Pioneering statewide genomic screening in South Carolina

February 23 2024

Banner for In Our DNA SC, a statewide genomic screening program launched by the Medical University of South Carolina. Credit: Medical University of South Carolina

Physicians and scientists have known for many years that our genetics

not only shape our physical features like our smiles but also our risk for developing certain diseases.

The problem? Most people don't know if they have inherited these genetic risk factors for disease.

In Our DNA SC, a statewide genomic screening program launched by the Medical University of South Carolina (MUSC), aims to change that.

The program, the first and only program of its kind in the Southeast, is led by Caitlin G. Allen, Ph.D., a public health researcher and co-investigator, and Daniel P. Judge, M.D., a cardiovascular genetics specialist and principal investigator of In Our DNA SC.

"Population-wide genomic screening is an innovative approach to improving population health by allowing us to identify people who are at higher risk for developing certain diseases based on their genetics," said Allen.

Fourteen months after launching, the In Our DNA SC team has enrolled the first 20,000 participants of their overall 100,000-participant goal and published their findings to date in the *American Journal of Human Genetics.*

Their paper not only highlights the information provided to participants about their genetic risk factors but also shares what the researchers have learned about implementing a population-wide genomic screening program.

"It is essential that participants have as much information as possible about their genetics and their health," said Allen. "In addition, the knowledge we gain from implementing a population-wide genetic screening program could help the community and researchers within

South Carolina and beyond."

Across the U.S., dozens of other health systems are conducting population-wide genomic screening programs for diseases.

"The growth of this type of screening has been exponential," said Allen. "The field is shifting to gathering evidence about how to conduct these programs successfully and spread it across institutions so you're not reinventing the wheel every time a new program is established."

In Our DNA SC has embraced this shift, and its lessons learned from the first 20,000 participants can serve as a roadmap for future screening efforts.

Genetic risk for diseases

In Our DNA SC provides no-cost genetic screening for three conditions: hereditary breast and ovarian cancer, a hereditary colorectal cancer known as Lynch Syndrome and genetic high cholesterol or familial hypercholesterolemia.

Perhaps best-known of the mutations for which the program screens are the BRCA1 and BRCA2 gene variants, which can dramatically increase the risk of ovarian and breast cancer. Compared with a woman in the general population, those with a harmful, or pathogenic, BRCA1/2 variant have four to six times higher risk for breast cancer and 10 to 40 times higher risk for ovarian cancer. Similarly, mutations associated with the other two conditions also raise the risk of disease dramatically.

Screening for such mutations can be costly or difficult to access for those in underserved communities. Allen and Judge want In Our DNA SC to reach racial and ethnic minorities and rural residents who are often underscreened.

"We want to meet people where they are, ask them to participate in spaces with which they are familiar and really empower communities to understand the value of this type of research," said Allen.

Power for participants

The goal of routine screening efforts, such as lab tests, mammograms and colonoscopies, is to catch a problem early. For Allen, genetic screening could join the ranks of these routine screening tests.

"We could take people who may not have a known family history for a disease and provide them with genetic testing," she said.

Judge agrees that genetic screening could be a game changer for preventing disease.

"Knowledge is power when it comes to disease prevention," said Judge. "We can find the genetic problem before it manifests as cancer or heart disease and intervene."

Thanks to genetic screening by In Our DNA SC, 137 people thus far have been identified as having a variant of concern. Most of those identified agreed to be referred for genetic counseling and four out of five of those referred completed the counseling. During genetic counseling, participants learn more about the meaning of their results and discuss preventive measures.

Power for researchers

Allen and her team used implementation science, the study of how to put an intervention into practice, to develop and improve their program. Implementation science has become a vital tool for studying population

health, enabling researchers to improve health outcomes and equity, optimize use of resources and make important changes to screening methods.

"Implementation science helps us to ask questions about why something worked or did not work, then make informed decisions about how to improve," said Allen.

She is proud that it has been a centerpiece of the screening effort in South Carolina.

"In Our DNA SC is among the first population screening programs to integrate implementation science practices from the start so that it is infused throughout the whole program," she said.

Thanks to these practices, the team realized that some of their greatest successes occurred outside the clinic. For example, community events enrolled the most racially diverse participants while at-home collection resulted in the highest rate of samples returned.

"Even the most promising programs can fail to achieve their intended impact without effective implementation," said Allen. "By studying implementation of In Our DNA SC, we have been able to identify challenges and best practices in setting up large-scale population-based screening programs that we are excited to share with others."

More information: Caitlin G. Allen et al, Using implementation science to evaluate a population-wide genomic screening program: Findings from the first 20,000 In Our DNA SC participants, *The American Journal of Human Genetics* (2024). DOI: 10.1016/j.ajhg.2024.01.004