

Kuan-lin Huang, Ph.D.

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MISSION

Empower every person to live healthily to 100-year-old by providing accurate & personalized prediction of disease risk, effective treatment, and health outcome.

APPOINTMENT/EMPLOYMENT

Assistant Professor, Genetics and Genomic Sciences 2018 October-Present
Faculty, Mount Sinai Center for Transformative Disease Modeling
Faculty, Icahn Genomics Institute
Associate Member, Tisch Cancer Institute
Icahn School of Medicine at Mount Sinai, NY, USA

Address: 1399 Park Avenue, 4 - 420-C, New York, NY 10029

Tel: (212) 824-6134 (x56134)

Lab website: www.ComputationalOmicsLab.org

Co-Founder and Chief Unboxer, Open Box Science (OpenBoxScience.Org) 2020 May-Present
Open Box Science (OBS) is a 501c3 non-for-profit that I started to provide a worldwide, open platform for science education and communication. The OBS community has hosted >200 webinars, breaking barriers to scientific knowledge and advances. My involvement with OBS is approved by Mount Sinai FCOI committee, so long I receive zero compensation from OBS.

GAPS IN EMPLOYMENT

Not applicable

EDUCATION

Postdoctoral Fellow, Department of Medicine, Washington University in St. Louis, MO 2018 September
Advisor: Dr. Li Ding (Multi-Omics Algorithms)

Ph.D., Genetics and Genomics, Washington University in St. Louis, MO 2018 February
Dissertation: *Multi-omics Portraits of Cancer*
Advisor: Dr. Li Ding (Cancer Genomics, 2014-2018)
Drs. Alison M. Goate and Carlos Cruchaga (Statistical Genetics & Alzheimer's Disease, 2013-2014)
Awards: Taiwanese Ministry of Education Ph.D. Scholarship / Lucille P. Markey Pathway in Human Pathobiology

B.A., Wesleyan University, Middletown, CT 2012 May
High Honors. Thesis research in Molecular Biology & Biochemistry (Advisor: Dr. Scott G. Holmes)
Honors. Thesis installation exhibition in Studio Art (Advisor: Jeffrey Schiff)
Awards: Freeman Full-Ride Four-Year Scholarship (1~2 Taiwanese students/year) / Howard Hughes Summer Research Fellowships / Scott Biomedical Prize / the only person in class awarded 2 *Honors*

CERTIFICATION

Not applicable

LICENSURE

Not applicable

HONORS/AWARDS

1. **Mount Sinai Alzheimer's Disease Research Center (ADRC) Scholar** [Institutional] 2023
2. **NIGMS Maximizing Investigators' Research Award (MIRA) for ESI** [National] 2020-2025
3. **Alzheimer's Association International Conference (AAIC) Fellowship** [International] 2016
4. **First Place Winner, Skandalaris Healthcare Hackathon** [Regional] 2015
5. **Taiwanese Ministry of Education Ph.D. Scholarship** [National] 2014-2015

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| 6. Lucille P. Markey Pathway in Human Pathobiology [Institutional] | 2014 |
| 7. Honors (Studio Art) & High Honors (Molecular Biology) Dissertations, Wesleyan University [Institutional] | 2012 |
| The only person in the 2012 class to receive two <i>honors</i> designations | |
| 8. Scott Biomedical Prize, Wesleyan University [Institutional] | 2012 |
| Awarded to one graduating senior showing strong promise in biomedical research | |
| 9. Howard Hughes Undergraduate Research Scholarship [National] | 2010, 2011 |
| Awarded two terms to support full-time summer undergraduate research in molecular genetics | |
| 10. Freeman Asian Full-Ride Scholarship for Wesleyan University, CT [International] | 2008-2012 |
| Merit-based, full-ride scholarship awarded to 1~2 Taiwanese students per year to attend Wesleyan University | |
| 11. Math and Science Gifted Program at Chien-Kuo Senior High School, Taiwan [National] | 2005-2008 |
| Admitted to the most competitive academic program (60 students/yr) in the lowest-acceptance-rate high school in Taiwan | |
| 12. Youth Athlete-Diplomat Representing Taiwan for the 2006 Asian Games, Doha, Qatar [National] | 2006 |
| Chosen as 1 of the 4 Youth Athlete-Diplomat representing Taiwan for the 2006 Asian Games | |

PATENTS

Not applicable

OTHER ENTREPRENEURIAL ACTIVITIES

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|---|--------------|
| 1. Co-Founder and Chief Unboxer , Open Box Science 501(c)(3) | 2020-Present |
| 2. Co-Founder and CEO , DeepGene LLC | 2016-2017 |

OTHER PROFESSIONAL ROLES

Active Peer-Reviewer: average 15 manuscripts/year, * indicates multiple times

Grant Proposals

Ad Hoc Peer-Review: Arizona Alzheimer's Disease Core Center Grant* / Wellcome Trust International Fellowships

Research Manuscripts

Guest Associate Editor: Frontiers in Aging Neuroscience (Topic: Advancing Clinical Neuroscience by Multi-Omic Driven Approaches Towards Personalized Medicine: Opportunity, Challenges, and the Future)

Ad Hoc Peer-Review: Bioinformatics* / BMC Bioinformatics / BMC Genomics / BMC Neurology / BMJ Medicine / Cell Reports / Cell Reports Medicine* / Clinical Proteomics / Communications Medicine / EMBO Molecular Medicine / Frontiers in Genetics / Genome Medicine* / Human Molecular Genetics / iScience / Molecular and Cellular Proteomics / Nature Cancer / Nature Communications* / Nature Medicine / npj Digital Medicine / PLOS Computational Biology

Active Memberships for Professional Societies: American Association for Cancer Research (AACR)/ International Society to Advance Alzheimer's Research and Treatment (ISTAART)/ International Society for Computational Biology (ISCB)

RESEARCH PROFILE

I am a human genomicist with track records in statistical genetics, DNA-sequencing (DNA-Seq) studies, proteomic biomarkers, multi-omic algorithm and software development. My research aims to provide accurate risk assessment and treatment options based on each individual's genome. In my Ph.D. and post-doc, I served as the lead analyst for multiple national/international consortium projects, including IGAP (GWAS), TCGA PanCanAtlas Germline Working Group (DNA-Seq), and CPTAC (Proteomics). My first-author contributions (Huang et al.) include genome-wide association studies (GWAS) of complex diseases (Nature Neuro 2017), large-scale DNA-Seq analyses of cancer (Cell 2018), proteogenomics integration for patient-derived models (Nature Comm 2017), and software for analyzing multi-omic data (MCP 2019, Nature Comm 2021). The genomic datasets I processed and the bioinformatic tools I developed have been cited and used in thousands of other studies.

I started my independent laboratory at ISMMS in Oct 2018, and my research program is guided by three principles: (1) be inclusive of diverse populations, (2) be comprehensive of multi-modal big data, and (3) catch it early for personalized prevention and interception of disease etiologies. As of March 2023, I have published 16 last-author research articles since ISMMS, and have an H-index: 34, Citations: 10,650 based on Google Scholar. My lab's primary contribution to science includes unveiling pathogenic variants in diverse populations, developing statistical software for multi-omics big data, identifying genetic predictors of treatment outcomes, and constructing machine-learning (ML) models to predict disease risks and treatments using personalized multi-omics.

CLINICAL PROFILE

Not applicable

MENTORSHIP PROFILE

Since I started my lab at ISMMS, I have served as primary mentors for 4 postdoc fellows, 1 hem/onc fellow, 1 PhD student, and 5 MS students whose primary affiliation is at ISMMS. My lab also hosts summer students (volunteer or DEI programs) and MD students for research project each year. External trainees who have completed first-author papers for which I am the primary mentor (last-author) include 1 high-school student, 2 undergrads, 2 MD students, 2 PhD students, 1 postdoc. Additionally, I serve on thesis committees for 5 PhD students and 3 MS students at ISMMS. My trainees come from diverse backgrounds, countries, and religions; internal/external trainees have gone on to successful careers in world-renowned companies and academic institutions after training, including Harvard, Columbia, Stanford, St. Jude, ISMMS, and Genentech.

I continue to hone my mentorship skills through reading, feedback from trainees, and mentors. I see mentorship as how I can contribute to growing a forest for the larger scientific community. Trees reach for the sky, like how budding researchers explore and advance in their scientific pursuits. As a tree planter, my job is to help them thrive: identify and amplify directions where they are growing best while helping to avoid dead ends. To provide the needed nutrients and water, but also guide them to where they could derive needed resources independently.

DIVERSITY AND INCLUSION IMPACT

I advocate for diversity, equity, and inclusion (DEI) at my lab, institution, and non-for-profit. In 2018, the initial 8 postdocs, graduate, and medical students in my lab represent 7 different nationalities and 6 major religion groups. I establish lab guidelines to ensure everyone's voice is heard. For example, in each lab meeting, the presenter warm-up the floor by asking an open question, where every member is required to provide a perspective. In any presentation, everyone is required to speak up at least once. At the institutional level, I promote these values by actively participating in departmental and/or institutional DEI initiatives, including hosting and lecturing for DEI scholars each year.

More, I am passionate about addressing cross-country DEI challenges in science. To provide some context, the US ranks 10th in 198 countries in GDP (\$76,027), compared to the bottom 47 countries with GDP of less than \$5,000. In 2020, I recruited two friends and co-founded an 501(c)(3) non-profit for open science & communication. In two years, the NGO has hosted 200+ free talks for a worldwide audience spanning over 70 countries. The OBS community follows a guideline of inviting early-career researchers (i.e., first authors) from diverse backgrounds, and we constantly broadcast our free events to discipline-specific interest groups in Latin America, Asia, Europe, African, and Australia.

Together, we aim to build a future where scientists have equal access to knowledge, training, and can communicate seamlessly across boundaries.

OVERALL IMPACT

As a human genomicist, my research goal is to provide accurate risk assessment and treatment options based on an individual's genome. My first-author contribution to science include GWAS, DNA-Seq, proteogenomic studies of complex diseases (cancer and neurodegeneration) as well as development of several software for analyzing multi-omic data. The genomic datasets I processed and the bioinformatic tools I developed have been cited and used in thousands of other studies. Since starting my independent laboratory at ISMMS in 2018, I have published 16 last-author research articles, with an H-index of 34 and 10,650 citations based on Google Scholar. I have also mentored a diverse group of trainees who have gone on to successful careers in world-renowned companies and academic institutions. Additionally, I am passionate about promoting diversity, equity, and inclusion (DEI) in science, and have co-founded a non-profit for open science and communication, which has hosted over 200 free talks for a worldwide audience spanning over 70 countries.

GRANTS, CONTRACTS, FOUNDATION SUPPORT

PAST GRANTS

Funding Source/Project Title	Role in Project	Dates	Total Costs	Supplemental Info
Google Cloud Pilot Grant (Huang) A Web-based Simulator for Pandemic Containment Strategies	PI	4/1/2020 - 10/1/2020	\$14,000	
Mount Sinai Genetics and Genomic Sciences Pilot Grant (Gang) A Novel Epigenetic Mark in Alzheimer Disease and Aging	Co-I	10/1/2019 - 10/1/2020	\$50,000	

CURRENT GRANTS

Funding Source/Project Title	Role in Project	Dates	Total Costs	Supplemental Info
American Cancer Society RSG-22-115-01-DMC (Huang)	PI	01/01/2023-12/31/2026	\$757,857	

Pan-ancestry Identification of Pathogenic Variants affecting DNA Repair				
Mount Sinai FBI/ADRC Mount Sinai ADRC Scholar Award (Elahi, Huang, Goate) Unbiased Discovery of Early Molecular Dysregulations in Alzheimer's Disease	M-PI	01/01/2023-12/31/2023	\$60,000	
NIH NIGMS R35 GM138113 (Huang) Integrative Approaches for Identifying Causal Gene-Cell Type Pairs of Complex Disease	PI	9/15/2020-7/31/2025	\$2,115,345	
NIH NINDS R01 NS116006 (Raj) The Role of Myeloid Cells in Parkinson's Disease	Co-I	4/1/2021-3/31/2026	\$3,398,901	
NIH NIA R01 AG072300 (Castellano) Mechanisms of TIMP2-mediated hippocampal revitalization in Alzheimer's disease	Co-I	5/1/2021-04/31/2024	\$1,518,274	

PENDING GRANTS

Funding Source/Project Title	Role in Project	Dates	Total Costs	Supplemental Info
NIH NIA U01 AG079849 (Goate, Renton, Marcora, O'Reilly) Elucidation of brain cell type-specific Alzheimer's disease genetic risk mechanisms in diverse populations	Co-I	7/1/2023 - 6/30/2028	\$12,865,629	
NIH NHGRI R01HG013085 (Do, Itan) Towards building an atlas of variant function for clinical interpretation	Co-I	7/1/2023 - 6/30/2028	\$4,209,637	
NIH NCI U01CA282267 (Huang, Lucas, O'Reilly) Genetics-informed Biomarker Strategy for Pancreatic Cancer Detection	M-PI (contact)	7/1/2023 - 6/30/2028	\$5,062,276	
NIH NLM R01LM14391 (Song) Multi-scale modelling of cross-modal, high-resolution molecular data to dissect aberrant disease mechanisms in tissue ecosystem	Co-I	12/1/2023 - 11/30/2027	\$1,679,230	
NIH NCI R01CA286073 (Dominguez-Sola) Genetics-informed Biomarker Strategy for Pancreatic Cancer Detection	Co-I	9/1/2023 - 8/31/2028	\$3,776,455	
NIH NIA R01AG (O'Reilly, Huang) A pathway-driven approach to stratified medicine for AD	M-PI	12/1/2023 - 11/30/2028	\$4,205,575	

CLINICAL TRIALS PARTICIPATION

Not applicable

TRAINEES

Name	Level of Trainee	Role in Training [Date]	Training Venue	Trainee's Immediate Outcome
Jordan Rosen, BS	MS student	Master's research mentor [2022-]	ISMMS	
Nathalie Ramos, BS	MS student	Master's research mentor [2022-]	ISMMS	
Megan Wojciechowicz, BS	PhD student	PhD advisor [2022-]	ISMMS	
Tefike Okotete	Undergraduate student	Research mentor for intern projects [2022]	ISMMS (trainee at NYU)	Continuing undergraduate studies at NYU

Raj Vaza, BS	MS student	Master's research mentor [2022-]	ISMMS	
Yuqi Liu	Undergraduate student	Research mentor for volunteer projects [2021-]	ISMMS (trainee at NYU)	
Michael Wang, MD	PhD student	Research mentor for visiting scholar [2019-2022]	ISMMS (trainee at PUMC)	Resident Surgeon, Peking Union Medical College
Tzipora Weinberger, BS	Master's student	Master's research mentor [2020-2022]	ISMMS	Data Scientist, Hackensack Meridian Health
Rikhiya Ghosh, PhD	Postdoc	Postdoc mentor [2021-]	ISMMS	
Jimmy Zhang	High school student	Research mentor [2019-2021]	ISMMS (trainee at Queens High School for the Sciences)	Undergraduate, Columbia University
Tomi Jun, MD	Hem/onc Fellow	Research mentor [2019-2021]	ISMMS	Medical Director, SEMA4 Genentech (current)
Zishan Wang, PhD	Postdoc	Postdoc mentor [2019-]	ISMMS	
Jing Wang, PhD	Postdoc	Postdoc mentor [2019]	ISMMS	Data Scientist, Amazon
Abdulkadir Elmas, PhD	Postdoc	Postdoc mentor [2019-]	ISMMS	
William Lee, BS	Master's student	Master's research mentor [2019-2020]	ISMMS	Bioinformatician, ISMMS (w/ Andrew Sharp)
Tao Qing, PhD	Postdoc	Advised on collaborative projects with Dr. Lajos Pustzai [2019-2022]	ISMMS (trainee at Yale)	Senior Scientist, Freenome
Katherine Houlahan, PhD	PhD student	Advised on collaborative projects with Dr. Paul Boutros [2019-2021]	ISMMS (trainee at UCLA)	Postdoc, Stanford (w/ Christina Curtis)
Guanlan Dong, BS	Undergraduate student	Research mentor for volunteer projects [2019-2020]	ISMMS (trainee at WashU)	Harvard B.I.G. PhD program
Ninad Oak, PhD	PhD student	Advised on collaborative projects with Dr. Sharon Plon [2018-2020]	ISMMS (trainee at Baylor)	Scientist, St. Jude Children's Hospital

PhD Dissertation Committee:

Yihan Zhao (Genetics and Genomic Sciences)/**Nicole Zatorski** (Pharmacology and Therapeutics Discovery)/**Miriam Saffern** (Immunology)/**Aanay Shah** (Clinical and Translational Research)/**Julia Zhao** (Genetics and Genomic Sciences) /**Katrin Bauer** (external, University Medical Center Mainz)

MS Dissertation Committee:

Nile Rizvi (Biomedical Sciences) / **Gauri Ganesh** (Biomedical Sciences) / **Andy Yang** (Biomedical Sciences)

PhD Rotation Students:

Miriam Saffern (Immunology, 2020)/ **Rebecca Signer** (Genetics and Genomic Sciences, 2021)/ **Hannah Kittrell** (Genetics and Genomic Sciences, 2022)/ **Megan Wojciechowicz** (Pharmacology and Therapeutics Discovery, 2022)/**Eric Park** (Genetics and Genomic Sciences, 2022)

Medical Students (mostly second semester of M1 ~ first semester of M2):

Prag Batra (2018-2020), **Serena Tharakan** (2019-2020), **Alexandra Capellini** (2019-2020), **Matthew Williams** (2019-2020), **Daniel Fulop** (2019-2022), **Makda Zewde** (2020-), **Suraj Jaladanki** (2019-2021), **Gabriel Santos Malave** (2020-2021), **Chang Moon** (2020-2021), **Luo Song** (2021-2022), **Varun Subramaniam** (2022-), **Kevin Spehar** (2022-), **Yash Lahoti** (2023-)

TEACHING ACTIVITIES

1. **Teaching Assistant and Guest Lecturer**, Washington University in Saint Louis, MO 2014-2017
2. **Community Health Project Founder and Leader**, Matibabu Foundation, Kenya 2013
3. **English Instructor** for compulsory substitute military service, Zhuwei Elementary School, Taiwan 2012-2013
4. **Teaching Assistant**, Wesleyan University, CT 2010-2011

ADMINISTRATIVE LEADERSHIP APPOINTMENTS

Abbreviations: Mount Sinai GGS (Department of Genetics and Genomic Sciences), Mount Sinai TDM (Center for Transformative Disease Modeling)

EDUCATION

Co-lead, GGS Departmental Cancer Genomics WIP, Icahn School of Medicine at Mount Sinai 2021-Present

GENERAL ADMINISTRATION

Member, GGS Faculty Search Committee , Icahn School of Medicine at Mount Sinai	2022-Present
Member, GGS Faculty Engagement Committee , Icahn School of Medicine at Mount Sinai	2021-Present
Member, GGS Faculty Mentorship Committee , Icahn School of Medicine at Mount Sinai	2021-Present
Member, Postdoc Advisory Committee , Icahn School of Medicine at Mount Sinai	2020-2022

PUBLICATIONS

H-index: 34 (accessed 3/25/2023), Citations: 10,650 (Google Scholar)

Peer-Reviewed Original Contributions [*Co-first authors. #Co-corresponding authors. %Consortium authorship]:

Summary of peer-reviewed research articles: 5 first-author (Nat Comm 2017, Nat Neuro 2017, Cell 2018, MCP 2019, Nat Comm 2021), 2 co-first author (Neoplasma 2016, Cancer Cell 2020). Since ISMMS appointment in 2018, 16 last-author research articles.

1. Yeast Tdh3 (glyceraldehyde 3-phosphate dehydrogenase) is a Sir2-interacting factor that regulates transcriptional silencing and rDNA recombination.
Ringel AE, Ryznar R, Picariello H, **Huang KL**, Lazarus AG, Holmes SG.
PLoS Genet. 2013;9(10):e1003871. doi: 10.1371/journal.pgen.1003871. Epub 2013 Oct 17. PMID: 24146631
2. Patterns and functional implications of rare germline variants across 12 cancer types.
Lu C*, Xie M*, Wendl MC*, Wang J*, McLellan MD*, Leiserson MD*, **Huang KL**, Wyczalkowski MA, Jayasinghe R, Banerjee T, Ning J, Tripathi P, Zhang Q, Niu B, Ye K, Schmidt HK, Fulton RS, McMichael JF, Batra P, Kandath C, Bharadwaj M, Koboldt DC, Miller CA, Kanchi KL, Eldred JM, Larson DE, Welch JS, You M, Ozenberger BA, Govindan R, Walter MJ, Ellis MJ, Mardis ER, Graubert TA, Dipersio JF, Ley TJ, Wilson RK, Goodfellow PJ, Raphael BJ, Chen F, Johnson KJ, Parvin JD, Ding L.
Nat Commun. 2015 Dec 22;6:10086. doi: 10.1038/ncomms10086. PMID: 26689913 **[Covered by 17 news outlets]**
3. Pan-cancer methylation and expression profiling of adenocarcinomas revealed epigenetic silencing in the WNT signaling pathway.
Li J*, **Huang KL***, Zhang T, Li H, Zhao J, Wang H.
Neoplasma 2016; 63 (2), 208-214
4. Systematic discovery of complex insertions and deletions in human cancers.
Ye K, Wang J, Jayasinghe R, Lameijer EW, McMichael JF, Ning J, McLellan MD, Xie M, Cao S, Yellapantula V, **Huang KL**, Scott A, Foltz S, Niu B, Johnson KJ, Moed M, Slagboom PE, Chen F, Wendl MC, Ding L.
Nat Med. 2016 Jan;22(1):97-104. doi: 10.1038/nm.4002. Epub 2015 Dec 14. PMID: 26657142 **[Covered by 9 news outlets]**
5. Proteogenomics connects somatic mutations to signalling in breast cancer.
Mertins P, Mani DR, Ruggles KV, Gillette MA, Clauser KR, Wang P, Wang X, Qiao JW, Cao S, Petralia F, Kawaler E, Mundt F, Krug K, Tu Z, Lei JT, Gatzka ML, Wilkerson M, Perou CM, Yellapantula V, **Huang KL**, Lin C, McLellan MD, Yan P, Davies SR, Townsend RR, Skates SJ, Wang J, Zhang B, Kinsinger CR, Mesri M, Rodriguez H, Ding L, Paulovich AG, Fenyö D, Ellis MJ, Carr SA; NCI CPTAC.
Nature. 2016 Jun 2;534(7605):55-62. doi: 10.1038/nature18003. Epub 2016 May 25. PMID: 27251275 **[Covered by 4 news outlets]**
6. Chitinase-3-like 1 protein (CHI3L1) locus influences cerebrospinal fluid levels of YKL-40.
Deming Y, Black K, Carrell D, Cai Y, Del-Aguila JL, Fernandez MV, Budde J, Ma S, Saef B, Howells B, Bertelsen S, **Huang KL**, Sutphen CL, Tarawneh R, Fagan AM, Holtzman DM, Morris JC, Goate AM, Dougherty JD, Cruchaga C.
BMC Neurol. 2016 Nov 10;16(1):217. doi: 10.1186/s12883-016-0742-9. PMID: 27832767
7. Proteogenomic integration reveals therapeutic targets in breast cancer xenografts.
Huang KL*, Li S*, Mertins P*, Cao S, Gunawardena HP, Ruggles KV, Mani DR, Clauser KR, Tanioka M, Usary J, Kavuri SM, Xie L, Yoon C, Qiao JW, Wrobel J, Wyczalkowski MA, Erdmann-Gilmore P, Snider JE, Hoog J, Singh P, Niu B, Guo Z, Sun SQ, Sanati S, Kawaler E, Wang X, Scott A, Ye K, McLellan MD, Wendl MC, Malovannaya A, Held JM, Gillette MA, Fenyö D, Kinsinger CR, Mesri M, Rodriguez H, Davies SR, Perou CM, Ma C, Reid Townsend R, Chen X, Carr SA, Ellis MJ, Ding L.
Nat Commun. 2017 Mar 28;8:14864. doi: 10.1038/ncomms14864. PMID: 28348404 **[Covered by 8 news outlets]**
8. Genome-wide association study identifies four novel loci associated with Alzheimer's endophenotypes and disease modifiers.
Deming Y, Li Z, Kapoor M, Harari O, Del-Aguila JL, Black K, Carrell D, Cai Y, Fernandez MV, Budde J, Ma S, Saef B, Howells B, **Huang KL**, Bertelsen S, Fagan AM, Holtzman DM, Morris JC, Kim S, Saykin AJ, De Jager PL, Albert M, Moghekar A, O'Brien R, Riemenschneider M, Petersen RC, Blennow K, Zetterberg H, Minthon L, Van Deerlin VM, Lee VM, Shaw LM, Trojanowski JQ, Schellenberg G, Haines JL, Mayeux R, Pericak-Vance MA, Farrer LA, Peskind ER, Li G, Di Narzo AF; Alzheimer's Disease Neuroimaging Initiative (ADNI); Alzheimer Disease Genetic Consortium (ADGC); Kauwe JS, Goate AM, Cruchaga C.

- Acta Neuropathol. 2017 May;133(5):839-856. doi: 10.1007/s00401-017-1685-y. Epub 2017 Feb 28. PMID: 28247064
9. GenomeVIP: a cloud platform for genomic variant discovery and interpretation.
Mashl RJ, Scott AD, **Huang KL**, Wyczalkowski MA, Yoon CJ, Niu B, DeNardo E, Yellapantula VD, Handsaker RE, Chen K, Koboldt DC, Ye K, Fenyö D, Raphael BJ, Wendl MC, Ding L.
Genome Res. 2017 Aug;27(8):1450-1459. doi: 10.1101/gr.211656.116. Epub 2017 May 18. PMID: 28522612
 10. A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease.
Huang KL, Marcora E, Pimenova AA, Di Narzo AF, Kapoor M, Jin SC, Harari O, Bertelsen S, Fairfax BP, Czajkowski J, Chouraki V, Grenier-Boley B, Bellenguez C, Deming Y, McKenzie A, Raj T, Renton AE, Budde J, Smith A, Fitzpatrick A, Bis JC, DeStefano A, Adams HHH, Ikram MA, van der Lee S, Del-Aguila JL, Fernandez MV, Ibañez L; International Genomics of Alzheimer's Project; Alzheimer's Disease Neuroimaging Initiative; Sims R, Escott-Price V, Mayeux R, Haines JL, Farrer LA, Pericak-Vance MA, Lambert JC, van Duijn C, Launer L, Seshadri S, Williams J, Amouyel P, Schellenberg GD, Zhang B, Borecki I, Kauwe JSK, Cruchaga C, Hao K, Goate AM.
Nat Neurosci. 2017 Aug;20(8):1052-1061. doi: 10.1038/nn.4587. Epub 2017 Jun 19. PMID: 28628103 **[Covered by 10 news outlets]**
 11. Breast tumors educate the proteome of stromal tissue in an individualized but coordinated manner.
Wang X, Mooradian AD, Erdmann-Gilmore P, Zhang Q, Viner R, Davies SR, **Huang KL**, Bomgardner R, Van Tine BA, Shao J, Ding L, Li S, Ellis MJ, Rogers JC, Townsend RR, Fenyö D, Held JM.
Sci Signal. 2017 Aug 8;10(491):eaam8065. doi: 10.1126/scisignal.aam8065. PMID: 28790197
 12. Perspective on Oncogenic Processes at the End of the Beginning of Cancer Genomics.
Ding L, Bailey MH, Porta-Pardo E, Thorsson V, Colaprico A, Bertrand D, Gibbs DL, Weerasinghe A, **Huang KL**, Tokheim C, Cortés-Ciriano I, Jayasinghe R, Chen F, Yu L, Sun S, Olsen C, Kim J, Taylor AM, Cherniack AD, Akbani R, Suphavilai C, Nagarajan N, Stuart JM, Mills GB, Wyczalkowski MA, Vincent BG, Hutter CM, Zenklusen JC, Hoadley KA, Wendl MC, Shmulevich L, Lazar AJ, Wheeler DA, Getz G; Cancer Genome Atlas Research Network.
Cell. 2018 Apr 5;173(2):305-320.e10. doi: 10.1016/j.cell.2018.03.033. PMID: 29625049 **[Covered by 21 news outlets]**
 13. Pathogenic Germline Variants in 10,389 Adult Cancers.
Huang KL, Mashl RJ, Wu Y, Ritter DI, Wang J, Oh C, Paczkowska M, Reynolds S, Wyczalkowski MA, Oak N, Scott AD, Krassowski M, Cherniack AD, Houlahan KE, Jayasinghe R, Wang LB, Zhou DC, Liu D, Cao S, Kim YW, Koire A, McMichael JF, Huchtagowder V, Kim TB, Hahn A, Wang C, McLellan MD, Al-Mulla F, Johnson KJ; Cancer Genome Atlas Research Network; Lichtarge O, Boutros PC, Raphael B, Lazar AJ, Zhang W, Wendl MC, Govindan R, Jain S, Wheeler D, Kulkarni S, Dipersio JF, Reimand J, Meric-Bernstam F, Chen K, Shmulevich I, Plon SE, Chen F, Ding L.
Cell. 2018 Apr 5;173(2):355-370.e14. doi: 10.1016/j.cell.2018.03.039. PMID: 29625052 **[Covered by 28 news outlets]**
 14. Pan-cancer analysis of somatic mutations across 21 neuroendocrine tumor types.
Cao Y, Zhou W, Li L, Wang J, Gao Z, Jiang Y, Jiang X, Shan A, Bailey MH, **Huang KL**, Sun SQ, McLellan MD, Niu B, Wang W, Ding L, Ning G.
Cell Res. 2018 May;28(5):601-604. doi: 10.1038/s41422-018-0019-5. Epub 2018 Mar 5. PMID: 29507395
 15. Mass Spectrometry-Based Proteomics Reveals Potential Roles of NEK9 and MAP2K4 in Resistance to PI3K Inhibition in Triple-Negative Breast Cancers.
Mundt F, Rajput S, Li S, Ruggles KV, Mooradian AD, Mertins P, Gillette MA, Krug K, Guo Z, Hoog J, Erdmann-Gilmore P, Primeau T, Huang S, Edwards DP, Wang X, Wang X, Kawaler E, Mani DR, Clauser KR, Gao F, Luo J, Davies SR, Johnson GL, **Huang KL**, Yoon CJ, Ding L, Fenyö D, Ellis MJ, Townsend RR, Held JM, Carr SA, Ma CX.
Cancer Res. 2018 May 15;78(10):2732-2746. doi: 10.1158/0008-5472.CAN-17-1990. Epub 2018 Feb 22. PMID: 29472518
 16. Integrative omics analyses broaden treatment targets in human cancer.
Sengupta S*, Sun SQ*, **Huang KL**, Oh C, Bailey MH, Varghese R, Wyczalkowski MA, Ning J, Tripathi P, McMichael JF, Johnson KJ, Kandoth C, Welch J, Ma C, Wendl MC, Payne SH, Fenyö D, Townsend RR, Dipersio JF, Chen F, Ding L.
Genome Med. 2018 Jul 27;10(1):60. doi: 10.1186/s13073-018-0564-z. PMID: 30053901
 17. Framework for microRNA variant annotation and prioritization using human population and disease datasets.
Oak N, Ghosh R, **Huang KL**, Wheeler DA, Ding L, Plon SE.
Hum Mutat. 2019 Jan;40(1):73-89. doi: 10.1002/humu.23668. Epub 2018 Nov 8. PMID: 30302893 **[Covered by 7 news outlets]**
 18. CharGer: clinical Characterization of Germline variants.
Scott AD, **Huang KL**, Weerasinghe A, Mashl RJ, Gao Q, Martins Rodrigues F, Wyczalkowski MA, Ding L.
Bioinformatics. 2019 Mar 1;35(5):865-867. doi: 10.1093/bioinformatics/bty649. PMID: 30102335 **[Covered by 1 news outlet]**
 19. Functional analysis of BARD1 missense variants in homology-directed repair and damage sensitivity.
Adamovich AI, Banerjee T, Wingo M, Duncan K, Ning J, Martins Rodrigues F, **Huang KL**, Lee C, Chen F, Ding L, Parvin JD.
PLoS Genet. 2019 Mar 29;15(3):e1008049. doi: 10.1371/journal.pgen.1008049. eCollection 2019 Mar. PMID: 30925164
 20. Regulated Phosphosignaling Associated with Breast Cancer Subtypes and Druggability.

Huang KL[#], Wu Y, Primeau T, Wang YT, Gao Y, McMichael JF, Scott AD, Cao S, Wendl MC, Johnson KJ, Ruggles K, Held J, Payne SH, Davies S, Dar A, Kinsinger CR, Mesri M, Rodriguez H, Ellis MJ, Townsend RR, Chen F, Fenyö D, Li S, Liu T, Carr SA, Ding L[#].

Mol Cell Proteomics. 2019 Aug;18(8):1630-1650. doi: 10.1074/mcp.RA118.001243. Epub 2019 Jun 13. PMID: 31196969

21. Mannose Phosphate Isomerase and Mannose Regulate Hepatic Stellate Cell Activation and Fibrosis in Zebrafish and Humans. DeRossi C, Bambino K, Morrison J, Sakarin I, Villacorta-Martin C, Zhang C, Ellis JL, Fiel MI, Ybanez M, Lee YA, **Huang KL**, Yin C, Sakaguchi TF, Friedman SL, Villanueva A, Chu J. Hepatology. 2019 Dec;70(6):2107-2122. doi: 10.1002/hep.30677. Epub 2019 May 24. PMID: 31016744

***For reference 22-42, I was acknowledged as part of the ICGC Pan-Cancer Analysis of Whole Genomes Consortium (PCAWG) based on my work to help test and establish the WashU variant-calling pipeline that helped generate the PCAWG genomic mutation data going into these projects. Otherwise, for most these papers, I did not directly contribute to other parts.

22. Pan-cancer analysis of whole genomes.

ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium[®].

Nature. 2020 Feb;578(7793):82-93. doi: 10.1038/s41586-020-1969-6. Epub 2020 Feb 5. PMID: 32025007 [**Covered by 44 news outlets**]

23. Patterns of somatic structural variation in human cancer genomes.

Li Y, Roberts ND, Wala JA, Shapira O, Schumacher SE, Kumar K, Khurana E, Waszak S, Korbel JO, Haber JE, Imielinski M; PCAWG Structural Variation Working Group; Weischenfeldt J, Beroukhim R, Campbell PJ; **PCAWG Consortium**[®].

Nature. 2020 Feb;578(7793):112-121. doi: 10.1038/s41586-019-1913-9. Epub 2020 Feb 5. PMID: 32025012

24. The evolutionary history of 2,658 cancers.

Gerstung M, Jolly C, Leshchiner I, Dentro SC, Gonzalez S, Rosebrock D, Mitchell TJ, Rubanova Y, Anur P, Yu K, Tarabichi M, Deshwar A, Wintersinger J, Kleinheinz K, Vázquez-García I, Haase K, Jerman L, Sengupta S, Macintyre G, Malikic S, Donmez N, Livitz DG, Cmero M, Demeulemeester J, Schumacher S, Fan Y, Yao X, Lee J, Schlesner M, Boutros PC, Bowtell DD, Zhu H, Getz G, Imielinski M, Beroukhim R, Sahinalp SC, Ji Y, Peifer M, Markowitz F, Mustonen V, Yuan K, Wang W, Morris QD; PCAWG Evolution & Heterogeneity Working Group; Spellman PT, Wedge DC, Van Loo P; **PCAWG Consortium**[®].

Nature. 2020 Feb;578(7793):122-128. doi: 10.1038/s41586-019-1907-7. Epub 2020 Feb 6. PMID: 32025013

25. Analyses of non-coding somatic drivers in 2,658 cancer whole genomes.

Rheinbay E, Nielsen MM, Abascal F, Wala JA, Shapira O, Tiao G, Hornshøj H, Hess JM, Juul RI, Lin Z, Feuerbach L, Sabarinathan R, Madsen T, Kim J, Mularoni L, Shuai S, Lanzós A, Herrmann C, Maruvka YE, Shen C, Amin SB, Bandopadhyay P, Bertl J, Boroevich KA, Busanovich J, Carlevaro-Fita J, Chakravarty D, Chan CWY, Craft D, Dhingra P, Diamanti K, Fonseca NA, Gonzalez-Perez A, Guo Q, Hamilton MP, Haradhvala NJ, Hong C, Isaev K, Johnson TA, Juul M, Kahles A, Kahraman A, Kim Y, Komorowski J, Kumar K, Kumar S, Lee D, Lehmann KV, Li Y, Liu EM, Lochovsky L, Park K, Pich O, Roberts ND, Saksena G, Schumacher SE, Sidiropoulos N, Sieverling L, Sinnott-Armstrong N, Stewart C, Tamborero D, Tubio JMC, Umer HM, Uusküla-Reimand L, Wadelius C, Wadi L, Yao X, Zhang CZ, Zhang J, Haber JE, Hobolth A, Imielinski M, Kellis M, Lawrence MS, von Mering C, Nakagawa H, Raphael BJ, Rubin MA, Sander C, Stein LD, Stuart JM, Tsunoda T, Wheeler DA, Johnson R, Reimand J, Gerstein M, Khurana E, Campbell PJ, López-Bigas N; PCAWG Drivers and Functional Interpretation Working Group; PCAWG Structural Variation Working Group; Weischenfeldt J, Beroukhim R, Martincorena I, Pedersen JS, Getz G; **PCAWG Consortium**[®].

Nature. 2020 Feb;578(7793):102-111. doi: 10.1038/s41586-020-1965-x. Epub 2020 Feb 5. PMID: 32025015

26. The repertoire of mutational signatures in human cancer.

Alexandrov LB, Kim J, Haradhvala NJ, Huang MN, Tian Ng AW, Wu Y, Boot A, Covington KR, Gordenin DA, Bergstrom EN, Islam SMA, Lopez-Bigas N, Klimczak LJ, McPherson JR, Morganella S, Sabarinathan R, Wheeler DA, Mustonen V; PCAWG Mutational Signatures Working Group; Getz G, Rozen SG, Stratton MR; **PCAWG Consortium**[®].

Nature. 2020 Feb;578(7793):94-101. doi: 10.1038/s41586-020-1943-3. Epub 2020 Feb 5. PMID: 32025018

27. Genomic basis for RNA alterations in cancer.

PCAWG Transcriptome Core Group; Calabrese C, Davidson NR, Demircioğlu D, Fonseca NA, He Y, Kahles A, Lehmann KV, Liu F, Shiraishi Y, Soulette CM, Urban L, Greger L, Li S, Liu D, Perry MD, Xiang Q, Zhang F, Zhang J, Bailey P, Erkek S, Hoadley KA, Hou Y, Huska MR, Kilpinen H, Korbel JO, Marin MG, Markowski J, Nandi T, Pan-Hammarström Q, Pedamallu CS, Siebert R, Stark SG, Su H, Tan P, Waszak SM, Yung C, Zhu S, Awadalla P, Creighton CJ, Meyerson M, Ouellette BFF, Wu K, Yang H; PCAWG Transcriptome Working Group; Brazma A, Brooks AN, Göke J, Rättsch G, Schwarz RF, Stegle O, Zhang Z; **PCAWG Consortium**[®].

Nature. 2020 Feb;578(7793):129-136. doi: 10.1038/s41586-020-1970-0. Epub 2020 Feb 5. PMID: 32025019

28. Genomic footprints of activated telomere maintenance mechanisms in cancer.

- Sieverling L, Hong C, Koser SD, Ginsbach P, Kleinheinz K, Hutter B, Braun DM, Cortés-Ciriano I, Xi R, Kabbe R, Park PJ, Eils R, Schlesner M; PCAWG-Structural Variation Working Group; Brors B, Rippe K, Jones DTW, Feuerbach L; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):733. doi: 10.1038/s41467-019-13824-9. PMID: 32024817
29. Combined burden and functional impact tests for cancer driver discovery using DriverPower.
Shuai S; PCAWG Drivers and Functional Interpretation Working Group; Gallinger S, Stein LD; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):734. doi: 10.1038/s41467-019-13929-1. PMID: 32024818
30. Divergent mutational processes distinguish hypoxic and normoxic tumours.
Bhandari V, Li CH, Bristow RG, Boutros PC; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):737. doi: 10.1038/s41467-019-14052-x. PMID: 32024819
31. High-coverage whole-genome analysis of 1220 cancers reveals hundreds of genes deregulated by rearrangement-mediated cis-regulatory alterations.
Zhang Y, Chen F, Fonseca NA, He Y, Fujita M, Nakagawa H, Zhang Z, Brazma A; PCAWG Transcriptome Working Group; PCAWG Structural Variation Working Group; Creighton CJ; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):736. doi: 10.1038/s41467-019-13885-w. PMID: 32024823
32. Reconstructing evolutionary trajectories of mutation signature activities in cancer using TrackSig.
Rubanova Y, Shi R, Harrigan CF, Li R, Wintersinger J, Sahin N, Deshwar AG; PCAWG Evolution and Heterogeneity Working Group; Morris QD; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):731. doi: 10.1038/s41467-020-14352-7. PMID: 32024834
33. Inferring structural variant cancer cell fraction.
Cmero M, Yuan K, Ong CS, Schröder J; PCAWG Evolution and Heterogeneity Working Group; Corcoran NM, Papenfuss T, Hovens CM, Markowitz F, Macintyre G; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):730. doi: 10.1038/s41467-020-14351-8. PMID: 32024845
34. Integrative pathway enrichment analysis of multivariate omics data.
Paczkowska M, Barenboim J, Sintupisut N, Fox NS, Zhu H, Abd-Rabbo D, Mee MW, Boutros PC; PCAWG Drivers and Functional Interpretation Working Group; Reimand J; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):735. doi: 10.1038/s41467-019-13983-9. PMID: 32024846
35. A deep learning system accurately classifies primary and metastatic cancers using passenger mutation patterns.
Jiao W, Atwal G, Polak P, Karlic R, Cuppen E; PCAWG Tumor Subtypes and Clinical Translation Working Group; Danyi A, de Ridder J, van Herpen C, Lolkema MP, Steeghs N, Getz G, Morris QD, Stein LD; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):728. doi: 10.1038/s41467-019-13825-8. PMID: 32024849
36. Pathway and network analysis of more than 2500 whole cancer genomes. R
Reyna MA, Haan D, Paczkowska M, Verbeke LPC, Vazquez M, Kahraman A, Pulido-Tamayo S, Barenboim J, Wadi L, Dhingra P, Shrestha R, Getz G, Lawrence MS, Pedersen JS, Rubin MA, Wheeler DA, Brunak S, Izarzugaza JMG, Khurana E, Marchal K, von Mering C, Sahinalp SC, Valencia A; PCAWG Drivers and Functional Interpretation Working Group; Reimand J, Stuart JM, Raphael BJ; **PCAWG Consortium**[®].
Nat Commun. 2020 Feb 5;11(1):729. doi: 10.1038/s41467-020-14367-0. PMID: 32024854
37. Cancer lncRNA Census reveals evidence for deep functional conservation of long noncoding RNAs in tumorigenesis.
Carlevaro-Fita J, Lánzó A, Feuerbach L, Hong C, Mas-Ponte D, Pedersen JS; PCAWG Drivers and Functional Interpretation Working Group; Johnson R; **PCAWG Consortium**[®].
Commun Biol. 2020 Feb 5;3(1):56. doi: 10.1038/s42003-019-0741-7. PMID: 32024996
38. Butler enables rapid cloud-based analysis of thousands of human genomes.
Yakneen S, Waszak SM; PCAWG Technical Working Group; Gertz M, Korbel JO; **PCAWG Consortium**[®].
Nat Biotechnol. 2020 Mar;38(3):288-292. doi: 10.1038/s41587-019-0360-3. Epub 2020 Feb 5. PMID: 32024987
39. Comprehensive molecular characterization of mitochondrial genomes in human cancers.
Yuan Y, Ju YS, Kim Y, Li J, Wang Y, Yoon CJ, Yang Y, Martincorena I, Creighton CJ, Weinstein JN, Xu Y, Han L, Kim HL, Nakagawa H, Park K, Campbell PJ, Liang H; **PCAWG Consortium**[®].
Nat Genet. 2020 Mar;52(3):342-352. doi: 10.1038/s41588-019-0557-x. Epub 2020 Feb 5. PMID: 32024997
40. Pan-cancer analysis of whole genomes identifies driver rearrangements promoted by LINE-1 retrotransposition.
Rodríguez-Martin B, Alvarez EG, Baez-Ortega A, Zamora J, Supek F, Demeulemeester J, Santamarina M, Ju YS, Temes J, Garcia-Souto D, Detering H, Li Y, Rodríguez-Castro J, Dueso-Barroso A, Bruzos AL, Dentro SC, Blanco MG, Contino G, Ardeljan D, Tojo M, Roberts ND, Zumalave S, Edwards PA, Weischenfeldt J, Puiggròs M, Chong Z, Chen K, Lee EA, Wala JA, Raine KM, Butler A, Waszak SM, Navarro FCP, Schumacher SE, Monlong J, Maura F, Bolli N, Bourque G, Gerstein M, Park PJ, Wedge DC, Beroukhim R, Torrents D, Korbel JO, Martincorena I, Fitzgerald RC, Van Loo P, Kazanian HH, Burns KH; PCAWG Structural Variation Working Group; Campbell PJ, Tubio JMC; **PCAWG Consortium**[®].

- Nat Genet. 2020 Mar;52(3):306-319. doi: 10.1038/s41588-019-0562-0. Epub 2020 Feb 5. PMID: 32024998
41. Disruption of chromatin folding domains by somatic genomic rearrangements in human cancer.
Akdemir KC, Le VT, Chandran S, Li Y, Verhaak RG, Beroukhir R, Campbell PJ, Chin L, Dixon JR, Futreal PA; PCAWG Structural Variation Working Group; **PCAWG Consortium**[®].
Nat Genet. 2020 Mar;52(3):294-305. doi: 10.1038/s41588-019-0564-y. Epub 2020 Feb 5. PMID: 32024999
 42. Comprehensive analysis of chromothripsis in 2,658 human cancers using whole-genome sequencing.
Cortés-Ciriano I, Lee JJ, Xi R, Jain D, Jung YL, Yang L, Gordenin D, Klimczak LJ, Zhang CZ, Pellman DS; PCAWG Structural Variation Working Group; Park PJ; **PCAWG Consortium**[®].
Nat Genet. 2020 Mar;52(3):331-341. doi: 10.1038/s41588-019-0576-7. Epub 2020 Feb 5. PMID: 32025003
 43. Comprehensive Analysis of Genetic Ancestry and Its Molecular Correlates in Cancer.
Carrot-Zhang J*, Chambwe N*, Damrauer JS*, Knijnenburg TA*, Robertson AG*, Yau C*, Zhou W*, Berger AC*, **Huang KL***, Newberg JY*, Mashl RJ, Romanel A, Sayaman RW, Demichelis F, Felau I, Frampton GM, Han S, Hoadley KA, Kemal A, Laird PW, Lazar AJ, Le X, Oak N, Shen H, Wong CK, Zenklusen JC, Ziv E; Cancer Genome Atlas Analysis Network; Cherniack AD, Beroukhir R.
Cancer Cell. 2020 May 11;37(5):639-654.e6. doi: 10.1016/j.ccell.2020.04.012. PMID: 32396860
 44. Ancestry-specific predisposing germline variants in cancer.
Oak N, Cherniack AD, Mashl RJ; TCGA Analysis Network; Hirsch FR, Ding L, Beroukhir R, Gümüş ZH, Plon SE, **Huang KL**.
Genome Med. 2020 May 29;12(1):51. doi: 10.1186/s13073-020-00744-3. PMID: 32471518
 45. Genotype concordance and polygenic risk score estimation across consumer genetic testing data.
Batra P, **Huang KL**.
Ann Hum Genet. 2020 Jul;84(4):352-356. doi: 10.1111/ahg.12389. Epub 2020 May 21. PMID: 32436989
 46. Sex differences in oncogenic mutational processes.
Li CH, Prokopec SD, Sun RX, Yousif F, Schmitz N; PCAWG Tumour Subtypes and Clinical Translation; Boutros PC; **PCAWG Consortium**[®].
Nat Commun. 2020 Aug 28;11(1):4330. doi: 10.1038/s41467-020-17359-2. PMID: 32859912
 47. Retrospective evaluation of whole exome and genome mutation calls in 746 cancer samples.
Bailey MH, Meyerson WU, Dursi LJ, Wang LB, Dong G, Liang WW, Weerasinghe A, Li S, Li Y, Kelso S; MC3 Working Group; PCAWG novel somatic mutation calling methods working group; Saksena G, Ellrott K, Wendl MC, Wheeler DA, Getz G, Simpson JT, Gerstein MB, Ding L; **PCAWG Consortium**[®].
Nat Commun. 2020 Sep 21;11(1):4748. doi: 10.1038/s41467-020-18151-y. PMID: 32958763
 48. Risk prediction of late-onset Alzheimer's disease implies an oligogenic architecture.
Zhang Q, Sidorenko J, Couvy-Duchesne B, Marioni RE, Wright MJ, Goate AM, Marcora E, **Huang KL**, Porter T, Laws SM; Australian Imaging Biomarkers and Lifestyle (AIBL) Study; Sachdev PS, Mather KA, Armstrong NJ, Thalamuthu A, Brodaty H, Yengo L, Yang J, Wray NR, McRae AF, Visscher PM.
Nat Commun. 2020 Sep 23;11(1):4799. doi: 10.1038/s41467-020-18534-1. PMID: 32968074
 49. Cooperation Between Distinct Cancer Driver Genes Underlies Intertumor Heterogeneity in Hepatocellular Carcinoma.
Molina-Sánchez P, Ruiz de Galarreta M, Yao MA, Lindblad KE, Bresnahan E, Bitterman E, Martin TC, Rubenstein T, Nie K, Golas J, Choudhary S, Bárcena-Varela M, Elmas A, Miguela V, Ding Y, Kan Z, Grinspan LT, **Huang KL**, Parsons RE, Shields DJ, Rollins RA, Lujambio A.
Gastroenterology. 2020 Dec;159(6):2203-2220.e14. doi: 10.1053/j.gastro.2020.08.015. Epub 2020 Aug 16. PMID: 32814112
 50. Shared Immunogenic Poly-Epitope Frameshift Mutations in Microsatellite Unstable Tumors.
Roudko V, Bozkus CC, Orfanelli T, McClain CB, Carr C, O'Donnell T, Chakraborty L, Samstein R, **Huang KL**, Blank SV, Greenbaum B, Bhardwaj N.
Cell. 2020 Dec 10;183(6):1634-1649.e17. doi: 10.1016/j.cell.2020.11.004. Epub 2020 Nov 30. PMID: 33259803
 51. AeQTL: eQTL analysis using region-based aggregation of rare genomic variants.
Dong G, Wendl MC, Zhang B, Ding L, **Huang KL**.
Pac Symp Biocomput. 2021;26:172-183. PMID: 33691015
 52. Spatially interacting phosphorylation sites and mutations in cancer.
Huang KL[#], Scott AD, Zhou DC, Wang LB, Weerasinghe A, Elmas A, Liu R, Wu Y, Wendl MC, Wyczalkowski MA, Baral J, Sengupta S, Lai CW, Ruggles K, Payne SH, Raphael B, Fenyö D, Chen K, Mills G, Ding L[#].
Nat Commun. 2021 Apr 19;12(1):2313. doi: 10.1038/s41467-021-22481-w. PMID: 33875650
 53. Diverse immune response of DNA damage repair-deficient tumors.
Qing T, Jun T, Lindblad KE, Lujambio A, Marczyk M, Pusztai L[#], **Huang KL**[#].
Cell Rep Med. 2021 May 18;2(5):100276. doi: 10.1016/j.xcrm.2021.100276. eCollection 2021 May 18. PMID: 34095878

54. The Functional Hallmarks of Cancer Predisposition Genes.
Capellini A, Williams M, Onel K#, **Huang KL**#. *Cancer Manag Res.* 2021 Jun 1;13:4351-4357. doi: 10.2147/CMAR.S311548. eCollection 2021. PMID: 34103990
55. Genetic dependency of Alzheimer's disease-associated genes across cells and tissue types.
Jaladanki SK, Elmas A, Malave GS, **Huang KL**. *Sci Rep.* 2021 Jun 8;11(1):12107. doi: 10.1038/s41598-021-91713-2. PMID: 34103633 **[Covered by AlzForum]**
56. Phenome-wide and expression quantitative trait locus associations of coronavirus disease 2019 genetic risk loci.
Moon CY, Schilder BM, Raj T, **Huang KL**. *iScience.* 2021 Jun 25;24(6):102550. doi: 10.1016/j.isci.2021.102550. Epub 2021 May 18. PMID: 34027315
57. Analysis of sex-specific risk factors and clinical outcomes in COVID-19.
Jun T, Nirenberg S, Weinberger T, Sharma N, Pujadas E, Cordon-Cardo C, Kovatch P, **Huang KL**. *Commun Med (Lond).* 2021 Jun 30;1:3. doi: 10.1038/s43856-021-00006-2. eCollection 2021. PMID: 35602223 **[Covered by Mount Sinai Press Release & 23 other news outlets]**
58. Prediction of individual COVID-19 diagnosis using baseline demographics and lab data.
Zhang J, Jun T, Frank J, Nirenberg S, Kovatch P, **Huang KL**. *Sci Rep.* 2021 Jul 6;11(1):13913. doi: 10.1038/s41598-021-93126-7. PMID: 34230510
59. Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes.
Wang Z, Fan X, Shen Y, Pagadala MS, Signer R, Cygan KJ, Fairbrother WG, Carter H, Chung WK, **Huang KL**. *Genome Med.* 2021 Sep 9;13(1):147. doi: 10.1186/s13073-021-00964-1. PMID: 34503567
60. Genomic Determinants of Homologous Recombination Deficiency across Human Cancers.
Qing T, Wang X, Jun T, Ding L, Puzstai L#, **Huang KL**#. *Cancers (Basel).* 2021 Sep 12;13(18):4572. doi: 10.3390/cancers13184572. PMID: 34572800
61. Pan-cancer proteogenomic investigations identify post-transcriptional kinase targets.
Elmas A, Tharakan S, Jaladanki S, Galsky MD, Liu T, **Huang KL**. *Commun Biol.* 2021 Sep 22;4(1):1112. doi: 10.1038/s42003-021-02636-7. PMID: 34552204
62. Genomic and molecular features distinguish young adult cancer from later-onset cancer.
Lee W, Wang Z, Saffern M, Jun T, **Huang KL**. *Cell Rep.* 2021 Nov 16;37(7):110005. doi: 10.1016/j.celrep.2021.110005. PMID: 34788626 **[Covered by Mount Sinai Press Release & 9 other news outlets]**
63. Analytical protocol to identify local ancestry-associated molecular features in cancer.
Carrot-Zhang J, Han S, Zhou W, Damrauer JS, Kemal A; **Cancer Genome Atlas Analysis Network**; Cherniack AD, Beroukhir R. *STAR Protoc.* 2021 Sep 20;2(4):100766. doi: 10.1016/j.xpro.2021.100766. eCollection 2021 Dec 17. PMID: 34585150
64. Proteomic Analyses Identify Therapeutic Targets in Hepatocellular Carcinoma.
Elmas A, Lujambio A, **Huang KL**. *Front Oncol.* 2022 Mar 30;12:814120. doi: 10.3389/fonc.2022.814120. eCollection 2022. PMID: 35433463
65. Modeling COVID-19 dynamic using a two-strain model with vaccination.
de León UA, Avila-Vales E, **Huang KL**. *Chaos Solitons Fractals.* 2022 Apr;157:111927. doi: 10.1016/j.chaos.2022.111927. Epub 2022 Feb 16. PMID: 35185299
66. Multiethnic Investigation of Risk and Immune Determinants of COVID-19 Outcomes.
Jun T, Mathew D, Sharma N, Nirenberg S, Huang HH, Kovatch P, Wherry EJ, **Huang KL**. *Front Cell Infect Microbiol.* 2022 Jul 22;12:933190. doi: 10.3389/fcimb.2022.933190. eCollection 2022. PMID: 35942057
67. Analysis of germline-driven ancestry-associated gene expression in cancers.
Chambwe N, Sayaman RW, Hu D, Huntsman S; **Cancer Genome Analysis Network**%; Kemal A, Caesar-Johnson S, Zenklusen JC, Ziv E, Beroukhir R, Cherniack AD. *STAR Protoc.* 2022 Jul 31;3(3):101586. doi: 10.1016/j.xpro.2022.101586. eCollection 2022 Sep 16. PMID: 35942349
68. Using EGFR amplification to stratify recurrent glioblastoma treated with immune checkpoint inhibitors.
Friedman JS, Jun T, Rashidipour O, **Huang KL**, Ellis E, Kadaba P, Belani P, Nael K, Tsankova NM, Sebra R, Hormigo A. *Cancer Immunol Immunother.* 2023 Jan 28. doi: 10.1007/s00262-023-03381-y. Online ahead of print. PMID: 36707424

Other Peer Reviewed Publications:

1. Ten Simple Rules for landing on the right job after your PhD or postdoc.
Huang KL. *PLoS Computational Biology* 2020. [PLoS ten simple rules series]: **8,000+ reads counting only the first year of publication.**

Books:

2. Solve It Yourself: Fix the World's Problem with Science.

Huang KL. 2021. [E-book, Paperback, Hardcover]: **Amazon #1 best-seller in Civics, #2 in motivational self-help in April 2021. Author interviews on Podcast, Radio, and TV shows.**

Non-Peer Reviewed Publications:

3. Most popular public searches on gene names.

Huang KL. *Nature* 2018. [Correspondence]

INVITED PRESENTATIONS

1. Hosted/co-hosted 10+ **Open Box Science** research seminars/career panels/science communication symposiums.
2. Universitätsmedizin der Johannes Gutenberg-Universität Mainz, DIASyM research core, Germany, 2023.
3. University of Alabama at Birmingham (UAB), Department of Genetics, USA, 2023.
4. National Taiwan University Hospital, Precision Medicine Research Group, Taipei, Taiwan 2023.
5. National Sun Yet-Sun University, School of Medicine, Kaohsiung, Taiwan 2022.
6. The 5th Global Conference on Biomedical Engineering (GCBME 2022), Taipei, Taiwan 2022.
7. 2nd CASMS Virtual Conference, USA, 2022.
8. Discussion Leader for Gordon Research Conference on Human Genetic Variation and Disease, USA 2022.
9. Charleston Conference on Alzheimer's Disease (CCAD), Hawaii, USA 2022.
10. VCCC Alliance Monday Livestream, Australia, 2022.
11. Drug Discovery News (DDN) Webinar, USA, 2021.
12. Perlmutter Cancer Center Research Seminar Series, NYU Langone Health, New York, USA, 2021.
13. The Lloyd Sherman Scholars program, Center for Excellence in Youth Education (CEYE), New York, USA, 2021.
14. TCI Big Data Cancer Retreat, Icahn School of Medicine at Mount Sinai, New York, USA, 2021.
15. Sage Bionetwork, Seattle, Washington, USA, 2021.
16. National Taiwan University Hospital (NTUH), Taipei, Taiwan, 2021.
17. National Central University, Taiwan, Taoyuan, Taiwan, 2021.
18. National Health Research Institutes (NHRI), Miaoli, Taiwan, 2021.
19. The European Conference on Machine Learning and Principles and Practice of Knowledge Discovery in Databases, Ghent, Belgium (virtual), 2020. [[Keynote](#)]
20. NGS Teleconference, Kaohsiung Medical University, Kaohsiung, Taiwan (virtual), 2020. [[Keynote](#)]
21. Charleston Conference on Alzheimer's Disease (CCAD)[Competition Finalist], South Carolina, USA, 2020.
22. Academia Sinica, Institute of Statistical Science, Taipei, Taiwan, 2020.
23. National Yang-Ming University, Institute of Biomedical Informatics, Taipei, Taiwan, 2020.
24. National Tsing Hua University, Institute of Biomedical Engineering, Hsinchu, Taiwan, 2020.
25. National Yang-Ming University, School of Dentistry, Taipei, Taiwan, 2020.
26. Icahn School of Medicine at Mount Sinai, Department of Genetics and Genomic Sciences, New York, USA, 2019.
27. Wesleyan University, Department of Molecular Biology and Biochemistry, Middletown, USA, 2019.
28. Icahn School of Medicine at Mount Sinai, Center for Transformative Disease Modeling, New York, USA, 2018.
29. The Scripps Research Institute, Skaggs Institute for Molecular Biology, San Diego, USA, 2018.
30. UT Southwestern, Department of Bioinformatics, Dallas, USA, 2018.
31. Oregon Health and Science University, The Knight Cancer Institute, Portland, USA, 2018.
32. UCLA Institute for Quantitative and Computational Biosciences, Los Angeles, USA, 2018.
33. Novartis Institutes for Biomedical Research, Cambridge, USA, 2018.
34. Calico Life Sciences, South San Francisco, USA, 2017.
35. 23andMe, Mountain View, USA, 2017.
36. NCI CPTAC Steering Committee Meeting, Bethesda, USA, 2017.
37. Academia Sinica, IBMS Seminar, Taipei, Taiwan, 2017.
38. TCGA PanCanAtlas Face-to-face Meeting, Houston, USA, 2016.
39. Alzheimer's Association International Conference (AAIC), Toronto, Canada, 2016.
40. American Association for Cancer Research Annual Meeting (AACR), NCI Cloud Pilot Section, New Orleans, USA, 2016.
41. NCI CPTAC Meeting, Bethesda, USA, 2015.

MEDIA RESOURCE / EDUCATIONAL MATERIALS

Through my non-profit Open Box Science, we have made 200+ interactive webinars openly available. These are mainly comprised of research talks by diverse, early-career researchers presenting their first-author publications. As of 2023/03, the OBS YouTube channel has accumulated 38.5k views with over 3.7k watch times from audience across 70+ countries.

<https://www.youtube.com/openboxscience>

VOLUNTARY PRESENTATIONS

Not applicable