



The University of Alabama at Birmingham

Department of Genetics

QUARTERLY
Genetics Update
SPRING 2023

Department of Defense Awards 10-Year Extension to Consortium



Bruce Korf M.D., Ph.D., as part of the Neurofibromatosis (NF) Clinical Trials Consortium, recently received a \$25 million award from the Department of Defense (DoD) that will extend research for an additional 10 years. The consortium, initially formed in 2006, will continue funding for clinical trials for all forms of neurofibromatosis, which aims to find options for new treatment opportunities as it relates to neurofibromatosis.

The consortium currently has 24 clinical sites across the United States and Australia, with nearly 20 clinical trials conducted involving over 500 study participants.

Dr. Might's Grant Application Selected by NIH



Matthew Might, Ph.D., in collaboration with researchers from various universities, recently had a grant application selected by the National Institute of Health (NIH), with funding to be approved by the National Institute of Neurological Disorders and Stroke (NINDS). The application, Diagnosing the Unknown for Care and Advancing Science (DUCAS), aims to advance diagnostic success and biological understandings through the implementation of computational models that predict the likelihood of diagnosis while expanding membership in the undiagnosed and rare disease ecosystem.

Along with Dr. Might, research was conducted by representatives from Harvard University, Stanford University, Washington University of St. Louis, the University of Utah, and the Morehouse School of Medicine. The award was issued on April 19th, and the NIH program team will meet with DUCAS principle investigators to begin discussions on establishing the new network and ideas for leveraging DUCAS to meet the FY2023 Omnibus Appropriations Bill directives.

Dr. Dutta Joins Association of American Physicians



Recently, **Anindya Dutta, M.D., Ph.D.** was elected to join the Association of American Physicians (AAP), an honorary medical society that advocates for the advancement of scientific and practical medicine through professional and social interaction among researchers.

[Read more](#)

Joint Venture to Create New Genomic Diagnostic Lab



The Board of Trustees of the University of Alabama System has further approved joint plans to create a state-of-the-art genomic diagnostic lab, marking a significant milestone in the development of the joint endeavor between UAB and Southern Research.

[Read more](#)

UAB Genetics Personnel Attend ACMG Meeting



The 2023 American College of Medical Genetics (ACMG) Annual Clinical Genetics meeting was held from March 14th – 18th in Salt Lake City, Utah.

Members of the Department of Genetics were well-represented, with trainees and faculty making key contributions/presentations, including: Jacob Greenway, Pongtawat Lertwilaiwittaya, Jennifer Cassady Hayek, Laboratory Genetics and Genomics (LGG) fellow Yunjia Chen, Anna Hurst, M.D., Program Director of the Medical Genetics Residency Program, and Nathaniel Robin, M.D., Director of Clinical Services.

Brewer to Remain on Alabama Board of Genetic Counseling



Fallon Brewer, MS, CGC will continue to serve as President of the Alabama Board of Genetic Counseling, which also includes several other UAB faculty. To date, the board has licensed over 300 Genetic Counselors from over 35 states.

Former UAB Resident to Join Clinical Division



In September, we will be welcoming back our former resident **Caterina Abdala, M.D.** as an Assistant Professor in the Clinical Division.

Dr. Abdala completed Medical School and her Pediatrics residency at the University of Puerto Rico School of Medicine. She then spent 2 years as a Genetics resident, graduating in 2022, going on to a Medical Biochemical fellowship at The Ohio State University/Nationwide Children's Hospital.

Genetic Counselors and Residents to Join UAB



Three new genetic counselors will join UAB this summer. The confirmed new hires are: **Macy Calvasina** from the Wake Forest Genetic Counseling Program, **Morgan Bryars** from the UAB Genetic Counseling Training Program, and **Catherine Smith**, who will be joining as the inaugural Cancer Genetics fellow. Two new residents will also join UAB this upcoming year. We accepted **Lindsey Jenkins** from Wayne State University to our Pediatrics-Medical Genetics residency program, and **Grace Raines, MD**, into the categorical Genetics residency program.

Dr. Raines is a graduate of the UAB Heersink School of Medicine, and completed her pediatrics residency at the San Antonio Uniformed Services Health Education Consortium, a US Army program.

Department Spotlight: Jon Daniel Sharer, Ph.D.



Dr. Sharer obtained his Ph.D. in Biochemistry and Molecular Biology from Rutgers University and performed postdoctoral training at the National Cancer Institute and Emory University.

[Read More](#)

Recent Faculty Publications

Lessons in Genetics: Common Diseases Occur in Patients with a Genetic Syndrome. American Journal of Medical Genetics. 2023 Feb; Vol 191, Issue 2: p. 634-635. Robin N. doi: 10.1002/ajmg.a.63046

Targeting Intracellular Neu1 for Coronavirus Infection Treatment. iScience. 2023 Feb 17. Vol 26, Issue 2. doi: 10.1016/j.isci.2023.106037.

How Cancer Hijacks the Body's Homeostasis through the Neuroendocrine System. Trends Neuroscience. 2023 Feb 16; S0166-2236(23)00019-X. Slominski R, Ramac C, **Chen J**, Slominski A. Online ahead of print. doi: 10.1016/j.tins.2023.01.003

Deciphering the Exact Breakpoints of Structural Variations Using Long-sequencing Reads with DeBreak. Nature Communications. 2023 Jan 17. Vol 14, Issue 1. p. 283. doi: 10.1038/s41467-023-35996-1. Chen Y, Wang A, Barkley C, Zhang Y, Zhao X, Gao M, Edmonds M, **Chong Z**

The Hitchhikers' Guide to RNA Sequencing and Functional Analysis. Brief Bioinformatics. 2023 Jan 19. Vol 24, Issue 1. doi: 10.1093/bib/bbac529. Chen J, Shrestha L, Green G, **Leier A**, **Marquez-Lago T**

Gene Fusion Detection and Characterization in Long-Read Cancer Transcriptome Sequencing Data with FusionSeeker. Cancer Res. 2023 Jan 4. Vol 83, Issue 1. pp. 28-33. Chen Y, **Chong Z**, Song Y, Tan Z, Wang Y, Chen W, Chen H, Human Genome Structural Variation Consortium. doi: 10.1158/0008-5472.CAN-22-1628

Differential impact of PI3K/AKT/mTOR signaling on tumor initiation and progression in animal models of prostate cancer. Prostate. 2023 Jan. Vol 83, Issue 1. pp. 97-108. Wang S, Zhang C, Xu Z, Chen M, Yu H, **Wang L**, **Liu R**. doi: 10.1002/pros.24441

Six New Cases of CRB2-related Syndrome and a Review of Clinical Findings in 28 Reported Patients. Clin Genet. 2023 Jan. Vol 103, Issue 1. pp. 97-102. Adutwum M, **Hurst A**, Mirzaa G, Kushner J, Rogers C, Khalek N, Cristancho A, Burrill N, Seifert M, Scarano M, Schunr R, Slavotinek A. doi: 10.1111/cge.14222.

Compound Heterozygous Variants within Two Conserved Sialyltransferase Motifs of ST3GAL5 Cause GM3 Synthase Deficiency. JIMD Rep. 2022 Nov 29. Vol 64, Issue 2. pp. 138-145. Rudy N, Aoki K, Ananth A, Holloway L, Skinner C, **Hurst A**, Tiemeyer M, Steet R. doi: 10.1002/jmd2.12353.

Cryptic lncRNA-encoded ORFs: A Hidden Source of Regulatory Proteins. J Clin Invest. 2023 Mar 1. Vol 133, Issue 5. **Dutta A**, Li H, Abounader R. doi: 10.1172/JCI167271.

Recent Faculty Grant Awards

PI: Deeann Wells

Sponsor: Gilbert Family Foundation

Title: Exon Skipping to Treat NF1

Award Amount: \$1,235,850

Dates: 3/1/2023 to 2/28/2026

PI: Lizhong Wang

Sponsor: Department of Defense (DoD)

Title: Role of TUBB4A in Racial Disparities, Tumor Metastasis, and Chemotherapy Resistance

Award Amount: \$371,250

Dates: 5/1/2023 to 4/30/2024

PI: Lizhong Wang

Sponsor: National Institute of Health (NIH)

Title: SEMA6D-mediated breast cancer disparity, metastasis, and tumor-immune interaction

Award Amount: \$2,341,658

Dates: 6/1/2023 to 5/31/2028

PI: Andre Leier

Sponsor: DoD

Title: SMaRT-Based Repair of NF1 Pre-mRNA

Award Amount: \$148,500

Dates: 8/1/2023 to 7/31/2025

Welcome to the Department

New Staff	New Post-Docs	New Faculty/Researchers
Lynae Hanks	Dr. Uzzal Kumar	Saba Tufail
Sable Washington	Acharjee	Dr. Yunjia Chen
	Dr. Aleena Francis	
	Dr. Ali Cem Kucukdagli	



The Harbert Institute for Innovation and Entrepreneurship recently announced a new fund to serve early-stage UAB technologies. The Blazer Bridge Fund is intended to identify and assist in the development of promising ideas, discoveries, innovations, and/or technologies from UAB faculty and staff that have commercial potential. The BBF expects to fund about six projects per award round at a maximum of \$50,000 per project. Submissions are due on or before May 31, 2023. For more information, visit go.uab.edu/blazer-bridge-fund.

Innovation Office Hours

A promotional graphic for Innovation Office Hours. The left side has a dark green background with white and yellow text. The right side has a medium green background with a yellow lightbulb icon, the text 'INNOVATION OFFICE HOURS', and three circular portraits of staff members.

**WONDERING
ABOUT
INTELLECTUAL
PROPERTY?
WE'RE HERE
TO CHAT.**

Talk with one of our licensing experts about technology transfer, commercialization, patenting, or anything relating to intellectual property protection.

**INNOVATION
OFFICE
HOURS**

**Virtual Office Hours:
Wednesdays, 1-2 p.m.
Via Zoom**

UAB BILL L. HARBERT INSTITUTE FOR
INNOVATION AND ENTREPRENEURSHIP
The University of Alabama at Birmingham

Do you have questions about intellectual property, technology transfer, or the commercialization process? Are you curious about how to patent an invention, or if you have an invention to begin with?

Are you a first-time inventor and you're not sure where to start? Talk with a licensing associate from the UAB Bill L. Harbert Institute for Innovation and Entrepreneurship during our virtual drop-in office hours!

Find event info and registration link here:

https://calendar.uab.edu/event/innovation-office-hours?utm_campaign=widget&utm_medium=widget&utm_source=The+University+of+Alabama+at+Birmingham#.Y87ic3bMJMs

Important E&M Documentation & Coding Changes

In recent months, many of you and your teams may have heard Dr. Stephen Stair, Brian Bates, and/or members of the UAB Medicine Compliance team discussing the changes to the Evaluation and Management (E&M) Guidelines that went into effect Sunday, January 1, 2023. We want to remind you about the changes to the provider documentation guidelines that will be in place for ALL E&M services.

Overall, we feel that these changes are long overdue and are positive. Instead of harping on missing elements of history (ex., review of systems; past, family, social history) and/or physical exam, we will continue to work with our Providers to best document their medical decision making and/or total time spent in the calendar day caring for a patient.

Links below include key information about these changes:

- Education Slides – Inpatient 2023 New E&M Guidelines
- Compliance Alert - 2023 AMA CPT E&M Changes
- Pearls of Success – 2023 E&M Guidelines, documentation and coding
- AMA CPT MDM Table 2023

If you have any questions, please do not hesitate to the UAB Medicine Compliance team at ComplianceBillingInquiry@uabmc.edu or (205) 731-9863.



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Editor: [Ryan McCain](#)

If you have content you would like included in the quarterly newsletter, please contact [Ryan McCain](#).

