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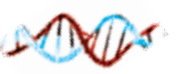


A New Tool for Primary Care

Nurses lead effort to evaluate, educate, and implement

SITTING IN A GENETIC COUNSELING session at Duke Primary Care's Pickett Road Clinic, Sharon Graffagnino received news she never thought she'd hear. She was at high risk for diabetes. It was news that caught her completely by surprise.

"It was really shocking for me to get such strong and bad genetic information," said Graffagnino, 51. "I always thought my numbers would be normal and in the middle."

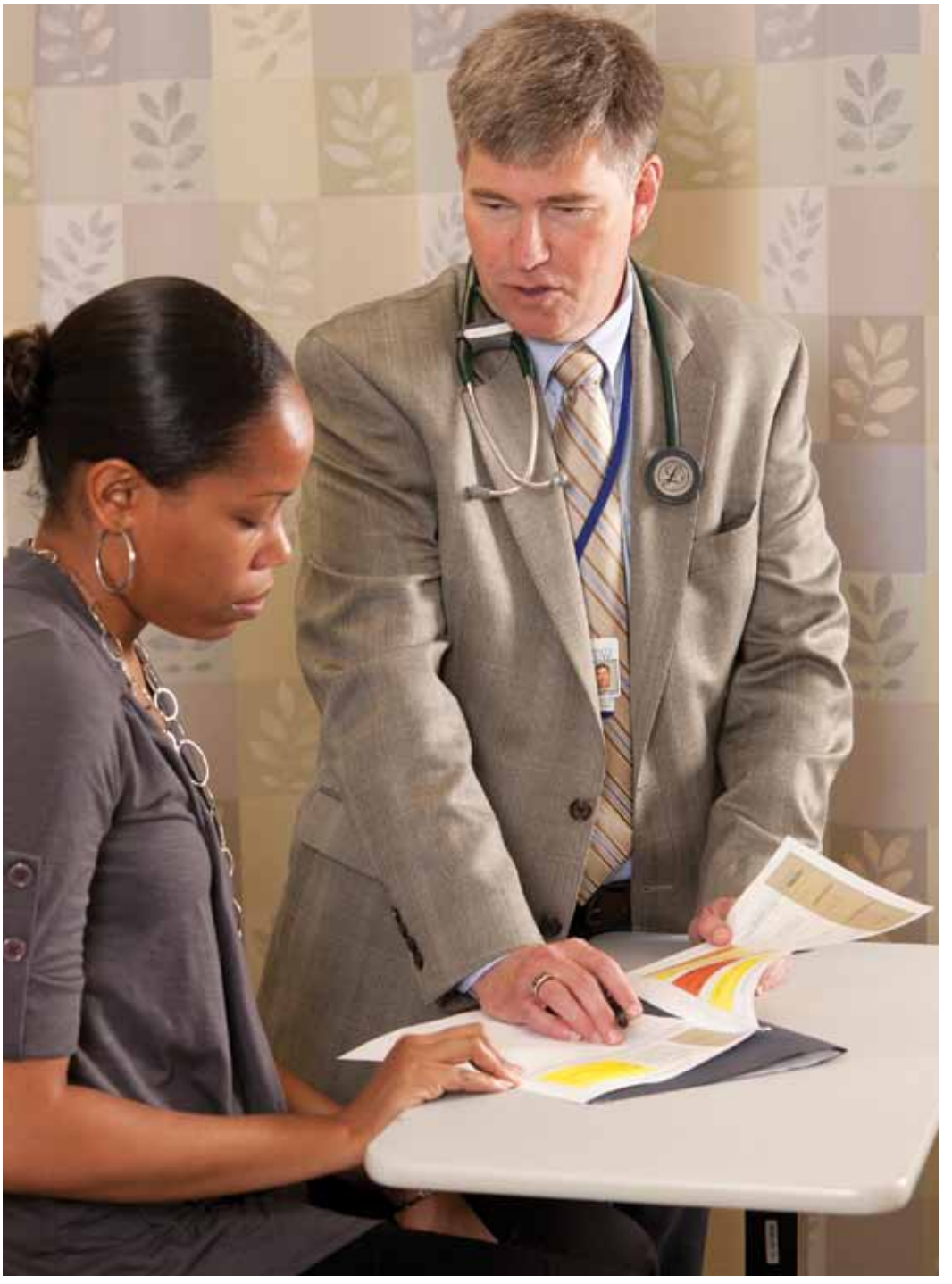
Graffagnino was familiar with diabetes and its serious health effects. Her brother weighs 400 pounds and takes insulin four times a day; her sister teeters on the border of full-blown diabetes; and her father battled against it until he passed away. But, as someone who always tried to eat well and exercise, she thought she was protected.

She learned she had a strong genetic risk for developing diabetes when she chose to participate in Effects of Type 2 Diabetes Genetic Risk Information on Health Behaviors and Outcome study through the Duke University School of Nursing. The result: Graffagnino now pays more attention to her food choices.

"I definitely hear the little voice in the back of my head if I'm overdoing it with sweets or starches or not getting enough exercise," she said. "I'm totally aware and take it seriously. That doesn't mean I always do really well, but I'm trying."

This study, which tests whether identifying and relaying genetic risk factors can positively change health behaviors, is part of a larger push within the Duke University Health System (DUHS) toward personalized medicine. This emerging health care model fuses a patient's genetic and genomic information

by Whitney L.J. Howell



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MICHAEL SCOTT

with family history to provide tailored care. Nursing faculty are leading an interdisciplinary team focused on initiating a paradigm shift in patient care.

Personalized Medicine and Nurses

Nurses have a longstanding reputation as being effective in identifying and managing illness and treatment-related symptoms. Knowledge of genetic factors that put some patients at higher risk for intense symptom experiences is rapidly developing. Nurse researchers are working on studies that help build this knowledge.

"If nurses are aware of genetic markers that make adults or children more or less susceptible to symptom distress, they are better prepared to make decisions and recommendations that will greatly benefit the patient," said associate professor Sharron Docherty, PhD, CPNP-AC/PC, RN. "Understanding individual patients' genetic risk for key symptomatology, such as cognitive dysfunction or fatigue, will allow them to more efficiently and accurately assess patients and intervene during treatment."

It's also critical to study the use of personalized genetic information to increase knowledge about the impact of whole genome sequencing results on patients and families.

"The role of nurses is to help broker information to the patient and help families understand the difficult concepts that come with this information," she said. "Nurses today and in the future must be able to understand whole genome sequencing, risk levels, and be able to help families interpret results."

Sharron Docherty consults with a family in the Neonatal Intensive Care Unit

Researching the Impact of Genetic Knowledge

Interest in how personalized medicine can touch clinical care sparked at Duke in 2010 with the creation of the Duke Center for Personalized Medicine. This group, led by genomics clinical director Geoffrey Ginsburg, MD, PhD, fuses resources and expertise from a broad array of players, including the Institute for Genome Sciences & Policy (IGSP), the schools of medicine and nursing, the Sanford School of Public Policy, and the Fuqua School of Business. Through these partnerships, faculty and students are searching for the best ways to bring personalized medicine to all aspects of health care.

The Center for Personalized Medicine (CPM) is also pursuing a policy agenda that will consider reimbursement, legal, and ethical issues related to genomic testing when assessing potential research projects, said assistant professor Allison Vorderstrasse, DNSc, APRN, CNE.

While these multidisciplinary investigations are complex and must be deftly designed, nurses are, in many ways, the keystones in these projects.

"Clinically, nurses tend to take a personalized, holistic approach with patients without even realizing it. They're all well trained to provide a lot of patient interaction and education," said Vorderstrasse, who serves on several personalized medicine working groups. "It's a goal, through CPM's educational, clinical and research initiatives, to ensure that nurses, as providers, are up-to-date on health care advances and that they can translate what it all means to patients."

But there's not much existing evidence to validate the proposed benefits of the individualized approach to health care. This flimsy body of research is why studies, such as the previously mentioned diabetes study, are critical, said assistant professor Michael Scott, DNP, FNP-BC. This work provides valuable information about how genetic factors can be used clinically.

"With this study, we wanted to explore the value of genetic testing as it pertains to the predisposition to chronic diseases, in this case Type 2 diabetes, and how feasible it is to do this type of testing in a primary care clinic," said Scott, who provided risk counseling to study participants. "We wanted to see if personal genomic information is likely to benefit patients through motivation of lifestyle behavioral change."

Scott's colleagues from the IGSP recruited participants from the Pickett Road Clinic and Pickens Family Medicine who had no reported history of diabetes and had never been genetically tested. Overall, 317 participants completed the full study.

To gather genetic material, researchers scraped the inside of each participant's cheek with a tongue blade, and a genetic testing company analyzed samples for certain genes associated with a risk for Type 2 diabetes. Investigators also collected information about other factors influencing diabetes risk, such as family history, blood sugar, body mass index, age, and ethnicity. All participants had the opportunity to meet with Scott for risk counseling, and those who were randomly selected for testing received counseling rooted in their individual genetic results.

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ALLISON VORDERSTRASSE

Based on study results, 26 percent of participants were pre-diabetic with blood sugar levels between 100 and 125; 68 percent were either overweight or obese; and 57 percent had a family history of diabetes. In addition, 92 percent returned for counseling sessions. Preliminary analysis is ongoing, but early results have shown both groups—those who were genetically tested and those who weren't—made minimal improvement in overall health status, especially with weight loss.

"We're thinking just the possibility of sitting down with me to talk about risk in the way a clinician doesn't have time to do may have made an impact. Weight loss in both groups doesn't mean genetic testing isn't necessary," Scott said. "These are just the early results, but whether the participant was genetically tested, it's clear simple involvement with a health care professional gets people motivated."

The Center for Personalized Medicine has other ongoing medicine studies, as well. Vorderstrasse, who sits on the center's leadership team charged with creating criteria that will determine which future studies win funding, is also a co-principal investigator on a pilot study. This study is currently recruiting patients and is led by co-principal investigators Ruth Wolever, PhD, of Duke Integrative Medicine and Alex Cho, MD. It is designed to determine whether genomic risk assessment can be effectively added to standard risk assessments for heart disease. The goal, she said, is to develop risk assessment tools and behavioral support interventions (health coaching) for implementation in primary care.

Educating Nurses to Play Their Role

Determining the efficacy of discussing genetic results with individual patients could drastically change the health care delivery model, and all providers must be ready to implement such a strategy. This spring, the School of Nursing introduced an interdisciplinary pilot elective class focused on genomic fundamentals and how they apply to personalized medicine.

Having a solid understanding of how genomics can impact patients' response to treatments and medications could greatly enhance the quality of care, said assistant professor Jennifer Dungan, PhD, RN.

"If nurses truly understand how genomics impacts health risk at the forefront and know how to evaluate it, they can be much better at approaching these topics with patients," said Dungan, who co-teaches the course with Sara Katsanis, MS, an associate in research in the Duke IGSP. "Rather than being reactive to patients, they can proactively reach out and determine if this information is important to patients, what it all means, and what to focus on."

During the first seven weeks of the seven-student class, the curriculum focused on fundamental genomic principles, such as human genetic variation, bioinformatics, ethical and social issues, and the principles of genomic testing. But it's the second half of the semester where students saw how genomic test results can directly impact health care.

In addition to hearing clinical and research experts discuss these topics, students had the option, but weren't required, to complete a free, anonymous 23andMe genetic testing panel. After receiving the results, they used skills they mastered during earlier weeks to evaluate their own genetic risk through the

Web site of the direct-to-consumer company that analyzed the tests. Students who didn't submit samples used data from mock patients.

This activity gave students the opportunity to experience what it would be like to learn about genetic risk from the patient's perspective. They discussed how to convey genetic results and how to put them into proper context. It's the knowledge of how and where to find additional information in existing literature, however, that is one of the class's most valuable lessons.

"I think getting the resources is one of the biggest take aways from the class. It's having the knowledge of where to turn," said Ann Miller, an accelerated BSN student in Dungan's class. "A lot of physicians and nurses don't know about this stuff. Being the only person or one of a few that know and understand is a huge asset."

Sara Katsanis and Jennifer Dungan teach a new interdisciplinary course on genomic fundamentals and personalized medicine.

Tailoring health services to the individual patient is the next phase of medical care, she said, and a nursing workforce well-educated in genetics and genomics will be better prepared to advocate for their patients' needs.

This type of class also introduces nurses and nursing students to more scientific aspects of health care, said Sarah Timberlake, an accelerated BSN student who has a microbiology and biomedical research background. The more exposure nursing students have to genetics and genomics in the classroom, she said, the greater opportunities they will have to influence how this knowledge is integrated into personalizing health care.

Educational efforts aren't solely focusing on students, however. According to Dungan, the School of Nursing is investigating ways to provide genetics and genomics education for practicing nurses, physical therapists, and other health professionals through short courses, workshops, or continuing education modules. The Center for Personalized Medicine leadership group is also discussing a similar educational model for physicians, including online modules and webinars.

Next Steps in Personalized Medicine

Although much has been done over the past two years, work still remains before personalized medicine can be effectively implemented across the Duke University Health System. In the coming year, Vorderstrasse said, working groups intend to establish an official agenda and priorities. The long-term goal is to develop partnerships both within and outside of Duke.

In addition, there are extensive curricular revisions underway that will thread genetic and genomic information through bachelor's, master's, and doctoral tracks in nursing, Dungan said. Weaving instruction through all programs would eliminate the need for an elective class and would turn nursing graduates into some of the most knowledgeable and effective providers in their communities, she said.

Nursing faculty and students view these efforts as necessary in order to provide the highest quality care possible, and support for the personalized medicine model continues to grow throughout the health system. But, most importantly, patients are beginning to grasp the importance of having health care providers who understand how genetics and genomics can directly impact individual health outcomes.

"When you look at American culture, having a doctor or nurse who can explain genetic risk is extremely important. We see so many people innocently unaware of the lifestyle choice they're actually making as they pull up for fast food, for example," Graffagnino said. "Most people don't know that their genetics are a big part of how your health will play out over the years. They don't understand how medically tied in everything is."