

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

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Lab Website: <https://yamamotoflylab.org>

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 and Human Genetics
(primary), Graduate Program in Developmental Biology, BCM
- 2017 – 2024 *Assistant Professor (tenure track)*, Department of Molecular and 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 and 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 and 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 and Genomics”, a recognition for co-directing the *Method and Logic* Course in AY2021, GSBS, BCM
- 2022 “Outstanding Lecturer Award for Graduate Program in Genetics and 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

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 and 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 Drs. Jennifer Posey, Ross Poché, and Joshua Wythe.

LECTURES (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-present *Anatomy of the Nervous System*, Ph.D. level course. One lecture and one lab. BCM
- 2017-present *Genetics Ethics Session*. M.D. level course. One discussion session. BCM

2017-present *Genetics Journal Club*. Ph.D. level course. One-three discussion sessions.
 2017-present *Responsible Conduct of Research (Ethics) training*, Postdoc level course. One lecture. BCM
 2017-present *Methods and Logic*, Ph.D. level course. Three-four lectures. BCM
 2019 *TBMM Bench to Bedside-Neurodegeneration Journal Club*. Ph.D. level course.
 One lecture, BCM
 2019-2020 *Powerful Presentations*, Ph.D. level course. Three lectures + 1:1 student meetings. BCM
 2019-present *Concepts in Genetics and Genomics*, Ph.D. level course. One lecture.
 MD Anderson Cancer Center
 2020-present *Model Systems in Developmental Biology and Disease*. Ph.D. level course. 4 lectures. BCM
 2020 CNS Diseases (BIOH 441), B.S. level course, University of Montana
 2021-present *Animal Models of Human Disease*, Ph.D. level course. One lecture. BCM
 2022-present *CTR-CAQ Lecture Series*, Clinical Translational Research Certificate of Added Qualification,
 Ph.D. level course. One lecture. BCM

LECTURES (POST-GRADUATE EDUCATION LEVEL)

2023-present *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>

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 Reports; Cell Stress; Clinical and Translational Medicine; Current Biology; Circulation; Development; Developmental Biology; Developmental Cell; Disease Models and Mechanisms; eLife; European Journal of Medical Genetics (EJMG); Experimental Cell Research, 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; Mutation Research; Nature Cell Biology; Nucleic Acid Research, PLoS Biology; PLoS Genetics; PLoS One; Proceedings of the National Academy of Sciences (PNAS); 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 (UK, 2016, 2019); Cerebral Palsy Alliance Research Foundation (Australia, 2019); Czech Science Foundation (Czech Republic, 2020); European Research Counsel (European Commission, 2022), 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-CMND Study Section (USA, 2021 & 2023); National Institutes of Health-GHD Study Section (USA, 2024); Swiss National Science Foundation (Switzerland, 2022-2024); 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” 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-present)

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”, *Jikken Igaku (Experimental Medicine)*, Yodosha (Japan) (2021)

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

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

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

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

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

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

Editor of the book “*Drosophila* in Fundamental and Translational Neurobiology”, Editors: Shinya Yamamoto and Herman A. Dierick, Elsevier (Netherlands) (2025...Scheduled)

FUNDING

CURRENT SUPPORT

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

U54 OD030165 (ORIP) Heaney/Lee/Milosavljevic (PD/PI) 9/15/2020-9/14/2025

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: Co-I of the whole proposal, and Project Co-Lead of Resource and Services Section

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

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

#2023-32824 (CHAN ZUCKERBERG INITIATIVE) Gibbs/King/Bellen (PD/PI) 08/01/2023 - 07/31/2025

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 NS132415-02S01 subaward (NINDS) Kohane (PD/PI) 06/01/2024 - 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

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.**

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**U54 NS093793-07S1 (NINDS)**

Bellen (PD/PI) 7/01/2020-6/30/2021

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**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**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**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**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

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, Horii 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, Horii 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 1 β 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, Eiriksdottir 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, Tiffet 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. PMCID: 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**)
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, PMCID: 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, Cheadle-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, Tiffit 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, Willemssen 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, Faucher 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).
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, Dymont 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, Augsburg 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, Karkanas 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 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 Reports*. 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, Öunap 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 Et (2023). A comprehensive *Drosophila* genetic resource to study SARS-CoV-2 virus-host interactions in vivo. *Cell Reports*. 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. PMCID: 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, Grinton 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. 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*. doi: 10.1136/jmg-2024-110015 (Online ahead of print). PMID: 39327041. PMCID: N/A. (**†corresponding authors**).
94. 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, Batkovskyste 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*. doi: 10.1038/s41588-024-01941-1. Online ahead of print. PMID: 3943389. PMCID: N/A.

ARTICLES IN REVISION, SUBMITTED & IN PREPARATION

95. Deal SL, Bei D, Gibson SB, Delgado-Seo H, Fujita Y, Wilwayco K, Seto ES, **Yamamoto S†** (2024). RNAi-based screen for cuticle pigmentation in *Drosophila melanogaster* reveals novel regulators of brain dopamine. Submitted to *PLoS Biology*. Uploaded to *bioRxiv*. <https://doi.org/10.1101/2023.07.20.549932>. PMID: In Progress. PMCID: In Progress. (**†corresponding author**).
96. Park YJ, Lu TC, Jackson T, Goodman LD, Ran L, Chen J, Liang CY, Harrison E, Ko C, Hsu AL, **Yamamoto S**, Qi Y, Bellen HJ, Li H (2024). Whole organism snRNA-seq reveals systemic peripheral changes in Alzheimer's Disease fly models. In Review, *Cell*. Uploaded to *bioRxiv* <https://www.biorxiv.org/content/10.1101/2024.03.10.584317>. PMID: 38559164. PMCID: In Progress.
97. Mok J-W, Gibson SB, Dostalík SA, **Yamamoto S†** (2024). Functional assays in *Drosophila* as potential clinical tests to resolve variants of unknown significance found in people living with rare diseases. In Review, *Genome Research*. PMID: In Progress. PMCID: In Progress. (**†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/1XUsbagdutexAi/bibliography/public/>

-In addition, Shinya Yamamoto is listed as a consortium member/collaborator on the following additional papers published from the 'Undiagnosed Diseases Network' and 'Texome Project' in PubMed.

PMIDs: 27693232, 28132692, 28157539, 30773277, 31155284, 31327508, 31585109, 32197074, 33508234, 34450031, 34529933, 37451268, 37467750, 37451268, 38181735, 38387458 (*Am J Hum Genet*), 33184947, 33438828, 33847457, 33949769, 34042254, 34668327, 37246601, 37622199, 37743782, 38511854 (*Am J Med Genet A*), 37194416, 38311799, 38504481 (*Ann Clin Transl Neurol*), 34185323 (*Ann Neurol*), 35868845 (*Ann Rheum Dis*), 31443933 (*Biol Psychiatry*), 30134969 (*BMC Health Serv Res*), 31615419 (*BMC Bioinformatics*), 38279154 (*BMC Genomics*), 32761064, 36718090 (*Brain*), 35218524 (*Cerebellum*), 33963760 (*Clin Genet*), 31299614 (*Clin Imaging*), 29970384, 33811063 (*Cold Spring Harb Mol Case Stud*), 37565267 (*Dis Model Mech*), 30682224 (*Epilepsia*), 37500725 (*Eur J Hum Genet*), 33724412 (*Genetics*), 28914269, 29907797, 30568308, 33093671, 33239752, 33580225, 33833410, 34163037, 34230636, 35482014, 3641303, 37191094, 37947183, 38431799, 38436216, 38767059 (*Genet Med*), 33944996 (*Hum Genet*), 35913761, 37552066 (*Hum Mol Genet*), 31264822, 33239752, 33580225 (*Hum Mutat*), 33630084 (*JAMA Netw Open*), 33771552 (*J Allergy Clin Immunol*), 34009343 (*J Am Med Inform Assoc*), 35998814 (*J Biomed Inform*), 30706981, 30964584, 33108040, 34096130, 34115423, 34374469, 37005744 (*J Genet Couns*), 35621276 (*J Inherit Metab Dis*), 35318459 (*J Hum Genet*), 37316189 (*J Med Genet*), 37271495 (*J Pediatr*), 36984839 (*Metabolites*), 34702355 (*Mol Autism*), 30993913, 33159716, 33350591, 33955715, 33960148, 35247231 (*Mol Genet Genomic Med*), 34412939, 35144859 (*Mol Genet Metab*), 34211179 (*Nat Genet*), 33664507, 31160820 (*Nat Med*), 37653044 (*Nat Metab*), 37560121 (*Neurol Genet*), 36765070 (*NPJ Genom Med*), 28416019, 33971915, 36624503, 37032333, 37667351 (*Orphanet J Rare Dis*), 31439813 (*Postgrad Med J*), 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 through 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 (IDDR) 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 Galiko 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: Hiroataka 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, CureSATB2 & 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 and 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: *BMP2* 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 (2023/11/10)
- 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...scheduled)
- “Using *Drosophila* to discover and study new human disease genes”, Rice University, (Host: Kathleen Beckingham), 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)
- “Facilitating rare disease diagnosis through functional studies using *Drosophila*”, 2nd Undiagnosed Hackathon Tools Workshop, Radboud UMC, Nijmegen, the Netherlands (6/26/2024)
- “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 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 (Host: Dr. Kanae Ando), 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)
- 2025 “Using *Drosophila* to discover and study new human developmental disease genes”, 2025 Gordon Research Conference: Developmental Biology, Pomona, CA (2025/3/30...scheduled)
- “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 (2024/4...scheduled)

MENTORSHIP OF STUDENTS AND TRAINEES

GRADUATE (PHD) STUDENTS

- 2015-2021 Jose L. Salazar (Program in Molecular & Human Genetics, BCM)
Subsequent position: Technical Services Scientist, Oxford Nanopore Technologies
- 2018-2021 J. Michael Harnish (Program in Molecular & Human Genetics, BCM)
Subsequent position: Life Science Consultant, Clearview Healthcare Partners
- 2017-2023 Samantha Deal (Program in Developmental Biology, BCM)
Subsequent position: IRACDA Postdoctoral Fellow,
Amita Sehgal's lab, University of Pennsylvania & HHMI
- 2020- Shelley Gibson (Genetics and Genomics Graduate Program, BCM)
- 2022- Harmin Delgado-Seo (Neuroscience Graduate Program, BCM)
- 2023- Haley A. Dostalick (Genetics and Genomics Graduate Program, BCM)

POSTDOCTORAL FELLOWS

- 2016 Sumit Saurabh, Ph.D.
Subsequent position: Teaching Faculty, Loyola University
- 2019-2022 Sheng-An Yang, Ph.D.
Subsequent position: Staff Scientist, ClinGen Project, BCM
- 2021- Jung-Wang Mok, Ph.D.
- 2022- Mikiko Oka, Ph.D.
- 2022- Hirokazu Hashimoto, Ph.D.
- 2022-2024 Rei Yasuda, M.D., Ph.D.

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 Brown (PREP, 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)

RESEARCH ROTATIONS

- 2013 Yi-Chen Hsieh (Graduate Student Rotation, BCM)
- 2014 Krystal English (PREP Student Rotation, BCM)

2015 Valencia Potter (Graduate Student Rotation, BCM)
 2015 Yingyao Shao (Graduate Student Rotation, BCM)
 2017 Fatma Isleyen (Graduate Student Rotation, BCM)
 2018 Angad Jolly (Graduate Student Rotation, BCM)
 2018 J. Michael Harnish (Graduate Student Rotation, BCM)
 2018 Brooke Hull (PREP Student Rotation, BCM)
 2019 Morgan Stephens (Graduate Student Rotation, BCM)
 2019 Shelley Gibson (Graduate Student Rotation, BCM)
 2021 Julie Ann Goddard (Graduate Student Rotation, BCM)
 2022 Devine Jackson (Graduate Student Rotation, BCM)
 2022 Harim Delgado-Seo, Harim (Graduate Student Rotation, BCM)
 2022 Danielle Mendonca (Graduate Student Rotation, BCM)
 2022 Ellen Thompson (Graduate Student Rotation, BCM)
 2023 Haley A. Dostalík (Graduate Student Rotation, BCM)

THESIS ADVISORY COMMITTEES

2014-2018 Chang-Ru Tsai (PI: Michael J Galko, Program in Developmental Biology, BCM)
 2017-2018 Mumine Senturk (PI: Hugo J Bellen, Program in Developmental Biology, BCM),
 as ex-officio member
 2017-2019 Julia Wang [PI: Hugo J 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 J Bellen, Program in Molecular & Human Genetics, BCM)
 2018-2023 Daniel Sutton (PI: Andrew K Groves, Program in Molecular & Human Genetics, BCM)
 2018-2023 Meigen Yu (PI: Joshua M Shulman, Program in Neuroscience, BCM)
 2019-2024 Matthew Avalos (PI: Juan Botas, Program in Molecular & Human Genetics, BCM)
 2019-2022 Liping Wang (PI: Hugo J 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 and Genomics Graduate Program, BCM)
 2020- Megan Mair (PI: Juan Botas, Program in Molecular & Human Genetics, BCM)
 2020- Morgan C. Stephens (PI: Juan Botas, Genetics and Genomics Graduate Program, BCM)
 2020- Emily Leptich (PI: Rachel Arey, Neuroscience Graduate Program, BCM)
 2020- Laura Deus Ramírez (PI: Christoph Herman, Genetics and 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- Jennifer Deger (PIs: Joshua Shulman & Hugo J Bellen, Neuroscience Graduate Program, BCM)
 2022-2024 Guo Hu (PI: Meng Wang, Genetics and Genomics Graduate Program, BCM),
 as ex-officio member
 2022- Tyler Jackson (PI: Hongjie Li, Cancer and Cell Biology Graduate Program, BCM)

2022 Nirav Shah (PI: Jennifer Posey, Genetics and Genomics Graduate Program, BCM)
 2023- Jinghan Zhao (PI: Joshua Shulman, Neuroscience Graduate Program, BCM)
 2023- Ye-Jin Park (PIs: Hongjie Li & Hugo Bellen, DDMT Graduate Program, BCM)
 2023- Bing Xie (PI: James Martin, Genetics and Genomics Graduate Program, BCM)
 2023- Brandon Garcia (PI: Jennifer Posey, Genetics and Genomics and MSTP Programs, BCM)
 2024- Ellen Thompson (PI: Susan Rosenberg, DDMT Graduate Program, BCM)
 2024- Guo-Teng Liang (PI: Steven Boeynaemes, Neuroscience Graduate Program, BCM)
 2024- Keyang Yu (PI: Aleksandar Milosavljevic, Genetics and 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- Colleen Strohlein (PIs: Huda Zoghbi & Joshua Shulman, Neuroscience Graduate Program,
 BCM)
 2024- Sofia Ivana Aramburu [PI: Susan Rosenberg, DDMT Graduate Program, BCM]

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 and 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 and 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 and Human Genetics, BCM)
 2019 Megan Mair (Program in Molecular and Human Genetics, BCM)
 2020 Alice Wen (Program in Genetics and Genomics, BCM)
 2020 Kristy Jay (Program in Genetics and Genomics, BCM)
 2021 Emily Leptich (Program in Neuroscience, BCM, BCM)
 2021 Gary Huang (Program in Genetics and Genomics, BCM)
 2021 Fangfei Guo (Program in Genetics and Genomics, BCM)
 2022 Brooke Horist (Program in Genetics and Genomics, BCM)
 2022 Julie Ann Goddard (Program in Genetics and Genomics, BCM)
 2023 Jackson Tyler (Program in Cancer and Cell Biology, BCM)
 2023 Pragati Kore (Program in Genetics and 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 and Genomics, BCM)
 2024 Grace Pina (Program in Genetics and Genomics, BCM)

-Updated on 12/13/2024