Department of Human Genetics

Spotlight on Global REACH

2014 Annual Newsletter

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I really enjoy the opportunities I’ve had to interact with scientific colleagues in other countries who share my passion for discovery and education. I have had the pleasure of working with trainees from India, Korea, Japan, Russia, China, Lebanon, Brazil, France, Argentina, Hungary, Persia and the UK, and enjoyed learning about their customs and culture. That knowledge has given me a new perspective on American style. I have continued to collaborate with some former trainees after they established independent careers, and these long-term relationships have been mutually beneficial and fulfilling. Despite the financial pressures academic researchers are experiencing in the US, we are in a privileged position when it comes to research infrastructure and resources. I am inspired by the effort and creativity that scientists in many other countries devote to setting up technologies that we take for granted. It is rewarding to establish collaborations that transfer technologies and education, and make discovery science a global enterprise.

Graduate Student Fellowships

The University of Michigan established a global REACH program several years ago in order to promote international collaborations for our faculty and students in research, education and health care (http://globalreach.med.umich.edu/about). To be successful, the collaborations established through this mechanism must be mutually beneficial. Several faculty members in the Department of Human Genetics have been engaged in the global REACH program as scientific collaborators and as educators. Read more about our faculty engagement in many different countries around the world in this newsletter’s spotlight on global activities.

The University of Michigan recently launched a $4 billion dollar fundraising campaign aimed at three priority areas: student support, engaged learning, and bold ideas. Through action and words, our leaders, faculty members, alumni and friends have emphasized their commitment to guaranteeing that a diverse group of the world’s brightest students are able to study at the University of Michigan, by making an investment in the discovery of new knowledge. Former University President, Mary Sue Coleman, announced the goal of raising $1 billion for student support, and committed $25 million in University funds as a matching opportunity to encourage donations. I am delighted to share the news that Professor Emeritus of Human Genetics and Internal Medicine, Dr. George Brewer and Mrs. Lucia Brewer have donated $100,000 to endow a scholarship for physician-scientist trainees pursuing research in human genetics. This was matched by $25,000 in funds from the President’s office. We are eager to inaugurate and expand this scholarship because of the importance of training individuals who understand the pressing clinical questions and have the scientific training to address them.
Our new University President, Mark Schlissel, MD, PhD, emphasized his personal commitment to fundraising for graduate education during his recent inauguration as the 14th President of our nearly 200-year-old institution. He pledged to embrace the University’s mission to seek out, encourage and value all voices, and to create an open and accessible, diverse and democratic community. I attended the inauguration festivities with HG PhD student from Estonia, Kärt Tomberg. The event was truly inspiring.

Ora Pescovitz, MD, our most recent Executive Vice President for Medical Affairs, was a leader and driver for supporting basic science in the fundraising campaign. She personally gave generously and recruited others to donate to the EBS’s Endowment for the Development of Graduate Education (EDGE) fund (http://ebs.med.umich.edu/edge), which supports students who are in the final stages of completing their dissertation research in Human Genetics; Matt Avenarius, Jake Higgins, and Bill Law have all been supported by this fund for their research in hearing, prostate cancer, and congenital birth defects, respectively.

We are grateful to all who have given generously to support graduate education and research.

With best regards,

Sally A. Camper, Ph.D.
Human Genetics faculty members are engaged in teaching, technology development and collaborative research activities across the world, through the University’s Global REACH (Research, Education, and Collaboration in Health), especially at sites where the University of Michigan has established joint institutes intended to facilitate and support long term interactions. Cristen Willer’s work on heart health with collaborators in Nord-Tronndelag, Norway was profiled in Medicine at Michigan (http://www.medicineatmichigan.org/magazine/2014/summer/limeight). Read here about

AFRICA

David Burke’s primary global engagement effort is part of a NIH-funded human genetics research initiative, “Human Health and Heredity in Africa” = H3Africa. He is also involved in an eye-health technology development effort that includes a clinic in rural Jamaica and health care delivery in Vanuatu, in the South Pacific. These activities are in alignment with his local leadership in “Distributed Health Technologies,” (http://ncrc.umich.edu/research/initiatives/distributed-health) which is designed to promote broad distribution of affordable health care solutions globally.

Africa in the 21st Century is clearly a region of great vitality and growth, and Dr. Burke relates being powerfully struck as being one of the oldest people around wherever he goes! Africa is dominated by the young -- and youth is the human fuel for change. He is enjoying being part of it.

There are, of course, significant challenges to working in African-based laboratories. The level of “infrastructure” is low in many areas, with cellular telephones being the major exception. Technologies must be robust to avoid the need for replacement or repair. Burke has emphasized very low-capital investment strategies (less than $5000), with all components being available from multiple suppliers. To paraphrase a line from an old movie: “Cheap is good!”

H3Africa is a 5-year initiative from NIH and the Wellcome Trust Foundation in the UK (http://h3africa.org/). The program emphasizes the “study of genomics and environmental determinants of common diseases, with the goal of improving the health of African populations.” H3Africa provides a significant investment in infrastructure development within underdeveloped countries and in training of African-based scientists. Dr. Burke joined Dr. Mike Boehnke (Biostatistics) and Dr. Akinlolu Ojo (Internal Medicine/Nephrology) at the University of Michigan to assist in infrastructure development and training associated with a grant application led by Dr. Dwomoa Adu, at the University of Ghana, in Accra. Burke emphasizes the development of inexpensive applications of state of the art technologies and hands-on training of African-based genetics laboratory researchers. Dr. Burke and Dr. Nicki Tiffin at the University of the Western Cape, South Africa, are emphasizing the computational and analytical training necessary to understand the genetic and environmental factors that contribute to higher incidence and earlier onset kidney disease in Africa and in African-Americans.
This project called “H3Africa Kidney Disease Research Network”, has clinical components in Ghana, Nigeria, Kenya, and Ethiopia. Individuals and families with kidney disease are examined at each site, and blood and saliva samples are collected. Burke’s effort is to train local scientists to perform low-cost, sustainable techniques to process these biological samples, extract genomic DNA, and perform molecular genotyping of sequence variants from the material. The goal is to obtain publication-quality genomic DNA data from samples with no need to export DNAs from the collection site.

Burke assesses the local infrastructure constraints at each site in Africa and trains laboratory workers in analysis methods. The preliminary trial-and-error work is done in his Ann Arbor laboratory at the North Campus Research Complex. The human DNA samples are analyzed and archived within the communities of origin, with the local scientists and physicians performing the work, writing the papers, and getting the international recognition they deserve. He hopes the methods that emerge from this global collaboration will be transferred rapidly among genetics researchers in other low-infrastructure areas.

**BRAZIL**

Partnering on the genetics of short stature and endocrine disorders, Sally Camper formed one of her most valuable collaborations just hoping to “give back.” “I spent a year abroad in college,” she says, “and it totally changed my life. So I decided to become involved in the International Endocrine Scholars Program to help others have this same opportunity.”

During one of the society’s international meetings, she met a potential postdoc from Brazil, Luciani Carvalho. Dr. Carvalho, a member of the Faculty of Medicine at the University of São Paulo, works in the short stature clinic. Both women were looking at the genetic basis of severe short stature caused by pituitary dysfunction, and what each brought to the table excited the other.

“Across Brazil, people with severe short stature come to São Paulo for treatment with growth hormone,” says Camper. “So they have a catchment of patients from the entire country, which was exciting to me. Dr. Carvalho had been working to identify genes that cause short stature from these patients. She wanted to learn from my lab how to model these in mice to study the pathophysiology of disease, as well as how to test the function of variants you’d find in people.” This is important, says Camper, “because it’s not always obvious whether a variant found in a person of short stature is one that is tolerated or whether it’s causing the problem.”

Carvalho spent two years in Camper’s lab, learning to do cell culture assays and work with genetically modified mice. They have continued to collaborate on the causes of hypopituitarism in patients and have also begun exploring the role of stem cells in pituitary development and disease. Camper recently won renewal of a longstanding NIH grant based in part on data she’s gathered with Carvalho and other collaborators at the short stature clinic.

They have turned their attention to next-generation sequencing technology, where Camper feels she can bring additional value. “It is time to move beyond the single gene test and screen the entire genome.” Initially, the plan is for the Brazilian researchers to identify patients, get informed consent, and carry out treatment, while the DNA sequencing and bioinformatic analysis are done at U-M. The two groups will collaborate to study the significance of the genetic variants. The collaboration is ideal because the Faculty of Medicine at University of São Paulo are eager to build capacity in high throughput sequencing, bioinformatics analysis, and follow-up functional studies in vertebrates.

The potential ripple effects of this excite Camper since the techniques can be used to study so many medical problems, from birth defects to cancer. And, just as importantly, she hopes the results of this research will ultimately help patients answer life-altering questions like the risks to their children and the likely progression of their disease.
**BRAZIL**

**Faculty Member Assists in Establishing Zebrafish Facility in São Paulo, Brazil**

Dr. Carvalho recently hosted **Tony Antonellis** in São Paulo so that he could share his expertise in modeling human disease in zebrafish. He presided over a week long workshop for faculty and students. Each day, Dr. Antonellis gave a lecture on how to use zebrafish to study human genetic disease, directed a journal club given by students covering recent articles on zebrafish research, and provided hands-on training in zebrafish husbandry and experimentation. Gratifyingly, by the end of his visit the facility was fully operational. Breeding fish yielded hundreds of embryos each day, and workshop participants successfully injected and analyzed zebrafish embryos. In fact, one student in the class decided to switch to a zebrafish model system for her research based on the approaches and techniques that she had learned! The potential for additional collaboration on genetic causes of neurological diseases and epilepsy emerged from the visit.

**CHINA**

**Margit Burmeister** first went to China in 2009, as part of a sabbatical leave during which she taught an introductory biology course at Shanghai Jiao Tong University (SJTU) Joint Institute. This joint venture began in 2006 as part of the Global REACH initiative. Burmeister made contacts with Professor Lin He, the Director of the Bio-X Institute at SJTU, and an authority on genome wide association studies and psychiatric disorders. She has visited the Bio-X Institute several times since then, with funding from the Chinese government to complete further research in complex disorders such as psychiatric genetics.

She is an advisor for the China Super Brain project, a TV series featuring “geniuses” with supernormal abilities that compete in a fashion similar to the American TV program, “American Idol.” The participants take part in genetic research and electrophysiological tests.

**Jun Li**’s cancer research study with colleagues in China illuminates the value of studying different populations to understand disease risk. He is collaborating with a team in the School of Oncology at the site of another joint institute with University of Michigan: the Peking University Health Science Center. Their goal is to study intra-tumor heterogeneity and mutation signatures in esophageal squamous cell carcinoma (ESCC). Esophageal cancer is a top ten cause of cancer; China accounts for ~50% of cases worldwide. Parts of northern China have the highest incidence levels of esophageal cancer in the world (50 per 100,000 individuals per year). This disease is population specific: among Caucasians, the primary subtype is esophageal adenocarcinoma (EAC), with gastro-esophageal reflux disease as the major risk factor; however among Asians, ESCC is the dominant subtype, and environmental exposures such as tobacco and alcohol use or HPV infection might contribute. The actual genetic and environmental factors driving such remarkable geographic variation are currently unknown.

To better understand esophageal cancer, the team has characterized intra-tumoral genetic heterogeneity by comparing alterations in different sectors of the same tumor, and contrasting them with surrounding normal tissues as well as the metastatic tumors in nearby lymph nodes. The results will identify major genes and pathways affected in each patient, potentially revealing the temporal progression of tumorigenic events from normal tissue to primary tumor, and finally to metastasis.
The Peking group developed a powerful clinical resource, with a 10,000-subject cohort study and an existing collection of tumor samples and genomic data. Dr. Li’s group at UM brings expertise in statistical analysis. This study is expected to generate new insights into the genomic evolution of ESCC, and it provides a rare opportunity to be complemented by concurrent studies of EAC in Caucasians that are ongoing at University of Michigan.

**INDIA**

The University of Michigan is actively engaged in collaborative efforts with several academic institutions India, most extensively with the All India Institute of Medical Sciences (AIIMS) in Delhi and Maharashtra University of the Health Sciences (MUHS) in central India. UM faculty visits to AIIMS over the past few years have focused on trauma care, and a Memorandum of Understanding with AIIMS was signed in 2011. In 2012, collaborations were formalized with MUHS through the joint Obama-Singh 21st Century Knowledge Initiative award.

In November 2013, President Mary Sue Coleman led a delegation from the University of Michigan to New Delhi, India. Dr. Joseph Kolars, Senior Associate Dean for Education and Global Initiatives, and more than 20 faculty from the Medical School accompanied her. The goal of the visit was to develop a robust platform for collaborations with AIIMS. Dr. M.C. Mishra, Director of AIIMS, and Indian dignitaries representing several government research agencies, were in attendance at the official inauguration of the collaboration between AIIMS and UM. The leadership of AIIMS and UM are providing institutional support to overcome administrative barriers and cultural hurdles in order to enhance international scientific teamwork geared to identify the genetic causes of human disease.

**Stephanie Bielas** represented the Department of Human Genetics on this trip. Her lab focuses on neurogenetic disease, especially brain malformations. Due to its prominence in India, AIIMS attracts many unusual cases which are rarely encountered elsewhere. Dr. Bielas accompanied physicians in clinics and observed patients with rare neurodevelopmental disorders and their families. The personal connection gave her a newfound appreciation for the impact of her genetic research. Productive collaborations have enhanced both research and educational exchange. For example, Dr. Bielas’ lab hosted Indian clinicians interested in using next-generation sequencing for disease gene identification that would further basic understanding of brain development. Ultimately all collaborators expect that discovering the causes will lead to improved care for patients on both continents.

Dr. Bielas and members of The Pediatric Genetics Unit (lead by Madhulika Kabra, to the Left of Dr. Bielas) at AIIMS, Delhi, India.
Graduate Education

This year (2014-2015) there are 37 graduate students in the Department of Human Genetics, including 19 doctoral students, two of whom are MD, PhD students; 5 MS in Human Genetics and 13 MS in Genetic Counseling. In addition, there are 10 PhD students in other graduate programs who are being mentored by Human Genetics primary faculty, including 5 in Bioinformatics and 5 in Cell & Molecular Biology (CMB).

New Students

PhD Program in Human Genetics
Feichen Shen, Zhejiang University, China (Kidd Lab)
Christina Vallianatos, University of Michigan (Iwase Lab)
Yifan Wang, University of Maryland, College Park (Mills Lab)

Master’s Program in Human Genetics
Rakhee Bajaj, B.S. Michigan State University
Yan-Cheng Chao, B.S., Fudan University, China
Amanda Moccia, B.S. Western Michigan University
Kathleen Wang, B.S. University of Michigan

Master’s Program in Genetic Counseling
Shenin Dettwyler, B.S. Indiana University of Pennsylvania, PA
Carol Ko, B.S. UCLA Huntington Beach, CA
Mary Oliver, B.A. Kalamazoo College, MI
Lynn Pais, M.S. Vellore Institute of Technology Mangalore
Parker Read, B. S. Texas A & M, TX
Linford Williams, B.S. University of Michigan, MI

Introducing New Postdoctoral Fellows

Rosie Bunton, Ph.D. (Meisler Lab) is a native of the UK. She received her BSc degree from the University of London and her MSc from King’s College, London. In July, she defended her Ph.D. thesis at the Institute of Neurology, University College, London on the role of superoxide dismutase in mouse models of Alzheimer’s Disease. In the Meisler lab, she will be working on molecular regulation of the sodium channel gene SCN8A and its role in epilepsy.

Matthew Oetjens Ph.D. (Kidd Lab) is originally from Michigan and received his B.S. from Evergreen State College in Olympia, WA, and Ph.D. from Vanderbilt University. Dr. Oetjens’s graduate research focused on the genetics of drug efficacy and drug response. In the Kidd lab, he will characterize evolution and population genetics utilizing genome wide sequencing data.

Amanda Pendleton Ph.D. (Kidd Lab) is originally from Florida and received her B.S. in Botany and Ph.D. in Plant Molecular and Cellular Biology from the University of Florida. Dr. Pendleton’s graduate research focused on genome evolution in pathogenic fungi. In the Kidd lab, Dr. Pendleton will study genome evolution and phenotypic diversity in canines.

Qingxuan Song, Ph.D. (Li Lab) received his M.A in Statistics from UM’s Department of Statistics. He is a recent Ph.D. graduate from the Department of Molecular, Cellular & Developmental Biology, where he applied Next-Gen Sequencing to pooled segregated yeast strains to understand the mechanisms of filamentous growth of yeast cells. In the laboratory of Jun Li, he is integrating results from genome and transcriptome analysis to better understand mechanisms of breast and esophageal squamous cancers.

Yao-Chang Tsan, Ph.D. (Bielas Lab) was born in Taiwan and received his B.S. in Zoology from the National Taiwan University. He came to the University of Michigan for graduate work with Dr. Sue O’Shea in the Department of Cell and Developmental Biology. In the Bielas lab, Dr. Tsan is using both embryonic and induced pluripotent stem cells as models to study human brain development.
Recent Graduates

Congratulations to graduates in the Department of Human Genetics for completing their programs and moving forward in their careers:

Ph.D. Program

Lesley Everett, Ph.D. (Ginsburg Lab) March, 2014 – “Characterization of the Function of the LMAN1 ER Cargo Receptor in vivo.” Lesley resumed her medical training as part of the Medical Scientist Training Program, and will graduate with an M.D., Ph.D. in May, 2016.

Cheryl Jacobs Smith, Ph.D. (Sekiguchi Lab) March, 2014 – “Mechanisms in Suppressing Chromosomal Translocations and Maintaining Genome Stability.” Cheryl is now a postdoctoral fellow in Cancer Health Disparities at the National Cancer Institute at the National Institute of Health, Bethesda, Maryland.

Serina Mazzoni, Ph.D. (Fearon Lab) August, 2014 – “A role for AXIN2 in oncogenesis.” Serina plans to finish her experiments in the Fearon Lab before pursuing a postdoctoral position studying the genetics of nitrogen utilization in plants.

Human Genetics Master’s Program

Christine Hong, M.S. May, 2014 – Christine Hong will be working as a research assistant with Dr. Stephen Gruber at the USC Norris Comprehensive Cancer Center at the Keck School of Medicine of the University of Southern California.

Adam Helms, M.D., Ph.D., received a M.S. in Human Genetics and is now a clinical lecturer in cardiology in internal medicine. His research interests are in inherited cardiomyopathy and arrhythmia disorders.

Genetic Counseling Program

Julie Frank, M.S.G.C., May, 2014 – University of Maryland Medical Center, Pediatric Genetics, Baltimore, Maryland.


Deanna Julian, M.S.G.C., May, 2014 – Baylor Medical College, Adult Genetics, in Houston, Texas.

Emily Moe, M.S.G.C., May, 2014 – Children’s Hospital of Wisconsin, Milwaukee, Wisconsin.


Caroline Weipert, M.S.G.C., May, 2014 – Brigham and Women’s Hospital and Harvard Medical School Division of Genetics, Boston, Massachusetts

Human Genetics Student Awards

Lesley Everett - 2014 James V. Neel Doctoral Fellowship Award
Lauren Hipp - 2014 James V. Neel Genetic Counseling Fellowship Award
William Law - 2014 Basic Science EDGE Award (Endowment for Development of Graduate Education)
Emily Maclary - Rackham Predoctoral Fellowship; NIH Ruth L. Kirschstein National Research Service Award; 2014 James V. Neel Doctoral Fellowship Award

Christina Valliantos - 2014 Anita and Howard Cramer Scholarship Award
Kate Partynski - 2014 Michigan Association of Genetic Counselors Student Research Award; 2014 Jane Engelberg Memorial Fellowship Student Research Award from National Society of Genetic Counselors
Kärt Tomberg - American Heart Association Fellowship
Faculty Awards, Honors and Promotions

Promotions
Tony Antonellis was promoted to Associate Professor, with tenure, effective September 1, 2014.

Awards, Honors

- **Vivian Cheung**: Vice-President elect of American Society for Clinical Investigation; she will serve as President-elect for one year, and then, President.
- **Eric Fearon**: Elected to the Institute of Medicine of the National Academies; Chair of the Biomedical Science Scholars Program in 2014-2015.
- **Santhi Ganesh**: Heart of a Champion (University of Michigan, Cardiovascular Center); Doris Duke Clinician Scientist Development Award (fibromuscular dysplasia and aneurysmal disease).
- **David Ginsburg**: Token of Appreciation from Medical Students, Award; 2013 Louis and Artur Lucian Award for Research in Circulatory Diseases, McGill University.
- **Jeff Innis**: Named Top Resident Teacher, Department of Pediatrics.
- **Shigeki Iwase**: March of Dimes Foundation Basil O’Connor Starter Scholar Research Award.
- **Catherine Keegan**: Inducted into American Society for Clinical Investigation.
- **Jun Li**: Dean’s Basic Research Award, University of Michigan.
- **Monica Marvin**: 2014 National Society of Genetic Counselors Strategic Leader Award.
- **Diane Robins**: Elected as a fellow of the American Association for the Advancement of Science; Elected to Endocrine Society Council; Sydney Ingbar Award for Distinguished Service to the Endocrine Society; Award for Excellence in Urological Research from the Society for Basic Urology Research and Society for Women in Urology.
- **Tom Wilson**: Environmental Mutagenesis and Genomics Society Service Award.

New Faculty

- **Jacob Kitzman, Ph.D.**, joined the faculty of the Department of Human Genetics in July 2014. He also holds an Assistant Professor appointment in the Department of Computational Medicine and Bioinformatics. The “Michigan Genomics Initiative” led by Dr. Gonçalo Abecasis, in the School of Public Health assisted with this recruitment. Kitzman received his bachelor’s degree in computer science, with a minor in biology, and a master’s degree in engineering from MIT. He worked for Roche NimbleGen Systems for two years prior to entering the Ph.D. program in the Department of Genome Sciences at the University of Washington, Seattle. He completed his thesis work in the laboratory of Dr. Jay Shendure in 2013. Kitzman’s research uses genome-scale technologies for the discovery and interpretation of human genetic variation, which includes extending whole-genome sequencing to capture haplotype phase information, developing clinical applications, such as noninvasive prenatal whole-genome sequencing, and innovating new, massively parallel approaches to assay functional impacts of variation in clinically relevant genes.

- **Jacob Mueller, Ph.D.**, joined the faculty as an Assistant Professor in Human Genetics in February, 2014 as part of an interdisciplinary cluster hire in Reproductive Sciences. Dr. Mueller received his B.A. in Biology and Economics from Gettysburg College in 1998. He conducted doctoral studies with Dr. Mariana Wolfner at Cornell University and was a postdoctoral fellow with Dr. David C. Page at the Whitehead Institute at MIT. During his postdoctoral research, Dr. Mueller discovered large, nearly identical segmental duplications, or amplicons, which are prominent features of both the mouse and human X chromosomes. His laboratory now explores the biological significance of these poorly understood structures and their role in infertility.

- **Saher Sue Hammoud, Ph.D.**, has accepted an offer and will join the Human Genetics faculty as Assistant Professor in November, 2014. She is an Organogenesis Scholar and also one of the Reproductive Sciences Cluster hires. Dr. Hammoud received her B.S. in Biological Science at Wayne State University in 2005, and her Ph.D. from the University of Utah in 2011. She conducted research on sperm chromatin and its role in fertility under the guidance of Douglas Carrell and Bradley Cairns. As a postdoctoral fellow in the Department of Oncological Science at the Huntsman Cancer Institute in Salt Lake City, she was mentored by David Jones and Brad Cairns. She was awarded a Helen Hay Whitney Research Foundation-HHMI fellowship to explore the relationship between stem/progenitor cells and cancer. Her future research will focus on epigenetic regulation of gonadal stem cells and male infertility.
Department Events

2014 James V. Neel Lecture in Human Genetics

Richard P. Lifton, M.D., Ph.D., gave the fourteenth James V. Neel Lecture in Human Genetics at the University of Michigan on May 12, 2014. Lifton is the Chair of the Department of Genetics at Yale University School of Medicine and an Investigator in the Howard Hughes Medical Institute. He is well known for his work on the genetic underpinnings of hypertension and other common disorders. Cristen Willer Ph.D. shares his interest in the genetics of cardiovascular disease, and hosted his visit.

This annual lectureship honors James V. Neel, M.D., Ph.D., 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. In 1956, Neel established the first academic department of human genetics in the United States at the University of Michigan Medical School, which he chaired for 25 years.

The lecture also included the presentation of the James V. Neel Fellowship Awards, which recognize outstanding academic and research achievements of human genetics graduate students pursuing Ph.D. degrees and M.S. in Genetic Counseling. Recipients were Emily Maclary and Leslie Everett from the Ph.D. program, and Lauren Hipp, M.S., from the genetic counseling program. Following the lecture, faculty, students and guests attended a poster session and reception for the awardees and Dr. Lifton.

Genetics Training Program

The annual retreat for the Genetics Training Program was held on Thursday, June 5, 2014, at the BSRB Kahn Auditorium. Dr. Miriam Meisler delivered the keynote address entitled, “Genetic Approaches to Neurological Disease: 20 Years of Student Discoveries.” Dr. Meisler traced the course of sodium channel research in her laboratory from the discovery of the SCN8A gene in a mutant mouse in 1995 to the identification of de novo mutations in epilepsy patients in 2012. Her talk highlighted the important contributions by Human Genetics graduate students Nicholas Plummer (Ph.D. 1998), David Buchner (Ph.D. 2003) and Janelle O’Brien (Ph.D. 2013). It was fascinating to hear how the discovery unfolded over 20 years.

Each of the DHG graduates also presented an update of their current research activities. Nicholas Plummer, a Staff Scientist at the NIH/NIEHS in North Carolina, discussed his development of an elegant series of transgenic mouse lines, “New mouse strains for manipulating genetically defined subpopulations of central norepinephrine neurons.” David Buchner, Assistant Professor of Genetics at Case Western University, described his quantitative genetic analysis of “The complex genetic basis of obesity and glucose homeostasis.” Janelle O’Brien, Postdoctoral Research Scholar at the University of Iowa, described her development of viral vectors for “Treatment of retinal degeneration in a mouse model of Bardet-Biedl Syndrome.” A common thread in the presentations of these Genetics Training Grant alumni was the way in which their training in the Department of Human Genetics contributed to their subsequent careers.

Dr. Meisler recently stepped down as Director of the Training Grant after 14 years of service and a successful competing renewal. Drs. John Moran and Tony Antonellis have taken the helm as Director and Associate Director, with assistance from Karen Grahl.
Annual Diane Baker Alumnae Lecture in Genetic Counseling

The Department of Human Genetics is excited to announce that Meg Hefner (UM GC Class of 1980) as recipient of the 5th Annual Diane Baker Alumnae Award. Meg is an Adjunct Associate Professor in the Division of Medical Genetics at the Saint Louis University School of Medicine. She is an internationally recognized expert in CHARGE syndrome and has dedicated most of her professional life to helping patients and families affected by this diagnosis. CHARGE is a multiple congenital anomaly that is the leading cause of congenital neurosensory loss of vision and hearing. The acronym stands for coloboma of the eye, heart defects, atresia of the nasal choanae, retardation of growth, genital abnormalities, and ear abnormalities and deafness. In many cases the disorder is caused by mutations in CHD7, a chromatin remodeling protein, studied by Dr. Donna Martin.

Meg has demonstrated excellence in activism as the founder of the CHARGE Syndrome Foundation, co-author of the book CHARGE Syndrome, and developer of family-oriented educational materials. Meg founded the first U.S. CHARGE Syndrome clinic at the Cardinal Glennon Children’s Medical Center in St. Louis, and developed an international database and registry, an invaluable resource for research that will increase our understanding of this condition.

This annual Alumnae Lecture honors Diane Baker, who led the program for many years and shaped it with her vision and commitment to the profession of genetic counseling. This year’s lecture will take place on November 18, 2014 at 3:00 p.m., in rooms 3813/3817, Medical Science Building II.

2014 Human Genetics Summer Picnic

Department family and friends enjoyed a wonderful afternoon and evening on June 19 at Gallup Park on the Huron River. Activities included face painting and games for children and families, the ever popular Tug of War contest, won by the Bielas Lab (with a little help from their friends!), and a catered picnic dinner. Special thanks to Dave Burke, Karen Grahl, Ryan Mills, Sue Tarle, Alison Martin and all those who planned and assisted in making this event a success.

The Department of Human Genetics 26th Annual Retreat

The Annual Department of Human Genetics retreat was held on September 26-27, 2014 at the Kellogg Biological Station in Hickory Corners, Michigan. Research talks, posters, and special activities were enjoyed by over 100 faculty, postdocs, students and staff. Special thanks to Jeff Kidd for orchestrating this year’s event, and to our guest speaker, Dr. David M. Bodine, who joined us from the Biology Branch of the National Human Genome Research Institute. Dr. Bodine is known for his work on genetic disorders of the blood that range from anemia to malignancy, which arise during hematopoiesis, the process of producing specialized cell types found in blood from stem cells. He has been a pioneer in studying epigenetic changes that occur during hematopoiesis, extending the application of the The ENCYclopedia Of DNA Elements (ENCODE) Project to purified cells from blood. Dr. Bodine received the 2012 Division of Intramural Research Annual Mentorship Award in recognition of outstanding achievements in the scientific training and mentoring of research fellows.
In June 2014, representatives of U of M’s Basic Science Departments met with alumni in the downtown Chicago area. This was the second regional gathering in Chicago. It was a great opportunity to take advantage of our “big blue network” and make connections between generations of alums across the departments. Nicole Scott received her Ph.D. in 2010 and is finishing her postdoctoral training at Argonne National Labs in microbial ecology. She expects to take a job in metagenomics in the near future. Jenny Mason (Ph.D. 2010) is carrying out postdoctoral studies in the Department of Radiation and Cellular Oncology, at the University of Chicago. Amy Hulme (Ph.D. 2007) is at Northwestern University studying HIV. Our next alumni gathering will be at the American Society of Human Genetics meeting in San Diego. We hope to see you there.

Alumni Gathers in Chicago and San Diego

In June 2014, representatives of U of M’s Basic Science Departments met with alumni in the downtown Chicago area. This was the second regional gathering in Chicago. It was a great opportunity to take advantage of our “big blue network” and make connections between generations of alums across the departments. Nicole Scott received her Ph.D. in 2010 and is finishing her postdoctoral training at Argonne National Labs in microbial ecology. She expects to take a job in metagenomics in the near future. Jenny Mason (Ph.D. 2010) is carrying out postdoctoral studies in the Department of Radiation and Cellular Oncology, at the University of Chicago. Amy Hulme (Ph.D. 2007) is at Northwestern University studying HIV. Our next alumni gathering will be at the American Society of Human Genetics meeting in San Diego. We hope to see you there.

Annual Chili Cook-Off for HG Graduate Education

The 2014 Annual Chili Cook-off for Graduate Education is an annual Department of Human Genetics event that raises funds for graduate education in the department. Last year’s competition included faculty, staff, postdoc and student entries in both chili and corn bread categories. Winning recipes included, “Tough Guys Oatmeal”, “Three-B’s Roadhouse Chili”, “Moe’s Hobo Chicken Chili” and “A-maizing Blue Chili”. Cornbread top finishers were “Sweet & Tasty Cornbread”, “Sweet Nothings” and “Korney Cornbread”. The 2014 competition is scheduled for November 24th and will include a special “Turkey Award” in honor of the Thanksgiving holiday.

ASHG Future Meeting Dates

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<td>2014</td>
<td>San Diego, CA</td>
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<tr>
<td>2015</td>
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<td>2016</td>
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Champions for Basic Research and Education

The Department of Human Genetics is extremely grateful to those who have given generously to support the missions of our department and this University in the area of genetic research and graduate education. Some of our special champions are highlighted below.

Dr. George Brewer and Mrs. Lucia Brewer have endowed a new scholarship for physician-scientist trainees pursuing research in human genetics. They recognize the importance of training individuals who will be able to discover the etiology of disease and use that knowledge to develop treatments for patients. Dr. Brewer is best known for his work on Wilson’s disease, a genetic disorder that can cause toxic accumulation of copper. Dr. Brewer discovered a life-saving treatment for this disorder, and devoted himself to developing increasingly effective, FDA-approved treatments for this and other ‘orphan’ diseases. His recent, ongoing research investigates the role of copper in common diseases like Alzheimer’s. The generous gift that George and Lucia have made will encourage students to pursue dual training in medicine and research. We look forward to the inauguration of this fellowship on November 7, 2014. George will deliver the keynote address on “The critical role of the physician-scientist in translational research and biomedical entrepreneurship.”

Drs. Peter and Janice Farrehi have established a research fund to promote a better understanding and potential treatment for X-linked intellectual disability. Pediatric Geneticists like Dr. Catherine Keegan, working with the University of Michigan Medical Genetics Laboratory, use genome sequencing to diagnose disorders. Often, knowing the cause of a disorder marks the end of a diagnostic odyssey, and gives families access to information about the development of other individuals with the same diagnosis. Yet, discovering the genetic cause of a medical condition is just the first step towards developing effective treatments. X-linked intellectual disability has many causes, including Fragile X and mutations in SMCX, a gene that regulates gene transcription by demethylating histones. Dr. Iwase discovered the function of the SM CX gene in chromatin modification and developed a mouse model for this disorder. With support from the Farrehi Fund, Dr. Iwase and his team are investigating the role of this gene in brain development and behavior, and they plan to use his animal model for testing possible treatments.

Dr. Stephen Fletcher established a research fund to find a cure for the neuropathy associated with Charcot Marie Tooth disease. Dr. Fletcher, a Houston-area pediatric neurosurgeon, is committed to finding a cure for his granddaughter Caroline, who was diagnosed with a hereditary motor neuropathy. Her disorder is named for the physicians who first described it in the late 1800’s, Drs. Charcot, Marie, and Tooth (CMT). Despite the unusual name, it is a common disease with no current treatment options. Dr. Fletcher identified several leading researchers around the country, and made a generous commitment to support research by Dr. Anthony Antonellis on this disorder, which has symptoms similar to ALS. After Dr. Fletcher’s generous gift in July 2014, his daughter issued an ice bucket challenge, piggy backing on the ALS fundraising effort. This is continuing to bring additional donations to the Fletcher fund.

Identifying the genetic causes of this disease provides the foundation for research to understand how mutations cause disease pathophysiology. We expect that this will lead to better therapeutic design and ultimately, benefit patients. Dr. Antonellis identified numerous mutations in aminoacyl-tRNA synthetases (ARS) that cause CMT, and Dr. Miriam Meisler identified mutations in a polyphosphoinositide phosphatase. The fundamental goal of research supported by the Fletcher fund is to determine how mutations in a large family of enzymes critical for the generation of all proteins can cause this movement disorder characterized by impaired walking, reduced sensation, and respiratory distress. Understanding how ARS mutations cause these diseases will lead to therapeutic design for treating patients.

Research supported by the Fletcher fund has already: (1) identified the causal ARS mutation in multiple patients; (2) demonstrated that these mutations cause a loss of function in enzyme activity; and (3) revealed that the mutations alter the proper localization of the protein within neuronal cells. Ongoing work will identify neuronal proteins that are affected by ARS mutations and approaches for improving the function of ARS enzymes in patient populations.

In memory of Dr. Myron Levine, the Department of Human Genetics established a fund to support genetic research. Dr. Levine’s early work on genetic regulation of latent infection by herpes virus evolved to exploiting the virus as a carrier for gene therapy, taking advantage of the natural tropism of the virus for neuronal ganglia. Clinical trials are now testing a gene therapy for chronic pain, using the herpes vector. This illustrates how an idea spawned by a basic research discovery can develop into a potential treatment for patients.
Department of Human Genetics 2013-2014 Donations

Contribute today to help support named professorships, lectureships, fellowships and research funds.

For online giving go to: [http://hg.med.umich.edu/giving](http://hg.med.umich.edu/giving).

Thank you to those who made donations this past year:

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2014 Human Genetics Faculty

Back row: Jacob Kitzman, Jacob Mueller, Tom Wilson, John Moran, David Kohrman, David Burke, Ryan Mills, Guy Lenk
Middle row: Gilbert Omenn, Jeff Kidd, Jun Li, Stephanie Bielas, Margit Burmeister, Ken Kwan, Sundeep Kalantry, Miriam Meisler, David Ginsburg, Tom Glover, Julie Douglas
Front row: Wendy Uhlmann, Donna Martin, Monica Marvin, Diane Robins, JoAnn Sekiguchi, Sally Camper, Santhi Ganesh, Catherine Keegan, Beverly Yashar
Not pictured: Tony Antoellis, Martin Arlt, Ray Chan, Vivian Cheung, Ernest Chu, Eric Fearon, Tom Gelehrter, Jeff Innis, Shigeki Iwase, John Kim, Hüira Kopera, Jane Schuette, Charles Sing, Richard Tashian, Cristen Willer

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