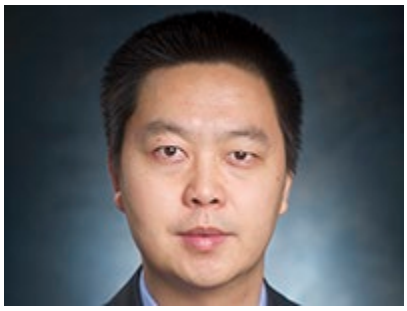
UAB HEERSINK SCHOOL OF MEDICINE
The University of Alabama at Birmingham

QUARTERLY GENETICS UPDATE

SPRING 2022

Researchers pioneering long-read sequencing studies explain why long reads matter



With long-read sequencing, read lengths can average 10,000 to 30,000 base pairs, with a maximum length over 1 megabase (1 million base pairs) on an Oxford Nanopore device, says **Zechen Chong, Ph.D.,** assistant professor in the Department of Genetics at the Heersink School of Medicine. Chong's lab is focused on developing new algorithms for the analysis of long-read sequencing data, and he is collaborating with several UAB researchers using long-read sequencing.

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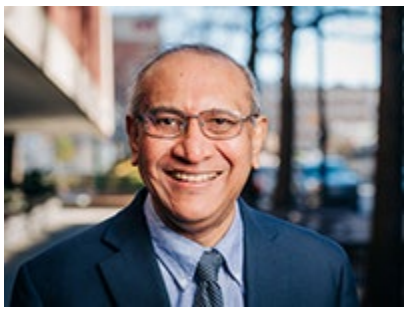
UAB Cardiogenomics Clinic transforms cardiac care through genomic medicine

"The approach to understanding the risk of developing potentially fatal heart diseases can vary, from using the latest advances in genetics to simply talking with our grandparents," said **Ali Al-Beshri, M.D.**, an assistant professor in the UAB Department of Genetics. "As technology has evolved, we have gained an improved understanding of what to do and how to manage individuals who are at risk for inheriting cardiovascular diseases. Furthermore, the latest advances in genetic medicine such as gene-based therapeutics are becoming increasingly common in the clinical pipeline."

Read more

Distinct structural domains in MUNC long non-coding RNA regulate gene expression



In a paper in Cell Reports, **Anindya Dutta, M.D., Ph.D.**, and colleagues detail the first structure-function study of a long non-coding RNA, or lncRNA, called MUNC lncRNA. Their findings point out the importance of experimentally determining the structure of an lncRNA through a chemical method — rather than computer modeling — to identify structural domains that turn out to have multiple different functions.

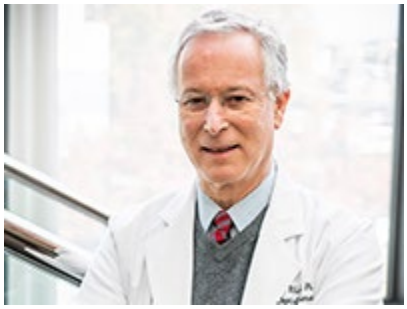
Read more

How a game-changing transplant could treat dying organs

Scientists have long known that defective mitochondria can cause biological chaos. "Mitochondrial dysfunction is a universal driver of disease," says **Keshav Singh**, a mitochondria researcher at the University of Alabama Birmingham who founded the Mitochondria Research and Medicine Society in the U.S. and India. Whether tissue damage is caused by disease or even space travel, faulty mitochondria are frequently involved.

Read more

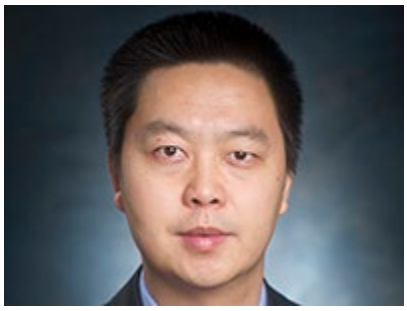
Robustly collaborative projects funded through annual AMC21 Multi-PI Awards; Lee, Pollock, and Korf advance cutting-edge research



The Heersink School of Medicine has awarded the annual Multi-Investigator Program Award to three research investigators for the 2021-2022 cycle: Timmy Lee, M.D., **Bruce Korf, M.D., Ph.D.**, and David Pollock, Ph.D. Dr. Korf was awarded a Multi-PI Award for his study UAB Gene Therapy Neurofibromatosis Program.

Read more

Dr. Chong selected for Featured Discovery

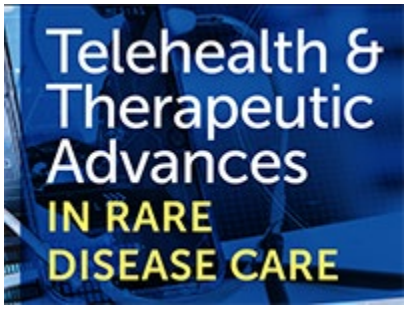


Zechen Chong, Ph.D., assistant professor in the Department of Genetics, is the latest winner of the Heersink School of Medicine's Featured Discovery. This initiative celebrates important research from Heersink faculty members.

Chong's study, "Accurate long-read de novo assembly evaluation with Inspector," was published in Genome Biology.

Read more

Telehealth and therapeutic advances are focus of 2022 Rare Disease Genomics Symposium



The University of Alabama at Birmingham presented the ninth annual Rare Disease Genomics Symposium on March 4 virtually. The symposium is held every year in conjunction with World Rare Disease Day and is hosted by UAB, Children's of Alabama, Alabama Rare and the Alabama Genomic Health Initiative, and the UAB Center for Clinical and Translational Science. This year's event focused on science- and research-related telehealth and therapeutic advances. Participants heared from experts in the fields of genetics, telemedicine, precision medicine and more.

Read more

All of Us research program releases first genomic data, creating unprecedented platform for health discoveries

The National Institutes of Health's All of Us Research Program has made its initial genomic data available, bringing nearly 100,000 whole genome sequences onto its data analysis platform, the Researcher Workbench. As the All of Us Southern Network leader with sites across Alabama,

Mississippi, and Louisiana, UAB Medicine has played a significant role in this milestone, nurturing relationships with diverse participants from throughout the region to ensure substantial diversity in the dataset.

Read more

Recent Faculty Publications

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Recent Faculty Grant Awards

PI: Michael Crowley

Sponsor: NIH-S10

Title: NovaSeq 6000 Sequencing System

Amount: $945,108

Dates: 3-1-2022 to 2-28-23

PI: Bruce Korf

Sponsor: Children's Tumor Foundation

Title: NFCTC Data Management Support

Amount: $50,936

Dates: 2-1-2022 to 1-31-23

PI: Bruce Korf

Sponsor: NF2 Biosolutions

Title: NF2 BioSolutions Blood and Tumor Banking Services for the

Neurofibromatosis Clinical Trials Consortium (NFCTC)
Amount: $15,000
Dates: 2-26-2022 to 2-25-2023

PI: Robert Kesterson
Sponsor: Gilbert Family Foundation
Title: NF1 Gene Rescue in Translational Animal Models
Amount: $400,000
Dates: 1-1-2022 to 12-31-22

PI: Keshav Singh
Sponsor: NIH-R21
Title: mtDNA Depleter Mouse for Decoding Mitochondrial Regulation of Diverse Organs
Amount: $200,475
Dates: 3-15-2022 to 2-28-23

PI: Zhangli Su
Sponsor: NIH-K99
Title: The Role of Small RNA Modifications in Glioma
Amount: $110,646
Dates: 3-11-2022 to 2-28-23

PI: Deeann Wallis
Sponsor: Gilbert Family Foundation
Title: Targeted Delivery of Full Length NF1 cDNA using Non-viral Vectors
Amount: $386,697
Dates: 1-1-2022 to 12-31-22

PI: Deeann Wallis

Sponsor: Gilbert Family Foundation
Title: RAFT-Polymerized Nanoparticles for Delivery of NF1 cDNA to Schwann Cells
Amount: $28,480
Dates: 10-1-2021 to 9-30-22

PI: Deeann Wallis
Sponsor: Gilbert Family Foundation
Title: Exon Skipping to Treat NF1
Amount: $347,082
Dates: 2-1-2022 to 1-31-23

Department Acknowledgements

Dan Sharer, Ph.D., professor, assumed the role of Interim Director of Genetics Research in January 2022.

Anna Hurst, M.D., assistant professor, was named associate editor of the American Journal of Medical Genetics.

Bruce Korf, M.D., Ph.D., professor, was elected to the American Association of Physicians.

Yulong Fu, Ph.D., assistant professor, named interim director of the Medical Genomics Laboratory.

Steve Doran, named space and facilities manager.

Thank you to **Leigh Ann Withers** and **Shelia Coleman** for all of their work on faculty recruitment recently.

Congratulations to the **UAB Laboratory Genetics and Genomics (LGG) ABMGG Fellowship** on being accredited by the ACGME.

Welcome to the Department

New Staff	New Post-Docs	New Students
Collin Ainslie	Shruti Kapoor	Logan Brewer
Elizabeth Michelle Bird	Pradeep Kumar	Amy Lamore
Rose Missildine	Sarmad Mehmood	Violet Morin
Annah Bochaberi Oigo	Nongyao Nonpanya	Danya Midani
Isaac Andres Segura Rueda		Holland Powell
		Brayden Rohr
Xiaoxia Zhang		Kylee Spencer

Upcoming Events

Save the Date: Department Faculty and Staff Retreat - Wednesday, May 25

UAB Genetics | News & Events

Editor: Heather Watts

If you have content you would like included in the quarterly newsletter, please contact Heather Watts.