Bringing the Promise of Precision Medicine to All Americans

Minoli Perera, PharmD, PhD

Through her research, Minoli Perera, PharmD, PhD, associate professor of Pharmacology, works to bring pharmacogenomics to African-American populations. Pharmacogenomics, part of the precision medicine movement, involves using a patient's genetic information to predict drug response, such as whether the medication will be effective or if it might lead to adverse effects.

Her lab specifically focuses on anticoagulants, the genomics of drug metabolism and the pharmacogenomics of inflammatory bowel disease.

Perera, who joined the Feinberg faculty earlier this year, is the principal investigator on a $7.5-million grant from the National Institute on Minority Health and Health Disparities to study genetic variation and drug response within African-American patients.

Q&A

What are your research interests?
My lab focuses on the discovery and translation of pharmacogenomics findings in African-Americans. The field of pharmacogenomics, which is relatively young, looks to predict drug response in patients by using their genome (i.e. genomics biomarkers). Discovery and translation in this field has accelerated with President Obama's Precision Medicine Initiative. However, African-Americans remain under-represented in these studies.

Our lab has focused on medications used in thrombotic conditions because 1) these drugs are dangerous and must be used within a tight therapeutic window and 2) African-Americans suffer from thrombotic diseases at a higher rate than other populations.

What is the ultimate goal of your research?
The rapid progress in pharmacogenomics has meant that many academic hospitals around the country have begun incorporating genetic biomarkers into the medical record and using this information to guide therapy. However, most of these studies have been conducted in populations of European descent, meaning that many times these predictive genomics biomarkers are uninformative to other populations, such as African-Americans. Our ultimate goal is to bring the promise of precision medicine to all Americans.

How does your research advance medical science and knowledge?
Right now, there is an open question of what genotypes we should be using to translate findings from pharmacogenomics into clinical care. Our lab is dedicated to finding and translating those biomarkers that are relevant to patients of African descent. This is of paramount importance as we find that some of the now-incorporated genetic biomarkers lack predictive power in African-Americans. Practically, this means we are using the wrong genetic information in African-Americans to guide their therapy.

African-Americans carry many population-specific genetic mutations, which may predispose this population to both disease and altered drug response. However, these unique genetic mutations can only be discovered in studies that target African-Americans. By conducting these types of studies, our lab has discovered several unique and previously unknown genetic biomarkers of drug response that specifically influence African-Americans.

How is your research funded?
We have received funding and genotyping support from several federal and non-federal agencies. The ACCOuNT (African-American Cardiovascular Pharmacogenetic CONsorTium), as well as work we are doing to discover regulators of drug metabolism in African-Americans, have been funded by the NIH's National Institute on Minority Health and Heath Disparities.

I have previously received funding from the American Heart Association, and I have a postdoctoral fellow funded by the NIH's National Heart, Lung and Blood Institute. In collaboration with the NIH Pharmacogenomics Research Network and the RIKEN Center for Integrative Medical Sciences in Japan, I have received genotyping services that help to generate needed data in pharmacogenomics.

(continued on page 9)

Precision Medicine

(continued from page 3)

Where have you recently published papers?
Earlier this year, we published a paper that identified novel genetic variants associated with venous thromboembolism (VTE) risk in African-Americans. This paper was really important to medicine, not only in the new biology that it uncovered, but also in the impact it made on African-American genomics discovery science.

African-Americans suffer disproportionately from VTE, with a 30 to 60 percent higher incidence than other populations. There are clinically actionable, genomic tests available to assess the increased risk of VTE. But the genetic variants used in these tests are not present in the African-American population. This means that clinicians could order this genetic test, and it would come back as negative in African-Americans, even though they have a higher risk of the disease. By looking specifically at African-American genomes, we discovered a new genetic variant in the gene thrombomodulin, which increases the risk of VTE by over twofold. This is a common genetic mutation, with 37 percent of African-Americans carrying at least one copy.

The take-home message of this paper is that by specifically studying African-Americans we were able to identify an important genetic biomarker that predisposes individuals to clots. Since this gene has not been implicated in VTE previously, this is also a potential therapeutic target for treatment of VTE.

What do you enjoy about teaching/mentoring young scientists in the lab?
The work that we do is scientifically interesting and important, but it also carries a social justice mission. This speaks to many young scientists. I am proud that many of the people who join my lab are inspired by the opportunity to work toward equality in precision medicine. This is especially true for minority students and trainees. I hope this work will become an avenue to bring more diversity into academia and science research in general.

Follow Feinberg Social Media

Funding

2017 NYSCF Robertson Stem Cell Investigator Award

More information

Sponsor: The New York Stem Cell Foundation

Submission deadline: Feb. 22, 2017

Upper Amount: $1.5M over five years

Synopsis: NYSCF is soliciting applications from early career investigators for Innovator Awards to be used for exploring the basic biology and translational potential of stem cells. The goal of this initiative is to foster bold and innovative scientists with the potential to transform the field of stem cell research and advance understanding and use of stem cells in the development of treatments for human disease. In addition to providing funding, NYSCF partners with investigators to advance and translate their research.

Military Medical Photonics Program

More information

Sponsor: United States Department of Defense, Department of the Air Force and Air Force Office of Scientific Research

Submission deadline: Dec. 31, 2016

Upper Amount: $1M

Synopsis: This project seeks unclassified proposals for broad-based research and development aimed at using lasers and other light source technology to develop applications in medicine, photobiology, surgery and closely related materials sciences, with applications to combat casualty care and other military medical problems. The efforts proposed may be basic or applied research, and must have direct relevance to combat casualty care or other military medical priorities.

Limited Submission: Precision Medicine Initiative Cohort Program Participant Technologies Center

More information

Sponsor: United States Department of Health and Human Services, National Institutes of Health, National Heart, Lung, and Blood Institute

Submission deadline: Jan. 17, 2017 (letter of intent)

Upper Amount: $8M

Synopsis: The purpose of this funding announcement award is to provide support for a Participant Technologies Center for the Precision Medicine Initiative® Cohort Program. The goal of the program is to build a research cohort of one million or more U.S. volunteers who are engaged as partners in a longitudinal, long-term effort to transform the understanding of factors contributing to individual health and disease.

View more funding opportunities