnosis and collaborative research for rare diseases.
Role: PI

U2C NS132415-03S01 subaward (NINDS) Kohane (PD/PI) 04/01/2025 - 03/31/2026
The major goal of this subcontract is to pursue functional studies in *Drosophila* to support the diagnostic mission of the Phase III UDN.
Role: Co-I

Mark A. Wallace Endowment Award (Texas Children's Hospital) **Yamamoto** (PI) 7/1/2025-6/30/2026
This internal funding is used to study the role of KCJN2 in psychiatric diseases using *Drosophila*.
Role: PI

Raynor Funds (Texas Children's Hospital) **Yamamoto** (PI) 7/1/2025-6/30/2026
This philanthropic fund is used to support functional studies in *Drosophila* to study variants and genes of uncertain significance found in patients with rare and undiagnosed diseases.
Role: PI

R21OD038417 (ORIP) **Yamamoto** (PD/PI) 7/15/2025-6/30/2027
The major goal of this project is to sequence and map ~700 EMS-induced lethal mutants on the *Drosophila* X-chromosome using the state-of-the-art-genomic and genetic technology. These mutants were left unmapped from the X-chromosome screen project (Yamamoto et al., 2014) and this collection is known to be enriched for mutants that affect evolutionarily conserved genes that are linked to rare diseases in human.

#2025-367725 (CHAN ZUCKERBERG INITIATIVE) LIU (PD/PI) 11/01/2025 - 10/31/2027
The major goal of this grant is to on project Benchmarking for Variant Interpretation to generate high-quality, real-world training and benchmarking datasets for interpreting rare disease variants and to extend and enhance AI MARRVEL into a more scalable, comprehensive variant prioritization platform.
Role: Co-PI

Anonymous fund (Texas Children's Hospital) **Yamamoto** (PI) 1/1/2026-12/31/2026
This philanthropic fund from an anonymous doner to the Jan and Dan Duncan Neuorological Reseearch Institute is designed to facilitate the study the functions of new candidate genes and variants that are linked to neurodevelopmental and neurodegenerative diseases using *Drosophila*.
Role: PI

PAST SUPPORT

Nakajima Foundation Fellowship (Nakajima Foundation) **Yamamoto** (Fellow) 5/1/2005-4/30/2010
This competitive fellowship is awarded to 3-4 Japanese students annually to pursue graduate studies in the life science field in a foreign country. Fellows receive a generous stipend for 5 years.
Role: Pre-Doctoral Fellow

NRI Fellowship (Texas Children's Hospital) **Yamamoto** (PI) 3/1/2013-2/28/2017
The NRI Fellowship is designed to recruit and support candidates that have just received their final degrees (Ph.D.) to start up an independent laboratory at the Jan and Dan Duncan Neurological Research Institute

(NRI) at TCH. This support was used to perform a forward genetic screen to identify new genes involved in Dopamine synthesis, secretion, and metabolism in *Drosophila*.
Role: PI

R13 HD080297-01 (NCI & NICHD) Blacklow (PI) 5/10/2014-4/30/2015
This grant supported a Gordon Research Conference (GRC) and Seminar (GRS) on "Notch Signaling in Development, Regeneration & Disease" held in July 19-25, 2014 at Bates College, Maine.
Role: Chair of GRS.

Targeted-Functional Screen of Autism-Associated Variants Award
(Simons Foundation) Wangler/**Yamamoto** (Co-PIs) 8/1/2015-7/31/2018
The major goal of this project is to perform functional analyses of rare variants in conserved Autism genes found in the Simons Simplex Cohort using *Drosophila*.
Role: Co-PI

U54 NS093793 (NINDS & CommonFund). Bellen (PD/PI) 9/15/2015-6/30/2023
The major goal of this project is to establish and operate a Model Organisms Screening Center (MOSC) that will provide valuable *in vivo* functional information of conserved genes that are likely to be involved in rare human diseases by performing genetic experiments in *Drosophila* and Zebrafish as part of the larger Undiagnosed Disease Network (UDN).
Role: Co-I of overall, Project Lead of *Drosophila* Core

New Investigator Research Grant (Alzheimer's Association) **Yamamoto** (PI) 10/1/2015-9/30/2017
The major goal of this project is to understand the functional effect of a late-onset Alzheimer's disease associated variant in *TM2D3* using *Drosophila*. **Role: PI.**

R24 OD022005 (ORIP) Bellen (PD/PI) 06/01/2016 - 05/31/2025 (NCE)
The goal of this study is to generate a large library of human cDNA expressing constructs and transgenic *Drosophila* strains to facilitate the use of fruit flies in biomedical research.
Role: Co-I

R01 DC014932 (NIDCD) Groves (PI) 12/1/2016-11/30/2022
The goal of this proposal is to study the role of the ubr3 gene in regulating proteins involved in usher syndrome, the most common form of deaf-blindness, and myh9-related disease syndromes that can also cause deafness. In addition, we will perform genetic screens to identify additional genes involved in hearing in insects and mice.
Role: Co-I

Junior Faculty Seed Funding Award (Naman Family Fund for Basic Research and Caroline Wiess Law Fund for Research in Molecular Medicine) **Yamamoto** (PI) 07/01/2017-06/30/2018
The major goal of this project is to understand the function of proteins and non-coding RNAs encoded in the Zika viral genome using *Drosophila* to understand the mechanism by which this virus cause microcephaly in humans.
Role: PI

Nancy Chang, Ph.D. Award for Research Excellence **Yamamoto** (PI) 07/01/2019-06/30/2020
The major goal of this project is to understand the function of TM2D family proteins in the context of Notch signaling and late-onset Alzheimer's disease using *Drosophila*.
Role: PI

U01 HG007530-07 subaward (NHGRI) Kohane/McCray/Might (PD/PI) 7/1/2020-6/30/2021

The major goal of this administrative supplement is to study the function of *GLS*, a gene identified as a in the Undiagnosed Disease Network as a disease-causing gene, using *Drosophila*.
Role: Project Lead

U54 NS093793-06S1 (NINDS) Bellen (PD/PI) 7/01/2020-6/30/2021
The major goal of this administrative supplement is to establish a global matchmaking system between scientist and clinicians called ModelMatcher to facilitate rare and undiagnosed disease research.
Role: Project Lead

R24 OD022005-05S1 (ORIP) Bellen (PD/PI) 07/01/2020 - 06/30/2022
The major goal of this administrative supplement is to establish a genetic tool kit of SARS-CoV-2 and human cDNAs to study the functional interaction of host-protein proteins involved in COVID-19.
Role: Co-I

U54 NS093793-07S1 (NINDS) Bellen (PD/PI) 7/01/2021-6/30/2022
The major goal of this administrative supplement is to increase the utility of MARRVEL and ModelMatcher by integrating these two online resources to facilitate rare and undiagnosed disease research.
Role: Project Lead

RESEARCH AWARD (SATB2 Gene Foundation) **Yamamoto** (PD/PI) 12/1/2021-2/28/2023
The major goal of this project is to study the function of pathogenic variants identified in SATB2-associated syndrome and to further functionally characterize the role of this gene in the nervous system using *Drosophila*.
Role: PI

R24 OD022005-07S1 (ORIP) Bellen (PD/PI) 06/01/2022-05/31/2025 (NCE)
The major goal of this administrative supplement is to expand and characterize a genetic tool kit of SARS-CoV-2 and human cDNAs to study the functional interaction of host-protein proteins involved in COVID-19.
Role: Co-I

U54 NS093793-07S2 (NINDS & CommonFund). Bellen (PD/PI) 7/1/2022-6/30/2023
The major goal of this project is to continue to perform genetic experiments in *Drosophila* and Zebrafish in the Model Organisms Screening Center (MOSC) to support the mission of the larger Undiagnosed Disease Network (UDN).
Role: Co-I of overall, Project Lead of *Drosophila* Core

U2C NS132415-01S01 subaward (NINDS) Kohane (PD/PI) 07/01/2023 - 03/31/2024
The major goal of this subcontract is to pursue functional studies in *Drosophila* to support the diagnostic mission of the Phase III UDN.
Role: Co-I

U2C NS132415-02S01 subaward (NINDS) Kohane (PD/PI) 04/01/2024 - 03/31/2025
The major goal of this subcontract is to pursue functional studies in *Drosophila* to support the diagnostic mission of the Phase III UDN.
Role: Co-I

PUBLICATIONS

ARTICLES PUBLISHED & IN PRESS (CHRONOLOGICAL ORDER)

1. Qin J, Takahashi Y, Imai M, **Yamamoto S**, Takakura K, Noda Y, Imakawa K (2003) Use of DNA array to screen blastocyst genes potentially involved in the process of murine implantation. ***Journal of Reproduction and Development***, 49(6):473-484. PMID: 14967898, PMCID: N/A.

2. Qin J, Takahashi Y, Isuzugawa K, Imai M, **Yamamoto S**, Hirai Y, Imakawa K (2005) Regulation of embryo outgrowth by a morphogenic factor, epimorphin, in the mouse. ***Molecular Reproduction and Development***, 70:455-463. PMID: 15685636, PMCID: N/A.

3. Qin J, Diaz-Cueto L, Schwarze JE, Takahashi Y, Imai M, Isuzugawa K, **Yamamoto S**, Chang KT, Gerton GL, Imakawa K (2005) Effects of progranulin on blastocyst hatching and subsequent adhesion and outgrowth in the mouse. ***Biology of Reproduction***, 73:434-442. PMID: 15901638, PMCID: N/A.

4. **Yamamoto S**, Isuzugawa K, Takahashi Y, Murase Y, Iwata M, Arisawa T, Nakano H, Nishimura N, Yamato S, Ohta M, Ina K, Murata T, Hori M, Ozaki H, Imakawa K (2005) Intestinal gene expression in TNBS treated mice using genechip and subtractive cDNA analysis: implications for Crohn's disease. ***Biological and Pharmaceutical Bulletin***, 28:2046-2053. PMID: 16272687, PMCID: N/A.

5. Takahashi Y, Isuzugawa K, Murase Y, Imai M, **Yamamoto S**, Iizuka M, Akira S, Bahr GM, Momotani E, Hori M, Ozaki H, Imakawa K (2006) Up-regulation of NOD1 and NOD2 through TLR4 and TNF-alpha in LPS-treated murine macrophages. ***The Journal of Veterinary Medical Science***, 68:471-478. PMID: 16757890, PMCID: N/A.

6. Andrews HK, Giagtzoglou N, **Yamamoto S**, Schulze KL, Bellen HJ (2009) Sequoia regulates cell fate decisions in the external sensory organs of adult *Drosophila*. ***EMBO Reports***, 10:636-641. PMID: 19444309, PMCID: PMC2711842.

7. **Yamamoto S***, Charng W-L*, Bellen HJ (2010) Endocytosis and intracellular trafficking of Notch and its ligands. ***Current Topics in Developmental Biology***, 92: 165-200 **(*equal contribution)**. PMID: 20816395, PMCID: PMC6233319.

8. Giagtzoglou N*, **Yamamoto S***, Zitserman D, Graves HK, Schulze KL, Wang H, Klein H, Bellen HJ (2012) dEHBP1 controls exocytosis and recycling of Delta during asymmetric divisions. ***Journal of Cell Biology***, 196: 65-83 **(*equal contribution)**. PMID: 22213802, PMCID: PMC3255984.

9. **Yamamoto S**, Charng W-L, Rana NA, Kakuda S, Jaiswal M, Bayat V, Xiong B, Zhang K, Sandoval H, David G, Wang H, Haltiwanger RS, Bellen HJ (2012) A mutation in EGF repeat of Notch discriminates between Serrate/Jagged and Delta ligand families. ***Science***, 338: 1229-1232. PMID: 23197537, PMCID: PMC3663443.

10. Xiong B, Bayat V, Jaiswal M, Zhang K, Sandoval H, Charng W-L, Li T, David G, Haueter C, **Yamamoto S**, Bellen HJ (2012) Crag, a GEF for Rab11, is required for rhodopsin trafficking and the maintenance of *Drosophila* photoreceptor cells. ***PLoS Biology***, 10: e1001438. PMID: 23226104, PMCID: PMC3514319.

11. Zhang K, Li Z, Jaiswal M, Bayat V, Xiong B, Sandoval H, Charng W-L, David D, Haueter C, Graham BH, **Yamamoto S**, Bellen HJ (2013) The C8ORF38 homologue Sicily is a cytosolic chaperone for a mitochondrial complex I subunit. ***Journal of Cell Biology***, 200:807-820. PMID: 23509070, PMCID: PMC3601355.

12. **Yamamoto S**, Bayat B, Bellen HJ, Tan C (2013) Protein Phosphatase 1ß limits ring canal constriction during *Drosophila* germline cyst formation. ***PLoS One***, 8:e70502. PMID: 23936219, PMCID: PMC3723691.

13. Giagtzoglou N, Li T, **Yamamoto S**, Bellen HJ (2013) dEHBP1 regulates Scabrous secretion during Notch mediated lateral inhibition. ***Journal of Cell Science***, 126:3686-3696. PMID: 23788431, PMCID: PMC3744027.

14. **Yamamoto S**, Bellen HJ (2014) Preface: Notch Signaling. ***Methods in Molecular Biology***, 1187:v. PMID: 25187920, PMCID: N/A.

15. **Yamamoto S†**, Schulze KL, Bellen HJ† (2014) Introduction to Notch signaling. ***Methods in Molecular Biology***, 1187:1-14. PMID: 25053477, PMCID: N/A. **(†corresponding authors)**.

16. **Yamamoto S†**, Seto ES (2014) Dopamine dynamics and signaling in *Drosophila*: an overview of genes, drugs and behavioral paradigms. ***Experimental Animals***, 63:107-119. PMID: 24770636, PMCID: PMC4160991. **(†corresponding author)**.

17. Charng W.-L, **Yamamoto S**, Jaiswal M, Bayat V, Xiong B, Zhang K, Sandoval H, David G, Gibbs S, Lu H.-C, Chen K, Giagtzoglou N, Bellen HJ (2014) *Drosophila* Tempura, a novel protein prenyltransferase-alpha subunit, regulates Notch signaling via Rab1 and Rab11. ***PLoS Biology***, 12: e1001777. PMID: 24492843, PMCID: PMC3904817.

18. Wang S, Tan KL, Agosto MA, Xiong B, **Yamamoto S**, Sandoval H, Jaiswal M, Bayat V, Zhang K, Charng W-L, David G, Duraine L, Venkatachalam K, Wensel TG, Bellen HJ (2014) The retromer complex is required for rhodopsin recycling and its loss leads to photoreceptor degeneration. ***PLoS Biology***, 12:e1001847. PMID: 24781186, PMCID: PMC4004542.

19. Charng W-L, **Yamamoto S**, Bellen HJ (2014) Shared mechanisms between *Drosophila* peripheral nervous system development and human neurodegenerative diseases. ***Current Opinion in Neurobiology***, 27:158-164. PMID: 24762652, PMCID: PMC4122633.

20. **Yamamoto S***, Jaiswal M*, Charng WL, Gambin T, Karaca E, Mirzaa G, Wiszniewski W, Sandoval H, Haelterman NA, Xiong B, Zhang K, Bayat V, David G, Li T, Chen K, Gala U, Harel T, Pehlivan D, Penney S, Vissers LE, de Ligt J, Jhangiani SN, Xie Y, Tsang SH, Parman Y, Sivaci M, Battaloglu E, Muzny D, Wan YW, Liu Z, Lin-Moore AT, Clark RD, Curry CJ, Link N, Schulze KL, Boerwinkle E, Dobyns WB, Allikmets R, Gibbs RA, Chen R, Lupski JR, Wangler MF, Bellen HJ (2014) A *Drosophila* genetic resource of mutants to study mechanisms underlying human genetic diseases. ***Cell***, 159:200-214 **(*equal contribution)**. PMID: 25259927, PMCID: PMC4298142.

21. Haelterman NA, Jiang L, Li Y, Bayat V, Sandoval H, Ugur B, Tan KL, Zhang K, Bei D, Xiong B, Charng W-L, Busby T, Jawaid A, David G, Jaiswal M, Venken KJT, **Yamamoto S**, Chen R, Bellen HJ (2014) Large-scale identification of chemically induced mutations in *Drosophila* melanogaster. ***Genome Research***, 24:1707-1718. PMID: 25258387, PMCID: PMC4199363.

22. Sandoval H, Yao CK, Chen K, Jaiswal M, Donti T, Lin YQ, Bayat V, Xiong B, Zhang K, David G, Charng W-L, **Yamamoto S**, Duraine L, Graham BH, Bellen HJ (2014) Mitochondrial fusion but not fission regulates larval growth and synaptic development through steroid hormone production. ***eLife***, 3:e03558. PMID: 25313867, PMCID: PMC4215535.

23. Liu L, Zhang K, Sandoval H, **Yamamoto S**, Jaiswal M, Sanz E, Li Z, Hui J, Graham BH, Quintana A, Bellen HJ (2015) Glial Lipid Droplets and ROS Induced by Mitochondrial Defects Promote Neurodegeneration. ***Cell***, 160:177-190. PMID: 25594180, PMCID: PMC4377295.

24. Wangler MF*, **Yamamoto S***, Bellen HJ (2015) Fruit Flies in Biomedical Research. ***Genetics***, 199(3):639-53. **(*equal contribution)**. PMID: 25624315, PMCID: PMC4349060.

25. Tian X, Gala U, Zhang Y, Shang W, Nagarkar-Jaiswal S, di Ronza A, Jaiswal M, **Yamamoto S**, Sandoval H, Duraine L, Sardiello M, Sillitoe RV, Venkatachalam K, Fan H, Bellen HJ, Tong C (2015) A voltage gated calcium channel regulates lysosomal fusion with endosomes and autophagosomes and is required for neuronal homeostasis. ***PLoS Biology***, 6;13(3):e1002103. PMID: 25811491, PMCID: PMC4374850.

26. Jaiswal M, Haelterman NA, Sandoval H, Xiong B, Donti T, Kalsotra A, **Yamamoto S**, Cooper TA, Graham BH, Bellen HJ (2015) Impaired mitochondrial energy production causes light induced photoreceptor degeneration independent of oxidative stress. ***PLoS Biology***, 13(7):e1002197. PMID: 26176594, PMCID: PMC4503542.

27. Bellen HJ, **Yamamoto S** (2015) Morgan's legacy: fruit flies and the functional annotation of conserved genes. ***Cell***, 163:12-14. PMID: 26406362. PMCID: PMC4783153.

28. David-Morrison G, Xu Z, Rui YN, Charng WL, Jaiswal M, **Yamamoto S**, Xiong B, Zhang K, Sandoval H, Duraine L, Zhang S, Bellen HJ (2016) WAC regulates mTOR activity through the TTT-Pontin/Reptin complex. ***Developmental Cell***, 36:139-151. PMID: 26812014. PMCID: PMC4730548.

29. Li T, Giagtzoglou N, Fan J, Jia J, **Yamamoto S**, Charng W-L, Jaiswal M, Sandoval H, Bayat V, Xiong B, Zhang K, David G, Wei W, Lewis MT, Groves AK, Bellen HJ (2016) The Ubr3 E3 ubiquitin ligase modulates Costal-2 levels and is necessary for Hedgehog signaling. ***PLoS Genetics***, 12(5):e1006054. PMID: 27195754, PMCID: PMC4873228.

30. Chen K, Lin G, Haelterman NA, Duraine L, Li Z, Graham BH, Jaiswal M, **Yamamoto S**, Bellen HJ (2015) Loss of frataxin affects mitochondrial function, causes iron depositions that induce sphingolipid synthesis and PDK1 phosphorylation, and both pathways synergize to promote neurodegeneration. ***eLife***, 5:e16043. PMID: 27343351, PMCID: PMC4956409.

31. Jakobsdottir J*, van der Lee SJ,*, Bis JC*, Chouraki V*, Li-Kroeger D*, **Yamamoto S***, Grove ML, Naj A, Vronskaya M, Salazar JL, DeStefano AL, Brody JA, Smith AV, Amin N, Sims R, Ibrahim-Verbaas CA, Choi SH, Satizabal CL, Lopez OL, Beiser A, Ikram MA, Garcia ME, Hayward C, Varga TV, Ripatti S, Franks PW, Hallmans G, Rolandsson O, Jansson JH, Porteous DJ, Salomaa V, Eirkisdottir G, Rice KM, Bellen HJ, Levy D, Uitterlinden AG, Emilsson V, Rotter JI, Aspelund T, Cohorts for Heart and Aging Research in Genomic Epidemiology consortium, Alzheimer's Disease Genetic Consortium, Genetic and Environmental Risk in Alzheimer's Disease consortium, O'Donnell CJ, Fitzpatrick AL, Launer LJ, Hofman A, Wang LS, Williams J, Schellenberg GD, Boerwinkle E, Psaty BM, Seshadri S, Shulman JM, Gudnason V, van Duijn CM (2016) Rare loss-of-function variant in *TM2D3* is associated with late-onset Alzheimer's disease. ***PLoS Genetics,***12: e1006327 PMID: 27764101 PMCID: PMC5072721 **(*equal contribution)**.

32. Yoon WH, Sandoval H, Nagarkar-Jaiswal S, Jaiswal M, **Yamamoto S**, Haelterman NA, Putluri N, Putluri V, Sreekumar A, Tos T, Aksoy A, Donti T, Graham BH, Ohno M, Nishi E, Hunter J, Muzny DM, Carmichael J, Shen J, Arboleda VA, Nelson SF, Wangler MF, Karaca E, Lupski JR, Bellen HJ (2017) Loss of Nardilysin, a Mitochondrial Co-chaperone for α-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. ***Neuron***, 93:115-131. PMID: 28017472. PMCID: PMC5242142.

33. Chao H-T, Davids M, Burke E, Pappas JG, Rosenfeld JA, McCarty A, Davis T, Wolfe L, Toro C, Tifft C, Xia F, Johnson TK, Warr CG, Members of the UDN, **Yamamoto S**, Adams D, Markello TC, Gahl WA, Bellen HJ, Wangler MF, Malicdan MCV (2017) A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in *EBF3*. ***AJHG***, 100, 128-137. PMID: 28017372. PMICD: PMC5223093.

34. Wang J, Al-Ouran R, Hu Y, Kim S-Y, Wan Y-W, Wangler MF, **Yamamoto S**, Chao H-T, Comjean A, Mohr SE, Members of UDN, Perrimon N, Liu Z, Bellen HJ (2017) MARRVEL: Integration of human and model organism genetic resources to facilitate functional annotation of the human genome. ***AJHG***, 100:843-853. PMID: 28502612. PMCID: PMC5670038

35. Cook M, Cazin C, Amoyel M, **Yamamoto S**, Bach E, Nystul T (2017) Neutral Competition for *Drosophila* Follicle and Cyst Stem Cell Niches Requires Vesicle Trafficking Genes. ***Development***. 206:1417-1428. PMID: 28512187. PMCID: PMC5500140

36. Luo X, Rosenfeld JA, **Yamamoto S**, Harel T, Zuo Z, Hall M, Wierenga KJ, Pastore MT, Bartholomew D, Delgado MR, Rotenberg J, Lewis RA, Emrick L, Bacino CA, Eldomery MK, Coban Akdemir Z, Xia F, Yang Y, Lalani SR, Lotze T, Lupski JR, Lee B, Bellen HJ, Wangler MF; Members of the UDN (2017) Clinically severe *CACNA1A* alleles affect synaptic function and neurodegeneration differentially. ***PLoS Genetics.*** 13:e1006905. PMID: 28742085. PMCID: PMC5557584.

37. Wangler MF*, **Yamamoto S***, Chao H-T, Posey JE, Westerfield M, Postlethwait J, Members of the UDN, Hieter P, Boycott KM, Campeau PM, Bellen HJ (2017) Model organisms in undiagnosed rare diseases. ***Genetics.*** 207(1):9-27. PMID: 28874452. PMCID: PMC5586389 **(*equal contributions)**

38. Salazar JL, **Yamamoto S†** (2018) Integration of *Drosophila* and Human Genetics to understand Notch signaling related diseases. ***Adv. Exp. Med. Biol.*** 1066:141-185. PMID: 30030826, PMCID: PMC6233323. **(†corresponding author)**.

39. Landrock KK, Sullivan P, Martini-Stoica H, Goldstein DS, Graham BH, **Yamamoto S**, Bellen HJ, Gibbs RA, Chen R, D'Amelio M, Stoica G (2018) Pleiotropic Neuropathological and Biochemical Alterations Associated with *Myo5a* Mutation in a Rat Model. ***Brain Res.*** 1679: 155-170. PMID: 29217155, PMCID: PMC7696654.

40. Oláhová M, Yoon WH, Thompson K, Jangam S, Fernandez L, Davidson JM, Kyle JE, Grove ME, Fisk DG, Kohler JN, Holmes M, Dries AM, Huang Y, Zhao C, Contrepois K, Zappala Z, Frésard L, Waggott D, Zink EM, Kim Y-M, Heyman HM, Stratton KG, Webb-Robertson BJM, Undiagnosed Diseases Network, Snyder M, Merker JD, Montgomery SB, Fisher PG, Feichtinger RG, Mayr JA, Hall J, Barbosa IA, Simpson MA, Deshpande C, Waters KM, Koeller D, Metz TO, Morris AA, Schelley S, Cowan T, Friederich MW, McFarland R, Van Hove JLK, Enns GM, **Yamamoto S**, Ashley EA, Wangler MF, Taylor RW, Bellen HJ, Bernstein JA, Wheeler MT (2018) Biallelic Mutations in *ATP5F1D*, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. ***AJHG***, 102:494-504. PMID: 29478781, PMCID: PMC6117612

41. Lee PT, Zirin J, Kanca O, Lin WW, Schulze KL, Li-Kroeger D, Tao R, Devereaux C, Hu Y, Chung V, Fang Y, He Y, Pan H, Ge M, Zuo Z, Housden BE, Mohr SE, **Yamamoto S**, Levis RW, Spradling AC, Perrimon N, Bellen HJ (2018) A gene-specific T2A-GAL4 library for *Drosophila*. ***eLife***. 22(7): e35574. PMID: 29565247, PMCID: PMC5898912.

42. Liu N, Schoch K, Luo X, Pena L, Bhavana VH, Kukolich M, Stringer S, Powis Z, Radtke K, Mroske C, Deak K, McDonald MT, McConkie-Rosell A, Markert ML, Kranz P, Stong N, Need AC, Bick D, Amaral MD, Worthey EA, Levy S, UDN, Wangler MF, Bellen HJ, Shashi V**†**, **Yamamoto S†** (2018) Functional variants in *TBX2* are associated with a syndromic cardiovascular and skeletal developmental disorder. ***Hum. Mol Genet.*** 27(14):2454-2465. PMID: 29726930, PMCID: PMC6030957. **(†corresponding authors)** …This disease was later named "Vertebral anomalies and variable endocrine and T-cell dysfunction" by OMIM based on our study)

43. Marcogliese PC, Shashi V, Spillmann RC, Stong N, Rosenfeld JA, Koenig MK, Martínez-Agosto JA, Herzog M, Chen AH, Dickson PI, Lin HJ, Vera MU, Salamon N, Ortiz D, Infante E, Steyaert W, Dermaut B, Poppe B, Chung H-L, Zuo Z, Lee P-T, Kanca O, Xia F, Yang Y, Smith EC, Jasien J, Kansagra S, Spiridigliozzi G, El-Dairi M, Lark R, Riley K, Koeberl DD, Golden-Grant K, Program for Undiagnosed Diseases (UD-PrOZA), Undiagnosed Diseases Network, **Yamamoto S**, Wangler MF, Mirzaa G, Hemelsoet D, Lee B, Nelson SF, Goldstein DB, Bellen HJ, Pena LDM (2018) Loss-of-function in *IRF2BPL* is associated with neurological phenotypes. ***AJHG.*** 103(2):245-260. PMID: 30057031, PMICD: PMC6081494.

44. Albrecht NE, Alevy J, Jiang D, Burger CA, Liu BI, Li F, Wang J, Kim SY, Hsu CW, Kalaga S, Udensi U, Asomugha C, Bohat R, Gaspero A, Justice MJ, Westenskow PD, **Yamamoto S**, Seavitt JR, Beaudet AL, Dickinson ME, Samuel MA (2018) Rapid and Integrative Discovery of Retina Regulatory Molecules. ***Cell Reports.*** 24(9):2506-2519. PMID: 30157441, PMCID: PMC6170014.

45. Deal SL, **Yamamoto S†** (2018) Unweaving the role of nuclear Lamins in neural circuit integrity. ***Cell Stress***, 2 (9): 219-224. PMID: 31223139, PMCID: PMC6558928. **(†corresponding author)**.

46. Splinter K, Adams D, Bellen HJ, Bernstein JA, Cheatle-Jarvela AM, Eng C, Esteves C, Gahl W, Rizwan Hamid, Jacob H, Kikani B, Koeller D, Kohane I, Loscalzo J, Luo X, Members of the Undiagnosed Diseases Network, McCray A, Metz TO, Mulvihill JJ, Nelson SF, Palmer C, Phillips JA III, Pick L, Postlethwait J, Shashi V, Sweetser D, Tifft C, Walley N, Wangler MF, Westerfield M, Wheeler M, Wise A, Worthey E, **Yamamoto S**, Ashley EA, Undiagnosed Diseases Network (2018) Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. ***NEJM***. 379:2131-2139. PMID: 30304647, PMCID: PMC6481166.

47. Deal SL, **Yamamoto S†** (2018) Unraveling novel mechanisms of neurodegeneration through a large-scale forward genetic screen in *Drosophila*. ***Frontiers in Genetics***. 9:700. PMID: 30693015, PMCID: PMC6339878. **(†corresponding author)**.

48. Kanca O, Andrews JC, Lee PT, Patel C, Braddock SR, Slavotinek AM, Cohen JS, Gubbels CS, Aldinger KA, Williams J, Indaram M, Fatemi A, Yu TW, Agrawal PB, Vezina G, Simons C, Crawford J, Lau CC; Undiagnosed Diseases Network, Chung WK, Markello TC, Dobyns WB, Adams DR, Gahl WA, Wangler MF, **Yamamoto S**, Bellen HJ, Malicdan MCV (2019) De Novo Variants in *WDR37* are Associated with Epilepsy, Colobomas and Cerebellar hypoplasia. ***AJHG.*** 105(2):413-424. PMID: 31327508, PMCID: PMC6699142.

49. Wang J, Undiagnosed Diseases Network, Liu Z, Bellen HJ, **Yamamoto S†** (2019) Navigating MARRVEL, a web-based tool that integrates human genomics and model organism genetics information. ***JoVE.*** (150):10.3791/59542. PMID: 31475990, PMCID: PMC7401700. **(†corresponding author)**.

50. Harnish JM, Deal SL, Undiagnosed Diseases Network, Chao HT, Wangler MF, **Yamamoto S†** (2019) In vivo functional study of disease-associated rare human variants using *Drosophila*. ***JoVE.*** (150): 10.3791/59658. PMID: 31498321. PMCID: PMC7418855. **(†corresponding author)**.

51. Wang J, Mao D, Fazal F, Kim SY, **Yamamoto S**, Bellen H, Liu Z (2019) Using MARRVEL v1.2 for Bioinformatics Analysis of Human Genes and Variant Pathogenicity. ***Curr Protoc Bioinformatics***. 67(1):e85. PMID: 31524990, PMCID: PMC6750039.

52. Guo H, Bettella E, Marcogliese PC, Zhao R, Andrews JC, Nowakowski TJ, Gillentine MA, Hoekzema K, Wang T, Wu H, Jangam S, Liu C, Ni H, Willemsen MH, van Bon BW, Rinne T, Stevens SJC, Kleefstra T, Brunner HG, Ijntema H, Long M, Zhao W, Hu Z, Colson C, Nicolas R, Schwartz C, Romano C,

Castiglia L, Bottitta M, Dhar SU, Erwin DJ, Emrick L, Keren B, Afenjar A, Zhu B, Bai B, Stankiewicz P, Herman K, University of Washington Center for Mendelian Genomics, Mercimek-Andrews S, Juusola J, Wilfert AB, Jamra RA, Büttner B, Mefford HC, Muir AM, Scheffer IE, Regan BM, Malone S, Gecz J, Cobben J, Weiss MM, Waisfisz Q, Bijlsma EK, Hoffer MJV, Ruivenkamp CAL, Sartori S, Xia F, Rosenfeld JA, Bernier RA, Wangler MF, **Yamamoto S**, Xia K, Stegmann APA, Bellen HJ, Murgia A, Eichler EE (2019) Disruptive mutations in *TANC2* define a neurodevelopmental syndrome associated with psychiatric disorders. ***Nat Commun.*** 10(1):4679. PMID: 31616000, PMCID: PMC6794285.

53. Kanca O, Zirin J, Garcia-Marques J, Knight SM, Yang-Zhou D, Amador G, Chung H, Zuo Z, Ma L, He Y, Lin WW, Fang Y, Ge M, **Yamamoto S**, Schulze KL, Hu Y, Spradling AC, Mohr SE, Perrimon N, Bellen HJ (2019) An efficient CRISPR-based strategy to insert small and large fragments of DNA using short homology arms. ***eLife.*** e51539. PMID: 31674908, PMCID: PMC6855806.

54. Bellen HJ**†**, Wangler MF, **Yamamoto S†** (2019) The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases. ***Hum Mol Genet.*** ;28(R2):R207-R214. PMID: 31227826, PMCID: PMC6872428. **(†corresponding authors)**.

55. Graves HK, Jangam S, Tan KL, Pignata A, Seto ES, **Yamamoto S†**, Wangler MF**†** (2020) A genetic screen for genes that impact peroxisomes in *Drosophila* identifies candidate genes for human disease. ***G3 (Bethesda)***. 10: 69-77. PMID: 31767637, PMCID: PMC6945042. **(†corresponding authors).**

56. Das P, Salazar JL, Li-Kroeger D, **Yamamoto S**, Nakamura M, Sasamura T, Inaki M, Masuda W, Kitagawa M, Yamakawa T, Matsuno K (2020) Maternal *almondex*, a neurogenic gene, is required for proper subcellular Notch distribution in early *Drosophila* embryogenesis. ***Dev Growth Differ.*** 62:80-93. PMID: 31782145, PMCID: N/A.

57. **Yamamoto S†** (2020) Making sense out of missense mutations: Mechanistic dissection of Notch receptors through structure-function studies in *Drosophila*. ***Dev Growth Differ.*** 62:15-34. PMID: 31943162. PMID: 31782145, PMCID: PMC7401704. **(†corresponding author).**

58. Chung HL, Wangler MF, Marcogliese PC, Jo J, Ravenscroft TA, Sadeghzadeh S, Li-Kroeger D, Schmidt R, Pestronk A, Rosenfeld JA, Burrage L, Herndon MJ, Undiagnosed Diseases Network, Lee B, Moser A, Jones R, Watkins P, Yoo T, Mar S, Bucelli, Choi M, **Yamamoto S**, Lee HK, Chae JH, Vogel TP, Bellen HJ (2020) Loss- or Gain-of-Function Mutations in *ACOX1* Cause Axonal Loss via Different Mechanisms. ***Neuron.*** 106: 589-606. PMID: 32169171, PMCID: PMC7289150.

59. Chung HL, Mao X, Wang H, Park YJ, Marcogliese PC, Rosenfeld JA, Burrage LC, Liu P, Murdock DR, **Yamamoto S**, Wangler MF; Undiagnosed Diseases Network, Chao HT, Long H, Feng L, Bacino CA, Bellen HJ, Xiao B (2020) De Novo Variants in *CDK19* Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. ***AJHG.*** 106: 717-725. PMID: 32330417, PMCID: PMC7212481.

60. Dutta D, Briere LC, Kanca O, Marcogliese PC, Walker MA, High FA, Vanderver A, Vögtle FN, Krier J, Carmichael N, Callahan C, Taft RJ, Simons C, Guy Helman G, The Undiagnosed Diseases Network, Wangler MF, **Yamamoto S**, Sweetser DA, Bellen HJ (2020) De novo mutations in *TOMM70*, a receptor of the mitochondrial import translocase, cause neurological impairment. ***HMG.*** 29: 1568-1579. PMID: 32356556. PMCID: PMC7268787.

61. Salazar JL, Yang SA, **Yamamoto S†** (2020) Post-Developmental Roles of Notch Signaling in the Nervous

System. ***Biomolecules.*** 10: 985. PMID: 32630239. PMCID: PMC7408554. **(†corresponding author).**

62. Barish S, Barakat TS, Michel BC, Mashtalir N, Phillips JB, Valencia AM, Ugur B, Wegner J, Scott TM, Bostwick B; Undiagnosed Diseases Network, Murdock DR, Dai H, Perenthaler E, Nikoncuk A, van Slegtenhorst M, Brooks AS, Keren B, Nava C, Mignot C, Douglas J, Rodan L, Nowak C, Ellard S, Stals K, Lynch SA, Faoucher M, Lesca G, Edery P, Engleman KL, Zhou D, Thiffault I, Herriges J, Gass J, Louie RJ, Stolerman E, Washington C, Vetrini F, Otsubo A, Pratt VM, Conboy E, Treat K, Shannon N, Camacho J, Wakeling E, Yuan B, Chen CA, Rosenfeld JA, Westerfield M, Wangler M, **Yamamoto S**, Kadoch C, Scott DA, Bellen HJ (2020). *BICRA*, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. ***AJHG.*** 107: 1096-1112. PMID: 33232675. PMCID: PMC7820627.

63. Harnish MJ, Link N, **Yamamoto S†** (2021) *Drosophila* as a model for infectious diseases. ***International Journal of Molecular Sciences.*** 22: 2724. PMID: 33800390. PMCID. PMC7962867. **(†corresponding author)**.

64. Baldridge D, Wangler MF, Bowman AN, **Yamamoto S**, UDN, Schedl T, Pak SC, Postlethwait JH, Solnica-Krezel L, Bellen HJ, Westerfield M (2021) Model Organisms Contribute to Diagnosis and Discovery in the Undiagnosed Diseases Network: the Current State and a Future Vision. ***Orphanet Journal of Rare Diseases.*** 16(1): 206. PMID: 33962631. PMCID: PMC8103593.

65. Luo X, Schoch K, Jangam SV, Bhavana VH, Graves HK, Kansagra S, Jasien J, Stong N, Keren B, Mignot C, Ravelli C, UDN, Bellen HJ, Wangler MF, Shashi V†, **Yamamoto S†** (2021) Rare deleterious *de novo* missense variants in *RNF2*/*RING2* are associated with a neurodevelopmental disorder with unique clinical features. ***Hum Mol Genet.*** 30(14):1283-1292. PMID: 33864376. PMCID: PMC8255132. **(†corresponding authors)**. (Cover of Issue)…This disease was named "Luo-Schoch-Yamamoto Syndrome" by OMIM based on our study)

66. Ravenscroft TA, Phillips JB, Fieg E, Bajikar SS, Peirce J, Wegner J, Luna AA, Fox EJ, Yan YL, Rosenfeld JA, Zirin J, Kanca O, UDN, Benke PJ, Cameron ES, Strehlow V, Platzer K, Osmond M, Licata T, Rojas S, Dyment D, Chong JSC, Lincoln S, Stoler JM, Postlethwait J, Wangler MF, **Yamamoto S**, Krier J, Westerfield M, Bellen HJ (2021). Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of *GDF11.* ***Genetics in Medicine.*** 23(10)1889-. PMID: 34113007. PMCID: PMC8487929.

67. Goodman LD, Cope H, Nil Z, Ravenscroft TA, Charng WL, Lu S, Tien AC, Pfundt R, Koolen DA, Haaxma CA, Kanca O, Moulton MJ, Pfundt R, Veenstra-Knol HE, Wassink-Ruiter JSK, Wevers MR, Jones M, Walsh LE, Klee VH, Theunis M, Legius E, Steel D, Barwick KES, Kurian MA, Mohammad SS, Dale RC, Terhal PA, van Binsbergen E, Kirmse B, Robinette B, Cogné B, Isidor B, Grebe TA, Kulch P, Hainline BE, Sapp K, Morava E, Klee EW, Macke EL, Trapane P, Spencer C, Si Y, Begtrup A, Moulton MJ, Dutta D, Kanca O, Undiagnosed Diseases Network; Wangler MF, **Yamamoto S**, Bellen HJ, Tan QKG (2021) *TNPO2* variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in *Drosophila*. ***AJHG***. 108(9):1669-1691. PMID: 34314705. PMCID: PMC8456166.

68. Manivannan SN, Roovers J, Smal N, Mefford H, Turkdogan D, Roelens F, Kanca O, Chung HL, MAE working group of EuroEPINOMICS RES Consortium, de Jonghe P, **Yamamoto S**, Weckhuysen S, Bellen HJ (2022) De novo *FZR1* loss-of-function variants cause developmental delay and epileptic

encephalopathies. ***Brain***. 145(5):1684-1697. PMID: 34788397. PMCID: PMC9166542.

69. Salazar JL, Yang SA, Lin YQ, Li-Kroeger D, Marcogliese PM, Deal SL, Neely GG, **Yamamoto S†** (2021) *TM2D* genes regulate Notch signaling and neuronal function in *Drosophila.* ***PLoS Genetics***. 17(12):e1009962. PMID: 34905536. PMCID: PMC8714088. **(†corresponding author)**.

70. Andrews JC, Wangler MF, **Yamamoto S†**, Posey JE† (2022) Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders. ***Encyclopedia of Behavioral Neuroscience***, 2nd edition (Academic Press). ISBN: 9780128196410 (Book Chapter). https://doi.org/10.1016/B978-0-12-819641-0.00145-6. **(†corresponding authors)**.

71. Marcogliese PC, Dutta D, Ray SS, Dang NDP, Zuo Z, Wang Y, Lu D, Fazal F, Ravenscroft TA, Chung H, Kanca O, Wan JJ, Douine ED, Undiagnosed Diseases Network, Pena LDM, **Yamamoto S,** Nelson SF, Might M, Meyer KC, Yeo NC, Bellen HJ (2022) Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling. ***Science Advances***. 8(3):eabl5613. PMID: 35044823. PMCID: PMC8769555.

72. Harnish JM, Li L, Rogic S, Poirier-Morency G, Kim S-Y, Undiagnosed Diseases Network, Boycott KM, Wangler MF, Bellen HJ, Hieter P, Pavlidis P, Liu Z, **Yamamoto S†** (2022) ModelMatcher: A scientist-centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research. ***Human Mutation***. 43(6):743-759. PMID: 35224820. PMCID: PMC9133126. **(†corresponding author)**.

73. Li H, Janssens J, De Waegeneer M, Kolluru SS, Davie K, Gardeux V, Saelens W, David FPA, Brbić M, Spanier K, Leskovec J, McLaughlin CN, Xie Q, Jones RC, Brueckner K, Shim J, Tattikota SG, Schnorrer F, Rust K, Nystul TG, Carvalho-Santos Z, Ribeiro C, Pal S, Mahadevaraju S, Przytycka TM, Allen AM, Goodwin SF, Berry CW, Fuller MT, White-Cooper H, Matunis EL, DiNardo S, Galenza A, O'Brien LE, Dow JAT; FCA Consortium§, Jasper H, Oliver B, Perrimon N, Deplancke B, Quake SR, Luo L, Aerts S, Agarwal D, Ahmed-Braimah Y, Arbeitman M, Ariss MM, Augsburger J, Ayush K, Baker CC, Banisch T, Birker K, Bodmer R, Bolival B, Brantley SE, Brill JA, Brown NC, Buehner NA, Cai XT, Cardoso-Figueiredo R, Casares F, Chang A, Clandinin TR, Crasta S, Desplan C, Detweiler AM, Dhakan DB, Donà E, Engert S, Floc'hlay S, George N, González-Segarra AJ, Groves AK, Gumbin S, Guo Y, Harris DE, Heifetz Y, Holtz SL, Horns F, Hudry B, Hung RJ, Jan YN, Jaszczak JS, Jefferis GSXE, Karkanias J, Karr TL, Katheder NS, Kezos J, Kim AA, Kim SK, Kockel L, Konstantinides N, Kornberg TB, Krause HM, Labott AT, Laturney M, Lehmann R, Leinwand S, Li J, Li JSS, Li K, Li K, Li L, Li T, Litovchenko M, Liu HH, Liu Y, Lu TC, Manning J, Mase A, Matera-Vatnick M, Matias NR, McDonough-Goldstein CE, McGeever A, McLachlan AD, Moreno-Roman P, Neff N, Neville M, Ngo S, Nielsen T, O'Brien CE, Osumi-Sutherland D, Özel MN, Papatheodorou I, Petkovic M, Pilgrim C, Pisco AO, Reisenman C, Sanders EN, Dos Santos G, Scott K, Sherlekar A, Shiu P, Sims D, Sit RV, Slaidina M, Smith HE, Sterne G, Su YH, Sutton D, Tamayo M, Tan M, Tastekin I, Treiber C, Vacek D, Vogler G, Waddell S, Wang W, Wilson RI, Wolfner MF, Wong YE, Xie A, Xu J, **Yamamoto S**, Yan J, Yao Z, Yoda K, Zhu R, Zinzen RP (2022) Fly Cell Atlas: A single-nucleus transcriptomic atlas of the adult fruit fly. ***Science***. 375(6584):eabk2432. PMID: 35239393. PMCID: PMC8944923.

74. Marcogliese PC, Deal SL, Andrews J, Harnish JM, Bhavana VH, Graves HK, Jangam S, Luo X, Liu N, Bei D, Chao YH, Hull B, Lee PT, Pan H, Bhadane P, Huang MC, Longley CM, Chao HT, Chung HL, Haelterman NA, Kanca O, Manivannan SN, Rossetti LZ, German RJ, Gerard A, Schwaibold EMC, Fehr S, Guerrini R, Vetro A, England E, Murali CN, Barakat TS, van Dooren MF, Wilke M, van Slegtenhorst

M, Lesca G, Sabatier I, Chatron N, Brownstein CA, Madden JA, Agrawal PB, Keren B, Courtin T, Perrin L, Brugger M, Roser T, Leiz S, Mau-Them FT, Delanne J, Sukarova-Angelovska E, Trajkova S, Rosenhahn E, Strehlow V, Platzer K, Keller R, Pavinato L, Brusco A, Rosenfeld JA, Marom R, Wangler MF†, **Yamamoto S†** (2022) *Drosophila* functional screening of *de novo* variants in autism spectrum disorders uncovers deleterious variants and facilitates discovery of rare neurodevelopmental diseases. ***Cell Rep.*** 38(11):110517. PMID: 35294868. PMCID: PMC8983390. **(†corresponding authors).**

75. Barish S, Senturk M, Schoch K, Minogue AL, Lopergolo D, Fallerini C, Harland J, Seemann JH, Stong N, Kranz PG, Kansagra S, Mikati MA, Jasien J, El-Dairi M; Undiagnosed Diseases Network, Galluzzi P, Ariani F, Renieri A, Mari F, Wangler MF, Arur S, Jiang YH, **Yamamoto S**, Shashi V, Bellen HJ. (2022) The microRNA processor DROSHA is a candidate gene for a severe progressive neurological disorder. ***Hum Mol Genet.*** 31(17):2934-2950. PMID: 35405010. PMCID: PMC9433733.

76. Yamaguchi M, **Yamamoto S** (2022) Role of *Drosophila* in Human Disease Research 2.0. ***Int J Mol Sci.*** 23(8):4203. PMID: 35457020. PMCID: PMC9027098.

77. Yang SA, Salazar JL, Li-Kroeger D†, **Yamamoto S†** (2022) Functional studies of genetic variants associated with human diseases in Notch signaling-related genes using *Drosophila*. ***Methods in Molecular Biology.*** 2472:235-276. PMID: 35674905 PMCID: PMC9396741. **(†corresponding authors)**.

78. Huang Y, Lemire G, Briere LC, Liu F, Wessels MW, Wang X, Osmond M, Kanca O, Lu S, High FA, Walker MA, Rodan LH; Undiagnosed Diseases Network; Care4Rare Canada Consortium, Wangler MF, **Yamamoto S**, Kernohan KD, Sweetser DA, Boycott KM, Bellen HJ (2022) The recurrent de novo c.2011C>T missense variant in MTSS2 causes syndromic intellectual disability. ***Am J Hum Genet.*** 109(10):1923-1931. PMID: 36067766. PMCID: PMC9606386.

79. Snijders Blok L, Verseput J, Rots D, Venselaar H, Innes AM, Stumpel C, Õunap K, Reinson K, Seaby EG, McKee S, Burton B, Kim K, van Hagen JM, Waisfisz Q, Joset P, Steindl K, Rauch A, Li D, Zackai EH, Sheppard SE, Keena B, Hakonarson H, Roos A, Kohlschmidt N, Cereda A, Iascone M, Rebessi E, Kernohan KD, Campeau PM, Millan F, Taylor JA, Lochmüller H, Higgs MR, Goula A, Bernhard B, Velasco DJ, Schmanski AA, Stark Z, Gallacher L, Pais L, Marcogliese PC, **Yamamoto S**, Raun N, Jakub TE, Kramer JM, den Hoed J, Fisher SE, Brunner HG, Kleefstra T (2023) A clustering of heterozygous missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder. ***HGG Adv.*** 4(1):100157. PMID: 36408368. PMCID: PMC9673101.

80. Keramidioti A, Golegou E, Psarra E, Paschalidis N, Kalodimou K, **Yamamoto S**, Delidakis C, Vakaloglou KM, Zervas CG (2022) Epithelial morphogenesis in the *Drosophila* egg chamber requires Parvin and ILK. ***Front Cell Dev Biol.*** 10:951082. PMID: 36531940. PMCID: N/A.

81. Nurmahdi H, Hasegawa M, Mujizah EY, Sasamura T, Inaki M, **Yamamoto S**, Yamakawa T, Matsuno K (2022) Notch Missense Mutations in *Drosophila* Reveal Functions of Specific EGF-like Repeats in Notch Folding, Trafficking, and Signaling by Folding, Trafficking, and Signaling. ***Biomolecules*** 12(12):1752. PMID: 36551180. PMCID: N/A.

82. Andrews JC, Mok JW, Kanca O, Jangam S, Tifft C, Macnamara EF, Russell BE, Wang LK; Undiagnosed Diseases Network; Nelson SF, Bellen HJ, **Yamamoto S**, Malicdan MCV, Wangler MF (2023) De Novo Variants in MRTFB have gain of function activity in *Drosophila* and are associated with a novel neurodevelopmental phenotype with dysmorphic features. ***Genet Med.*** 25(6):100833. PMID: 37013900.

PMCID: In Progress.

83. Tepe B, Macke EL, Niceta M, Hubsham MW, Kanca O, Schultz-Rogers L, Zarate YA, Schaefer B, Granadillo De Luque JL, Wegner DJ, Cogne B, Gilbert-Dussardier B, Guillou XL, Wagner EJ, Pais LS, Neil JE, Mochida GH, Walsh CA, Magal N, Drasinover V, Shohat M, Schwab T, Schmitz C, Clark K, Fine A, Lanpher B, Gavrilova R, Blanc P, Burglen L, Afenjar A, Steel D, Kurian MA, Prabhakar P. Gößwein S, Donato ND, Bertini ES, Undiagnosed Diseases Network(UDN), Wangler MF, **Yamamoto S**, Tartaglia M, Klee EW, Bellen HJ (2023) Biallelic variants in INTS11 are associated with a novel complex neurological disorder. ***Am J Hum Genet***. 110(5):774-789. PMID: 37054711. PMCID: PMC10183469.

84. Jangam SV, Briere LC, Jay KL, Andrews JC, Walker MA, Rodan LH, High FA; Undiagnosed Diseases Network; **Yamamoto S**, Sweetser DA, Wangler MF (2023) A de novo missense variant in EZH1 associated with developmental delay exhibits functional deficits in *Drosophila melanogaster*. ***Genetics*.** Online ahead of print. doi:10.1093/genetics/iyad110. PMID: 37314226. PMCID: In Progress.

85. Guichard A, Lu S, Kanca O, Bressan D, Huang Y, Ma M, Juste SS, Andrews JC, Jay KL, Sneider M, Schwartz R, Huang M-C, Bei D, Pan H, Ma L, Lin W-W, Auradkar A, Bhagwat P, Park S, Wan KH, Ohsako T, Takano-Shimizu T, Celniker SE, Wangler MF, **Yamamoto S†**, Bellen HJ†, Bier E† (2023). A comprehensive *Drosophila* genetic resource to study SARS-CoV-2 virus-host interactions in vivo. ***Cell Rep*.** 42(8):112842. PMID: 37480566. PMCID: In Progress. **(†corresponding authors)**.

86. **Yamamoto S***, Kanca O*, Wangler MF, Bellen HJ (2024). Integrating non-mammalian model organisms in the diagnosis of rare genetic diseases in humans. ***Nat Rev Genet*.** 25(1):46-60. doi: 10.1038/s41576-023-00633-6. PMID: 37491400. PMCID: N/A. **(*equal contributions)**.

87. Nil Z, Deshwar AR, Huang Y, Barish S, Zhang X, Choufani S, Le Quesne Stabej P, Hayes I, Yap P, Haldeman-Englert C, Wilson C, Prescott T, Tveten K, Vøllo A, Haynes D, Wheeler PG, Zon J, Cytrynbaum C, Jobling R, Blyth M, Banka S, Afenjar A, Mignot C, Robin-Renaldo F, Keren B, Kanca O, Mao X, Wegner DJ, Sisco K, Shinawi M; Undiagnosed Disease Network; Wangler MF, Weksberg R, **Yamamoto S**, Costain G, Bellen HJ (2023). Rare de novo gain-of-function missense variants in DOT1L are associated with developmental delay and congenital anomalies. ***Am J Hum Genet*.** 110(11):1919-1937. PMID: 37827158. PMCID: PMC10645550.

88. Pan X, Alvarez AN, Ma M, Lu S, Crawford MW, Briere LC, Kanca O, **Yamamoto S**, Sweetser DA, Wilson JL, Napier RJ, Pruneda JN, Bellen HJ (2023). Allelic strengths of encephalopathy-associated UBA5 variants correlate between in vivo and in vitro assays. ***Elife***. 12:RP89891. PMID: 38079206. PMCID: PMC10712953.

89. Link N**†**, Harnish MJ, Hull B, Gibson, S, Dietze M, Mgbike UE, Medina-Balcazar S, Shah PS, **Yamamoto†** (2024) A Zika virus protein expression screen in *Drosophila* to investigate targeted host pathways during development. ***Dis Model Mech*. (†corresponding authors)**. 17(2):dmm050297. PMID: 38214058. PMCID: PMC10924231 (Cover of Issue).

90. Dutta D, Kanca O, Shridharan RV, Marcogliese PC, Steger B, Morimoto M, Frost FG, Macnamara E; Undiagnosed Diseases Network; Wangler MF, **Yamamoto S**, Jenny A, Adams D, Malicdan MC, Bellen HJ (2024). Loss of the endoplasmic reticulum protein Tmem208 affects cell polarity, development, and viability. ***Proc Natl Acad Sci U S A***. 121(9):e2322582121. PMID: 38381787. PMC10907268.

91. Sutton DC, Andrews JC, Dolezal DM, Park YJ, Li H, Eberl DF, **Yamamoto S†**, Groves AK† (2024). Comparative exploration of mammalian deafness gene homologues in the Drosophila auditory organ shows genetic correlation between insect and vertebrate hearing. ***PLoS One.*** 19(2):e0297846. PMID: 38412189. PMCID: PMC10898740. **(†corresponding authors)**.

92. Pan X, Tao AM, Lu S, Ma M, Hannan SB, Slaugh R, Drewes Williams S, O'Grady L, Kanca O, Person R, Carter MT, Platzer K, Schnabel F, Abou Jamra R, Roberts AE, Newburger JW, Revah-Politi A, Granadillo JL, Stegmann APA, Sinnema M, Accogli A, Salpietro V, Capra V, Ghaloul-Gonzalez L, Brueckner M, Simon MEH, Sweetser DA, Glinton KE, Kirk SE; Baylor College of Medicine Center for Precision Medicine Models; Wangler MF, **Yamamoto S**, Chung WK, Bellen HJ (2024). De novo variants in FRYL are associated with developmental delay, intellectual disability, and dysmorphic features. ***Am J Hum Genet***. 111(4):742-760. PMID: 38479391. PMCID: PMC11023917.

93. Huang Y, Jay KL, Yen-Wen Huang A, Wan J, Jangam SV, Chorin O, Rothschild A, Barel O, Mariani M, Iascone M, Xue H; Undiagnosed Diseases Network; Huang J, Mignot C, Keren B, Saillour V, Mah-Som AY, Sacharow S, Rajabi F, Costin C, **Yamamoto S**, Kanca O, Bellen HJ, Rosenfeld JA, Palmer CGS, Nelson SF, Wangler MF, Martinez-Agosto JA (2024) Loss-of-function in RBBP5 results in a syndromic neurodevelopmental disorder associated with microcephaly. ***Genet Med***. 2024 Nov;26(11):101218. doi: 10.1016/j.gim.2024.101218. Epub 2024 Jul 19. PMID: 39036895. PMCID: PMC11648989.

94. Moulton MJ, Atala K, Zheng Y, Dutta D, Grange DK, Lin WW, Wegner DJ, Wambach JA, Duker AL, Bober MB, Kratz L, Wise CA, Oxendine I, Khanshour A; Undiagnosed Diseases Network; Wangler MF, **Yamamoto S**, Cole FS, Rios J, Bellen HJ (2024) Dominant missense variants in SREBF2 are associated with complex dermatological, neurological, and skeletal abnormalities. ***Genet Med***. 26(9):101174. PMID: 38847193. PMCID: PMC12358889.

95. Mao D, Liu C, Wang L, Ai-Ouran R, Deisseroth C, Pasupuleti S, Kim SY, Li L, Rosenfeld JA, Meng L, Burrage LC, Wangler MF, **Yamamoto S**; Undiagnosed Diseases Network; Santana M, Perez V, Shukla P, Eng CM, Lee B, Yuan B, Xia F, Bellen HJ, Liu P, Liu Z (2024) AI-MARRVEL - A Knowledge-Driven AI System for Diagnosing Mendelian Disorders. ***NEJM AI***. 1(5):10.1056/aioa2300009. PMID: 38962029. PMCID: PMC11221788.

96. den Hoed J, Hashimoto H, Khan M, Semmekrot F, Bosanko KA, Abe-Hatano C, Nakagawa E, Venselaar H, Quercia N, Chad L, Kurosaka H, Rondeau S, Fisher SE, **Yamamoto S†**, Zarate YA† (2024). Pathogenic SATB2 missense variants affecting p.Gly392 have variable functional implications and result in diverse clinical phenotypes. ***J Med Genet.*** 61(11):1062-1067. PMID: 39327041. PMCID: N/A. **(†corresponding authors)**.

97. Delgado-Vega AM, Cederroth H, Taylan F, Ekholm K, Ek M, Thonberg H, Jemt A, Nilsson D, Eisfeldt J, Bilgrav Saether K, Höijer I, Akgun-Dogan O, Asano Y, Barakat TS, Batkovskyte D, Baynam G, Bodamer O, Chetruengchai W, Corcoran P, Couse M, Danis D, Demidov G, Dohi E, Erhardsson M, Fernandez-Luna L, Fujiwara T, Garg N, Giugliani R, Gonzaga-Jauregui C, Grigelioniene G, Groza T, Gunnarsson C, Hammarsjö A, Hammond CK, Hatirnaz Ng Ö, Hesketh S, Hettiarachchi D, Johansson Soller M, Kirmani UA, Kjellberg M, Kvarnung M, Kvlividze O, Lagerstedt-Robinson K, Lasko P, Lassmann T, Lau LYS, Laurie S, Lim WK, Liu Z, Lysenkova Wiklander M, Makay P, Maiga AB, Maya-González C, Meyn MS, Neethiraj R, Nigro V, Nordgren F, Nordlund J, Orrsjö S, Ottosson J, Ozbek U, Özdemir Ö, Partin C, Pearce DA, Peck R, Pedersen A, Pettersson M, Pongpanich M, Posada de la Paz

M, Ramani A, Romero JA, Romero VI, Rosenquist R, Saw AM, Spencer M, Stattin EL, Srichomthong C, Tapia-Paez I, Taruscio D, Taylor JP, Tkemaladze T, Tully I, Tümer Z, van Zelst-Stams WAG, Verloes A, Västerviga E, Wang S, Yang R, **Yamamoto S**, Yépez VA, Zhang Q, Shotelersuk V, Wiafe SA, Alanay Y, Botto LD, Kirmani S, Lumaka A, Palmer EE, Puri RD, Wirta V, Lindstrand A, Buske OJ, Cederroth M, Nordgren A (2024). Pushing the boundaries of rare disease diagnostics with the help of the first Undiagnosed Hackathon. ***Nat Genet.*** 56(11):2287-2294. PMID: 39433890. PMCID: PMC12198426.

98. Booth KTA, Jangam SV, Chui MMC, Treat K, Graziani L, Soldano A, Ruan Y, Wan-Hei Hui J, White K, Christensen CK, Lynnes T, **Yamamoto S**, Kanca O, Tsang MHY, Lynch SA, Mullegama SV, Batista J, Iancu D, Joss SK, Wong SYY, Mak CCY, Kwong AKY, Bellen HJ, Conboy E, Sanges R, Leung AY, Wangler MF, Chung BHY, Vetrini F (2025) De novo and inherited variants in DDX39B cause a novel neurodevelopmental syndrome. ***Brain***. 148(8):2658-2670. PMID: 39918047. PMCID: PMC12316012.

99. Bereshneh AH, Andrews JC, Eberl DF, Bademci G, Borja NA, Bivona S, Undiagnosed Diseases Network, Baylor College of Medicine Center for Precision Medicine Models, Chung WK, **Yamamoto S**, Wangler MF, McKee S, Tekin M, Bellen HJ, Kanca O (2025) De novo variants in CDKL1 and CDKL2 are associated with neurodevelopmental symptoms. ***Am J Hum Genet***. 112(4):846-862. PMID: 40088891, PMCID: PMC12081231.

100. Mok JW, Mackay L, Blazo M, Mizerik E, Gecz J, Carroll R, Nizon M, Rondeau S, Joubert M, Cuinat S, Deb W, Sirias FV, Weisz-Hubshman M, Ketkar S, Polak U, Tran AA, Kearney D, Neil HA, Kanca O, Wangler MF, Bellen HJ, Lee BH, Baylor College of Medicine Center for Precision Medicine Models, **Yamamoto S†**, Keren Macholt (2025) C-terminal frameshift variants in GPKOW are associated with a multisystemic X-linked disorder. ***Genet Med.*** 27(7):101429. PMID: 40221893, PMCID: PMC12435177 **(†corresponding authors)**.

101. Oka M, Nakajima S, Suzuki E, **Yamamoto, S**, K Ando (2025) Glucose uptake in pigment glia suppresses tau-induced inflammation and photoreceptor degeneration in Drosophila. ***Dis Model Mech.*** 18(4):dmm052057. PMID: 40151148, PMCID: PMC12067088.

102. Yasuda R, Hashimoto H, Oka M, Mok JW, Waqar M, Dauwalder B, Kanca O, Mizuno T, **Yamamoto S†** (2025) Functional analysis of pathogenic variants in LAMB1-related leukoencephalopathy reveals genotype–phenotype correlations and suggests its role in glial cells. ***Hum Mol Genet.*** 34(11):990-999. PMID: 40237576. PMCID: PMC12085778 **(†corresponding author)**.

103. Park YJ, Lu TC, Jackson T, Goodman LD, Ran L, Chen J, Liang CY, Harrison E, Ko C, Chen X, Wang B, Hsu AL, Ochoa E, Bieniek KF, **Yamamoto S**, Zhu Y, Zheng H, Qi Y, Bellen HJ, Li H (2025). Distinct systemic impacts of Aβ42 and Tau revealed by whole-organism snRNA-seq. ***Neuron.*** 113(13):2065-2082.e8. PMID: 40381615. PMCID: PMC12245608.

104. Mok J-W, Gibson SB, Dostalik SA, **Yamamoto S†** (2025). Functional assays in Drosophila facilitate classification of variants of uncertain significance associated with rare diseases. ***Genome Res.*** 35(7):1473-1484. PMID: 40467338, PMCID: PMC12212082. **(†corresponding author)**.

105. Ma M, Zheng Y, Deng M, Lu S, Pan X, Luo X, Etoundi M, Li-Kroeger D, Worley KC, Burrage LC, Blieden LS, Allworth A, Chen WL, Merla G, Mandriani B, Otten CE, Blanc P, Rosenfeld JA, Dutta D, **Yamamoto S**, Wangler MF, Glass IA, Chen J, Blue E, Prontera P, Rosain J, Marlin S, Lalani SR, Bellen HJ; Undiagnosed Diseases Network (2025) Heterozygous variants in PLCG1 affect hearing, vision,

cardiac, and immune function. ***Elife***. 2025 13:RP95887. PMID: 40862571. PMCID: PMC12387771.

106. Jay KL, Gogate N, Hall PI, Ezell KM, Andrews JC, Jangam SV, Pan H, Pham K, German R, Gomez V, Jellinek-Russo E, Storch EA; Brain Gene Registry Consortium, Undiagnosed Diseases Network; **Yamamoto S**, Kanca O, Bellen HJ, Dierick HA, Cogan JD, Phillips JA, Hamid R, Cassini T, Rives L, Pruthi S, Chen HC, Posey JE, Wangler MF (2026). Resolving SLC6A1 variable expressivity with deep clinical phenotyping and Drosophila models. ***HGG Advances.*** 7(1):100541. PMID: 41174879, PMCID: PMC12681531.

107. Deal SL, Bei D, Gibson SB, Delgado-Seo H, Fujita Y, Wilwayco K, Seto ES, Sehgal A, **Yamamoto S†** (2026). RNAi-based screen for cuticle pigmentation in *Drosophila melanogaster* reveals novel regulators of brain dopamine and sleep. ***iScience.*** *29(1):114388*. PMID: 41561374 PMCID: PMC12814433. **(†corresponding author)**.

108. Tumiene B, Adams DR, Allaway R, Barrero MJ, Chan CH, Faundes V, Fear VS, Glezer P, Fuchs C, Groza T, Houwink EJF, Jamuar SS, Letinturier MCV, Lomash RM, Puri RD, Reichardt JKV, Mehrian-Shai R, van der Westhuizen FH, Varshney GK, **Yamamoto S**, Baynam G (2026) Translating multi-omics into healthcare: requisites for scalable and equitable implementation. ***Hum Genomics.*** 20(1):71. doi: 10.1186/s40246-025-00902-x. PMID: 41882744. PMCID: PMC13085408.

109. Bereshneh AH, Wilson KA, Pan X, Hannan SB, Cooper MA, Diaz J, Leon E, Moses TM, Azamian MS, Scott DA, Billie Au PY, Appendino JP, Scheffer IE, Kaspi A, Bahlo M, Hildebrand MS, Morgan AT, Ekure E; Baylor College of Medicine Center for Precision Medicine Models; Shulman JM, Hildebrandt F, Posey JE, Kruszka P, Vilain E, **Yamamoto S**, Kanca O, Berger S, Bellen HJ (2026). Rare heterozygous de novo variants in RAPGEF2 are associated with a neurodevelopmental disorder. ***Genet Med***. 28(4):101685. PMID: 41556274, PMCID: PMC13060567

110. Aceves-Ewing NM, Lanza DG, Marcogliese PC, Lu D, Hsu CW, Hashimoto H, Gonzalez M, Christiansen AE, Rasmussen TL, Ho AJ, Gaspero A, Sha C, Dickinson ME, Yuan B, Shayota BJ, Pachter S, Hu X, Day-Salvatore DL, Mackay L, Kanca O, Wangler MF, Potocki L, Rosenfeld JA, Lewis RA, Chao HT, Lee B, Blieden L, Wasserman BN, Bax DA, Ragge NK, Lee S; Undiagnosed Diseases Network; Baylor College of Medicine Center for Precision Medicine Models; **Yamamoto S**, Bellen HJ, Burrage LC, Heaney JD (2026) Uncovering phenotypic expansion in AXIN2-related disorders through precision animal modeling. ***Genet Med.*** 28(9):102648. PMID: 42429102. PMCID: In Progress (PMC11643287 for *bioRxiv* preprint).

111. Hu Y, Comjean A, Gao C, Veal A, Yamamoto S, Mohr SE, Perrimon N (2026) DIOPT: the DRSC Integrative Ortholog Prediction Tool, 2026 update. ***Genetics***. (Online ahead of print) doi: 10.1093/genetics/iyag194. PMID: 42504133. PMCID: In Progress (PMC13104911 for *bioRxiv* preprint).

112. Gibson SB, Deal SL, Park YJ, Sun B, Qi Y, Mok JW, Chung HL, Li H, **Yamamoto S†** (2026) Hippo signaling regulates cuticle pigmentation and dopamine metabolism in *Drosophila*. ***PLOS Genetics.*** 22(8): e1012260. PMID: 42627852. PMCID: PMCID: PMC13524344. **(†corresponding author)**.

113. Hwang S, Brown RM, Baldridge D, Baldwin EE, Beggs AH, Bernstein JA, Blue E, Borja NA, Botto LD, Briere LC, Cassini T, Esteves C, Fieg EL, Jarvik GP, Kobren SN, Levanto MP, Martínez-Agosto JA, Marwaha S, Nelson SF, Rosenfeld JA, Schedl T, Truong TK, Wambach JA, Wheeler MT, **Yamamoto S**; Undiagnosed Diseases Network; Shashi V, McCray AT, Adams DR, LeBlanc K (2026) Arriving at a

diagnosis: Effective strategies used by the Undiagnosed Diseases Network. ***Genet Med***. (Online ahead of print). doi: 10.1016/j.gim.2026.102718. PMID: 42720004. PMCID: In Progress.

114. Mok JW, Welch CL, Dostalik HA, Tan Z, Brautbar A, DesCartes M, Gray ET, Iglesias A, McLean SD, Nutter M, Daugherty O, Begtrup A, Wentzensen IM, Shen Y, Young DW, Matzuk MM, Baylor College of Medicine Center for Precision Medicine Models, Kanca O, Wangler MF, Bellen HJ, Chung WK†, **Yamamoto S†** (2026). A rare recurring gain-of-function variant in BMPR2 causes neurodevelopmental phenotypes in humans and flies. ***Am J Hum Genet***. (Online ahead of print) doi: 10.1016/j.ajhg.2026.08.019 PMID: 42767215. PMCID: In Progress. **(†corresponding authors)**.

ARTICLES IN REVISION, SUBMITTED & IN PREPARATION

115. Peterson W, Kobayashi T, Hu C, Wang H, Ishii K, Hata S, Kacenauskaite L, Hiramatsu K, Hasan MAM, Otsuka T, Nakamura M, Tsutsumi Y, Wada Y, He Z, Clement JE, Dalijono J, Wang J, Arenson J, Zhang C, McCann P, Kanazawa H, Nagasaka Y, Liu Y, Uechi H, Watanabe Y, Yamamura R, Hayashi M, Nakagawa Y, Qiu C, Bahbouh A, Huang K, Lisi F, Kanno H, Zhou Y, Ding T, Herbig M, Makino S, Nonaga S, Takami R, Tabata K, Amaya S, Furusawa K, Emoto K, Arai F, Cagan R, Di Carlo D, Igaki T, Kuranaga E, Kanca O, **Yamamoto S**, Bellen HJ, Komatsuzaki T, Sonoshita M, Goda K (2026) Flow zoometry of Drosophila. In revision for ***Nature Biotechnology***. Preprint uploaded to ***BioRxiv*** (doi: 10.1101/2024.04.04.588032). PMID: In Progress. PMCID: In Progress.

116. Townsley RE, Andrews JC, Srivastav S, Jangam SV, Hannan SB, Kanca O, Yamamoto S, Wangler MF (2026) NLGN3 autism variants have distinct functional impact on synapses and sleep behavior in Drosophila. In revision for ***PLoS Genet***. Preprint uploaded to ***bioRxiv*** (doi: 10.64898/2026.03.26.714389. PMID: 41929011. PMCID: PMC13041802)

117. Butler DF, Yuan W, Hashimoto H, Hoang H, Sheehan JH, Gomez V, Rosenfeld JA, Burrage LC, Silverman GA, Dickson P, Moeller KK, Emrick L, Ganapathi M; Undiagnosed Diseases Network; Baldridge D, Kanca O, Bacino C, Lee B, Bellen HJ, **Yamamoto S**, Chung WK, Schedl T, Wangler M, Yoo A, Pak SC (2026) Cross-species functional analysis of a de novo DCLK1 variant associated with a neurodevelopmental disorder. In revision for ***Nat Commun***. Preprint uploaded to ***Res Sq*** (doi: 10.21203/rs.3.rs-10425123/v1. PMID: 42539103).

118. Tumiene B, David AR, Allaway R, Barrero MJ, Chan C-H, Faundes V, Fear VS, Glezer P, Fuchs C, Groza T, Houwink EJF, Jamuar SS, Letinturier MCV, Lomash RM, Puri RD, Reichardt JKV, Shai R, van der Westhuizen F, Varshney GK, **Yamamoto S**, Tataru E-A, Baynam G (2026) The roadmap for the development of the Human Variome - Development of functional multi-omics analyses for the understanding of human biology. Preprint uploaded to ***zenodo*** (doi: 10.5281/zenodo.21980384)

119. **Yamamoto S*†**, Kanca O*, Wan KH, Johnson TK, Hashimoto H, Huang M-C, Graves HK, Bhavana VH, Pan H, Bei D, Jangam S, Bhadane P, Park SI, Fisher WW, Takashima Y, Ravani S, Tomaru M, Ohsako T, Wangler MF, Warr C, Takano-Shimizu T , Celniker SE**†**, Bellen HJ**†** (2026) A resource of Drosophila transgenic and mutant lines to study genetic variants in human protein coding genes. In preparation. **(*equal contribution, †corresponding authors)**.

120. Oka M, Imura E, Dupont J, Duraine L, Kanca O, Yamanaka N, **Yamamoto S†** (2026). A *Drosophila* model of *BUD13*-related progeroid syndrome reveals a previously unrecognized steroid hormone defect in human. In preparation. **(†corresponding author)**.

-Complete list of published work in PubMed can be found in the following ***NCBI MyBibliography*** page:
https://www.ncbi.nlm.nih.gov/myncbi/1XUsbagdutxAi/bibliography/public/

-Publications curated in ***Google Scholar*** can be found here:
https://scholar.google.com/citations?user=GCbIDKwAAAAJ

-In addition, Shinya Yamamoto is listed as a consortium member/collaborator on the following additional papers published from the 'Undiagnosed Diseases Network', 'Baylor College of Medicine Center for Precision Medicine Models' and 'Texome Project' in PubMed.

PMIDs: 41655254 (***Adv Sci (Weinh)***), 41411404 (***AMA J Ethics***), 27693232, 28132692, 28157539, 30773277, 31155284, 31327508, 31585109, 32197074, 33508234, 34450031, 34529933, 37451268, 37467750, 37541189, 37622199, 38181735, 38348595, 38387458, 38508193, 38531365, 38593811, 38723631, 38749428, 38834072, 39106866, 39706195, 39798569, 39978342, 40043707, 40088891, 40975062, 41075783, 41468891 (***Am J Hum Genet***), 33184947, 33438828, 33847457, 33949769, 34042254, 34668327, 37246601, 37622199, 37743782, 37897121, 38168088, 38511854, 38613168, 38924341, 39166407, 39166428, 39333051, 39400494, 39629753, 40317671, 40838347, 40936200 (***Am J Med Genet A***), 40780579 (***Am J Ophthalmol***), 37194416, 38311799, 38504481 (***Ann Clin Transl Neurol***), 34185323, 41257467 (***Ann Neurol***), 35868845 (***Ann Rheum Dis***), 39825393 (***BioData Min***), 31443933 (***Biol Psychiatry***), 30134969 (***BMC Health Serv Res***), 31615419 (***BMC Bioinformatics***), 38279154 (***BMC Genomics***), 41239373, 41491999 (***BMC Med Genomics***), 32761064, 36718090 (***Brain***), 35218524 (***Cerebellum***), 39963794 (***Circ Res***), 33963760, 40931319 (***Clin Genet***), 31299614 (***Clin Imaging***), 29970384, 33811063 (***Cold Spring Harb Mol Case Stud***), 37565267 (***Dis Model Mech***), 40862571 (***ELife***), 40859057 (***EMBO Mol Med***), 30682224 (***Epilepsia***), 37500725, 38467731, 38965372, 41145827, 41606215 (***Eur J Hum Genet***), 33724412 (***Genetics***), 28914269, 29907797, 30568308, 33093671, 33239752, 33580225, 33833410, 34163037, 34230636, 35482014, 3641303, 37191094, 37947183, 38431799, 38436216, 38767059, 38967101, 39891528, 40417743, 40576023, 41121346, 41653023 (***Genet Med***), 39486878, 40113264 (***Genome Res***), 41808409 (***HGG Adv***), 33944996, 38451290 (***Hum Genet***), 35913761, 37552066, 41467504 (***Hum Mol Genet***), 31264822, 33239752, 33580225 (***Hum Mutat***), 33630084, 40844783 (***JAMA Netw Open***), 33771552 (***J Allergy Clin Immunol***), 34009343 (***J Am Med Inform Assoc***), 35998814 (***J Biomed Inform***), 38780621 (***J Exp Med***), 30706981, 30964584, 33108040, 34096130, 34115423, 34374469, 37005744 (***J Genet Couns***), 39723123 (***JIMD Rep***), 35621276, 40563223 (***J Inherit Metab Dis***), 35318459 (***J Hum Genet***), 37316189, 39643435 (***J Med Genet***), 37271495 (***J Pediatr***), 36984839 (***Metabolites***), 34702355 (***Mol Autism***), 30993913, 33159716, 33350591, 33955715, 33960148, 35247231, 41407309 (***Mol Genet Genomic Med***), 34412939, 35144859, 37883914, 39742826, 40215727, 41232198 (***Mol Genet Metab***), 39435313 (***Mol Genet Metab Rep***), 37828001, 38413582, 39107278, 39174524, 39256359, 40770127 (***Nat Commun***), 34211179 (***Nat Genet***), 33664507, 31160820 (***Nat Med***), 37653044 (***Nat Metab***), 40788430 (***Neurogenetics***), 37560121, 39880924 (***Neurol Genet***), 40542121 (***NPJ Digit Med***), 36765070 (***NPJ Genom Med***), 28416019, 33971915, 36624503, 37032333, 37667351, 39543639 (***Orphanet J Rare Dis***), 39181022 (***Pediatr Neurol***), 40982570 (***PLoS Comput Biol***), 37934770 (***PLoS Genet***) 31439813 (***Postgrad Med J***), 39435313, 41438096 (***Rare***), 33268356, 33523931 (***Sci Adv***), 33883556 (***Sci Data***).

ORAL PRESENTATIONS

2008 "*Drosophila* EH Domain Binding Protein 1 is required for Notch signaling during external sensory organ development", BCM Program in Developmental Biology Annual Retreat, Cleveland, TX. (2008/02/23)

2009 "Identification of Novel Genes Involved in Neural Development, Physiology and Pathology on the *Drosophila* X chromosome through an F3 Forward Genetic Screen", BCM Program in Developmental Biology Annual Retreat, Galveston, TX. (2009/02/20)

2010 "Different EGF-repeats of Notch have Distinct Biological Functions: *in vivo* Structure Function Analysis of the *Drosophila* Notch Receptor", BCM Program in Developmental Biology Annual Retreat, Galveston, TX. (2010/02/12)

"Distinct modes of Notch signaling are mediated by different EGF repeats of Notch", 22nd Annual Graduate Studies Symposium, BCM, Houston, TX. (2010/10/28)

2011 "An Evolutionally Conserved Valine in EGF repeat 8 of Notch is Involved in Ligand Specificity", The Notch Meeting V, Athens, Greece. (2011/10/04)

"Novel Insights into the Notch Signaling Pathway though a Forward Genetic Screen on the *Drosophila* X Chromosome", Developmental Biology Unit, EMBL (Host: Dr. Anne Ephrussi), Heidelberg, Germany. (2011/09/30)

2012 "Identification of novel Notch alleles that affect intracellular trafficking", The Notch Meeting VI, Athens, Greece. (2012/10/01)

"Learning new things about an old gene: *in vivo* structure function analysis of Notch through a forward genetic approach", Developmental Biology Program, Memorial Sloan-Kettering Cancer Center (Host: Dr. Jennifer A Zallen), New York, NY. (2012/08/03)

2013 "Screening for key genes in nervous system development, function and maintenance", RIKEN Brain Science Institute (Host: Dr. Adrian Moore), Wako, Japan. (2013/04/10)

"Screening for key genes in nervous system development, function and maintenance", School of Veterinary Medicine, the University of Tokyo (Host: Dr. Kazuhiko Imakawa), Tokyo, Japan. (2013/04/11)

"Screening for key genes in nervous system development, function and maintenance", Department of Pharmacology, The University of Tokyo (Host: Dr. Taisuke Tomita, Tokyo, Japan. (2013/04/12)

"Screening for key genes in nervous system development, function and maintenance", School of Medicine, Kyoto University (Host: Dr. Makoto Mark Taketo), Kyoto, Japan. (2013/04/16)

"Screening for key genes in nervous system development, function and maintenance", Department of Biological Sciences, Osaka University (Host: Dr. Kenji Matsuno), Osaka, Japan. (2013/04/17)

"Screening for key genes in nervous system development, function and maintenance", School of Medicine, Kansai Medical University (Host: Dr. Ikuko Yao), Moriguchi, Japan. (2013/04/17)

"Screening for key genes in nervous system development, function and maintenance", School of Medicine, Hamamatsu Medical University (Host: Dr. Mitsutoshi Setou), Hamamatsu, Japan. (2013/04/18)

"Screening for key genes in nervous system development, function and maintenance", Department of Life Science, Rikkyo University (Host: Dr. Satoshi Goto), Tokyo, Japan. (2013/04/19)

"*In vivo* structure-function analysis of the Notch receptor in *Drosophila*", The 36th Annual Meeting of the Molecular Biology Society of Japan, Kobe, Japan. (2013/12/04)

"Can Flies Help Us Fight Cancer?", School of Medicine, Juntendo University (Host: Dr. Misa Imai), Tokyo, Japan. (2013/12/11)

"Identification of New Genes in Notch Signaling through Forward Genetic Approaches in *Drosophila*", Department of Biological Sciences, Tokyo Metropolitan University (Host: Dr. Aigaki Toshiro), Tokyo, Japan. (2013/12/12)

2014 "Using Fruit Flies to Understand Human Diseases: A Sophisticated Tool to Unravel Molecular Mechanisms and Promote Gene Discovery", Texas Children's Hospital (Host: Dr. Jimmy Holder), Houston, TX. (2014/01/22)

"A *Drosophila* resource to study the molecular pathology of human neurological disorders" University of Cambridge (Host: Dr. Sarah Bray), Cambridge, UK. (2014/03/05)

"Integration of *Drosophila* Phenotypic Screening and Human Exome Datasets to Identify New Human Disease Genes: Towards Identification of New Notch Signaling Disorders", 2014 Gordon Research Conference: Notch Signaling in Development, Regeneration & Disease, Lewiston, ME. (2014/07/23)

"Integration of Drosophila Phenotypic Screening and Large Human Exome Datasets to Identify and Study New Human Disease Genes", Houston Fly Club Monthly Meeting, Rice University, Houston (2014/7/30).

"Using *Drosophila* to identify and study human disease genes", Center for Interdisciplinary Cardiovascular Sciences, Brigham and Women's Hospital and Harvard Medical School (Host: Dr. Masanori Aikawa), Boston, MA. (2014/10/28)

2015 "Using *Drosophila* to Identify and Study of New Human Disease Genes", BCM Program in Developmental Biology Annual Retreat, Galveston, TX. (2015/02/06)

"Using *Drosophila* to Identify and Study of New Human Disease Genes", Alberta Children's Hospital Research Institute 2015 Symposium, Calgary, Canada. (2015/04/15)

"Using *Drosophila* to Identify and Study of New Human Disease Genes", 2015 Texas Children's Hospital Seminar Series, Houston, TX. (2015/04/23)

"Using *Drosophila* to identify and study human disease genes", Houston Fly Club Monthly Meeting, Rice University, Houston (2015/8/26).

"Using *Drosophila* to Understand Notch-related Human Diseases", The Notch Meeting IX, Athens, Greece. (2015/10/06)

"Using *Drosophila* to Identify and Study Human Diseases", Venetian Institute of Molecular Medicine and University of Padova (Host: Dr. Luca Scorrano), Padova, Italy. (2015/10/12)

"Using *Drosophila* to Identify and Study Human Diseases", University of Rome-La Sapienza (Host: Dr. Maurizio Gatti), Rome, Italy. (2015/10/14)

"Using *Drosophila* to Probe the Function of Genes and Variants Associated with Human Diseases", Intellectual and Developmental Disability Research Center (IDDRC) Director's Meeting, UC Davis Medical School, Sacramento, CA. (2015/11/12)

"Functional Genomics through Integration of *Drosophila* and Human Genetics", 2015 Department of Molecular & Human Genetics Seminar Series, BCM (Host: Dr. Huda Zoghbi), Houston, TX. (2015/12/15)

2016 "Functional Genomics through Integration of *Drosophila* and Human Genetics", Department of Genetics, MD Anderson Cancer Center (Hosts: Drs. Michael Galko and George Eisenhoffer), Houston, TX. (2016/01/20)

"Functional Genomics through Integration of *Drosophila* and Human Genetics", Division of Developmental Biology, Cincinnati Children's Hospital Medical Center (Host: Dr. Raphael Kopan), Cincinnati, OH. (2016/02/09)

"Functional Genomics through Integration of *Drosophila* and Human Genetics", Institute of Systems Genetics, New York University Langone Medical Center (Host: Dr. Jef Boeke), New York, NY. (2016/02/17)

"MiMIC and CRIMIC: Introducing versatile artificial exons for gene function studies in *Drosophila*" The Allied Genetics Conference (TAGC2016, Genetics Society of America), Orlando, FL. (2016/07/16)

"National & International Collaborations to Facilitate the Diagnosis & Study of Undiagnosed Diseases", Undiagnosed Diseases Network In-Person Steering Committee Meeting, Washington, DC. (2016/07/26)

"CRIMIC: Introducing versatile artificial exons via CRISPR for gene function studies in *Drosophila*" Symposium on Genome Editing (BCM, MD Anderson, University of Texas Health Science Center at Houston, Rice University), Houston, TX. (2016/11/08)

"Genetic model organisms to study disease associated variants", The 4th Conference of Undiagnosed Diseases Network International, Tokyo, Japan. (2016/11/17)

2017 "The Baylor College of Medicine-University of Washington Model Organisms Screening Center for the Undiagnosed Diseases Network", Undiagnosed Diseases Network Grand Rounds (CME Credit Approved Lecture), Online Webinar (2017/2/8)

"Using *Drosophila* to discover new genes involved in human neurological disorders", 27th Record Neuroscience Forum, Galveston, TX. (2017/02/11)

"*Drosophila* in Undiagnosed Disease Research", 4th Asia-Pacific *Drosophila* Research Conference, Osaka, Japan. (2017/05/09)

"Using *Drosophila* to discover and study human disease genes", Kobe University Inter-Genomics Seminar Series (Host: Dr. Yoshiko Aihara), Kobe, Japan. (2017/05/12)

"Using *Drosophila* to discover and study human disease genes", RIKEN Center for Developmental Biology (Host: Dr. Mitsuru Morimoto), Kobe, Japan. (2017/05/15)

"Using *Drosophila* to discover and study human disease genes", Advanced Insect Research Promotion Center at Kyoto Institute of Technology (Host: Drs. Masamitsu Yamaguchi and Toshiyuki Takano), Kyoto, Japan. (2017/12/19)

"Using *Drosophila* to discover and study human disease genes", RIKEN Center for Life Science Technologies (Host: Dr. Piero Carninci), Yokohama, Japan. (2017/12/20)

"Using *Drosophila* to discover and study human disease genes", RIKEN Brain Science Institute (Host: Dr. Adrian Moore), Wako, Japan. (2017/12/21)

2018 "Obtaining a Ph.D. and Running an Independent Lab in the USA", University of Tokyo Veterinary Medical School. (Host: Kazuhiko Imakawa), Tokyo, Japan (2018/2/21)

"The Model Organisms Screening Center for the Undiagnosed Diseases Network", IRUD-Beyond Symposium: Model organism research for human rare diseases, Tokyo, Japan (2018/2/22)

"Using *Drosophila* to discover and study new human disease genes", Jikei University School of Medicine (Host: Hirotaka Kanuka), Tokyo, Japan (2018/3/1)

"TM2D family genes in Notch signaling and Alzheimer 's disease pathogenesis", 2018 Alzheimer's Association Research Symposium on Alzheimer's Disease & Related Dementias, Methodist Research Institute, Houston, TX (2018/5/2)

"Model organisms facilitate rare disease diagnosis and therapeutic research", Undiagnosed Diseases Network Grand Rounds (CME Credit Approved Lecture), Online Webinar (2018/6/14)

"The Model Organism Screening Center for the Undiagnosed Diseases Network", 2018 INFRAFRONTIER / IMPC Stakeholder Meeting, Munich, Germany (2018/12/3)

2019 "*Drosophila* as a discovery tool for rare human disease causing genes", University of Texas Health Science Center at Houston School of Public Health, Houston, TX (2019/1/28)

"Informatics analysis workflow at the BCM-UO UDN Model Organisms Screening Center", Tool Building Coalition Committee Meeting, Undiagnosed Diseases Network In-person Steering Committee Meeting, Washington, DC (2019/1/30)

"Phenotypic expansion of known disease causing genes identified in the Undiagnosed Disease Network", 2nd International Symposium for Model Organism Research on Human Rare Diseases, Tokyo, Japan (2019/3/4)

"Using *Drosophila* to discover and study new human disease genes", National Institute of Genetics (Host: Kuniaki Saito), Mishima, Japan (2019/3/6)

"The Undiagnosed Disease Network in the USA", Japan Agency for Medical Research and Development (AMED, Host: Yoshihiko Izumida & Noriaki Imanishi), Tokyo, Japan (2019/3/12)

"The Undiagnosed Diseases Network *Drosophila* Core", Advanced Insect Research Promotion Center at Kyoto Institute of Technology (Host: Drs. Toshiyuki Takano), Kyoto, Japan (2019/3/14)

"Workshop Organizer: Collaborating with clinical researchers -expanding opportunities for *Drosophila* biologists in rare disease diagnosis and therapeutic research", 60th Annual *Drosophila* Research Conference, Dallas, TX (2019/3/29)

"A damaging missense de novo variant in *RNF2* is associated with neurological symptoms", Undiagnosed Diseases Network Grand Rounds (CME Credit Approved Lecture), Online Webinar (2019/6/13)

"Peculiar behavior of human *NOTCH* transgenes in vivo in *Drosophila*", Poster Teaser Session, The Notch Meeting XI, Athens, Greece. (2019/10/07)

"Informatics analysis in the UDN MOSC to prioritize candidate genes and variants", INFRAFRONTIER Rare Disease Data Integration Workshop, Satellite meeting of the Annual Meeting of the American Society of Human Genetics, Houston, TX (2019/10/15)

"Damaging de novo missense variants in *TOMM70* cause a variable white matter disease with neurological phenotypes", UDN Steering Committee In-Person Meeting, Washington DC (2019/12/06)

2020 "Using *Drosophila* to discover and study new human disease causing genes", Kyoto Prefectural University of Medicine (Host: Toshiki Mizuno), Kyoto, Japan (2020/1/21)

"Using *Drosophila* to discover and study new human disease causing genes", Kyoto Institute of Technology University (Host: Toshiyuki Takano), Kyoto, Japan (2020/1/22)

"Using *Drosophila* to discover and study new human disease causing genes", Kyoto University (Host: Tatsushi Igaki), Kyoto, Japan (2020/1/23)

"Using *Drosophila* to discover and study new human disease causing genes", Osaka University (Host: Masahito Ikawa), Osaka, Japan (2020/1/24)

"Using *Drosophila* to discover and study new human disease causing genes", International University of Health and Welfare (Host: Motoo Kitagawa), Narita, Japan (2020/1/27)

"Using *Drosophila* to discover and study new human disease causing genes", Radboud UMC (Host: Annette Schenck), Nijmegen, the Netherlands (2020/2/6)

"Functional Study Working Group of the Undiagnosed Disease Network International", 8th Conference of UDNI (Undiagnosed Disease Network International), Nijmegen, the Netherlands (2020/1/27)

"Strategies and resources to facilitate direct collaborations between clinicians and model organism researchers on a global scale", TAGC2020 Online (Genetics Society of America), Virtual Conference via ZOOM (2020/4/24)

"Bringing MARRVEL to the Next Level", UDN Steering Committee Virtual In-Person Meeting, Virtual Conference via ZOOM (2020/7/30)

"Using *Drosophila* to discover and study new human disease causing genes", RARE TAMU [Student chapter of NORD (National Organization for Rare Diseases) at Texas A&M University] seminar series, Texas A&M University, Virtual Presentation via ZOOM (2020/9/15)

"Facilitating collaborations between clinicians & *Drosophilists* in the USA and around the world", MBSJ2020 Online (Molecular Biology Society of Japan), Virtual Conference (2020/12/4)

2021 "The Baylor College of Medicine-University of Washington Model Organisms Screening Center for the Undiagnosed Diseases Network", Undiagnosed Diseases Network Grand Rounds (CME Credit Approved Lecture), Online Webinar (2021/1/14)

"Using *Drosophila* to discover and study new human disease genes", Dalhousie University (Host: Jamie Kramer), Online Webinar (2021/3/10)

"The roles of *TM2D* genes in late-onset Alzheimer's disease and Notch signaling", University College London, Institute of Healthy Aging Virtual Symposium, Online Webinar (2021/3/17)

"Tools and strategies to identify new disease causing human genes and variants using *Drosophila*", 62nd Annual *Drosophila* Research Conference, Online Webinar (2021/3/29)

"ModelMatcher: a matchmaking platform for scientists and clinicians", the Alliance of Genome Resources PI meeting, Virtual Presentation via ZOOM (2021/4/9)

"Functional Study Working Group of the Undiagnosed Disease Network International", 9th Conference of UDNI (Undiagnosed Disease Network International), Virtual Presentation via ZOOM (2021/4/11)

"ModelMatcher and MARRVEL: Two integrative tools for the rare and undiagnosed diseases community", UDN PEER (Undiagnosed Diseases Network Participant Engagement and Empowerment Resource) group meeting, Virtual Presentation via ZOOM (2021/4/12)

"MARRVEL: Mining genetic and genomic data across MO and human", GSA webinar series "Exploring gene function across humans and model organisms", Genetic Society of America, Online Webinar (2021/9/20)

"New Neurological Disease Gene Discovery through Functional Studies and Matchmaking", University of Texas McGovern Medical School (Host: Kartik Venkatachalam), Houston, TX (2021/10/25)

"New Neurological Disease Gene Discovery through Functional Studies and Matchmaking", Tsukuba University (Host: Niwa Ryosuke), Online Webinar (2021/10/28)

2022 "Studying Human Notch Proteins using *Drosophila melanogaster*", Osaka University (Host: Kenji Matsuno), Online Webinar (2022/1/18)

"Using *Drosophila* to discover and study new human disease genes", Sam Houston State University (Host: Mardelle Atkins), Online Webinar (2022/3/3)

"ModelMatcher: a scientist-centric online platform for rare and undiagnosed diseases research", GREGoR Consortium Functional Study Working Group Meeting, Online Webinar (2022/3/15)

"ModelMatcher: a scientist-centric online platform for rare and undiagnosed diseases research", Matchmaker Exchange Steering Committee Meeting, Online Webinar (2022/3/16)

"The role of model organisms in the Undiagnosed Diseases Network", Future of the Undiagnosed Diseases Network Meeting, Panelist, Boston, MA (2022/7/14)

"ModelMatcher: a scientist-centric online platform for rare and undiagnosed diseases research", IMPC Rare Disease Working Group Meeting, Online Webinar (2022/7/26)

"Using *Drosophila* to discover and study new human disease genes", Florida State University (Host: Mischelle Arbeitman and Edrem Bangi), Tallahassee, FL (2022/10/12)

"Fruit flies and mice facilitate undiagnosed diseases research in the BCM CPMM", 11th UDNI Conference Scientific Meeting, Vienna, Austria (2022/11/7)

"Functional Study Working Group Report", 11th UDNI Conference Business Meeting, Vienna, Austria (2022/11/8)

2023 "Using *Drosophila* to discover and study new human disease genes), MD Anderson Cancer Center Research Exchange Lecture Series (Host: Shih-Han Lee & Peter Van Loo), Houston, TX (2023/3/22)

"Developing Informatic Tools to Facilitate Rare Disease Diagnosis and Collaborative Research", Pediatrics Research Symposium Workshop, BCM and TCH, Houston, TX (2023/4/4)

"Collaborations between scientists and clinicians facilitate undiagnosed disease research", UDNI and Wilhelm Foundation, Undiagnosed Day 2023, Online Webinar (2023/4/29)

"Functional Studies in Evaluation of Undiagnosed and Rare Conditions", Harvard Medical School Continuing Education, Online CME Credit Course (2023/5/10)

"Informatic Tool Development in the UDN MOSC to Facilitate Rare Disease Diagnosis and Research", Longwood Bioinformatics Lecture Series, Harvard Medical School, Online Webinar (2023/5/19)

"Functional analysis of *SATB2* and disease-associated variants using *Drosophila*", 2023 SATB2 Research Roundtable, *Cure*SATB2 & SATB2 Europe, Online Webinar (2023/6/20)

"Navigating MARRVEL, an integrated search engine for rare and undiagnosed disease research", 1st Undiagnosed Hackathon Tools Workshop, Karolinska Institute, Stockholm, Sweden (2023/06/17)

"Functional studies of disease associated genes and rare variants using state-of-the-art technologies in *Drosophila melanogaster*", 2023 Gordon Research Conference: Human Genetics & Genomics, Waterville Valley, NH (2023/7/10)

"A comprehensive Drosophila resource to identify key functional interactions between SARS-CoV-2 factors and host proteins", Houston Fly Club Monthly Meeting, Houston, TX, Virtual Presentation via ZOOM (2023/7/27).

"MARRVEL and ModelMatcher: Platforms & Tools for Rare Disease Genomics Collaboration", Science in Society Annual Meeting 2023, Chan Zuckerberg Initiative (CZI), Newport Beach, CA (2023/09/22)

"Functional studies using *Drosophila* support clinical diagnosis and phenotypic expansion: *BMPR2* in neurodevelopmental disorders", 12th Conference of UDNI Scientific Meeting, Tbilisi, Georgia (2023/10/22)

"Functional Study Working Group Report", 12th Conference of UDNI (Undiagnosed Disease Network International) Business Meeting, Tbilisi, Georgia (2023/10/23)

"Functional studies in *Drosophila* facilitate rare disease diagnosis and research", National Institute of Genetics of Japan International Symposium, Mishima, Japan (2023/11/09)

"Humanization *Drosophila* genes for rare disease diagnosis and research", TT2023 (The 18th Transgenic Technology Meeting), Houston, TX (2023/11/13)

"Precision Medicine *Drosophila* Models for Undiagnosed and Rare Disease", 2023 KOMP2 & IMPC Fall Meeting, Houston, TX (2023/11/27)

2024 "MARRVEL and ModelMatcher: publicly available web services that facilitate collaborative research on rare diseases", TAGC2024 (Genetics Society of America), National Harbor, MD (2024/3/4)

"Using *Drosophila* to discover and study new human disease genes", Rice University, (Host: Kathleen Beckingham), Houston, TX (2024/4/1)

"Bioinformatic tool development to facilitate rare disease diagnosis and research", Genetics Faculty Short Talk Series, Baylor College of Medicine, Houston, TX (2024/4/1)

"Facilitating collaborations through the Undiagnosed Hackathon & informatic tool development", Undiagnosed Day event at Harvard Medical School, Boston, MA (2024/4/29)

"Functional Studies in Evaluation of Undiagnosed and Rare Conditions", Harvard Medical School Continuing Education, Online CME Credit Course (2024/5/13)

"Facilitating rare disease diagnosis through functional studies using *Drosophila*", 2nd Undiagnosed Hackathon Tools Workshop, Radboud UMC, Nijmegen, the Netherlands (2024/6/26)

"Facilitating rare disease diagnosis through functional studies using *Drosophila*", 13th Conference of UDNI Scientific Meeting, Seoul, Korea (2024/9/6)

"Functional Study Working Group Report", 13th Conference of UDNI Business Meeting, Seoul, Korea (2024/9/7)

"Facilitating rare disease diagnosis through functional studies using *Drosophila*", Department of Biological Sciences, Tokyo Metropolitan University Drosophila Symposium: New Horizons in *Drosophila* Models, Tokyo, Japan (2024/11/5)

"Combining state-of-the-art technologies in *Drosophila* to discover novel rare disease-causing genes and variants in human", 97th Annual Meeting of the Japanese Biochemical Society, Yokohama, Japan (2024/11/7)

"Medical research using *Drosophila*: potential application to the veterinary field", Alumni Reunion, Veterinary Medical School, University of Tokyo, Tokyo, Japan (2024/11/9)

"Facilitating rare disease diagnosis through functional studies using *Drosophila*", 2nd Rare Diseases & Rare Cancers in the Era of Systems Biology Symposium (hybrid conference), Medical College of Wisconsin, Milwaukee, WI (2024/11/14)

"Empowering clinically relevant research projects through the BCM Center for Precision Medicine Models (CPMM)", Inaugural NRI Faculty Retreat, Texas Children's Hospital, Houston, TX (2024/12/9)

2025 "Facilitating rare disease diagnosis through functional studies using *Drosophila*", Department of Genetics, Perelman School of Medicine at the University of Pennsylvania (Host: Drs. Dan Rader and Matthew Kayser), Philadelphia, PA (2025/3/13)

"Functional Studies in Rare Disease Diagnosis", Red de Expertos en Salud de Enfermedades Raras (RIBERSER, a.k.a Iberoamerican Health Expert Network for Rare Diseases), Online Webinar (2025/3/15)

"Pushing the limits of *Drosophila*", NeuroRadio (https://neuroradio.tokyo/, Hosts: Drs. Kenta Hagihara & Takeyuki Miyawaki), Online Podcast in Japanese (Episodes 86 & 87, 2025/4/2 & 7)

"Using *Drosophila* to discover and study new human developmental disease genes", 2025 Gordon Research Conference: Developmental Biology, Pomona, CA (2025/4/2)

"Using *Drosophila* to discover and study new human disease-causing genes", GSA Award Seminar Series, Online Webinar (2025/7/24)

"Alumni Lightning Talks", Science Careers Conference, BCM, TX (2025/9/9)

"Using *Drosophila* to discover and study new human disease-causing genes", Keck Seminar, Rice University, Houston, TX (2025/9/12)

"FlyBase & the Biomedical Model Organism Database Ecosystem, Japanese Young *Drosophila* Researcher's Club (Hosts: Drs. Yuta Takada & Mr. Yuma Tsukasa), Online Webinar (2025/10/10)

"Using *Drosophila* to discover and study new human disease-causing genes", College of Sciences, The University of Texas at San Antonio (Host: Dr. Jeffrey Vedanayagam), San Antonio, TX (2025/11/10)

2026 "Using *Drosophila* to discover and study new human disease-causing genes", Emory University School of Medicine (Host: Dr. Jian Zhou), Atlanta, GA (2026/3/23)

"Using *Drosophila* to discover and study new human disease-causing genes", Zebra Talk Series, Dell Medical School, The University of Texas at Austin (Host: Drs. Ankit Sabharwal and Stephen Ekker), Austin, TX (2026/4/23)

"Facilitating rare disease diagnosis using fruit flies through teamwork", RNDF Spring Assembly, Rare Neurological Disorder Foundation, Online Webinar (2026/5/1)

"Functional Studies using Model Organisms", Online Training Course: "Evaluation of Undiagnosed and Rare Conditions (https://undiagnosed.hms.harvard.edu/events/course/)", Undiagnosed Diseases Network (2026/5/13)

"Research on rare and undiagnosed diseases using *Drosophila*", NISTEP Seminar Series, NISTEP (National Institute of Science and Technology Policy), Online Webinar in Japanese (2026/7/3)

"Using *Drosophila* to discover and study new human disease-causing genes and variants", 51st Serendipity Seminar, The University of Tokyo (Host: Dr. Keisuke Goda), Tokyo, Japan (2026/7/30)

"Using *Drosophila* to discover and study new human disease-causing genes", NIH Neuroscience Seminar Series, National Institutes of Health (Host: Dr. Benjamin White), Bethesda, MD (2026/9/14)

"Using *Drosophila* to discover and study new human disease-causing genes", Rutgers University (Host: Dr. Kenneth D. Irvine), Princeton, NJ (2026/10/6)

"Using *Drosophila* to discover and study new human disease-causing genes", Quantitative & Computational Biology Seminars, Princeton University (Host: Dr. Stanislav Y. Shvartsman), Princeton, NJ (2026/10/7)

"A *Drosophila* model of *BUD13*-related progeroid syndrome reveals a previously unrecognized steroid hormone defect in human", Drosophila Camp (organized by Dr. Keisuke Goda at University of Tokyo), Sendai, Japan (11/1/2026)…Scheduled

"A Rare Recurring Gain-of-Function Variant in *BMPR2* Causes Neurodevelopmental Phenotypes in Humans and Flies", The 17th Japanese Drosophila Research Conference, Tokyo, Japan (11/6/2026)…Scheduled

2027 "Using *Drosophila* to discover and study new human disease-causing genes", Swiss 3R Day conference, University of Geneva (Host: Dr. Vladimir Katanaev), Geneva, Switzerland (2027/7/29)…Scheduled

MENTORSHIP OF STUDENTS AND TRAINEES

GRADUATE (PHD) STUDENTS

2015-2021 Jose L. Salazar (Program in Molecular & Human Genetics, BCM)
Current position: Field Applications Scientist, Oxford Nanopore Technologies

2018-2021 J. Michael Harnish (Program in Molecular & Human Genetics, BCM)
Current position: Business Development Lead, Author Health

2017-2023 Samantha Deal (Program in Developmental Biology, BCM)
Current position: Assistant Professor, Mercer University

2020-2026 Shelley Gibson (Genetics & Genomics Graduate Program, BCM)
Current position: Postdoctoral Fellow, Eunice Kennedy Shriver National Institute of Child Health (NICHD, Mentor: Dr. Mihaela Serpe)

2022- Harmin Delgado-Seo (Neuroscience Graduate Program, BCM)
2023- Haley A. Dostalik (Genetics & Genomics Graduate Program, BCM)
2025- Jasmine O. Brown (Genetics & Genomics Graduate Program, BCM)

GRADUATE (MS) STUDENTS

2026- Isabella Tandy (Master of Science in Biomedical Sciences Graduate Program, BCM)

POSTDOCTORAL FELLOWS

2016 Sumit Saurabh, Ph.D.
Current position: Assistant Professor, Chicago State University
2019-2022 Sheng-An Yang, Ph.D.
Current position: Clinical Variant Scientist, ARUP Laboratories
2021- Jung-Wang Mok, Ph.D.
2022- Mikiko Oka, Ph.D.
2022- Hirokazu Hashimoto, Ph.D.
2022-2024 Rei Yasuda, M.D., Ph.D.
Current position: Assisatnt Professor, Kyoto Prefectural University of Medicine

POST-BAC STUDENTS

2015-2016 Samantha L. Deal (Post-Baccalaureate student, BCM)
Subsequent position: Ph.D. student in BCM
2015-2016 Matthew Lagarde (Post-Baccalaureate student, BCM)
Subsequent position: M.D. student in G. McGovern Medical School at UTHealth
2015 Matthew Feiock (Post-Baccalaureate student, BCM)
Subsequent position: Pathology Laboratory Aide,
Florida Cancer Specialists & Research Institute
2018-2019 Brooke Hull [PREP (Post-baccalaureate Research Education Program), BCM]
Subsequent position: Ph.D. student in Princeton University
2023 Jasmine O. Brown (PREP, BCM)
Subsequent position: Ph.D. student in BCM

UNDERGRADUATE STUDENTS

2013 David Wang (Rice University, NEUR310 course)
Subsequent position: M.D. student in McGovern Medical School at UTHealth
2018-2020 Ashley Phillips (Rice University, NEUR310 course)
Subsequent position: M.P.H. student at NYU
2019-2020 Frank Shi (Rice University, NEUR310 course)
2019-2020 Victoria Lee (Rice University, BIOC310 course)
2022 Jean-Luc Shimizu (Cornell University, summer student)
2022-2023 Fernanda Valle Sirias (Rice University, NEUR310 course)
2025- Swayam Rath (Rice University, NEUR310 course)

RESEARCH ROTATIONS

2013 Yi-Chen Hsieh (PhD Student Rotation, BCM)
2014 Krystal English (PREP Student Rotation, BCM)
2015 Valencia Potter (PhD Student Rotation, BCM)
2015 Yingyao Shao (PhD Student Rotation, BCM)
2017 Fatma Isleyen (PhD Student Rotation, BCM)
2018 Angad Jolly (PhD Student Rotation, BCM)
2018 J. Michael Harnish (PhD Student Rotation, BCM)

2018 Brooke Hull (PREP Student Rotation, BCM)
2019 Morgan Stephens (PhD Student Rotation, BCM)
2019 Shelley Gibson (PhD Student Rotation, BCM)
2021 Julie Ann Goddard (PhD Student Rotation, BCM)
2022 Devine Jackson (PhD Student Rotation, BCM)
2022 Harim Delgado-Seo (PhD Student Rotation, BCM)
2022 Danielle Mendonca (PhD Student Rotation, BCM)
2022 Ellen Thompson (PhD Student Rotation, BCM)
2023 Haley A. Dostalik (PhD Student Rotation, BCM)
2025 Jasmine O. Brown (PhD Student Rotation, BCM)
2025 Shanice James (MS Student Rotation, BCM)
2025 Isabella Tandy (MS Student Rotation, BCM)
2025 Adrian Bafor (MS Student Rotation, BCM)
2025 Ashlee Prejean (MS Student Rotation, BCM)
2025 Mariyah Sion (MS Student Rotation, BCM)
2026 Carlee Marquez (PhD Student Rotation, BCM)

THESIS ADVISORY COMMITTEES

2014-2018 Chang-Ru Tsai (PI: Michael Galko, Program in Developmental Biology, BCM)
2017-2018 Mumine Senturk (PI: Hugo Bellen, Program in Developmental Biology, BCM), as ex-officio member
2017-2019 Julia Wang [PI: Hugo Bellen, Program in Developmental Biology and MSTP (Medical Scientist Training Program), BCM]
2017-2020 Amanda Gervaise (PI: Swathi Arur, Program in Developmental Biology, BCM)
2017-2020 Tarik Onur (PI: Juan Botas, Program in Molecular & Human Genetics, BCM)
2017-2021 Nicholas Albrecht (PI: Melanie Samuel, Program in Translational Biology and Molecular Medicine, BCM)
2018-2021 Thomas Ravenscroft (PI: Hugo Bellen, Program in Molecular & Human Genetics, BCM)
2018-2023 Daniel Sutton (PI: Andrew Groves, Program in Molecular & Human Genetics, BCM)
2018-2023 Meigen Yu (PI: Joshua Shulman, Program in Neuroscience, BCM)
2019-2025 Matthew Avalos (PI: Juan Botas, Program in Molecular & Human Genetics, BCM)
2019-2022 Liping Wang (PI: Hugo Bellen, Program in Developmental Biology, BCM)
2019-2023 Jiayang Li (PI: Juan Botas, Quantitative & Computational Biosciences Program, BCM)
2020-2021 Sean Dooling (PI: Mauro Costa-Mattioli, Program in Molecular & Human Genetics, BCM), as ex-officio member
2020-2023 Dillon R. Shapiro (PI: Olivier Lichtarge, Program in Molecular & Human Genetics, BCM), as ex-officio member
2020-2023 Ruizhi (Vince) Duan (PI: James Lupski, Program in Molecular & Human Genetics, BCM)
2020-2024 Kristy Jay (PI: Michael Wangler, Genetics & Genomics Graduate Program, BCM)
2020-2026 Megan Mair (PI: Juan Botas, Program in Molecular & Human Genetics, BCM)
2020-2025 Morgan Stephens (PI: Juan Botas, Genetics & Genomics Graduate Program, BCM)
2020-2026 Emily Leptich (PI: Rachel Arey, Neuroscience Graduate Program, BCM)
2020- Laura Deus Ramírez (PI: Christoph Herman, Genetics & Genomics Graduate Program)
2021-2022 Jamie Reyes (PI: Margaret Goodell, Program in Molecular & Human Genetics, BCM), as ex-officio member
2021-2024 Kamryn Gerner-Mauro (PI: Jichao Chen, DDMT Graduate Program, BCM)
2022-2025 Jennifer Deger (PIs: Joshua Shulman & Hugo J Bellen, Neuroscience Graduate Program and

MSTP, BCM)
2022-2024 Guo Hu (PI: Meng Wang, Genetics & Genomics Graduate Program, BCM), as ex-officio member
2022- Tyler Jackson (PI: Hongjie Li, Cancer and Cell Biology Graduate Program, BCM)
2022,2025- Nirav Shah (PI: Jennifer Posey, Genetics & Genomics Graduate Program, BCM) as ex-officio member
2023- Jinghan Zhao (PI: Joshua Shulman, Neuroscience Graduate Program, BCM)
2023- Ye-Jin Park (PIs: Hongjie Li & Hugo Bellen, DDMT Graduate Program, BCM)
2023-2026 Bing Xie (PI: James Martin, Genetics & Genomics Graduate Program, BCM)
2023-2025 Brandon Garcia (PI: Jennifer Posey, Genetics & Genomics Program and MSTP, BCM)
2024- Michal Tyrlik (PI: Hugo Bellen, Genetics & Genomics Graduate Program and MSTP, BCM)
2024- Ellen Thompson (PI: Susan Rosenberg, DDMT Graduate Program, BCM)
2024- Guo-Teng Liang (PI: Steven Boeynaemes, Neuroscience Graduate Program, BCM)
2024-2026 Keyang Yu (PI: Aleksandar Milosavljevic, Genetics & Genomics Graduate Program, BCM), as ex-officio member
2024- Meredith Williams (PI: Rachel Arey, DDMT Graduate Program, BCM)
2024 Shania Pintor [PI: Margot Williams, Cancer & Cell Biology (CCB) Graduate Program, BCM]
2024- David Quispe Parra (PI: Ross A. Poché, Genetics & Genomics Graduate Program, BCM)
2024- Colleen Strohlein (PIs: Huda Zoghbi & Joshua Shulman, Neuroscience Graduate Program, BCM)
2024- Sofia Ivana Aramburu (PI: Susan Rosenberg, DDMT Graduate Program, BCM)
2024- Yan Li (PI: Huda Zoghbi, Genetics & Genomics Graduate Program, BCM) as reporting member
2024- Alex Larson (PIs: Joshua Shulman & Steven Boeynaemes, Neuroscience Graduate Program, BCM)
2025- Grace Pina (PI: Juan Botas, Genetics & Genomics Graduate Program, BCM)
2025- Paige Hall (PI: Michael Wangler, Genetics & Genomics Graduate Program, BCM) as reporting member
2025- Guantong Qi (PI: Zhandong Liu, Genetics & Genomics Graduate Program, BCM) as reporting member
2025- Kai-Li Chang (PI: Zhandong Liu, Genetics & Genomics Graduate Program, BCM)
2025- Edward Pietryk (PI: Cheryl Walker, Genetics & Genomics Graduate Program, BCM) as ex-officio member
2025- Adya Verma (PI: Margot Williams, DDMT Graduate Program, BCM) as reporting member
2025- Mubtasim Akhyar (PI: Steven Boeynaemes, CCB Graduate Program, BCM)
2025- Brandon Baltelmie (PI: Jason Heaney, Genetics & Genomics Graduate Program, BCM) as reporting member
2025- Juwan Copeland (PI: Andrew Groves, Genetics & Genomics Graduate Program, BCM) as reporting member
2025- Adya Verma (PI: Margot Williams, DDMT Graduate Program, BCM)
2025 Marije Been (PI: Erik Strokebaum, Radboud University, Nijmegen, the Netherlands)
2026- Justin Dhindsa (PI: Nicholas Tran, Genetics & Genomics Program and MSTP, BCM) as reporting member
2026- Yifan Chen (PI: Ryan Dhindsa, Genetics & Genomics Graduate Program and MSTP, BCM) as reporting member
2026- Harita Mani (PI: Vishnu Cuddapah, Genetics & Genomics Graduate Program, BCM) as reporting member

2026- Chloe Sands (PI: Ryan Dhindsa, Genetics & Genomics Graduate Program, BCM)
as reporting member
2026- Alex Garza (PI: Benjamin Arenkiel, Genetics & Genomics Graduate Program, BCM)
as ex-officio member

QUALIFYING EXAM COMMITTEES

2014 Matthew Hill (Program in Developmental Biology, BCM)
2015 Rebecca Murdaugh (Program in Developmental Biology, BCM)
2016 Amanda Gervaise (Program in Developmental Biology, BCM)
2017 Sean Dooling (Program in Molecular & Human Genetics, BCM), as chair of committee
2017 Nicholas Albrecht (Program in Translational Biology and Molecular Medicine, BCM)
2018 Meigen Yu (Program in Neuroscience, BCM)
2018 Vera Hutchison (Program in Developmental Biology, BCM)
2018 Max Gao (Program in Developmental Biology, BCM)
2018 Mayuri Patel (Program in Developmental Biology, BCM)
2018 Grant Mangleburg (Program in Molecular & Human Genetics and MSTP, BCM), as chair
2019 Ivanshi Patel (Program in Developmental Biology, BCM)
2019 Mary Edgington (Program in Developmental Biology, BCM)
2019 Timothy Wu (Program in Molecular & Human Genetics, BCM)
2019 Megan Mair (Program in Molecular & Human Genetics, BCM)
2020 Alice Wen (Program in Genetics & Genomics, BCM)
2020 Kristy Jay (Program in Genetics & Genomics, BCM)
2021 Emily Leptich (Program in Neuroscience, BCM, BCM)
2021 Gary Huang (Program in Genetics & Genomics, BCM)
2021 Fangfei Guo (Program in Genetics & Genomics, BCM)
2022 Brooke Horist (Program in Genetics & Genomics, BCM)
2022 Julie Ann Goddard (Program in Genetics & Genomics, BCM)
2023 Jackson Tyler (Program in Cancer and Cell Biology, BCM)
2023 Pragati Kore (Program in Genetics & Genomics, BCM)
2024 Guo-Teng Liang (Program in Neuroscience, BCM)
2024 Rajashree Venkatraman (Program in DDMT, BCM)
2024 Ashweni Ramanah (Program in DDMT, BCM)
2024 Melanie Mew (Program in Genetics & Genomics, BCM)
2024 Grace Pina (Program in Genetics & Genomics, BCM)
2025 Colleen Strohlein (Program in Neuroscience, BCM)
2025 Alex Larson (Program in Neuroscience, BCM)
2025 Meredith Williams (Program in DDMT, BCM)
2025 Shania Pinto (Program in Cancer and Cell Biology, BCM)
2025 Aanya Subramaniam (Program in Genetics & Genomics, BCM)
2025 Harita Mani (Program in Genetics & Genomics, BCM)
2026 Martha Chan (Program in Genetics & Genomics, BCM)
2026 Jessica Mason (Program in Genetics & Genomics, BCM)

-Updated on 10/13/2026