The OU College of Medicine Genetics Laboratory has expanded. Not only did it gain more physical space (an additional 2,500 square feet), but it has expanded the diagnostic tests it offers. With generous funding from the Children’s Hospital Foundation and under the supervision of Shibo Li, MD and XinJing Wang, MD, PhD (Laboratory Director and Associate Director, respectively) the Genetics Laboratory now offers Whole Exome Sequencing (WES) for clinical diagnosis to medical providers.

WES is a sophisticated technology that permits the detection of genetic defects affecting key portions of genes across the whole genome in a single test. It is a convenient way to identify disease-causing genetic variants to aid in the diagnosis of people with rare and unknown genetic disorders. Unlike other types of molecular testing which focus on a single gene at a time, and are often time-consuming and cost prohibitive, WES queries all genes in the human genome.

The WES testing process involves three key steps: (a) wet bench work—sample preparation and DNA sequencing; (b) dry bench work—bioinformatics analysis and validation testing, and finally, (c) clinical correlation of the results. It takes up to six people, including lab technicians, lab directors, medical providers, and genetic counselors to process and analyze samples and to ultimately relay the result to the patient / family. According to Li, “it takes about a teaspoon of blood” to be able to extract enough of a person’s DNA to run the WES testing.

Prior to the development of this technology, 25-30 percent of patients suspected of having a genetic disorder had no molecular diagnosis, even after exhaustive genetic testing. Even with cutting-edge
MSGC Leadership Continued

of genetic counselors for the emerging trends in the field.

When she was a trainee, Youngs hadn’t given much thought to the crucial role played by the program director to support the various components of the program and its students. However, as she began her clinical career in Kansas City, she realized the importance of academia and took advantage of opportunities to teach medical students and genetic counseling students.

As the leadership of the program transitions, Hassed, who plans to retire in 2018, will remain as a resource for Youngs until the end of the academic year. She will also assist in revising the clinical learning objectives and the varied training opportunities for MSGC students.

Master’s degrees from the MSGC program are conferred by the OUHSC Graduate College. It is the only degree-granting educational program in the Section of Genetics and is gaining national recognition for its quality of education and affordability. In the past 14 years, 100 percent of our MSGC program graduates have gone on to pass the Genetic Counseling Certifying Board Exam. A trend that Youngs will work diligently to maintain.

Hassed’s advice to Youngs is “Be calm. Stay in the boat and avoid the rocks.”

The MSGC program at the OUHSC is accredited by the Accreditation Council for Genetic Counseling through 2020.

Recent Poster Abstracts:

**Porter T., Anderson M., Jacobs N, Hassed S. From Adversity to Advocacy: Factors Influencing Parental Growth toward Advocacy following the Diagnosis of Down Syndrome in a Child. Presented at the 36th Annual Conference of the National Society of Genetic Counselors, September 15, 2017, Columbus, OH.**

**Rodden L.N. and Bidichandani S.I. A CRISPR-Cas9 based approach for targeted epigenetic modification in Friedreich ataxia. 2nd International Ataxia Research Conference, Pisa, Italy. September 27-30, 2017. **Winner of a FARA Student Travel Award**

Lab Continued

“Erin is the right person to take the MSGC program to the next level.”
Dr. Susan Hassed

testing such as WES, it is not always possible to link the genetic disorder with a causative variant in the genome. However, as this technology grows and the databases linking clinical presentations with causative variants expand, Li and Wang are excited about the potential for providing definitive diagnoses to patients with rare and unknown genetic disorders.

A unique challenge in WES testing is the difficulty of communicating relevant details from a 10 to 20 page lab report to medical providers, especially to those who have not specialized in genetics. Li and Wang will rely on the skills of genetic counselors to bridge potential communication gaps between the lab, the patient’s medical provider, and the affected patient / family.

A common question is whether health insurance will cover the cost of WES. The answer is yes, but with some limits. Testing is more likely to be approved for a patient who has a disorder where the genetic cause is currently unknown.

In addition to WES, the OU College of Medicine Genetics Laboratory continues to provide a variety of traditional genetic tests involving chromosome and molecular studies, CGH array testing, FISH for chromosomal anomalies and malignancies, and routine chromosomal karyotyping.

Wenfu Li, MS identifies and reviews Whole Exome Sequencing test results with XinJing Wang, MD, PhD.

For more information about the Genetics Section visit: [www.ouhsc.edu/genetics](http://www.ouhsc.edu/genetics)
Welcome the MSGC Class of 2019

Each fall new students from across the United States and beyond, join the 2-year, Master of Science in Genetic Counseling Program (MSGC). This August we welcomed the members of the MSGC Class of 2019:

- Ashlee Byrnes—Falls Church, VA—MS in Biochemistry and BS in Biology
- Jada Jackson—Edmond, OK—BS in Biology
- Cheryl Reeves—Wichita Falls, TX—BS in Biology and BS in Psychology
- Lauren Taylor—Coppell, TX—BA in Neuroscience
- Kelsey Watson—Atlanta, GA—BS in Biology

These five 1st year students join the five 2nd year students in classes, clinics, labs, seminars, case conferences and Genetics Journal Club for the next year. Many of our students also gain experience working as Graduate Research Assistants (GRA) in the Section of Genetics, Community Pediatrics or in research laboratories on campus.

Physician Assistant Joins Clinical Genetics Team

In September, Abby Moeller, PA-C joined the Genetics clinical team. She earned her Master of Health Science-Physician Associate degree from OU College of Medicine. Moeller previously worked in Urologic Oncology at the Stephenson Cancer Center.

Moeller joins a team of three clinical geneticists; physician assistant, Ashley Taylor; seven genetic counselors; three clinical laboratories; two genetic research laboratories; a nurse case manager; and a dietician. She will see patients in the OU Children's Physicians Specialty Clinic and participate in our training programs, including a ACGME Genetics Residency program; Master of Science in Genetic Counseling Program; and three ABMGG Laboratory Training Programs.
Out and About

Clockwise from Upper Left: Dr. Sanjay Bidichandani, Dr. Hua Wang and Dr. XinJing Wang smile during the MSGC Welcome potluck. Emily Teague, MSGC student, fills her plate during the potluck. Tavanna Porter, MSGC Class of 2017 presents her poster at the NSGC conference. Faculty, alumni and students gather for a reunion dinner at NSGC 2017 in Columbus, OH. Students and alumni have fun at the NSGC conference. Admin staff view the August 21, 2017 total solar eclipse. Patients and their families join the genetic counselors and students in preparation for a clinical correlation session for 1st year medical students. Trainees and Lab technicians review results in the Genetics laboratory. Ashley Taylor, Julie Boyd and Dr. Susan Palmer chat at the potluck. Center: Caleb Heid shows off his best Arnold Schwarzenegger impression.
Recent Publications:


John Mulvihill: was an invited grant reviewer for The Neurofibromatosis Therapeutic Acceleration Program, a new national effort to understand and control cutaneous neurofibromas. (September 2017)

Sanjay Bidichandani: served on NIH study section: NINDS / ZNS1 SRB-A 12, Clinical trial readiness for rare neurological and neuromuscular diseases - (U01) (PAR-16-020). Alexandria, VA; July 14, 2017

John Mulvihill: gave a research seminar in the morning, “Germ cell mutation in cancer survivors: Using solidarity and population registries,” at the Geisel School of Medicine Center for Genomic Medicine and, in the afternoon, a CME lecture, “Why my child: Mutation, neurofibromatosis, Denmark, and solidarity” at Dartmouth, NH. (September 15, 2017)

Sanjay Bidichandani: made a platform presentation: “Epigenetic silencing in Friedreich ataxia is caused by hypermethylation of the FXN CpG island shore.” International Ataxia Research Conference, Pisa, Italy. (September 27, 2017)

Sanjay Bidichandani: served on the selection committee to identify new inductees into the Academy of Teaching Scholars of the OU College of Medicine.

Klaas Wierenga: will serve as the co-Principal Investigator for the HRSA funded Heartland Regional Genetics Network. An aim of this new network is to expand genetics telemedicine capacity in Oklahoma. The anticipated project period is June 2017 to May 2020.

New Funding for Genetic Research

Dharambir Sanghera: received a PHF Team Science grant in the amount of $99,964 for a project entitled: Discovery of novel biomarkers of Acute Ischemic Stroke using metabolome and miRNA studies.

Sanjay Bidichandani: received a grant in the amount of $140,000 from BioMarin Pharmaceutical for a project entitled: FXN DNA methylation as a biomarker for response to HDAC inhibitor treatment in Friedreich ataxia.

Dr. Dharambir Sanghera

Dr. Sanjay Bidichandani

Genetics Journal Club

The Section of Genetics has hosted a weekly journal club for the past 18 years. The purpose of Genetics Journal Club is to provide a regularly-scheduled opportunity for faculty, staff and trainees to critically evaluate current journal articles in the field of human or medical genetics; to discuss the clinical or research applicability; and to collaborate in an intellectually stimulating environment.

The Genetics Journal Club is directed by Michelle Polan, MD; XinJing Wang, MD, PhD; and Erin Youngs, MS, LCGC.

Genetics Journal Club is held Thursdays at noon and anyone with an interest in genetics is welcome to attend.

For more information about the Genetics Section visit: www.ouhsc.edu/genetics