

Genetics Studies Have a Diversity Problem That Researchers Struggle To Fix

Close to 90% of genomics studies have been conducted using DNA from participants of European descent.

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When he recently walked into the dental clinic at the Medical University of South Carolina donning a bright-blue pullover with “In Our DNA SC” embroidered prominently on the front, Lee Moultrie said, two Black women stopped him to ask questions.

“It’s a walking billboard,” said Moultrie, a health care advocate who serves on the community advisory board for In Our DNA SC, a study underway at the university that aims to enroll 100,000 South Carolinians — including a representative percentage of Black people — in genetics research. The goal is to better understand how genes affect health risks such as cancer and heart disease.

Moultrie, who is Black and has participated in the research project himself, used the opportunity at the dental clinic to encourage the women to sign up and contribute their DNA. He keeps brochures about the study in his car and at the barbershop he visits weekly for this reason. It’s one way he wants to help solve a problem that has plagued the field of genetics research for decades: The data is based mostly on DNA from white people.

Project leaders in Charleston [told KFF Health News in 2022](#) that they hoped to enroll participants who reflect the demographic diversity of South Carolina, where just under 27% of residents identify as Black or African American. To date, though, they’ve failed to hit that mark. Only about 12% of the project’s participants who provided sociodemographic data identify as Black, while an additional 5% have identified as belonging to another racial minority group.

“We’d like to be a lot more diverse,” acknowledged Daniel Judge, principal investigator for the study and a cardiovascular genetics specialist at the Medical University of South Carolina.

Lack of diversity in genetics research has real health care implications. Since the completion more than 20 years ago of the Human Genome Project, which mapped most human genes for the first time, close to 90% of genomics studies have been conducted using DNA from participants of European descent, [research shows](#). And while human beings of all races and ancestries are more

than 99% genetically identical, even small differences in genes can spell big differences in health outcomes.

“Precision medicine” is a term used to describe how genetics can improve the way diseases are diagnosed and treated by considering a person’s DNA, environment, and lifestyle. But if this emerging field of health care is based on research involving mostly white people, “it could lead to mistakes, unknowingly,” said Misa Graff, an associate professor in epidemiology at the University of North Carolina and a genetics researcher.

In fact, that’s already happening. In 2016, for example, [research found](#) that some Black patients had been misdiagnosed with a potentially fatal heart condition because they’d tested positive for a genetic variant thought to be harmful. That variant is much more common among Black Americans than white Americans, the research found, and is considered likely harmless among Black people. Misclassifications can be avoided if “even modest numbers of people from diverse populations are included in sequence databases,” the authors wrote.

The genetics research project in Charleston requires participants to complete an online consent form and submit a saliva sample, either in person at a designated lab or collection event or by mail. They are not paid to participate, but they do receive a report outlining their DNA results. Those who test positive for a genetic marker linked to cancer or high cholesterol are offered a virtual appointment with a genetics counselor free of charge.

Some research projects require more time from their volunteers, which can skew the pool of participants, Graff said, because not everyone has the luxury of free time. “We need to be even more creative in how we obtain people to help contribute to studies,” she said.

Moultrie said he recently asked project leaders to reach out to African American media outlets throughout the Palmetto State to explain how the genetics research project works and to encourage Black people to participate. He also suggested that when researchers talk to Black community leaders, such as church pastors, they ought to persuade those leaders to enroll in the study instead of simply passing the message along to their congregations.

“We have new ideas. We have ways we can do this,” Moultrie said. “We’ll get there.”

Other ongoing efforts are already improving diversity in genetics research. At the National Institutes of Health, a program called “All of Us” aims to analyze the DNA of more than 1 million people across the country to build a diverse health database. So far, that program has enrolled more than 790,000 participants. Of these, more than 560,000 have provided DNA samples and about 45% identify as being part of a racial or ethnic minority group.

“Diversity is so important,” said Karriem Watson, chief engagement officer for the [All of Us research program](#). “When you think about groups that carry the greatest burden of disease, we know that those groups are often from minoritized populations.”

Diverse participation in All of Us hasn't come about by accident. NIH researchers strategically partnered with community health centers, faith-based groups, and Black fraternities and sororities to recruit people who have been historically underrepresented in biomedical research.

In South Carolina, for example, the NIH works with Cooperative Health, a network of federally qualified health centers near the state capital that serve many patients who are uninsured and Black, to recruit patients for All of Us. Eric Schlueter, chief medical officer of Cooperative Health, said the partnership works because their patients trust them.

"We have a strong history of being integrated into the community. Many of our employees grew up and still live in the same communities that we serve," Schlueter said. "That is what is part of our secret sauce."

So far, Cooperative Health has enrolled almost 3,000 people in the research program, about 70% of whom are Black.

"Our patients are just like other patients," Schlueter said. "They want to be able to provide an opportunity for their children and their children's children to have better health, and they realize this is an opportunity to do that."

Theoretically, researchers at the NIH and the Medical University of South Carolina may be trying to recruit some of the same people for their separate genetics studies, although nothing would prevent a patient from participating in both efforts.

The researchers in Charleston acknowledge they still have work to do. To date, In Our DNA SC has recruited about half of the 100,000 people it hopes for, and of those, about three-quarters have submitted DNA samples.

Caitlin Allen, a program investigator and a public health researcher at the medical university, acknowledged that some of the program's tactics haven't succeeded in recruiting many Black participants.

For example, some patients scheduled to see providers at the Medical University of South Carolina receive an electronic message through their patient portal before an appointment, which includes information about participating in the research project. But studies show that racial and ethnic minorities are less likely to engage with their electronic health records than white patients, Allen said.

"We see low uptake" with that strategy, she said, because many of the people researchers are trying to engage likely aren't receiving the message.

The study involves four research coordinators trained to take DNA samples, but there's a limit to how many people they can talk to face-to-face. "We're not necessarily able to go into every single room," Allen said.

That said, in-person community events seem to work well for enrolling diverse participants. In

March, In Our DNA SC research coordinators collected more than 30 DNA samples at a bicentennial event in Orangeburg, South Carolina, where more than 60% of residents identify as Black. Between the first and second year of the research project, Allen said, In Our DNA SC doubled the number of these community events that research coordinators attended.

“I would love to see it ramp up even more,” she said.

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