Diabetes, India and zebra fish. These words may seem unrelated, but to Dr. Dharambir Sanghera, Professor and Director of the Genetic Epidemiology Laboratory here at OUHSC, the three are definitely related.

For most of her career, Dr. Sanghera has focused on diabetes research and how it affects different populations on the Indian sub-continent. One fifth of the world’s population is in India and until the last few decades was relatively unaffected by Type II diabetes. She and other researchers across the globe have speculated that as the Indian culture has become more “westernized” the prevalence of Type II diabetes has increased across the Indian sub-continent and Southeast Asia. Most Indians don’t fit the stereotypical diabetes patient, most are thin, non-smokers or non-drinkers, and 50-60% are vegetarian. And Indians who are diagnosed with diabetes are often young, about 35 years old. This led to the idea that there might be unique genetic variants causing this surge in the diagnosis of diabetes.

For Dr. Sanghera, a genetic epidemiologist, the hunt began to find the possible genes causing this increased susceptibility to Type II diabetes in Indians. From the Presbyterian Health Foundation (PHF) to take the human genetic variant found in the Punjabi 6 and add it to the zebra fish eggs, to test and determine if, as the fish grows, will the fish’s liver exhibit signs of high triglycerides and then develop diabetes? The research hypothesis is that, if this is in fact the gene related to the development of diabetes in humans, then the fish will get diabetes and fatty liver disease. This exciting study is currently ongoing.

How will this type of research effect future treatment of Type II diabetes? Identifying genetic variants for different diseases, particularly complex and common disorders like diabetes, could eventually lead to improved personalized medicine, like prevention screenings or personalized therapeutics for disease treatment. Dr. Sanghera firmly believes that “family studies are key and fundamental to finding and studying genetic variants, which is directly relevant to the future of personalized medicine.”

Translating Research into Personal Medicine

University of Pennsylvania; and after screening almost 200,000 samples, eight genes were discovered that could be the cause for this sudden rise in diabetes.

The focus now is on one specific, but common, GCKR gene, which is found in the Punjabi 6, a distinct ethnic group in the Indian sub-continent. Through funding from the National Heart, Lung and Blood Institute (NHBLI), Dr. Sanghera and collaborators, sequenced DNA from 900 people and of those, 3 families were carrying the same genetic variant as the Punjabi 6.

How do the zebra fish play a role? Dr. Sanghera and her lab received funding from the Presbyterian Health Foundation (PHF) to take the human GCKR gene with 3 disruptive mutations identified in Sikh pedigrees.
In Dr. Sanjay Bidichandani’s research lab each day brings new questions or problems to solve which invigorates the lab staff to look beyond basic research and consider the possibilities of translational research which might one day produce an effective drug treatment for people affected with Friedreich ataxia (FA).

A recent breakthrough in the Bidichandani lab was the discovery of a new epigenetic silencing signal in the gene for FA which was identified through Next-Generation Sequencing (NGS). This analysis revealed a novel and dramatic difference in the silencing signal in the gene for FA in people with FA versus those who do not have FA. This discovery has the potential to identify a biomarker which could help predict the prognosis of FA. Excitingly, early evidence also indicates that this signal may help to predict which patients have a better response to gene reactivating drug treatments for FA. It is the characterization of this novel biomarker for drug response that Dr. Bidichandani and his lab staff are currently focused on.

In collaboration with researchers at the Children’s Hospital of Philadelphia (CHOP) and through a contract with BioMarin (a pharmaceutical company), the Bidichandani lab is conducting NGS based screening of FA patients’ cells to help identify mechanisms that will help predict a patient’s response to gene reactivating drugs.

FARA, the Friedreich’s Ataxia Research Alliance, recently awarded Dr. Bidichandani a grant to purchase a new NGS system for his lab. With the new NGS system the lab will be able to screen 100’s of DNA samples. For Dr. Bidichandani, “this will help to create a meaningful partnership between our lab, other academic researchers, drug development experts in pharmaceutical companies, and a non-profit patient advocacy group.”

Dr. Sanjay Bidichandani

To Yogesh Chutake, PhD and Layne Rodden, a PhD student in the Bidichandani lab, it is clear that it will take both basic science research and translational research to succeed in identifying therapies for FA. Dr. Chutake says “organizations like FARA are key to bridging the funding gap and help connect companies like BioMarin to researchers in academia.”

Ms. Rodden will be taking the lead in testing of potential drug therapies developed by companies like BioMarin. She is excited to see what possibilities will unfold for future treatments for FA and the hope it will bring to thousands of families, as the lab continues to be on the forefront of FA research.

Above: Dr. Chutake and Layne Rodden set-up the NGS system to analyze samples. Upper Right: Dr.Chutake prepares samples for analysis. Lower Right: Ms. Rodden selects samples for analysis.
Genetics Celebrates Dr. Terrence Stull’s Retirement

After 22 years as chairman of the Department of Pediatrics, Dr. Terrence Stull announced he was retiring from OUHSC. Dr. Stull was key to the recruitment of Dr. John Mulvihill in 1998 to fill the Kimberly V. Talley/CMRI endowed chair in genetics and Dr. Sanjay Bidichandani for the Claire Gordon Duncan/CMRI chair in genetics. Dr. Stull has been a great supporter of the Section of Genetics, a wonderful leader for Pediatrics and he will be missed. The Section of Genetics hosted a party at the home of Dr. Shibo Li, August 1st, to thank Dr. Stull for his service. Dr. ChaoYing Yan, Chairman of Pediatrics and Li Li Yang, Liaison for the International Office at the First Hospital of Jilin University in China, with whom Dr. Stull and Dr. Li have collaborated for several years to create an educational exchange program, were in attendance at the party.

(Above, L-R) Party attendees enjoy the food and the company of colleagues; Dr. Stull chats with Dr. Susan Palmer; and Dr. Sanjay Bidichandani talks with the Altshuler family.

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

(Clockwise from Top): 1. The Genetics Laboratory staff celebrate at Danielle Otis’ reception for Pediatrics Employee of the Year. 2. Dr. Hua Wang and Dr. Susan Hassed observe an interactive Genetics Journal Club on the topic of giving feedback, presented by Dr. Alix Darden. 3 & 4. Patients and families join the genetic counselors and genetic counseling students in preparation for two different clinical correlations sessions with OU medical students. 5. MSCG Class of 2018. 6. Genetics faculty and staff enjoy a potluck lunch welcoming the new MSGC students. 7. Genetics faculty and staff fill their plates during the MSGC Welcome Potluck.

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New Telemedicine Genetic Counseling Clinic

The Section of Genetics is pleased to announce the beginning of a new partnership with Oklahoma Cancer Specialists and Research Institute (OCSRI) in Tulsa. Members of the OUHSC genetic counseling team will provide genetic counseling services, via telemedicine, to patients in the Tulsa area who might have disease onset at a young age, high genetic predisposition or family history of cancer.

The new telemedicine clinic begins in October and will be offered on the 2nd Friday of the month.

OCSRI is an research affiliate of the Stephenson Cancer Center and certified member of the MD Anderson Cancer Network. This new clinic joins the growing number of genetic counseling service clinics provided by the OUHSC Section of Genetics.

Poster Abstracts:

**Fanning EA; Cratsenberg DM, Wierenga KJ.** Novel pathogenic variant in HNRNPK identified in a female with Au-Kline syndrome. National Society of Genetic Counselors 35th Annual Education Conference; 2016 Sep 28-Oct 1, Seattle, WA.

**Wadley AF, Hall M, Chen J, Fanning EA, Krumholz A, Wierenga KJ.** When negative turns positive: the experience of diagnostic exomes that were initially non-diagnostic. National Society of Genetic Counselors 35th Annual Education Conference; 2016 Sep 28-Oct 1, Seattle, WA.


**Samad D, Aston C, Jacobs N, Hassed SJ.** ASAP: Adult Screening Anticipatory Program; Is it Time for Screening of Adult-Onset Conditions? National Society of Genetic Counselors 35th Annual Education Conference; 2016 Sep 28-Oct 1, Seattle, WA.

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**Dr. John J. Mulvihill:** was invited to serve a two-year term on the Advisory Panel on Rare Diseases of the Patient-Centered Outcomes Research Institute (PCORI), an independent, non-profit organization authorized by Congress to fund research that will provide patients, their caregivers, and clinicians with the evidence-based information needed to make better-informed healthcare decisions.

**Dr. John J. Mulvihill:** served on the program committee and presented on the Round Table on Coalition Building at a Workshop on Unifying the Evaluation and Implementation of Genomic Medicine, sponsored by the Division of Genomic Medicine of the National Human Genome Research Institute, on August 18, 2016, in Bethesda, Maryland.

**Dr. John J. Mulvihill:** was invited to serve on the Pregnancy Working Group of PhenX (consensus measures for Phenotypes and eXposures), a project of the National Human Genome Research Institute, which is developing a catalog of recommended, standard measures of phenotypes and environmental exposures for use in biomedical research. [https://www.phenxtoolkit.org/](https://www.phenxtoolkit.org/)

**Dr. John J. Mulvihill:** presented on the Status of Training within the Undiagnosed Diseases Network at its Steering Committee meeting, in Washington, DC, July 26-27, 2016.

**Dr. John J. Mulvihill:** presented "Human Germ Cell Mutagenesis: A Clinical Update," at the annual meeting of the Environmental Mutagenesis and Genomics Society in Kansas City, MO on September 27, 2016.

**Dr. Susan Hassed:** was chosen to have images she created put on display in the Science-as-Art exhibit within the new Office of Research Administration office in University Research Park. The images are titled “Paired cells” and “Transition”.

**Shona Whitehead:** was recognized at the Sept 28, 2016 Academy of Teaching Scholars awards reception, for completing the inaugural Leadership for Program Administrators training curriculum, directed and facilitated by Dr. John Zubialde, College of Medicine, Senior Associate Dean.

**Dr. Sanjay Bidichandani:** served as an expert reviewer for the Brain Canada Hudson Translational Team Grant Competition.

**Dr. Sanjay Bidichandani:** served on the committee to review the Clinical Transitions and Reproductive System Courses in the College of Medicine.

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

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**In Memoriam**

Sharon Vaz, MS, RN and OUHSC MSGC Program alumnus (Class of 2005) passed away July 14, 2016. Born in India, raised in Kuwait and after coming to the US, Sharon worked as a nurse at Norman Regional Hospital. She was the Oklahoma State Genetics Coordinator and served as the Chief of Screening and Special Services at OSDH at the time of her untimely death. Sharon touched many lives through her fierce advocacy for newborn screening, genetics and public health. She will be missed by her friends, family and colleagues around the world.

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**Recent Publications:**