

## Research Activities at the UMGCP

The UMGCP Research Program is designed to allow each student to develop a research thesis based on his/her individual interests. Students have access to a wide range of experienced mentors, assistance in securing financial resources, and focused didactic instruction. The class of 2015 is finishing their analyses, presenting their discoveries to our local community, and preparing manuscripts for national publication.

**Caitlin Hale** investigated atypical phenotypes in CHARGE syndrome by carrying out a case review of 28 patients with a confirmed or suspected diagnosis of CHARGE syndrome identified at UM between 2003-2014, as well as a literature review for atypical CHARGE phenotypes. Her review identified a subset of patients in the UM cohort with skeletal and brain abnormalities, which have not previously been considered salient findings in CHARGE syndrome. These findings have led Caitlin and her study team to propose an expansion of the clinical diagnostic criteria for CHARGE syndrome. Based on the quality of Caitlin's work, she was chosen as the keynote speaker at the **12th International CHARGE Syndrome Conference** to be held in July, 2015.

**Michelle Jacobs** partnered with the Inherited Retinal Dystrophy Clinic at the UM Kellogg Eye Center to evaluate the knowledge, attitudes, and practices of a national cohort of ophthalmologists concerning genetic testing. Analysis of survey responses indicated that ophthalmologists are more likely to order genetic testing if 1) they know a genetic specialist or genetic counselor, 2) a patient asks about testing, or 3) they have strong genetics knowledge. These findings are consistent with previous research in oncology and general medicine. However, Michelle also found that ophthalmologists who have personally undergone genetic testing are more likely to order testing for their patients. Michelle's results highlight the need for improved training in genetic training for physicians from diverse disciplines and support a new educational initiative proposed by the NEI and the NIH.

**Diane Koeller**, a dual degree student in Genetic Counseling and Public Health, studied attitudes toward and utilization of genetic counseling among a large national cohort of early adopters of direct-to-consumer genetic testing via 23andMe and Pathway Genomics. Diane's analysis focused on consumer perspectives at three time points over a 6-month time period. Data from over 1400 participants indicate that many people sought genetic counseling after receiving their test results due to difficulty understanding the report or because they found the results to be upsetting. Phase two of Diane's study is focused on interviews exploring participants' motivations for testing and satisfaction with their results. This research will give genetic counselors a deeper understanding of how to interact with healthy patients who seek genetic counseling for their DTC results in a way that best meets their unique expectations and needs.



Poster session at 2014 Department of Human Genetics Retreat

**Kyle Salsbery** explored how genetic counselors address religious and spiritual topics in genetic counseling sessions. Six genetic counselors and three chaplains of Jewish and Christian faith participated in Kyle's focus groups. Kyle discovered that while genetic counselors recognize that religion and spirituality are important to their patients, they are often reluctant to address or explore these topics. Participants described the challenge they experience in balancing the provision of both medical information and emotional or spiritual support. Chaplains highlighted the important role that genetic counselors can play in identifying and referring patients that may benefit from the services of spiritual care providers. Kyle hopes that his research will foster increased assessment of a patient's religious and spiritual needs in the genetic counseling session and lead to increased collaboration between genetic counselors and spiritual care providers.

**Alex Yragui** explored how genetic counselors make decisions about what to discuss with patients about a prenatal diagnosis of Down syndrome. Her qualitative interviews follow up on the findings of Emily Moe (UMGCP Class of 2014), who identified variability in whether genetic counselors describe the lived experiences of families with a child with Down syndrome. Alex found that genetic counselors feel a tension between providing comprehensive information about the diagnosis, while also remaining client-focused and respectful of clients' decisions and attitudes. Genetic counselors appear to make real-time decisions within genetic counseling sessions about what information to discuss, most-often following the patient's lead. Demonstrating support for clients' decisions and maintaining a strong counselor-client relationship were important priorities. Alex's work is part of a larger national research agenda exploring the challenges of providing balanced and comprehensive information about a genetic diagnosis within a client-centered context.

For information on the research project carried out by Katlyn Partynski, visit the Fall 2014 UMGCP newsletter at <http://hg.med.umich.edu/gcweb/news-letters>.

## 2015 ABGC Board Examination: Continued Success!



Congratulations to recent alumni Caroline Weipert, Emily Moe, Lauren Hipp, Deanna Julian, Julie Frank, Dana Schlegel, and Lavania Sharma who recently joined the ranks of ABGC Certified Genetic Counselors. The UMGCP Class of 2014 joins many other UMGCP classes in achieving a 100% board

exam pass rate on their first attempt (compared to the 2015 national first time candidate pass rate of 80%). Additionally, the mean UMGCP candidates' scores were at or above 97% of the national mean for all 15 exam categories, and were at or above 103% of the national mean for 12 of 15 exam categories. Once again, great work!

# Expanding and Individualizing the Clinical Experience: Designer Rotations

During their final semester, UMGCP students have the unique opportunity to complete a designer rotation in a discipline they are highly interested in. This clinical internship allows them to gain a broader range of clinical experiences and in-depth knowledge that complements their genetic counseling training. By way of example, in 2015 Caitlin Hale's interest in pediatrics led her to develop a designer rotation focused on

the lived experiences and complex challenges impacting families with varied genetic conditions. Over the course of the semester, Caitlin interacted with families and healthcare providers in the Disorders of Sex Development Clinic, the Prader-Willi Clinic, the Adult Cystic Fibrosis Clinic, the Sickle Cell Clinic, and the Neurogenetics Clinic in the Michigan Health Care System. In these experiences Caitlin

learned about critical non-genetic aspects of patient care and associated multidisciplinary relationships. Other Designer Rotations have focused on Women's Reproductive Health and Oncology. Given the diversity of patients and expertise at the University of Michigan Health System, there are incredible opportunities for students to develop their own Designer Rotations with mentorship from program faculty.

## SAVE THE DATES!!!

### Monday, April 27, 2015

Genetic Counseling Program  
Universal Admissions Offer Date

### Friday, May 1, 2015

Genetic Counseling Program Graduation  
(Reception at Bev's Thursday night starting at 7pm)

### Monday May 19, 2015, 2:30 pm

James V. Neel Lecture in Human Genetics  
"A Potential Role for 'Jumping Genes' in Cancer"  
Kahn Auditorium. Speaker: Haig H. Kazazian Jr.  
University of Pennsylvania Perelman School of Medicine

### Friday May 29, 2015

Joint UM/Wayne State Genetic Counselors Supervisors Workshop  
Motivational Interviewing for Genetic Counselors  
Speaker: Ken Resnicow, PhD, University of Michigan, School of Public Health Ann Arbor, MI

### Wednesday, September 2, 2015

Fall Orientation for incoming students begins

### Tuesday, September 8, 2015

Fall classes start

### September 18-19, 2015

Department of Human Genetics Retreat  
Gull Lake, MI

### Friday October 9, 2015

An Introduction to the UM Genetic Counseling Program  
Intended Audience: Prospective Applicants  
More details coming this summer.

For more information contact:  
Monica Marvin at [monicama@umich.edu](mailto:monicama@umich.edu)



## Membership in the Greater Genetics Community: Michelle Jacobs and the 2015 ACMG Meeting

UMGCP students have the opportunity to attend and participate in national conferences relevant to the field of genetic counseling, largely due to the Carole McTague Genetic Counseling Enrichment Fund. This academic year, in addition to attending the 2014 Annual Education Conference of the NSGC, Michelle Jacobs and Alex Yragui (Class of 2015) attended the American College of Medical Genetics Conference in Salt Lake City, UT. While there, Michelle Jacobs presented her poster, "Characterization of 16p12.1 microdeletion and microduplication phenotypes in the setting of a second copy number variant." Michelle's interest in this particular chromosomal locus began when she was involved in the evaluation of a patient with

a 16p12.1 deletion and grew as she continued to study the literature. Ultimately, Michelle's enthusiasm and diligence culminated in her ACMG presentation that summarized her review of the literature and a series of UM patients with a microdeletion or microduplication of 16p12.1. Her findings support the hypothesis that microduplication or microdeletion at 16p12.1 may not be sufficient to produce a phenotype, and that a "second hit" of the other allele may be necessary. Her poster highlighted the challenges and complexities in genetic counseling for copy number variants. Kudos to Michelle for her important contribution to our body of genetics/genomics knowledge!

## Community Connections: Learning from Families

Each year, our students are fortunate to be able to learn directly from families affected by a variety of genetic conditions. During these family-centered experiences, parents come to our campus for the express purpose of sharing their stories. This year's visitors included representatives of the local Down Syndrome Support team (including a visit from mothers in the fall and one from fathers in the winter), the Fragile X Association of Michigan, Little People of America, and the Lynch syndrome community. These visits are critical opportunities for students to explore what life is like for families with these conditions, including understanding how grief, coping and adjustment

change over time. Through open and honest discussions with our visitors, students learn how to anticipate relevant questions and emotions families may have, as well as the individual variability in the lived experiences of the families. In addition, these visits help students understand the impact health care professionals can have on families learning about a new diagnosis and then living with that diagnosis. We are incredibly grateful to the families for generously contributing their time and their stories. They truly inspire us!



## UMGCP Alumni Visits

We were thrilled to welcome several alumni back to Ann Arbor for very special visits this year. We kicked off the year with a visit from Meg Hefner (Class of 1980), the 5th recipient of the Diane Baker Alumni Award. Meg shared her expertise regarding CHARGE syndrome and provided mentorship to students related to their research theses and career aspirations. Barbara Hamlington and Darcy Huismann (both from the Class of 2011) travelled from Colorado. Barbara discussed her experiences in a novel oncology-focused genetic counseling team and helped students explore ways of providing leadership in the workplace. Darcy shared her experiences working as a genetic counselor in adult genetics and as a clinical trials coordinator for the lysosomal storage disorders group at her center. Liz Kearney (Class of 1996) and Past President of NSGC travelled from California and provided a leadership workshop for second year genetic counseling students, encouraging students to begin their own leadership journey through examination of their own core values. Most recently, Lavania Sharma (Class of 2013) met with the second year students to discuss her experiences working for a commercial laboratory and the opportunities for genetic counselors to use their expertise and skill set in such settings. It's incredibly rewarding and energizing for faculty and students to hear from alumni. We look forward to seeing more visitors in the coming year!