In this Issue >>>

Core Genetics Lab Opening
MSGC Graduation

Volume #1
Issue #1
July 2013

A Newsletter of the OUHSC Department of Pediatrics, Section of Genetics

The *Gene Scene*

Gene Team >>>

Multiple summer scholars in genetics this summer:

- *American College of Medical Genetics Scholars program selects Jonathan Lee to conduct research with Drs. Klaas Wierenga and John Mulvihill*
- *SURE student, Zain Raza, conducting research with Dr. Bidichandani*
- *"Gene Team" also welcomes Suresh Kandavanam, Andrew Gessouroun, and Barat Venkataramany*

Genetics Residency >>>

Goodbye Marsha Pratt

Marsha Pratt, MD completed her genetics residency on June 30th. "I feel very fortunate to have joined the program." Marsha states. She plans to take the board exam in August and hopes to one day rejoin the OUHSC family.

Hello Esther Lee

Esther Lee, MD joined the program on July 1st following an Internal Medicine internship and one year of ophthalmology residency training.

Laboratory Grand Opening

Governor Mary Fallin cuts ribbon

Left to right: John Mulvihill, MD, Sanjay Bidichandani, MBBS, PhD, Shibo Li, MD, Hon. Gov. Mary Fallin, Terrence Stull, MD, Wade Christensen, Andrea Wierenga, PhD

The Core Genetics Laboratories celebrated its grand opening ceremony on May 30th. The event boasted attendance of nearly 100 faculty, staff, Children's Hospital Foundation board members, donors and advocates. Topping the list of attendees was the Honorable Governor Mary Fallin, who led the ribbon-cutting ceremony.

"Thanks to genetic research, scientists have made some remarkable, life-changing discoveries to improve the lives of children and their families," Gov. Fallin said. "This new lab space will help accelerate that work and give our physicians the tools they need to further advance pediatric care."

"The Core Genetics Laboratories are an integral part of the section of Pediatric Genetics, and greatly enhance our clinical service, research and educational missions," stated Sanjay Bidichandani, MBBS, PhD, CMRI Claire Gordon Duncan Chair in Genetics and Chief of the Section of Genetics.

"This increased lab space will allow us to greatly expand the repertoire of diagnostic tests leading to improved care for the

Continued on page 3...

Klaas Wierenga, M.D., Associate Professor, Pediatric Genetics, presented a poster at the 2013 American College of Medical Genetics Annual Meeting titled "Autosomal Recessive Intellectual Disability Caused by Homozygous Deleterious Mutations in TRMT1 Identified by Whole Exome Sequencing".

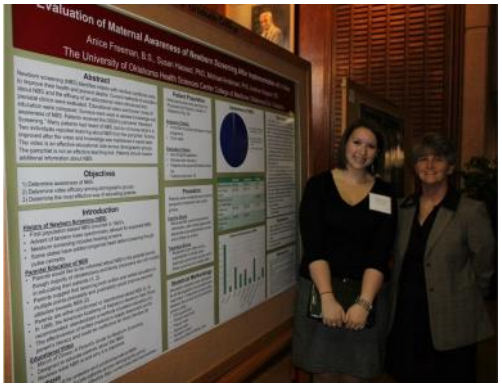
At the 2013 American College of Medical Genetics meeting in Phoenix, Arizona, March 19-23, **John J. Mulvihill, M.D.,** Professor, Pediatric Genetics, made an oral presentation on Emergency Preparedness among Newborn Screening Laboratories.

<<< Publications & Presentations >>>

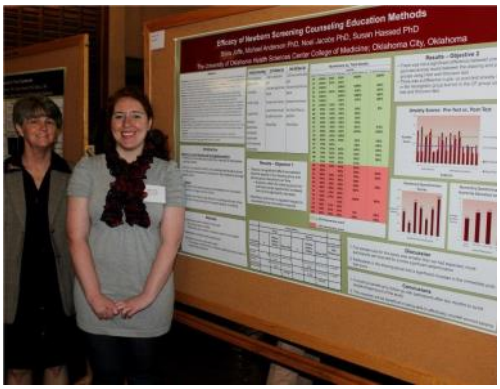
Sanjay Bidichandani, MBBS, Ph.D., Section Chief, Pediatric Genetics, presented a poster at the Keystone Symposium on RNA Silencing held in Whistler, BC, Canada from March 19-24.

Sanghera DK. et al. Genome-wide association study identifies a novel locus contributing to type 2 diabetes susceptibility in Sikhs of Punjabi Origin from India. *Diabetes*. 2013 May; 62(5):1746-55.

Graduation Day: Masters of Genetic Counseling



Graduates Anice Freeman (above) and Brina Joffe (below) with Dr. Susan Hassed



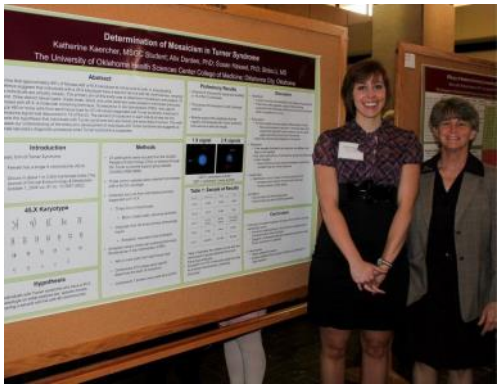
In May, we celebrated the graduation of the 2013 class of MSGC students. The two year program, directed by Susan Hassed, PhD, involves coursework and clinical experience. Additionally, the students conduct their own thesis research project which is presented at their masters defense.

Anice Freeman presented, "Evaluation of Maternal Awareness of Newborn Screening: Implementation of an Educational Video in Prenatal Clinic." Anice is currently interviewing in Atlanta, GA.

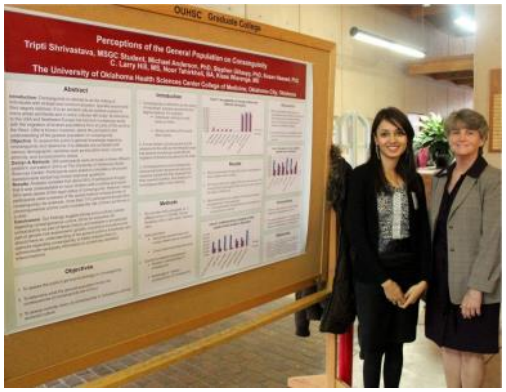
Brina Joffe's research project was entitled, "Efficacy of Newborn Screening Counseling Education Methods." Brina has accepted a pediatric genetic counseling (GC) position at University of Arkansas for Medical Sciences Medical Center in Little Rock, AR.

Kate Kaercher performed research entitled, "Turner Syndrome Mosaicism," and has accepted a pediatric GC position at Sanford Children's Specialty Clinic in Sioux Falls, SD.

Tripti Shrivastava's project was entitled, "Perceptions of American Population on Consanguinity." Tripti will be working as a cancer GC at US Oncology in El Paso, TX.



Dr. Susan Hassed stands alongside Kate Kaercher (above) and Tripti Shrivastava (below)



Section Happenings >>>

Shona Whitehead, Sponsored Program Coordinator, and husband, Chris, welcomed their son, John Stephen, to the world on June 8th.

Aimee Martin, Research Project Coordinator, was married to Albert Babarsky on June 22nd.

From the IRB >>>

Dr. Klaas Wierenga has two newly open studies both sponsored by BioMarin.

PKUDOS is a registry trial for patients with phenylketonuria (PKU). Prism 301 is a phase 3 study which looks at the safety and tolerability of BMN 165 in PKU patients. Both studies are actively recruiting!

Grand Rounds >>>

Sanjay Bidichandani, MBBS, PhD, discussed "Trends in Development of Targeted Therapies for Genetic Diseases" on April 10th.

Visiting guest, Virginia Kimonis, MD, presented "Novel Treatment Strategies in VCP Inclusion Body Myopathy, Paget Bone Disease and Dementia" on May 29th.

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

Varied Backgrounds Bring Diversity to the Section

Staff >>>

Fathia Jones; Administrative Coordinator

BS in Business Administration, Southern Nazarene University

Previously: Government Contractor, Federal Aviation Administration

Aimee Martin; Research Project Coordinator

BA in Chemistry, West Virginia University

Previously: Sponsored Programs Administrator, OUHSC

Bidichandani Lab >>>

Megan Brophy; Ph.D. Student

MA in Medical Science, Loyola University

Previously: Research Assistant, OMRF

Whitney Costello; Research Technician

BS in Physics, minor in Chinese, University of Oklahoma

Future plans: Graduate degree in biophysics

Christina Lam; Research Technician

BS in Biology, minor in Chemistry, University of Central Oklahoma

Previously: Research Assistant, UCO Department of Biology

Li Lab >>>

Lina Lin; Associate Research Scholar

Ph.D., Norman Bethune Medical Sciene of Jilin University, China

Research focus: Digestive diseases

Jing Ning; Associate Research Scholar

MS in Endocrinology, Second Hospital of Dalian Medical U., China

Previously: Vascular-cardiology Intern, Second Hospital of Jilin University

Sanghera Lab >>>

Praveen Natt; Research Assistant I

MS in Biology (focus: Cell & Molecular Biology), Illinois Institute of Technology

Previously: 2011 & 2012 Summer Internship, Dow Agrosciences

Anuradha Subramanian; Associate Research Scholar

MS in Molecular Medicine, University of Essex, UK

Previously: Cancer Research Intern, Apollo Specialty Hospital, India

Lab Opening (cont.)

families and children of Oklahoma."

The Clinical Genetics Laboratory began in 1999 under the direction of Dr. Shibo Li. The laboratory performs molecular and cytogenetic analyses and provides DNA sequencing for over 70 disorders and cytogenetic analysis for over 100 different chromosome anomalies seen in hematological and other disorders. Additionally, the lab also conducts research projects in various areas.

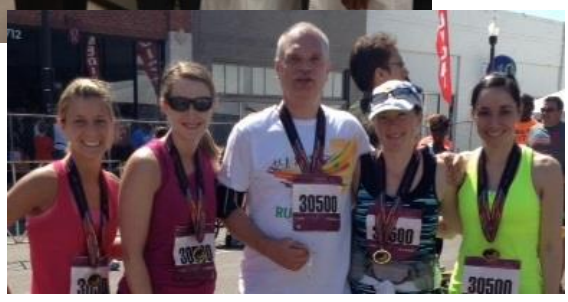
The Biochemical Genetics Laboratory began in the latter part of 2009 upon Dr. Andrea Wierenga's arrival to the university. The lab offers state-of-the-art testing services for inborn errors of metabolism and works closely with the Oklahoma State Newborn Screening Program. It performs a variety of tests, such as assaying amino acids, organic acids and Vitamin D.

The Children's Hospital Foundation (CHF) is a non-profit organization that supported the lab addition and renovation. Kathy McCraken, Executive Director of CHF, said, "Our state can be extremely proud of the improved care that is available for children fighting rare and genetic diseases, but these accomplishments happen only because of the unwavering support of generous donors."

Noteworthy >>>

- **Sanjay Bidichandani, MBBS, PhD,** Section Chief, Pediatric Genetics, served on grant review panels for the American Association for the Advancement of Science and the non-profit British Foundation Ataxia UK.
- In Hiroshima, Japan, **John J. Mulvihill, MD,** Professor, Pediatric Genetics, chaired the annual Science Advisory Committee for the Radiation Effects Research Foundation, which continues to study the survivors of the US atomic bombs.
- **Sanjay Bidichandani, MBBS, PhD,** Section Chief, Pediatric Genetics, presented, "Development of Targeted Therapies for Rare & Genetic Diseases: The New 'Brave New World'" in the Henry Turner Lecture Series on February 13.
- **Dharambir Sanghera, PhD, FAHA,** Associate Professor, Pediatric Genetics, was invited to serve as a reviewer for "Chronic Disease, Aging, and Genetics" [ZRG1 PSE-P (02) M] an NIH Study Section held on March 14, 2013.
- **Sanjay Bidichandani, MBBS, PhD,** Section Chief, Pediatric Genetics, served on grant review panel for MDA's translational research "Bridge-to-Industry" postdoctoral fellowship program.
- **John J. Mulvihill, MD,** Professor, Pediatrics Genetics, attended the Joint Conference of the Human Genome Meeting 2013 and the 21st International Congress of Genetics, April 13-18, in Singapore. Mulvihill gave an invited lecture on "Solidarity: A neglected principle in human genomics," and served as the only American on the Committee on Ethics, Law, and Society of the International Human Genome Organization.
- **Sanjay Bidichandani, MBBS, PhD,** Section Chief, Pediatric Genetics, served on the Medical Advisory Committee of the Muscular Dystrophy Association; Tucson, Arizona; May 23-24, 2013.
- **Dharambir Sanghera, PhD, FAHA,** Associate Professor, Pediatric Genetics, has been awarded an OCAST grant sub-contract AP12.2-022, Contract #: 8179 (PI Dr. Fabiola Spence, OCCC). The purpose of the award is to train students for Biotechnology Internship Program under OUHSC-OCCC partnership.
- **Sanjay Bidichandani, MBBS, PhD,** Section Chief, Pediatric Genetics, served as Chair of session on "Genetic Modifiers" at the annual MDA conference on neuromuscular disease in Washington DC (April 21-24, 2013).

Around Campus



Clockwise from top: Dr. Andrea Wierenga and her lab staff; Dr. Shibo Li and his lab team; MSGC students celebrate graduation; Genetics team competes in the Memorial Marathon Relay; Dr. John Mulvihill, Dr. Terrence Stull, and Dr. Sanjay Bidichandani attend the core genetics lab grand opening; Genetics team members host the "Race 4 Jase" galactosemia event.