

The University of Alabama at Birmingham

Department of Genetics

QUARTERLY

GENETICS UPDATE

WINTER 2023

Department Spotlight: Wioletta Czaja, Ph.D.



Wioletta Czaja, Ph.D., who joined the Department of Genetics in October 2022, comes to UAB with a fresh energy ready to immediately make contributions to the university as well as the department. She has arrived with a great knowledge of genetics and a tireless work ethic, with the intention of establishing partnerships and collaborations that lead to innovative discoveries.

Read more

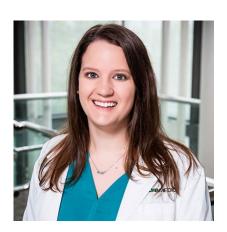
Rare Disease Symposium to Commemorate 10th Year



The 10th Annual Rare Disease Day Symposium will be a hybrid event, taking place on Zoom and at the Bradley Lecture Center. This year's Symposium will be held on March 10th from 8:30 a.m. to 3:30 p.m, and is sponsored by UAB, Children's of Alabama, Alabama Rare, and the Alabama Genomic Initiative.

Read more

Faculty Member Receives Esteemed Honor



Anna Hurst, MD, MS, Associate Professor and Medical Geneticist, was recently selected for induction as a faculty member of the UAB Hersink School of Medicine's Alpha Omega Alpha (AOA) Medical Society.

Established in 1902, AOA has elected over 150,000 members, with 3,000 students, alumni, and faculty elected each year. AOA is recognized as "a professional medical organization that recognizes and advocates for excellence in scholarship and the highest ideals in the profession of medicine."

With the organization's commintment to "scholarship, leadership, professionalism and

service," this is an outstanding accomplishment that is greatly recognized. Congratulations, Dr. Hurst!

Residency Programs Receive Positive Notice

Per the Accreditation Council for Graduate Medical Eduction (ACGME), both the medical genetics residency and fellowship programs received continued accreditation with ZERO citations or comments.

Congratualtions on the success and hard work of both of these programs, in order to make sure the Department of Genetics continues to excel!

Department Retreats Focus on Growth

Over the last quarter, the UAB Department of Genetics held multiple socials attended by various faculty and other department members. The purposes of the socials were to reflect on the successes of the past year, while also giving the opportunity to personnel members to unwind and interact with department and staff outside of the work environment.

In October, the department held a social at the Hilton Hotel in downtown Birmingham to discuss the overall strengths and weaknesses of the department, while assessing the overall opportunities for improvement within the department.

The annual faculty holiday dinner was held at The Club on December 14th, and gave department members an opportunity to celebrate the holiday season. Interactive conversations were conducted, with the intention of developing key strategies that would lead to the future success of the Department of Genetics.

Overall feedback for both events from participants proved to be positive, with face-to-face interactions proving to be an effective and useful tool with the creation of future collaborations within the department.



Recent Faculty Publications

Differential Requirement for DICER1 Activity during the Development of Mitral and Tricuspid Valves. **Yan S**, Peng Y., Lu J, Shakil S., Shi Y., Crossman DK, Johnson WH, Liu S., Rokosh DG, Lincoln J., Wang Q., Jiao K. *Journal of Cell Science*. 2022 Sep 1; Vol 135, Issue 17. doi: 10.1242/jcs.259783.

Hydroxyproline Stimulates Inflammation and Reprograms Macrophage Signaling in a Rat Kidney Stone Model. **Kumar P.**, Yang Z., Lever JM, Chávez MD, Fatima H., Crossman DK, Maynard CL, George JF, Mitchell T. *Biochimica et Biophysica Acta - Molecular Basis of Disease*. 2022 Sep 1; Vol 1868, Issue 9:166442. doi: 10.1016/j.bbadis.2022.166442. Epub 2022 May 10.

DOCKopathies: A Systematic Review of the Clinical Pathologies Associated with Human DOCK Pathogenic Variants. Human Mutation. 2022 Sep; Vol 43, Issue 9, pp. 1149-1161. doi: 10.1002/humu.24398. Epub 2022 May 20.

Phosphatase Control of Cytokine-mediated Overproduction of Galactose-Deficient IgA1, the Main Autoantigen in IgA Nephropathy. **Reily C.**, Rice T., Crossman DK, Rizk DV. *Journal of Autoimmunity*. 2022 Oct; 132:102883. doi: 10.1016/j.jaut.2022.102883. Epub 2022 Aug 17.

WINNER: A Network Biology Tool for Biomolecular Characterization and Prioritization. **Nguyen T**, Yue Z, **Slominski R.**, Welner R, Zhang J, **Chen JY**. *Front Big Data*. 2022 Nov 4; doi: 10.3389/fdata.2022.1016606. eCollection 2022.

Lessons in Genetics: Common Diseases Occur in Patients with a Genetic Syndrome. **Robin NH**. *American Journal of Medical Genetics*. 2022 Nov 16. doi: 10.1002/ajmg.a.63046.

Return of non-ACMG Recommended Incidental Genetic Findings to Pediatric Patients: Considerations and Opportunities from Experiences in Genomic Sequencing. **Bowling KM**, Thompson ML, Kelly MA, Scollon S, Slavotinek AM, Powell BC, Kirmse BM, Hendon LG, Brothers KB, Korf BR, Cooper GM, Greally JM, Hurst ACE. *Genome Medicine*. 2022 Nov 21; Vol 14, Issue 1, p. 131. doi: 10.1186/s13073-022-01139-2.

Low c-Kit Expression Identifies Primitive Therapy-resistant CML Stem Cells. **Shah M.**, Kumar H., Qiu S., Li H., Harris M., He J., Abraham A., Crossman DK, Paterson A., Welner RS, Bhatia R. *JCI Insight*. 2022 Nov 22:e157421. doi: 10.1172/jci.insight.157421. Online ahead of print.

Protein Arginine Methyltransferase 1 in the Generation of Immune Megakaryocytes: A Perspective Review. **Zhao X, Chong Z**, Chen Y, Zheng XL, Wang QF, Li Y. *Journal of Biological Chemistry*. 2022 Nov. Vol 298, Issue 11: 102517. doi: 10.1016/j.jbc.2022.102517. Epub 2022 Sep 21.

Autophagy Inhibition Impairs Leukemia Stem Cell Function in FLT3-ITD AML, but has Antagonistic Interactions with Tyrosine Kinase Inhibition. **Qiu S**, Kumar H, Yan C, Li H, Paterson AJ, Anderson NR, He J, Yang J, Xie M, Crossman DK, Lu R, Welner RS, Bhatia R. *Leukemia*. 2022 Nov. Vol 36, Issue 11: pp. 2621-2633. doi: 10.1038/s41375-022-01719-6. Epub 2022 Oct 11.

Neuroendocrine Signaling in the Skin with a Special Focus on the Epidermal Neuropeptides. **Slominski AT, Slominski RM**, Raman C, **Chen JY**, Athar M, Elmets C. *American Journal of Cell Physiology*. 2022 Dec. Vol. 323, Issue 6: C1757-C1776. doi: 10.1152/ajpcell.00147.2022. Epub 2022 Nov 1.

Editorial: Prenatal Genetic Testing. Jenkins M, Seasely AR, Subramaniam A, **Robin NH**. *Current Opinion in Pediatrics*. 2022 Dec 1. Vol 34, Issue 6: pp. 531-532. doi: 10.1097/MOP.00000000001175.

PIM3 Kinase Promotes Tumor Metastasis in Hepatoblastoma by Upregulating Cell Surface Expression of Chemokine Receptor CXCR4. **Marayati R**, Julson J, Bownes LV, Quinn CH, Stafman LL, Beierle AM, Markert HR, Hutchins SC, Stewart JE, Crossman DK, Hjelmeland AB, Mroczek-Musulman E, Beierle EA. *Clinical and Experimental Metastasis*. 2022 Dec. Vol 39, Issue 6: pp. 899-912. doi: 10.1007/s10585-022-10186-3. Epub 2022 Oct 31.

Differential Impact of PI3K/AKT/mTOR Signaling on Tumor Initiation and Progression in Animal Models of Prostate Cancer. **Wang S**, Zhang C, Xu Z, Chen MH, Yu H, Wang L, Liu R. *The Prostate*. 2023 Jan. Vol 83, Issue 1: pp. 97-108. doi: 10.1002/pros.24441. Epub 2022 Sep 26.

Return of non-ACMG Recommended Incidental Genetic Findings to Pediatric Patients: Considerations and Opportunities from Experiences in Genomic Sequencing. **Bowling KM**, Thompson ML, Kelly MA, Scollon S, Slavotinek AM, Powell BC, Kirmse BM, Hendon LG, Brothers KB, **Korf BR**, Cooper GM, Greally JM, **Hurst ACE**. *Genome Medicine*. 2022 Nov 21. Vol 14, Issue 1: 131. doi: 10.1186/s13073-022-01139-2.

Recent Faculty Grant Awards

PI: Amindya Dutta, Ph.D.

Sponsor: NIH

Title: Involvement of Noncanonical Short RNAs in Gene Repression through the RNA-

Induced Silencing Complex

PI: Jia Xu, Ph.D.

Sponsor: BCRF (Breast Cancer Research Foundation) of Alabama

Title: Developing Novel AKT Degraders to Selectively Inhibit the Growth of PI3K/AKT/PTEN

Pathway Mutant Breast Cancer

PI: Keshav Singh, Ph.D.

Sponsor: BCRF (Breast Cancer Research Foundation) of Alabama

Title: Intercellular Mitochondrial Trafficking as a Novel Mechanism in Breast Cancer

Progression and Metastasis

PI: Amindya Dutta, Ph.D.

Sponsor: BRCF (Breast Cancer Research Foundation) of Alabama

Title: FAM129B/NIBAN2 as a Biomarker for Therapy with NRF2 Inhibitors

Welcome to the Department

New Student Volunteers

New Faculty Yuechuan Chen Wioletta Czaja Chen Huang Sabiquan Nahar Jia Xu

Han Jing Shamza Manzoor Raventh Sanagapalli Tulya Rupa Sri Vankayala Pati Beining Tian

Yuwei Zhang

Anna Crider Wendy Wess Marcello Sable Washington

New Staff

UAB-HudsonAlpha Genomic Medicine PostDoctoral Training Program

This training grant is an Institutional Ruth L. Kirschstein
National Research Service Award (NRSA) T32 grant funded by the
NHGRI within the National Institutes of Health (NIH), created to prepare
postdoctoral trainees for academic careers in genomic medicine.

The overall goals of the Genomic Medicine Training Program are:

- To develop experimental competence in our trainees so that they can become productive and independent investigators;
- To develop skills in our trainees that will enable them to evaluate critically their own research endeavors as well as those of other laboratories/investigators;
- To provide our trainees with a background in genomics and bioinformatics with the intention of demonstrating how a genomic approach can be used to solve problems in prevention, diagnosis, and treatment of human disease;
- To familiarize trainees with the many modern tools of genomics research that are available in our laboratories, and to encourage cooperation and collaboration in solving experimental problems.

Who is appointed to this NHGRI T32 training grant?

We have structured a 2-year period of research and education for each of the postdoctoral fellows entering the training program. We expect to recruit trainees with three different types of backgrounds, and will aim to balance the roster of trainees equally among these groups:

- MDs who have completed clinical training in medical genetics and are eligible for certification by the American Board of Medical Genetics and Genomics;
- MDs, including those who have completed clinical training in medical genetics and are eligible for certification by the American Board of Medical Genetics and Genomics, or who have completed training in a non-genetics related discipline;
- PhDs or PharmDs who have decided to focus their careers on clinical or translational research related to genomic medicine.



How to Apply

Our fellowship accepts applications year-round. Candidates will have the opportunity to work either at The University of Alabama at Birmingham or HudsonAlpha Institute for Biotechnology at Huntsville.

We strongly encourage and solicit applications from women and under-represented minorities. We currently accept two trainees per year.

To learn more and to apply: https://www.uab.edu/medicine/genetics/t32/apply









The University of Alabama at Birmingham



UAB Genetics | News & Events

Editor: Ryan McCain

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