CLEMSON UNIVERSITY
CENTER FOR HUMAN GENETICS

The Transcript

Newsletter of the Center for Human Genetics at Clemson University
Fall, 2021

From the Director

We hope that the COVID pandemic will ultimately be winding down and that we can return to normal pre-pandemic conditions. Meanwhile, I want to express my appreciation for our faculty, students, and staff, who have diligently cooperated with the guidelines issued by the Clemson University administration and who have enabled the Center to maintain research productivity despite the operational restrictions. As the university is transitioning to in-person classes, I would like to remind everyone of the importance to continue to adhere to the mask mandate and regular COVID testing guidelines established by Clemson University.

Dr. Trudy F. C. Mackay, FRS, is the Self Family Endowed Chair of Human Genetics. She is a Fellow of the Royal Society of London, a member of the National Academy of Sciences of the USA, a member of the American Philosophical Society, and recipient of the 2016 Wolf Prize.

Earlier this year, we received a five-year $10,559,560 (direct cost) grant from the National Institute of General Medical Sciences to establish a Center of Biomedical Research Excellence (COBRE) in Human Genetics. The COBRE grant is in collaboration with the Greenwood Genetic Center. The objective of the COBRE grant program is to facilitate the professional development and transition to independent research support for young faculty at universities in states that are relatively underfunded in terms of NIH support. The first five-year grant is a Phase 1 funding period that can be renewed as a Phase 2 and ultimately a Phase 3 award, potentially providing up to 15 years of funding.

The goal of the COBRE in Human Genetics is to understand the genetic, genomic, and epigenetic mechanisms by which molecular genetic variation affects transcriptional and other molecular networks in health and disease. Unifying foci are the role of non-coding elements of the genome in gene regulation and computational approaches to improve genomic prediction to advance precision medicine. COBRE investigators leverage comparative genomic approaches using Drosophila, zebrafish, and human cell lines to gain insights into the mechanisms underpinning human diseases.

The COBRE in Human Genetics supports four project leaders, Drs. Andrei Alexandrov, Miriam Konkel, Fabio Morgante, and Heather Flanagan-Steet.

Dr. Andrei Alexandrov's research will provide insights into the roles of human long non-coding RNAs (lncRNAs) in risk for human disease and identify potential new therapeutic anti-cancer and anti-viral targets. Dr. Miriam Konkel's research mapping and defining characteristics of transposable element insertions in the human genome will yield valuable insight into this understudied but highly important source of variation and the role of transposable elements in rare and common disease. Dr. Fabio Morgante's research incorporating genotype by environment interaction and other context-dependent effects into statistical models predicting complex trait phenotypes from large scale genetic variation data will bring us closer to the goal of precision medicine as well as gain insight into the genetic architecture of cardiovascular disease. Dr. Heather Flanagan-Steet's research on the developmental effects and molecular genetic mechanisms that underlie the manifestation of congenital disorders of glycosylation will provide insights that may lead to novel therapeutic interventions to ameliorate patient's symptoms in this group of rare genetic disorders with variable penetrance. Together, these projects investigate the roles of gene regulation, post-translational modification and structural variation in common and rare diseases; develop more powerful models for genotype-phenotype prediction; and utilize systems genetics approaches in cellular and animal models to more fully understand the genome-wide impact of genetic variants on molecular networks and disease in human populations.

As part of the COBRE, the Center for Human Genetics has initiated a distinguished lecture series with prominent scientists. Thus far, we have been pleased to host Drs. Arpana Agrawal from Washington University in St. Louis, Carole Ober from the University of Chicago, and Teri Manolio from NHGRI as distinguished speakers.

The Center for Human Genetics welcomes its new graduate students: Rebecca Bishop, Bibhu Simkhada, Alex Froidceur, Austin Herbert, Naqing, Jamutai, Edward Mabry, Debarati Majumdar, Hui Ma, Baxton Munn, Chunming Liu, Jeffrey Che and Chia-Lun "Allen" Wu. Welcome also to Brian Kessler, our new molecular research technician, and postdoctoral fellow Kaya Yuan. A sad farewell and best wishes in the next stage in their careers to Marion Campbell, Miller Barksdale, Krista Knowles, and Lakshmi Sunkara.

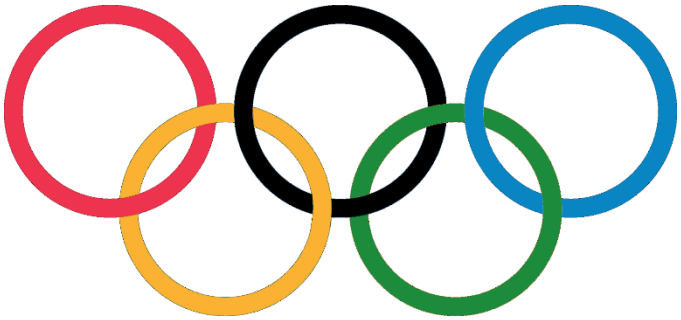
Finally, we congratulate Brandon Baker and Sneha Mokashi for finishing their doctoral degrees. Brandon has moved on to a position at IQVIA Biotech in Morrisville, NC. Sneha will stay nearby as a postdoctoral fellow with Dr. Heather Flanagan-Steet at the Greenwood Genetic Center

I am looking forward to a successful and productive Fall semester.

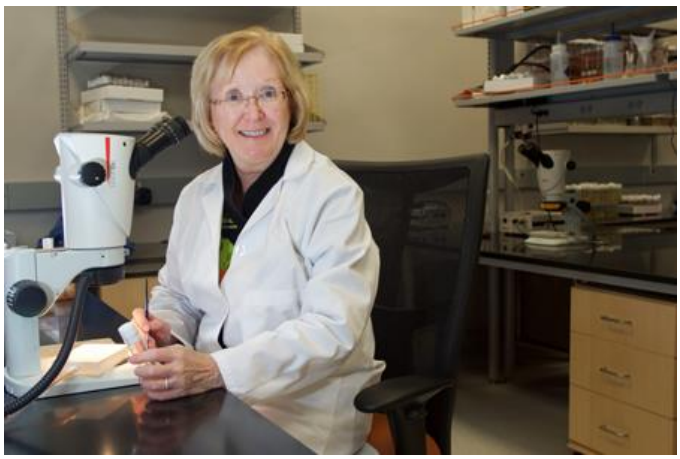
Celebrating an International Community

This year's summer Olympics were held in Tokyo. While enjoying the spectacle of international competitions in multiple sports, we should remember that the practice of science is perhaps the most significant international endeavor. Louis Pasteur proclaimed that "Science knows no country, because knowledge belongs to humanity, and is the torch which illuminates the world." His words ring true today; in a world where conflicts are commonplace, science knows no borders and scientists across the globe form an international collaborative community who all work in small or large ways to improve the human condition and quality of life for all inhabitants of our small planet.

The Center for Human Genetics values international diversity among its staff, students, and faculty. Since its inception in 2018, the Center has hosted among its members and visiting scholars individuals from 18 different national origins and four continents, promoting a vibrant academic as well as cultural community. "The most remarkable thing about our multinational community is that we do not realize that we are different," says Robert Anholt, "people are similar at a fundamental level and share the same aspirations, emotions and objectives for academic achievement." As the late Italian neuroscientist and Nobel laureate Rita Levi-Montalcini, who worked both in Italy and the US, said **"**My life has been enriched by excellent human relations, work and interests. I have never felt lonely."



Trudy Mackay elected to the American Philosophical Society



Dr. Trudy Mackay, Director of the Clemson Center for Human Genetics and Self Family Endowed Chair of Human Genetics, has been elected as member of the prestigious American Philosophical Society. Founded by Benjamin Franklin in 1743, the APS is the oldest learned society in the United States with a membership that is comprised of top scholars from a wide variety of academic disciplines. As of April 2020, only 5,710 members have been elected since 1743, and since 1900 260 members have received the Nobel prize.

Mackay's previous accolades include election to the Royal Society of London, the American Academy of Arts and Sciences, and the National Academy of Science of the USA as well as the Wolf Prize in Agriculture and the Dawson Prize.

Mackay is the first APS member at Clemson University and the only APS member in the Biological Sciences in the Carolinas.

Flies on Drugs

by J. Spencer Hatfield

Brandon Baker defended his Ph.D. dissertation on May 14th, 2021. His dissertation, titled "Systems genetics of cocaine and methamphetamine consumption in *Drosophila melanogaster*" culminated in four publications, including first author papers in *Genome Research* and *Proceedings of the National Academy of Sciences of the USA*. Brandon is the first doctoral candidate to graduate from the Clemson University Center for Human Genetics under the mentorship of Drs. Trudy Mackay and Robert Anholt.

Brandon's dissertation follows a cohesive theme of investigations into the biological underpinnings of drug consumption traits, connecting puzzle pieces at multiple resolutions to decipher the relationship between substance use disorders and genetic background. To start, he performed a genome-wide association study on 46 lines of the *Drosophila melanogaster* Genetic Reference Panel (DGRP), measuring levels of consumption and preference for both cocaine and methamphetamine. This study implicated over 700 candidate genes significantly associated with drug consumption phenotypes. Next, he selected 34 of these genes to target for knockdown using RNA-interference and subsequently validate as causal variants with behavioral testing. His data implicate the dopaminergic neurons, glia, and mushroom body of the fruit fly in drug consumption and preference. Diving deeper into this finding, he collaborated with other lab members to dissect the effects of acute cocaine exposure on the fly transcriptome at both the bulk RNA and single-cell RNA level.

To perform these studies, Brandon allowed flies to consume a set amount of cocaine, after which they were dissected (heads and bodies for the bulk-RNA project, brains for the single-cell project) and sequenced. Single-cell sequencing utilized the 10X Chromium microfluidics system to generate next-generation libraries. Analyses of the single-cell data were performed by Dr. Vijay Shankar and annotated by Sneha Mokashi. Together they uncovered differential expression clusters that implicate multiple brain regions as critical for the transcriptional response to acute cocaine exposure, including the Kenyon cells of the mushroom body, surface glia, and astrocytes. The next piece of the puzzle for Brandon was the question of the underlying genetic architecture of increased cocaine and methamphetamine

consumption. To investigate this, he created advanced intercross populations from the *Drosophila melanogaster* Genetic Reference Panel in which alternative alleles of candidate genes were fixed in random outbred populations.

These experimental populations allow for fine genetic mapping and more accurate estimates of quantitative trait loci. Brandon performed extensive phenotyping of cocaine and methamphetamine consumption on these populations and compared the gene sequences of the top 10% of drug-consuming flies to a random set of flies within the distribution. This comparison unveiled 988 drug- consumption-related genes in *Drosophila melanogaster*, with over 65% of these genes having an ortholog in humans. He again validated the 22 most interesting genes (all of which have a human ortholog) using RNAi-based knockdown and subsequent consumption testing. Over 75% of these genes showed a significant result in at least one sex. These results demonstrate translational potential to identify human orthologs of implicated Drosophila genes for future studies on the genetic and neurological underpinnings of substance use disorders.

Dr. Baker now works out of Cincinnati, Ohio as a Strategic Feasibility Associate for IQVIA Biotech. His work focuses on feasibility assessment of proposed clinical trials, an integral step in the translation of scientific research to medical care.

Of Single Nucleotides and Single Cells

by Rebecca MacPherson

On Thursday, July 29th, 2021, Sneha Mokashi defended her Ph.D. dissertation in Genetics, titled “Of single nucleotides and single cells: charting the genotype-phenotype map at high resolution using *Drosophila melanogaster*”. Sneha spent the first few years of her Ph. D. at North Carolina State University and transitioned with Drs. Mackay and Anholt when they moved their laboratories to Clemson University in the summer of 2018. Her doctoral research culminated in four publications.

Understanding the mechanisms by which genetic variation results in phenotypic variation is essential for understanding variation in complex traits. Sneha approached this question by exploring how individual genetic variants in a model system contribute to phenotypic variation, and how environmental perturbations influence gene expression in the brain at single cell resolution.

The first portion of her dissertation focused on the behavioral and transcriptomic effects of single nucleotide changes in the *D. melanogaster Obp56h* region. *Obp56h* is expressed in a subset of cells in the fruit fly brain and has been previously implicated in olfaction, mating behavior, lipid metabolism, and heat stress. Sneha selected naturally segregating single nucleotide allelic variants at the *Obp56h* locus and used CRISPR-Cas9 germline editing to create a series of lines that only differed at a single nucleotide in the *Obp56h* region. In these lines, she observed variant-specific effects on temperature- and starvation-stress phenotypes, as well as viability and activity. Transcriptional data also show variant-specific effects on the number of differentially expressed genes, as some lines show no differentially expressed genes compared to one another, whereas others show over 800. Protein-protein interaction networks based on RNA-seq data implicate mitochondrial electron transport and oxidative phosphorylation as a fundamental process that could result in variation in fitness phenotypes. In Sneha’s study, rare alleles and alleles in coding regions did not have larger effects than common or non-coding alleles. In contrast to the large effects observed when these alleles were expressed individually in a common genetic background, phenotypic effects of these alleles on variation in the same traits could not be detected in a wild-derived inbred population, likely due to context-dependence and suppressing epistatic interactions. Thus, her data indicate that the infinitesimal model, a longstanding model within quantitative genetics, is not sufficient to explain the biology underlying complex traits.

Next, Sneha examined the effects of cocaine and developmental ethanol exposure on the *D. melanogaster* brain at single-cell resolution. With the assistance of laboratory technician Rachel Hannah, she developed the

methodology for dissociating *D. melanogaster* brains into single cells for single cell RNA sequencing. She obtained high quality RNA sequencing data from brains of flies that were acutely exposed to cocaine, as well as from the brains of flies reared on food supplemented with ethanol as a model of Fetal Alcohol Spectrum Disorder. In each of these experiments, she used well established markers to identify the identities of individual clusters of cells bearing unique transcriptional signatures. These clusters represented almost all major neural cell types, including glia. Sneha observed extensive sexual dimorphism and several differentially expressed genes overlapped between acute cocaine exposure and chronic developmental ethanol exposure. Kenyon cells within the mushroom body (the center for experience-dependent modulation of behavior in the fly brain) and glia showed large changes in transcription in response to acute cocaine exposure, and genes associated with lipid, glutathione, glutamate, and GABA metabolism were differentially expressed in response to developmental ethanol exposure.

Sneha moved to a postdoctoral position with Dr. Heather Flanagan-Steet, Director of Functional Studies at the Greenwood Genetic Center in Greenwood, SC. Her new research focus is studying congenital disorders of glycosylation using a zebrafish (*Dania rerio*) model.

A Conversation with Richard Steet

Could you briefly describe your research program and interests?

My research interests lie at the intersection of glycobiology, lysosomal biology and human disease. The current focus of our research is aimed at defining the pathogenic mechanisms that underlie the lysosomal storage disorders and congenital disorders of glycosylation (CDG) using both zebrafish and cultured cells. We employ a powerful combination of genetic, biochemical, and microscopic methods to uncover the pathogenic cascades of these disorders and explore new ways to treat them. Another major part of my research program is centered on the functional characterization of novel and rare genetic variants, leveraging our expertise in defining disease mechanisms to improve diagnosis and management of genetic disorders.

Can you talk about the collaboration efforts between the GGC and the CHG?

Productive collaborations between GGC and the CHG began almost immediately after the collective arrival of myself and Heather, and Trudy and Robert, in Greenwood. Our collaborative efforts include the identification and characterization of modifiers for rare metabolic disorders (using both fruit flies and our zebrafish system), as well as the advanced genomic testing on undiagnosed patients at GGC. We expect these interactions to only grow in the future and that they will lead to the discovery of new disease/gene associations and important advances in human genetics.

How did you end up at the GGC in South Carolina?

While at the University of Georgia (just two hours away), I had the opportunity to meet and interact with several faculty at GGC and developed a strong professional and personal relationship with them. When the chance arose to relocate to Greenwood and lead the Research Division at GGC, I was able to make this decision with confidence knowing I shared the same motivation and goal to help patients with rare disorders with my current GGC colleagues.

What is the most rewarding part of genetics research?

To see that your efforts can directly impact patients with genetic disorders, by providing them with answers and hopefully improving their outcomes. On a more personal note, I find the challenge of unraveling the mechanisms that underlie these different conditions highly rewarding. Every day is a new adventure in genetics research.

Richard Steet is the Director of Research and Head of the JC Self Research Institute at the Greenwood Genetic Center and Adjunct Professor of Genetics and Biochemistry at Clemson University.

What do you see for the future of medicine and pharmaceutical therapies in the next 100 years?

I don't think any of us can fully grasp what medicine will look like in 100 years, but I would guess we will have effective therapies for countless genetic disorders. These therapies will capitalize on our ability to quickly identify the genetic basis of these conditions and intervene with gene replacement or editing strategies. I also believe we will see major advances in tissue engineering and the management of prevalent and chronic diseases that will extend the healthy lives of humans well past the age of 100.

How do you spend your spare time away from the lab? Any hobbies?

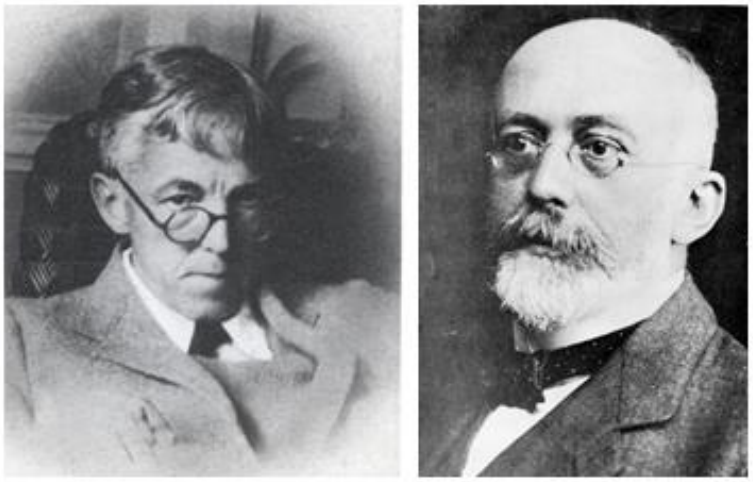
I "attempt" to play golf and enjoy life on the lake with my family. I'm also an avid fan of live music and enjoy cooking and entertaining.

What advice do you have for young genetics investigators?

Stay curious and always be ready to learn and incorporate new technologies. The field of genetics will require that you stay flexible and be willing to think big. This is a very exciting time to be an investigator in genetics, and the field needs intrepid and creative scientists like you!

Viewpoint: The Problem with Linkage Disequilibirum (LD)

by Robert Anholt



GH Hardy and Wilhelm Weinberg

In 1908, GH Hardy from Trinity College at Cambridge University wrote a one-page correspondence to *Science* in response to a comment by a certain Mr. Udny Yule that "if brachydactyly is dominant in the course of time one would expect, in the absence of counteracting factors, to get three brachydactylous persons to one normal" based on Mendel's principles. Hardy argued against that opinion and in the process developed his equation that indicate that in a large, random mating population allele frequencies would be stable. That same year a German obstetrician/gynecologist from Stuttgart, Wilhelm Weinberg, published his *article "Über den Nachweis der Vererbung beim Menschen"* which reached the same conclusions as GH Hardy's letter, and their concept became known as the Hardy-Weinberg principle. When allele frequencies in a population correspond to the Hardy-Weinberg predictions, the population is in equilibrium. However, the Hardy-Weinberg theory does not allow for any form of selection, gene flow, overlap among generations, introduction of mutations, or genetic drift, and for these reasons only rarely applies to realistic population scenarios. Non-random association of alleles in a population, as happens for example, when alleles are closely linked along a chromosome with little history of recombination, will show deviation from the Hardy-Weinberg predictions, commonly known as linkage disequilibrium (LD).

The human genome is composed of large blocks of LD and in human genetics LD is both a blessing and a curse. Because of extensive LD within a population, genome-wide association studies can rely on tagging SNPs, each of which represents a large fragment of the genome. This reduces the genotypic effort and cost and the number of statistical tests of genotype-phenotype association, but as a consequence it is challenging to assign causality with variation in a phenotype for any SNP in a linkage block. An additional complication is that there is variation in the LD structure of the genome between different ethnicities. The issue of LD is not unique to human populations but also pertains to populations that have gone through bottlenecks introduced by inbreeding, such as domestic dogs and inbred mouse and rat strains.

Invertebrate model organisms with short generation times, *i.e.* a long history of recombination events, have an advantage in that LD decays rapidly; in Drosophila LD decays within a couple of hundred base pairs. The average low extent of LD enables assigning causality to variants in genome wide association studies, but it requires full genome sequencing.

To address the issue of causality in human studies one approach is to examine gene expression in case-control study designs to identify genes with differential expression between the two groups that also have a variant in close proximity that was identified in the genome-wide association analysis. The problem here is that the relevant tissues for RNA extraction are more often than not unobtainable, thereby limiting this approach primarily to studies on lymphocytes, tumor cells or biopsy tissue.

Comparative genomics provides a powerful complement to studies on human populations. Fundamental biological processes are highly conserved from the nematode, fruit fly, zebrafish, and rodent to people. Each model system has its limitations, but each also offers considerable advantages. Combining studies on multiple genomic models with studies on human cell lines or populations can provide both causality and molecular context to genetic variants and provide a guide through the genotype-phenotype landscape. It is, however, rare for individual investigators to utilize multiple model systems at once, in part due to limited familiarity with these systems or because of logistics, *e.g.* managing a zebrafish facility and a mouse facility at the same time can be challenging. In addition, many human geneticists remain skeptical about the translational potential of model organisms for their area of expertise. The formation of collaborative groups that apply comparative genomic approaches along with studies on human populations needs to be encouraged to unleash an enormous potential to gain insights in a myriad of human traits in health and disease. As Alexander Graham Bell once said: "Great discoveries and improvements invariably involve the cooperation of many minds."

Robert Anholt is the Provost Distinguished Professor of Genetics and Biochemistry and Director of Faculty Excellence in the College of Science at Clemson University.

Grants

Lela Lackey received a Pilot Grant Award from the Alpha1 Foundation to study regulation of the mRNA that produces α-1-antitrypsin. Protein levels of α-1-antitrypsin impact the development of chronic obstructive pulmonary disease.

Lela Lackey received a Maximizing Investigator Research Area (MIRA) R35 grant from the National Institute of General Medical Sciences to study pre-mRNA intronic structures in trans factor binding and alternative splicing. This project focuses on the role of RNA structure in recognition of splice sites and other regulatory elements within RNA transcripts, such as SF3B regulated alternative splicing. SF3B mutations are common in several blood cancers. The Lackey laboratory will develop methods to identify pathogenic non-coding variants and develop RNA-centric therapeutics.

Rebecca MacPherson received an F31 Ruth Kirschstein National Research Service Award from the National Institute of General Medical Sciences to develop Drosophila models of rare Mendelian disorders of chromatin modification. Her project will focus on identifying genetic modifiers for fly orthologs of genes causing Coffin-Siris syndrome.

Jennifer Mason received a Maximizing Investigator Research Area (MIRA) R35 grant from the National Institute of General Medical Sciences to study the role of homologous recombination in the replication stress response. The proposed work will use genetics, molecular biology, and proteomics to understand how proteins in the DNA repair pathway function to protect genome stability. The completion of the proposed work will provide molecular insight into how disruption of recombination results in cancer and is essential for development of novel therapeutics that exploit chromosome instability.

Seminars

On Friday, **September 3**, at 2:30 pm **Dr. Avindra Nath**, Clinical Director of the Section of Infections of the Nervous System in the Division of Neuroimmunology and Neurovirology at the National Institute of Neurological Disorders and stroke, will present a seminar titled “The role of endogenous retroviruses in neurodegeneration.”

On Friday, **October 8**, at 2:30 pm **Dr. Harmit Malik**, Professor and Associate Director of the Basic Sciences Division at the Fred Hutchinson Cancer Research Center in Seattle, will present a seminar titled “Rules of engagement: molecular arms races between host and viral genomes.”

On Friday, **November 5**, at 2:30 pm **Dr. Sarah Tishkoff,** David and Lyn Silfen University Professor in the Departments of Genetics and Biology at the Perelman School of Medicine and Director of the Penn Center for Global Genomics & Health Equity at the University of Pennsylvania, will present a seminar titled “Genomic evolution and adaptation in Africa: Implications for health and disease.”

Publications

(affiliates of the Center for Human Genetics are in bold font)

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Greenwood Genetic Center
Assistant Research Scientist in Human Genetics

The Greenwood Genetic Center (www.ggc.org) is a nonprofit institute organized to provide clinical genetic services, diagnostic laboratory testing, educational programs and resources, and research in the field of medical genetics. With a focus on compassionate patient care and innovative scientific advancement, here at the Greenwood Genetic Center, we keep every patient and their families at the forefront of everything we do. Our researchers collaborate with GGC clinicians and laboratory faculty, as well as with colleagues around the world, to achieve significant advances in identifying causative genes, understanding disease mechanisms, and developing treatment and prevention strategies for genetic conditions. The Research Division maintains a state-of-the-art aquaculture facility and an Olympus FV3000 confocal microscope, along with extensive research space. Faculty development at the GGC is currently supported by an NIH COBRE grant in Human Genetics shared between the Greenwood Genetic Center and the Clemson Center for Human Genetics (https://scienceweb.clemson.edu/chg/) on the Greenwood campus. The grant offers mentorship opportunities and pilot project funding to new faculty, with the ability to become a Project Leader and obtain substantial research support for the development of an independent research program.

Job Description: Assistant Research Scientist in Human Genetics
This is a full-time faculty position in the Research Division at GGC, with primary responsibilities in independent research and functional analysis of genetic variants identified in our patient population. Outstanding opportunities exist for collaboration with basic, translational and clinical investigators. Successful candidates will have track records of creativity and productivity, a desire to integrate into our exciting, collaborative, and growing community of researchers, and a dedication to contribute to our values of diversity and openness. They will be expected to mount a productive and innovative research program, to obtain outside funding, and to participate actively in graduate and post-graduate teaching, training and mentorship. The specific areas of research interests we are seeking to recruit are: **1) transcriptional regulation; 2) RNA biology and human disorders involving RNA-related genes; 3) chromatin remodeling and epigenetics; 4) neurobiology and the mechanisms underlying neurodevelopmental disorders.**

Please contact GGC Director of Research, Dr. Richard Steet (rsteet@ggc.org) directly if interested to learn more about the application process and the positon.

Requirements:
Minimum Position Requirements (including years of experience, certifications, licenses, etc.):
- PhD in Genetics, Biochemistry, Molecular/Developmental Biology, or relevant biological field from a college or university of recognized standing.
- Experience in modern genetics, bioinformatics and molecular biology techniques.
- Must have demonstrated scholarly achievement with a record of publications in peer-reviewed journals.
- Must have demonstrated the ability to obtain extramural funding.
- Experience or expertise with zebrafish as a model organism is preferred.

Knowledge, Skills, and Abilities:
- Excellent oral and written communication skills
- Ability to work in a collaborative and multidisciplinary environment

Essential Duties and Responsibilities:
- Development of a robust, externally-supported research program
- Ability to perform functional studies on genetic variants within their area of expertise
- Participate in professional organizations and in internal case conferences and committees.
- Advise trainees and provide career guidance.
- Support and foster interdisciplinary efforts between GGC divisions, and between the GGC and the Clemson Center for Human Genetics.

Additional Information:
SALARY: A full time (12 month) salary is being offered for this position. The final salary will be based on rank and experience of the successful candidate.
CLOSING DATE: September 30, 2021

The Greenwood Genetic Center is an Equal Opportunity/Affirmative Action Employer. The Greenwood Genetic Center encourages applications for employment from persons who are members of groups that have been underrepresented based on race, color, national origin, gender, age, or disability.

Application Instructions:
Please submit the following documents to Kathleen Crowder (kcrowder@ggc.org); letters of support will only be requested for applicants who are invited to interview for the position.
- Cover letter
- Full Resume/CV
- Research Accomplishments and Interests (5 page limit)

Clemson University
Assistant Professor in Human Genetics

Clemson University invites applications for a tenure-track Assistant Professor as part of a Cluster Hire at the Center for Human Genetics, with an expected start date of August 2022. Clemson University offers competitive salaries, benefits and start-up funds.

The successful applicant will have an accomplished research record at the forefront of human genetics/genomics. Applicants whose research combines experimental laboratory work and computational approaches are especially desirable. Areas of special interest are the mechanisms by which variation in epigenetic modification, gene regulatory networks, chromatin conformation and nuclear architecture affect variation in human health and disease. However, all areas with the potential to significantly advance the field of human genetics will be considered.

The Center for Human Genetics (https://scienceweb.clemson.edu/chg/) is housed in Self Regional Hall, a 17,000-square-foot building located in Greenwood, South Carolina on the Greenwood Genetic Center Partnership Campus. The Center for Human Genetics provides a vibrant interactive research environment with state-of-the-art genomic and computational resources, and is ideally configured for collaborative research. The successful applicant will be part of a collaborative and interdisciplinary environment that includes the research, diagnostic and clinical geneticists at the Greenwood Genetic Center, the genetics, genomics, statistics and bioinformatics faculty at Clemson University, the USC School of Medicine in Greenville and the Prisma Health System. The home department at Clemson will be determined by the fit of the applicant's research interests with the mission of one of the departments in the College of Science (www.clemson.edu/science), including the Department of Genetics and Biochemistry (www.clemson.edu/science/departments/genetics-biochemistry/index.html), the School of Mathematical and Statistical Sciences (www.clemson.edu/science/departments/mathematical-sciences/index.html) and the Department of Biological Sciences (www.clemson.edu/science/departments/biosci/index.html).

Clemson University is committed to building a diverse and inclusive community of faculty scholars dedicated to working and teaching in a multi-cultural environment (http://www.clemson.edu/inclusion/). We encourage applications from women, minorities and individuals with a commitment to mentoring colleagues and students from demographic groups underrepresented in the sciences. We are also supportive of the needs of dual-career couples.

Successful candidates must hold a doctoral degree and have postdoctoral experience. Competitive candidates will demonstrate an ability to develop a vigorous and independent, externally funded and nationally recognized research program; demonstrate teaching excellence and a commitment to diversity inclusion; and participate in relevant undergraduate and graduate education programs.

Applicants should submit the following items via Interfolio at https://apply.interfolio.com/82505 : (1) cover letter; (2) Curriculum Vitae; (3) statement of research interests including future plans; (4) statement of teaching interests and experience; (5) statement describing past experience and/or future plans to promote diversity and inclusion; and (6) up to three reprints in one PDF. Applicants should also arrange, through Interfolio, the submission of three confidential letters of recommendation on their behalf.

Inquiries should be directed to Dr. Trudy Mackay (tmackay@clemson.edu).

For full consideration, applications should be submitted by October 1, 2021. Review will continue until the position is filled.

Clemson University is an AA/EEO employer and does not discriminate against any person or group on the basis of age, color, disability, gender, pregnancy, national origin, race, religion, sexual orientation, veteran status or genetic information. Clemson University is building a culturally diverse faculty and staff committed to working in a multicultural environment.