The Pediatric Genetics Department at the OUHSC has been building a solid foundation since Dr. John Mulvihill arrived at the OU College of Medicine in 1998. Upon his arrival he began a strategic plan of hiring a team of clinicians and researchers. One of his early recruits was Shibo Li, MD, in 1999, who now serves as Professor in Pediatrics and Director of the Clinical Cytogenetics and Molecular Laboratories.

In 2009, Andrea Wierenga, PhD, became a valuable member of the Genetics team as Director of the Biochemical Genetics Laboratory and Clinical Assistant Professor. Together, Drs. Li and Wierenga offer a dynamic panel of testing that complements the OUHSC Genetics Section multi-faceted mission: “To offer state-of-the-art genetic laboratory services for patients, family members and professional colleagues and to discover new knowledge about the genetic determinants of human disease ...”

The Clinical Cytogenetics, Molecular and Biochemical Genetics Laboratories form the Core Genetics Laboratory and all are accredited through the College of American Pathologists (CAP) and the Clinical Laboratory Improvement Amendments (CLIA). The CAP accreditation has stringent guidelines and examines quality control procedures, equipment, facilities, safety, overall management, etc. In order to be accredited by CLIA, laboratories first have to meet the guidelines and inspection process set by the CAP.

In 2014, The American Board of Medical Genetics and Genomics (ABMGG) granted all three labs with provisional accreditation as being the three new clinical laboratory training programs in Clinical Molecular Genetics, Clinical Cytogenetics and Clinical Biochemical Genetics. These are the flagship clinical lab training programs in medical genetics, and only 28 other schools in the US currently offer all three training programs!
Staff News >>>

Congratulations Shona on Staff Senate Employee of the Month!

Shona Whitehead, Sponsored Program Coordinator in the Pediatric Genetics Section was appointed Staff Senate Employee of the Month for January!

New Faces >>>

Sarah Hoss, Research Project Coordinator.

Background: MS in Biology from the University of Central Oklahoma. Previously: Clinical Laboratory Scientist at the OK State Dept. of Health.

“Shona has an aptitude to take a problem and look at it from many angles until it is solved. She stays focused and her turnaround time on virtually any project is amazing.”
- Genetics Section member

So Long Carrie Guy!

Carrie Guy, MS, LCGC, Clinical Professor and Assistant Director of the Master of Science in Genetic Counseling (MSGC) Program, recently left the OUHSC campus for Laramie, Wyoming.

She was an integral part of the leadership in the Core Genetics Lab as well as the MSGC Program.

Although she won’t be on campus any longer, she will continue to work for Genetics remotely throughout the transition. Good luck to Carrie and the Guy Family!

Matt Grim, Genetics Business Advisor, and staff present Carrie Guy with a photo of the OU Children’s hospital taken by Dr. Shibo Li, and autographed by the entire Genetics section.

For more information about the Genetics Section visit: www.oumedicine.com/genetics
residents specializing in Genetics, fellows from varied specialties such as Hemoc, Pediatrics, Endocrinology, Neurology, etc. Dr. Li also hosts a robust exchange program with Jilin University in China where trainees come from China to study and gain laboratory and research experience in the Molecular or Cytogenetics subspecialty. His lab has also welcomed high school students and undergraduates to tour the lab and view specimens through the microscope.

Although he directs two labs with different subspecialties, Dr. Li considers it to be one laboratory because most of his 17 member staff, comprised of a mixture of faculty, trainees, and students, cross-train and work in both. The Molecular Genetics lab specializes in the field of genetics that studies the structure and function of DNA and the number of chromosomes. Based on this, the lab along with the clinicians make a prognosis and determine treatment. Some of the molecular services offered include DNA sequencing and testing for disorders such as Huntington Disease, Sickle Cell, Spinal Muscular Atrophy and many others.

The Cytogenetics Laboratory focuses on the branch of genetics that is concerned with the study of the structure and function of the cell and the process of putting together karyotypes (or the complete set of chromosomes). Changes can happen in the cells as chromosomes can break, move, or duplicate, and can occur in newborns, prenatal, congenital, as a result of cancer, etc. Examples of cytogenetic testing available are chromosome analysis and the fluorescence in situ hybridization test or the “FISH” test which maps the genetic material in a person’s cells and can be used to visualize specific genes or portions of genes.

Together the Cytogenetics and Molecular Genetics lab, performed over 4,000 tests in 2014, a huge increase from the 200+ tests they averaged back in the first year of operation. Not only do the labs play an important role in testing for different departments at the OUHSC but they continue to partner with OU Norman, and perform testing and other services for in-state entities such as Baptist, Lawton, Tulsa, in addition to testing for international locations in Canada and other countries. In fact, they are one of the very few genetic testing facilities that test for Bartter’s Syndrome, a rare kidney disorder that affects approximately 1 per million.

For 2014: The Clinical Molecular and Cytogenetics Labs performed over 4,000 tests! The Biochemical Genetics Lab performed over 1,100 tests!

For 2015: The Clinical Molecular and Cytogenetics Labs performed over 4,000 tests! The Biochemical Genetics Lab performed over 1,100 tests!
Publications


>>> Klaas Wierenga, MD, Associate Professor, was featured in the Fall 2014 issue of the OU Medicine magazine in recognition for his groundbreaking collaborative research for Stormorken syndrome. The work appears in Proceedings of the National Academy of Sciences.

Presentations

Sanjay Bidichandani, MBBS, PhD, CMRI
Claire Gordon Duncan Chair of Genetics and Section Chief:

> Featured speaker, where he presented a talk entitled “Translating Research into Therapies,” at the MDA Uncork the Cure gala event. It was attended by 300 people and $300,000 was raised for neuromuscular disease research (Dallas, TX; Nov. 6, 2014).

> Presented a lecture entitled “Treating the Untreatable” at the general assembly of the Oklahoma School of Math and Sciences (Nov. 19, 2014).

Awards

>>> Yogesh Chutake, PhD, Postdoctoral Research Fellow in the Bidichandani lab won the competitive Postdoctoral Fellowship Award from the Penn Medicine Center for Orphan Disease Research and Therapy at the University of Pennsylvania.

For more information about the Genetics Section visit: www.oumedicine.com/genetics
people worldwide.

On the research front, Dr. Li’s lab stays active with thirteen publications in 2013 and eight in 2014. Collaborating with different groups in research such as OU Norman, University of Pittsburgh, etc. is very important to continue new genetic discoveries and projects. As new discoveries are found through research, educational opportunities abound.

He credits the success and growth of the lab to his entire team by saying “It’s due to their hard work— which needs to be appreciated and recognized.” He will continue to encourage his team to keep improving and striving for excellence, but remains in awe of everything they have and continue to accomplish.

Before coming to OUHSC Andrea Wierenga was at the University of Miami pursuing a PhD fellowship in metabolic nutrition. The idea of being able to establish a lab from the ground up that would provide much needed newborn screening services was a motivating challenge, so Dr. Wierenga set her sights on OU. Soon after, she became Director of the Biochemical Genetics Laboratory (BGL) and the other half of the Core Genetics Laboratory.

The nature of the testing and research that is performed in the BGL delves into the underlying mechanisms of inherited metabolic diseases. Biochemical genetics itself is the study of the fundamental relationship between genes, protein and metabolism. Wierenga and her team of two technologists and one postdoctoral research fellow, perform testing services for inborn errors of metabolism for newborn infants identified through the Oklahoma State Newborn Screening Program. As a result of contracting with the State Department of Health for assessments in 2013, the BGL is the sole testing site for all newborns in Oklahoma testing positive for metabolic diseases and disorders. Tests include amino acids, organic acids, Vitamin D screenings and many others.

Business continues to grow in the BGL. For the year of 2014, the BGL lab performed approximately 1,100 tests almost tripling their number from 2013, when they performed approximately 400. Increasing their testing capabilities isn’t their only focus. They anticipate continuing collaborations with other departments on campus such as a recent project with Nephrology, and entities outside of OUHSC.

With continued refinement of testing procedures and standardizing methods derived from research ventures, the BGL is proving their dedication to innovation by leveraging new ways to analyze their data. Partnering with OU medical student Jonathan Lee last year in a bioinformatics project to create a unique computer program to assist in the diagnosis of genetic conditions, helps to further the goal of the BGL. “Going forward we hope to continue collaborations that look at metabolism and nutrition in populations and to explore research that will help alleviate problems in management of inborn metabolic disorders,” said Dr. Wierenga.

Education and research remain at the forefront of the Core Genetics Laboratory’s mission. Upon granting accreditation the ABMGG commented on the Core Genetics Lab’s strengths as having “extensive educational opportunities” and a “variety of research opportunities” as one of the many qualifying factors in awarding accreditation. Garfield Simon, Postdoctoral Research fellow in the Biochemical Genetics Laboratory confirmed that assertion: “My experience here has been invaluable. Being a part of a team that employs varying bio-analytical skills to decipher metabolic profiles is quite fulfilling. I now have a greater knowledge of the different approaches employed in the diagnostic workup of pediatric genetic patients.”

Dr. Li and his team have solid plans to further develop whole genome sequencing for the lab in 2015, and Dr. Wierenga and the BGL are currently working on further development of enzyme assays and dried blood spot tests. Looking forward the future looks bright for the Core Genetics Laboratory, as each lab team continues to revamp itself with the emphasis of new technological breakthroughs.

Jianqin Zhang, visiting researcher, performing DNA isolation in the Molecular Lab; Garfield Simon, Postdoctoral Research Fellow, and Bryan Lettenmaier, Research Technician, discuss test findings in the Genetic Biochemical Lab.
Sanjay Bidichandani, MBBS, PhD, was appointed as Scientific Director on the Executive Committee of the Board of Directors of the Friedreich Ataxia Research Alliance (FARA), the world’s largest funder of research in Friedreich ataxia.

Susan Hassed, PhD, LCGC, will be featured in February’s issue of *Oklahoma Magazine* where she was interviewed about genetic testing.

Sanjay Bidichandani, MBBS, PhD, served on the abstract review committee that selected oral presentations for the International Ataxia Research Conference to be held in April 2015 in Windsor, UK.

Special Thanks to Shibo Li, MD; Andrea Wierenga, PhD; Carrie Guy, MS, LCGC; and Shona Whitehead for being an integral part in the accreditation for the ABMGG Clinical Laboratory Training Programs in Clinical Molecular, Clinical Cytogenetics and Biochemical Genetics!

Clockwise from top left: Genetics Section staff members show off their holiday sweaters; Genetics Section gathers for Holiday Potluck; Dr. Shibo Li, Matt Grim, and Dr. Sanjay Bidichandani survey Carrie Guy’s autographed photo; Genetics Section show off their Halloween costumes; (l to r) Drs. Marsha Pratt, Chimene Kesserwan, Bidichandani and Carrie Guy chat in the hallway.