MEDICAL SCHOOL
UNIVERSITY OF MICHIGAN

DEPARTMENT OF
HUMAN GENETICS

HUMAN GENETICS

ISSUE 22 • FALL 2019

BIG DATA
BIG QUESTIONS

WHAT IS BIG DATA?

The advent of new technologies for creating and studying biological data has created an avalanche of new information that is often beyond the capabilities of a single individual to analyze and interpret using standard approaches. The resulting "big data" reflects not only the vast amounts of information that is being stored (volume), but also the rate of data generation (velocity) and the complexity of the data (variety). This is reflected in studies of human genetics by the ever-increasing amounts of next generation sequence data being used to study and diagnose human diseases. Researchers are developing new strategies for leveraging this deluge of information to identify new and exciting patterns in the data that are furthering our understanding of how these diseases originate and progress.

DEPARTMENT OF HUMAN GENETICS

NEWSLETTER

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A SEASON OF CHANGE IN HUMAN GENETICS AND GENOMICS

THE CHAIR'S MESSAGE

GREETINGS FRIENDS AND COLLEAGUES OF THE DEPARTMENT OF HUMAN GENETICS!

Each Fall we get a colorful reminder of how the world around us changes and how this impacts our outlook and experiences. Over the past year we have witnessed many changes in our active and ever-evolving Department. As we begin a new academic year, we look forward with excitement to further positive changes as we strive to advance our Department's missions in education and research.

> **"ONE CRITICAL GOAL OF OUR DEPARTMENT IS TO TRAIN THE NEXT GENERATION OF GENETICISTS"**

One critical goal of our Department is to train the next generation of geneticists, which is driven by our Masters, Genetic Counseling, and Ph.D. graduate programs, and by our efforts to train post-doctoral fellows and research staff. Over the past year we successfully graduated 19 students in the above programs (see page 7), many of whom have gone on to excellent positions in genetics. This coming year we welcome five Masters students, ten Genetic Counseling students, and seven Ph.D. students to our Department, and we are eager to work with each of these students during their training. Another major change was in renaming our Ph.D. program to the "Genetics and Genomics Graduate Program" (see page 6). We feel this name better reflects the research of our faculty and the content of our curriculum, and we are hopeful that this will draw more doctoral students to laboratories in our Department.

An equally important goal is to generate knowledge in the area of genetic research. Impressively, in the past year our Department published over 120 manuscripts in excellent journals; many of these studies were directed and written by our students. As recognition for this productivity, many individuals were honored with prestigious awards (see pages 7-8). In the current era of genetic and genomic research, we have the ability to generate and analyze large data sets that assist us in defining genetic mechanisms. In this issue of our newsletter, we feature some exciting research in "Big Data" from Department labs and highlight the trainees that are driving these projects (see page 4).

Another change that we look forward to is our renewed efforts to recruit faculty members performing cutting-edge research in genetics and genomics. With the support of the Medical School and in coordination with units across UM,

> **"WE ARE DELIGHTED TO ANNOUNCE THAT WE HAVE MULTIPLE POSITIONS OPEN FOR FACULTY"**

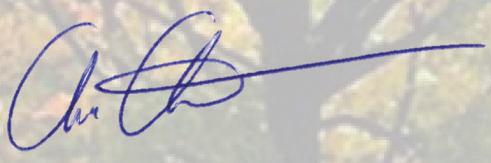
we are delighted to announce that we have multiple positions open for faculty to join our collaborative, tight-knit community. Indeed, Stephanie Moon, Ph.D. will join us this year as an Assistant Professor. Dr. Moon's research is on the mechanisms of RNA processing and how they relate to neurological disease (see page 9).

Finally, my role as Chair was another major change. I am humbled and honored to be charged with leading our Department and with maintaining our outstanding reputation.

> **"I OWE A HUGE DEBT TO MY IMMEDIATE PREDECESSORS SALLY CAMPER AND DAVE BURKE FOR THEIR LEADERSHIP"**

I owe a huge debt to my immediate predecessors - Sally Camper and Dave Burke - for their leadership and for helping with this transition. I have a lot to learn about being a Chair and about the needs of our faculty, trainees, and staff. However, as I navigate this challenge, I look forward to working with everyone at Michigan to advance our education and research missions.

So, enjoy the Fall and I look forward to seeing many of you in the coming year!



Tony Antonellis, Ph.D.
Chair, Department of Human Genetics

NEW APPROACHES TO ANCIENT PROBLEMS:

EXAMINING SPERMATOGENESIS THROUGH THE LENS OF EVOLUTION

HAMMOUD LAB & LI LAB COLLABORATION

Sperm are highly specialized cells that carry the genetic information from father to offspring and provide a continuous link between the past, present, and future of a species. Although essential for both survival and evolution of a species, we do not clearly understand how this process is regulated and how these factors may have changed over time. Extensive research in rodents has begun to shed light on the intrinsic (germ cell driven) and extrinsic (somatic cell driven) programs underlying spermatogenesis. Unfortunately, this knowledge often does not translate to humans, severely limiting our efforts to identify and treat causes of male infertility.

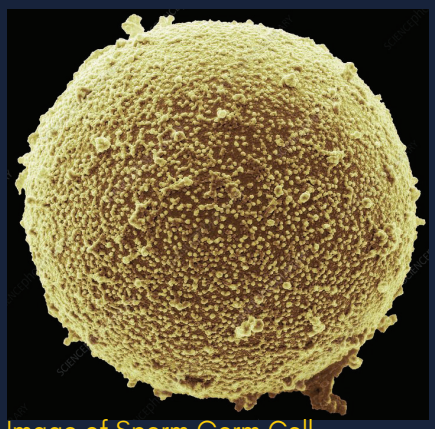
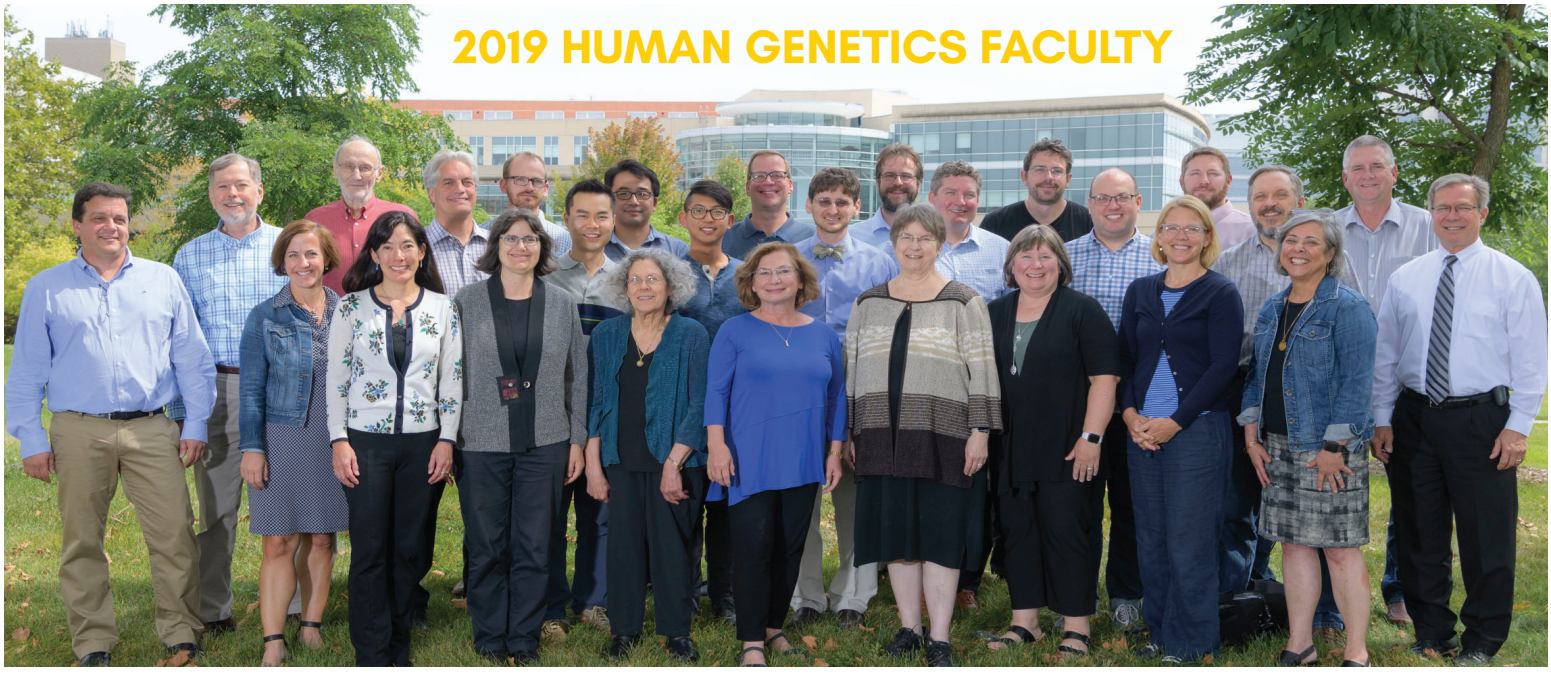


Image of Sperm Germ Cell
By Steve Gschmeissner

A collaboration between the Hammoud (Adrienne Niederriter Shami) and Li (Xianing Zheng and Qianyi Ma) labs seeks to make novel comparisons between primate and rodent testes to uncover conserved and diverged molecular programs required to produce functional sperm. Using single cell RNA-sequencing of mouse, human, and macaque testes, the team was able to capture thousands of developing germ cells and the somatic cells that support them, and use bioinformatics approaches to identify and compare analogous molecular states throughout the course of the spermatogenic program. This has uncovered striking differences between species on the cellular level through new populations, as well as on the gene level with subtler changes in expression and communication with nearby cells--indicating that some cells are not as similar between species as once thought. By identifying conserved and divergent regulators of spermatogenesis between primates and rodents, new insights can be used to develop new techniques to restore fertility, including efforts towards making germ cells in a dish.

Adrienne Niederriter Shami
Xianing Zheng,
Qianyi Ma



BIG DATA ALLOWS INFORMED PREDICTIONS

Sierra Nishizaki, Ph.D. Candidate
The Boyle Lab

If you were to take one of each of the 23 chromosomes in the human genome and line them up end-to-end it would be about 40 inches long, about as long as an average baseball bat. However, only 0.8 inches of this distance, about the knob of a baseball bat, encodes for genes and leads to functional protein products. If only 2% of the genome encodes proteins, what is the other 98% of the genome doing? This question is one that Sierra Nishizaki, a PhD student in the laboratory of Dr. Alan Boyle, is trying to answer using BIG DATA.


The Boyle Lab

Some of this 98% represents regulatory regions of the genome known to orchestrate gene expression. By examining these regions, we can identify places where mutations would disrupt normal gene expression - and possibly lead to human disease. However, regulatory regions represent 4x more of the genome than functional genes, comprising ~240 million potentially important bases to sift through. However, by using data from large consortium studies, such as ENCODE, Sierra is developing computational approaches to predict key mutations more likely to alter gene expression. By integrating big data containing genome-wide information on genomic accessibility and protein binding, we can build a global view of how individual mutations may affect protein binding and gene expression. While these computational approaches cannot replace experimental ones, they can help geneticists focus their research to more quickly advance our understanding of human disease. For more details, please see our recent article in the journal Bioinformatics.

BIG DATA FROM SMALL NUMBERS

Yifan Wang, Ph.D.
The Mills Lab

The search for genetic origins of disease phenotypes has led to the sequencing and analysis of DNA and RNA from hundreds of thousands of individuals. These large-scale projects consume massive amounts of storage and computing capacity and are rightly considered 'BIG DATA'. However, there is still much we do not know even within a single genome. The Mills Lab develops and applies computational approaches for diving deep into individual samples and cells to explore regions of the genome that have been previously overlooked and could potentially harbor mutations which are clinically relevant. They do this by applying and combining multiple technologies to tease apart complex and repetitive parts of the genome, which has resulted in hundreds of terabytes of data across a small number of families and the identification of many novel mutations.

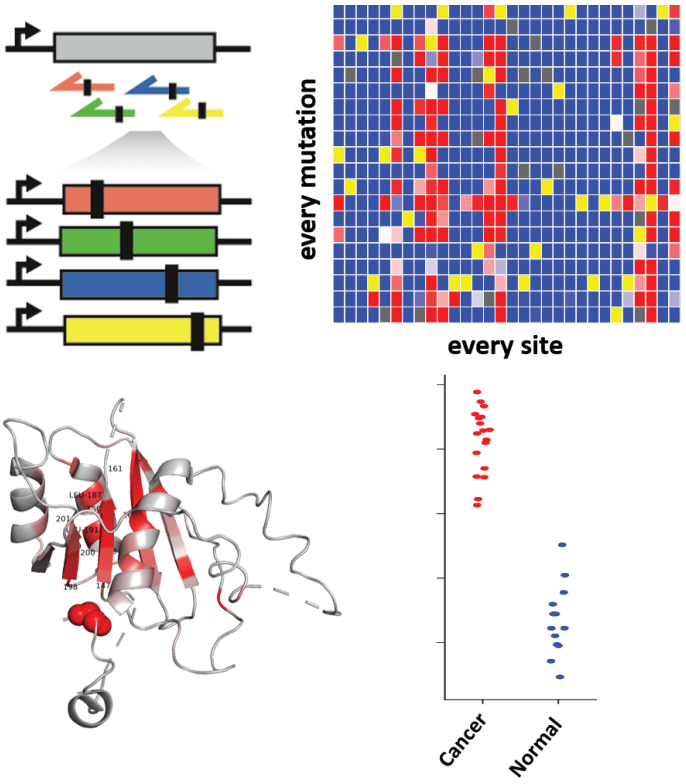
Although fruitful, this level of analysis was not enough for Dr. Yifan Wang, a former Human Genetics graduate student in the lab who has continued on as a postdoctoral fellow. Dr. Wang's research goes even deeper by investigating how the DNA in individual cells can differ from each other, and to do this she must examine thousands of distinct molecules from each cell. She is currently playing a major role in the Brain Somatic Mosaicism Network, a multi-institutional collaboration that seeks to map out these differences to determine what impact these mutations may have on neuropsychiatric disorders such as Schizophrenia. Her work is further providing a foundation for studies of mosaicism in other tissues and diseases.

TACKLING VARIANTS
OF UNCERTAIN SIGNIFICANCE

JACOB KITZMAN, Ph.D.
ASSISTANT PROFESSOR
HUMAN GENETICS

GIVEN THE UNIQUE SET OF VARIANTS FOUND IN AN INDIVIDUAL HUMAN GENOME, HOW CAN WE DETERMINE WHICH ARE PATHOGENIC AMONG A SEA OF BENIGN ONES?

This problem – variant interpretation – is one of the major challenges facing geneticists today. The lab of Dr. Jacob Kitzman, Assistant Professor of Human Genetics and Computational Medicine and Bioinformatics, aims to address this problem through functional assays which combine high-throughput experimental screens with data mining and statistical modeling. As a long term goal, these projects seek to measure the functional impact of every possible mutation in clinically important genes and regulatory elements. The resulting data can be mined to identify new pathogenic mutations before they are found in the clinic, potentially enabling early detection and steering the course of treatment or monitoring for carriers. Projects in Dr. Kitzman's lab address this problem at each level of the central dogma, dissecting how variants impact protein function, RNA splicing, and transcriptional regulation, across a variety of Mendelian and complex disorders.



THE MORAN LAB:
MITSUHIRO NAKAMURA
POSTDOCTORAL FELLOW

Mobile genetic elements (a.k.a., "jumping genes") are sequences that can move from one location in the genome to another. Dr. Nakamura studies a class of mobile genetic elements called LINE-1 retrotransposons. LINE-1-derived sequences comprise approximately 17% of the human genome and ongoing LINE-1 mediated insertions are responsible for approximately 1 out of 250 non-recurrent disease-producing human mutations.

Active LINE-1s encode two proteins (ORF1p and ORF2p) required for their mobility. Dr. Nakamura's research uses biochemical, genetic, and molecular biological approaches to determine how LINE-1 retrotransposons can move to new genomic positions.

Dr. Nakamura has generated evidence that ORF2p contains a previously undiscovered single strand endonuclease activity, which may facilitate LINE-1 integration at endogenous DNA lesions. A similar endonuclease activity also is contained within the telomerase protein, which elongates DNA ends to ensure chromosome stability. Because the reverse transcriptase domains within LINE-1 ORF2p and telomerase proteins are likely derived from a common ancestor, Dr. Nakamura and colleagues propose that this newly discovered ORF2p endonuclease activity originated early in eukaryotic evolution prior to the divergence of LINE-like retrotransposons and telomerase. Thus, Dr. Nakamura's research provides new insights into LINE-1 biology and highlights mechanistic similarities between LINE-1 mobility and the action of telomerase.

ACADEMIC PROGRAMS

HUMAN GENETICS MASTERS PROGRAM

The Masters Program (M.S.) in Human Genetics provides advanced classroom and research training in human genetics, genomics and molecular biology. In 2014, the program was redesigned to include a Research Track, which provides highly motivated students with the opportunity to conduct cutting edge genetics research and write a Masters thesis. The number of outstanding applicants to the HG M.S. program has steadily increased each year, and admission is highly competitive. Each matriculating class of MS students is comprised of students from diverse backgrounds with distinct career goals. Our MS students complete their degree with a strong foundation of knowledge in genetics, genomics, bioinformatics and computational genomics. Graduates have been highly successful in pursuing a broad range of career paths. Over the past three years, our 14 M.S. graduates have gone on to Ph.D. programs at Stanford, Michigan and Vanderbilt, the M.D. program at the University of Texas, senior research positions in university research labs and the biotechnology industry and faculty positions at the University of Wisconsin and Universities in Ghana and Nigeria. We are excited to welcome 5 new HG M.S. students this Fall and look forward to helping them to achieve their career goals.

GENETICS & GENOMICS GRADUATE PROGRAM

We are pleased to introduce the Genetics and Genomics Graduate Program! To reflect the research of our faculty, the content of our curriculum, and the evolving demands of Ph.D. students in the biological sciences, the Department approved a name change for our doctoral program. Students entering the Genetics and Genomics Graduate Program will have access to over 30 outstanding faculty members addressing diverse questions in genetics and genomics research, including: developmental genetics; molecular genetics; evolutionary and population genetics; genome structure and function; the etiology of complex traits and diseases; and the molecular basis of Mendelian disease. To enhance their research training, Ph.D. students will study in a literature-based curriculum that integrates well with other coursework available at Michigan. As part of the Program in Biomedical Sciences (PIBS), the Genetics and Genomics Graduate Program provides a unique scientific environment for students interested in any area of genetic and genomic research.

GENETIC COUNSELING PROGRAM

2019 marks the 40th year of our Genetic Counseling Program (UMGCP). Since its inception, the UMGCP has provided a comprehensive and rigorous curriculum that relies heavily on the strengths of the Department of Human Genetics, Michigan Medicine's clinical enterprise, and the broader UM community. Our graduates can be found using their expertise to provide clinical care, conduct research, shape public policy and inform commercial genomic applications.

40 years ago, our class sizes were 4 or less and our clinical training team in Ann Arbor consisted of fewer than 5 GCs in 2 clinical settings. This fall we welcomed 10 incoming students and 11 returning students. Our current student class surpasses our recent strategic goal of "20 students by 2020." This goal was designed to respond to demands for genetic counselors both locally and nationally.

Today our students have the opportunity to learn with greater than 30 physicians and 25 GCs at Michigan Medicine genetics clinics in cardiology, ophthalmology, internal medicine, pediatrics, oncology, and maternal fetal medicine. Michigan Medicine's workforce expansion allows us to stay true to our commitment to provide an individualized, integrated and supportive environment. Collaborations with partners across the university have allowed us to make important strides in increasing the diversity of our program. Nineteen percent of our student body is from traditionally underrepresented minorities, 25% were Pell grant recipients as undergraduates, and 20% are males.

In 2020, we will embark on strategic planning as part of a reaccreditation review by the Accreditation Council of Genetic Counseling. We look forward to hearing input from our alumni, faculty, students, and community members as we vision for the future and think about how we can continue to get better over time.

Prospective students apply: https://medicine.umich.edu/medschool/education/phd-programs/phd-admissions

GRADUATE STUDENT AWARDS

Elysa Bond, GC Program
2019-2020 Jane Engelberg Memorial Fellowship Student Research Award; 2019 James V. Neel Genetic Counseling Fellowship Award

Marissa Cloutier, Kalantry Lab
Career Training in Reproductive Biology Training Grant Fellowship; Van Andel Epigenomics Workshop Scholarship

Alyssa Kruger, Mueller Lab
James V. Neel Doctoral Fellowship; National Science Foundation Graduate Research Fellowship

Rebecca Meyer-Schuman Antonellis Lab
Excellence in Basic Science Award; 2019 Gordon Seminar Outstanding Speaker Award; Semi-finalist, ASHG/Epstein Trainee Award

Ilana Miller, GC Program
NSGC Genetics & Precision Medicine Fellowship

Amanda Moccia, Bielas Lab
EDGE Research Award; Global Research Engagement Opportunity Award

Yanchao Pan, M.S. Program
Anita and Howard Cramer Award for Excellence in Academics

Robert Porter, Iwase Lab
Excellence in Basic Science Award

Adrienne Shami, Hammoud Lab
George J. and Lucia Brewer Scholarship Award; Excellence in Basic Science Award

Elise Sobotka, GC Program
Jane Engelberg Memorial Fellowship Student Research Award; Public Health Genetics and Precision Medicine Fellowship

Megan Trotter, Kalantry Lab
Van Andel Epigenomics Workshop Scholarship; 1st Place Scientific Image, RNA Biomedicine Symposium

Elizabeth Werren, Bielas Lab
National Science Foundation Doctoral Dissertation Research Improvement Grant; Leakey Foundation Dissertation Research Grant

CONGRATULATIONS
2019 GRADUATES

Adelyn Beil, GC M.S.
Matthew Breneman, HG M.S.
Colby Chase, GC M.S.
Jason Keil, Ph.D.
Kelsey Lenhart, GC M.S.
Kelsie McVeety, GC M.S.
Mackenzie Mosera, GC M.S.
Katherine Ozelius, GC M.S.
Yanchao Pan, HG M.S.
Amanda Schaefer, GC M.S.
Lauren Schmitz, HG M.S.
Lauren Seeman, GC M.S.
Feichen Shen, Ph.D
Corrine Smolen, HG M.S.
Christina Vallianatos, Ph.D.
Arushi Varshney, Ph.D.
Natalie Waligorski, GC M.S.
Yifan Wang, Ph.D.

2019 GENETIC COUNSELING PROGRAM GRADUATES

FACULTY & POSTDOCTORAL . . .


Stephanie Bielas


Shigeki Iwase


Steven Parker


Cristen Willer


Stephanie Moon


Lev Prasov


Thomas Glover


John Moran


Jacob Mueller


Hironori Bando


Xiaoyan Isaac Jia


Laura Kirby


Elaine Ritter


Wenxi Yu

APPOINTMENTS

FACULTY PROMOTIONS

Anthony Antonellis, Ph.D.
appointed Chair of the Department of Human Genetics

Stephanie Bielas, Ph.D.
promoted to Associate Professor of Human Genetics

Shigeki Iwase, Ph.D.
promoted to Associate Professor of Human Genetics

Stephen Parker, Ph.D.
promoted to Associate Professor of Human Genetics

Cristen Willer, Ph.D.
named Co-Director of Cohort Development with U-M Precision Health

NEW FACULTY

Stephanie Moon, Ph.D.
Assistant Professor of Human Genetics and Faculty Scholar of the Center for RNA Biomedicine

Lev Prasov, M.D., Ph.D.
Assistant Professor of Ophthalmology and Visual Sciences with a joint appointment with Human Genetics

ACCOMPLISHMENTS

FACULTY AWARDS

Thomas Glover, Ph.D., FACMG
2018 Distinguished Faculty Lectureship Award in Biomedical Research, UM Medical School; 2018 Elected Fellow, American Association for the Advancement of Science

John Moran, Ph.D.
2019 Distinguished Faculty Lectureship Award in Biomedical Research

Jacob Mueller, Ph.D.
2019 Endowment for Basic Science (EBS) Teaching Award

POSTDOCTORAL AWARDS

Hironori Bando, Ph.D.
2019 Oustanding Abstract Award, The Endocrine Society's 101st Annual Meeting; Early Career Forum Travel Award

Xiaoyan Isaac Jia, Ph.D.
2019 Oustanding Speaker Award, Gordon Research Seminar on Human Genetics & Genomics

Laura Kirby, Ph.D.
elected to the Michigan Life Sciences Fellows Program

Elaine Ritter, Ph.D.
Sandra Davenport Fellowship, CHARGE Syndrome Foundation; Hearing, Balance, & Chemical Senses T32 Training Grant; Association for Research in Otolaryngology (ARO) Travel Award

Wenxi Yu, Ph.D.
elected into the American Epilepsy Society Fellows Program

RECENT ALUMNI

Peter Gergics, M.D., Ph.D., resident in Internal Medicine at St. Joseph Ann Arbor

Jacy Wagnon, Ph.D., faculty member at The Ohio State University

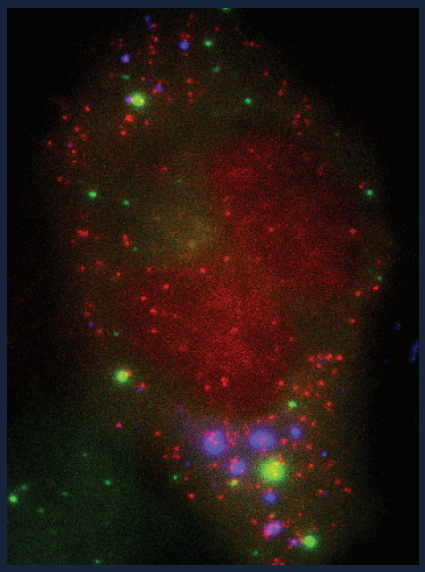
U-M BIOSCIENCES INITIATIVE

Stephanie Moon, Ph.D.

Assistant Professor of Human Genetics; Faculty Scholar of the Center for RNA Biomedicine

Advances in microscopy have allowed biomedical scientists to begin to explore the spatial and temporal dynamics of gene regulation in living cells virtually in real-time. These high-dimensional image-based datasets have the potential to yield information about the molecular underpinnings of disease. For instance, chronic activation of cellular stress response pathways is observed in tissues and cells in many different neurodegenerative disease contexts. Therefore, an important question to address is how cellular stress response pathways affect health and, when mis-regulated, contribute to disease.

As a first step towards answering this question, we used powerful fluorescence microscopy techniques to evaluate how the stress response affects, in real-time, messenger RNAs in living human cells. This method allowed us to simultaneously monitor the movement and translational status of mRNAs within the cell, while measuring the dynamics of their interactions with RNA-protein granules, called P-bodies and stress granules. By generating a high-dimensional dataset that includes the spatiotemporal behavior of three distinct components, we were able to ask questions and test hypotheses that would otherwise be inaccessible using traditional molecular biology approaches that measure population-level behavior in a single point in time. We now have a better understanding of how post-transcriptional gene regulation is dynamically altered during stress responses, and what role P-bodies and stress granules could play in mediating this process. Importantly, this work sets a foundation for our future research in examining how neurological diseases affect mRNA regulation and informs new therapeutic interventions.



Individual mRNAs (red) can be observed interacting with P-bodies (green) and stress granules (blue) during a stress response (Moon et al., 2019 Nat. Cell Biol.).

> **"We are thrilled to announce our first Biosciences Faculty hire, Dr. Stephanie Moon. We would like to take this opportunity to express our appreciation to the Biosciences Initiative, the Biological Sciences Scholars Program, the Neuroscience Scholars Program, and the Department of Human Genetics who partnered with us to attract such a talented candidate. We are very excited for her future with the University of Michigan and the Center for RNA Biomedicine."**
>
> SALLY CAMPER, PH.D.

THE DIVERSE ROLES OF RNA

Sundeep Kalantry, Ph.D.
Associate Professor, Human Genetics

The central dogma of molecular biology, established in the 1960's, states that DNA generates RNA, which in turn generates proteins. However, research, especially over the last twenty years, has made it abundantly clear that RNA is not merely an intermediate between DNA and proteins. It turns out that RNAs can regulate DNA, other RNAs, and also proteins. Current estimates show that three-quarters or more of our DNA is in fact transcribed in a temporally or tissue-specific manner. Much of this transcriptome is noncoding - the RNAs do not code for proteins.

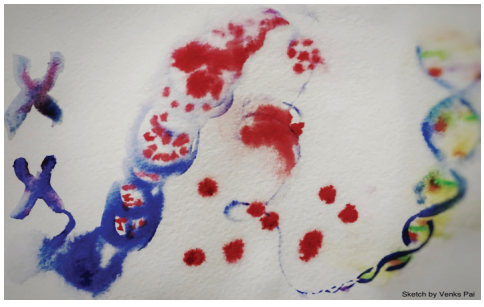


IMAGE OF UNRAVELING DNA

While genetic studies demonstrate that not all noncoding RNAs are functional, some have discrete regulatory roles. These roles include: recruiting proteins to DNAs to regulate DNA metabolism, from transcription to DNA repair; recruiting proteins to other RNAs, to degrade or prevent their translation; inhibiting or potentiating activity of bound proteins; RNAs themselves functioning as enzymes; and, generating cDNAs that integrate back into the DNA to alter and shape genomes. Although our appreciation for the diverse functions of RNA has grown, it would be hubris to think that we have uncovered the many lives of RNA beyond generating proteins! Surely, the future holds many exciting discoveries into how RNAs influence physiology.

UPCOMING ANNUAL LECTURES

9TH ANNUAL THOMAS D. GELEHRTER LECTURESHIP IN MEDICAL GENETICS

TUESDAY, OCTOBER 29, 2019
KAHN AUDITORIUM
BIOMEDICAL SCIENCE RESEARCH BUILDING

HELEN H. HOBBS, M.D.
INVESTIGATOR OF THE HOWARD HUGHES MEDICAL INSTITUTE;
PROFESSOR OF INTERNAL MEDICINE AND MOLECULAR GENETICS;
UNIVERSITY OF TEXAS SOUTHWESTERN MEDICAL CENTER

This annual lectureship honors Dr. Thomas D. Gelehrter, a leader within the human genetics community and internationally recognized as an expert in human genetics. Dr. Gelehrter joined the University of Michigan Medical School faculty in 1974 and served as chair of the Department of Human Genetics for 17 years, from 1987 to 2004. He is currently an active Professor Emeritus in the department. The Lectureship builds upon Dr. Gelehrter's career dedicated to promoting excellence in research, education, and care in medical genetics and will be an enduring legacy that continues to raise awareness about the importance of medical genetics and improve this vital field. A prominent scientist in the field of medical genetics who embodies the principles that inspired Dr. Gelehrter's outstanding contributions to human genetic research, teaching, and patient care is invited to speak at the annual lectureship.

20TH ANNUAL JAMES V. NEEL LECTURESHIP IN HUMAN GENETICS

ERIC LANDER, PH.D.
PRESIDENT AND FOUNDING DIRECTOR
BROAD INSTITUTE OF MIT AND HARVARD

TUESDAY, APRIL 21, 2020
FORUM HALL, PALMER COMMONS

James Van Gundia Neel, M.D., Ph.D., was a pioneer in the study of human genetics and one of the first to foresee its importance in the diagnosis and treatment of medical conditions. During his 39-year career in the U-M Medical School, Neel established one of the first clinics to evaluate and counsel people with hereditary diseases, as well as the first academic department of human genetics in the United States. During a distinguished career spanning more than 60 years, he made major contributions to our understanding of: (i) the genetics of several human diseases including diabetes mellitus, neurofibromatosis, and sickle cell anemia; (ii) the effects of atomic radiation on humans; and (iii) the genetic structure of Amerindian populations. The James V. Neel Lecture in Human Genetics honors the legacy of James Neel through lectures by prominent genetic researchers and fellowship awards to outstanding graduate students.

IN MEMORIAM

We are sad to share the passing of **Dr. Ernest (Ernie) Chu**, who was an integral member of the Department of Human Genetics.

Ernest H. Y. Chu
Emeritus Professor, Human Genetics
June 1927 - April 2019

Dr. Chu immigrated to the US from China and earned his Ph.D. in Genetics at University of California, Berkeley. He held positions at Yale and Oak Ridge National Laboratory in Tennessee prior to joining our faculty in 1972; he retired in 1992. Dr. Chu is considered one of the pioneers in the study of chromosomes. He and others in the field of somatic cell genetics were highly active in mapping genes and cellular traits to specific areas of human chromosomes. This was an exciting time in human genetics, and Dr. Chu's research laid the groundwork for the Human Genome Project, which was launched in 1990 and declared complete in 2003.

PHILANTHROPY

FARREHI FAMILY

THE FARREHI FAMILY EDUCATION FUND

Shigeki Iwase, Ph.D.
Associate Professor of Human Genetics

We would like to thank the Farrehi Family for their generous donation to the Iwase Lab. The support from the Farrehi Family has enabled us to study KDM5C - deficiency in a young male with autism & intellectual disability. This study is both time and resource consuming, yet an inevitable step for future therapeutic applications.

The most important aspect of the Farrehi's support is that it has created seriousness about finding the next step for therapeutic intervention. We recently published a research article in Frontiers in Molecular Neuroscience and have a second article in the process of publication. A collaboration between several labs in Michigan, led by Christina Valliantos, a recent graduate of the lab, found a suppressor gene for KDM5C deficiency. This fund supports current efforts to test a small inhibitor of KMT2A, a promising therapeutic target. We hope to start improving the compound to increase the penetrance of the blood brain barrier.

We greatly appreciate the support from the Farrehi Family and look forward to the day we can eventually intervene in the KD-M5C deficiency and related disorders.

> "IT HAS BEEN SUCH A REWARDING EXPERIENCE FOR TRAINEES IN THE LAB TO ACTUALLY SEE WHAT THEY DO MATTERS AND THAT THERE ARE PEOPLE WAITING FOR THE PROGRESS OF OUR RESEARCH."

FUNDS THAT SUPPORT OUR STUDENTS AND RESEARCH

The Anita and Howard Cramer Fellowship Fund.

The George J. and Lucia F. Brewer Scholarship Fund.

The James V. Neel Fellowship Fund.

The Jane S. Schultz Fund.

The Myron Levine Memorial Research Fund.

FUNDS THAT SUPPORT OUR GENETIC COUNSELING PROGRAM

The Carole McTague Genetic Counseling Enrichment Fund.

FUNDS THAT SUPPORT OUR ANNUAL LECTURES

The Diane Baker Alumni Lecture.

The Thomas D. Gelehrter Lectureship.

The James V. Neel Lectureship.

THANK YOU

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DEPARTMENT OF HUMAN GENETICS
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Ann Arbor, MI 48109-5618

RETURN SERVICE REQUESTED

PHILANTHROPIC SUPPORT FOR RESEARCH

Philanthropic donations play a vital role in supporting research and education in the Department of Human Genetics. These valued contributions help launch new research projects, provide scholarships for outstanding students, and support student travel to scientific meetings. Endowed lectureships bring renowned leaders in Human Genetics and Genetic Counseling to the University of Michigan to promote the importance of genetics, educate the medical community and the public about promising new research, and provide opportunities for discussion and collaboration with students and faculty. To learn more about the impact of your gift for basic research, please view a movie at https://vimeo.com/292778380.

THANK YOU FOR YOUR SUPPORT!

GIFTS OF ANY SIZE SUPPORT
HUMAN GENETICS
STUDENTS AND RESEARCH.

TO DONATE TO ANY OF OUR INITIATIVES
USE THE ONLINE GIVING SITE:

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UNIVERSITY OF MICHIGAN

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