

Researchers home in on roots of Caribbean populations using new DNA analysis method

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Those of us who want to learn about our ancestors—who they were, where they came from and how they mingled (or didn't) with others around them—often turn to historical records or elderly family members for answers. But a new study by researchers at the Stanford University School of Medicine and the University of Miami Miller School of Medicine indicates that the answers can also be found within our own genes.

The researchers compared patterns of [genetic variation](#) found in populations in and around the Caribbean, which has had a particularly tumultuous past since Christopher Columbus stumbled into the Bahamas in 1492. Not only did they identify an influx of European genes into the native population that occurred within a generation of Columbus' arrival, but they also discovered two geographically distinct pulses of African immigration that correspond to the beginning and height of the transatlantic slave trade.

The study demonstrates how deciphering genetic echoes from the distant past can illuminate human history. But it also helps explain why some populations, like Latinos, who may be classified by medical researchers as a single group, display marked differences among populations in susceptibility to diseases or responses to therapeutic drugs.

"If we don't understand the origin of our genetic variants, we won't be able to design personalized, or even population-level, medicine," said Andres Moreno-Estrada, MD, PhD, a life sciences research associate at

Stanford. "Until recently, Latinos have been considered as a single group of people, when in fact they are very heterogeneous. We wanted to know what are the roots of the Caribbean people. Where do they come from? Clearly the population history of the region is very complex."

Moreno-Estrada is the lead author of the study, which will be published Nov. 14 in *PLOS Genetics*. Carlos Bustamante, PhD, professor of genetics at Stanford, shares senior authorship with Eden Martin, PhD, of the University of Miami.

"Until recently, researchers have tried to extract this type of information from ancient DNA, which can be very difficult to find and to analyze, and can't show the full range of Caribbean diversity," Moreno-Estrada said. "We wanted to approach the question from the other end—starting from the present day and going back in time."

The group, led by Bustamante and Martin, documented genetic variants found in 251 people of Caribbean descent—representing Cuba, Puerto Rico, Haiti, the Dominican Republic, Honduras and Colombia—living in South Florida, and 79 Venezuelans representing three native South American tribes. They then compared the genetic variants with those found in more than 3,000 Native Americans, Europeans and Africans.

"For us, this is a very important project," Bustamante said.

"Hispanic/Latinos, are the second-largest ethnic group in the United States, with people tracing their national origins to more than two dozen countries. Yet they are largely under-represented in medical genetic studies. An often-cited reason for this is that we do not know enough about genetic differences within and among groups to effectively design multi- and trans-population studies. Dr. Martin and I were very fortunate to receive NIH funding for the GOAL project [Genetic Origins and Admixture of Latinos], which develops novel medical and population genetic approaches that we hope will improve the design and, ultimately,

the outcome of medical genetic studies in this group."

To conduct the research, the team devised a new way of analyzing DNA to infer genetic ancestry at a fine geographic scale. Using this approach, they were able to estimate not just what proportion of each individual's genome was derived from each continent, but also to determine the closest ancestral group at a more-regional level.

"We gathered genome-wide data from these populations and employed various analytical strategies to identify segments that looked like those found in Native Americans, others which were more African in origin, and so on," Moreno-Estrada said. "We then looked within those segments to more precisely determine their geographic source within those larger groups."

The approach allowed the researchers to categorize regions of DNA as not just European, for example, but Iberian. Or not just African, but West African. They could also estimate when each mixing event occurred by assuming longer segments had been incorporated more recently than shorter segments. That's because, over time, our chromosomes randomly swap regions during cell division, breaking apart and mixing up formerly long, contiguous stretches of DNA. The more time that passes, the greater the likelihood that any one piece will be disrupted by this recombination process.

The research confirmed much of what is known about the history of the Caribbean islands. But it also answered some long-standing questions about the ancestry of native Caribbean people, the impact of European colonization, and the timing and geographic origins of forced African immigration.

The researchers found, for example, that the Caribbean was first populated by people from inland South America about 2,500 years ago.

Their DNA mirrors that of Amazonian tribes in the interior of the continent, and this flow of genes matches what is known about how language spread across the region during that time.

The European component, which was introduced 16 to 17 generations ago (or about 500 years ago—roughly when Columbus reached the islands) matches, but does not exactly mirror, the range of genetic diversity in modern-day Iberia. This finding most likely indicates that a small number of Europeans settled in the Caribbean and contributed their DNA to future islanders. It also confirms that, after the initial colonization of the Caribbean islands, future waves of immigration from Europe primarily came to the mainland.

Finally, African genetic diversity was first introduced to the Caribbean population about 15 generations ago (about 1550), when the first enslaved Africans were brought to the islands. The second pulse occurred five to seven generations ago, during the late 18th century, at the height of the transatlantic slave trade. The origin of the first pulse arose from the north coast of West Africa, whereas the second originated from central coast of West Africa.

"The transatlantic slave trade involved the brutal and forced migration of over 12 million people," Bustamante said. "They were the ancestors to and kin of many people who now live in the Americas, Africa and throughout the world today. We have tried to understand for years what role, if any, DNA can play in reconstructing these voyages. Realistically, we are just scratching the surface and seeing that we can find genetics signals that corroborate historical research. We are cautiously optimistic that as technology improves we can delve deeper and help reclaim more of this critically important history."

As exciting as it is to use genetics to answer age-old historical mysteries, it's the potential contribution of this knowledge to medicine that has

captured the researchers' interest.

"All this affects what we call a genetic-mapping strategy to identify disease variants specific to population subgroups," Moreno-Estrada said. "For example, those individuals with more European influence may be at increased risk for certain diseases because that genetic contribution was made by only a few individuals. Or, perhaps Caribbeans with more African ancestry may share an increased risk of diseases with others from West Africa. We're not yet at the point where we are able to say which populations are most likely to have specific diseases, but now we can begin to figure out the important components."

Provided by Stanford University Medical Center

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