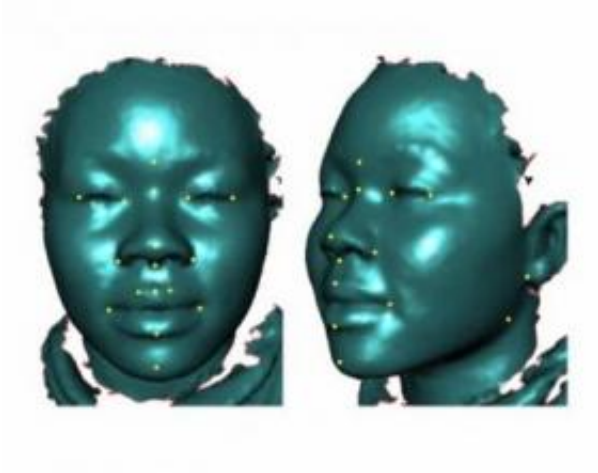
Mixed population provides insights into human genetic makeup

February 14 2009



These are three-dimensional images of a face with location points indicated. Credit: Mark Shriver, Penn State

Genetic diseases and genetically mixed populations can help researchers understand human diversity and human origins according to a Penn State physical anthropologist.

"We wanted to get to a strategy to predict what a face will look like," said Mark D. Shriver, associate professor of biological anthropology. "We want to understand the path of evolution that leads to that part of the selection process."

To pinpoint genes that influence the shape of the human face and head,

Shriver began with an online database of genes linked to disease -- Online Mendelian Inheritance of Man. If the symptoms of the disease involved the face or skull the gene implicated in the disease became a candidate for those facial traits.

This approach works because although Shriver looked at genes implicated in disease, those same genes in a healthy person may also influence the same physical trait -- length, width, shape, size -- but within the range normal for healthy individuals. Facial traits vary among humans, but do tend to group by population. For example, in general, West Africans have wider faces than Europeans and Europeans have longer faces than West Africans.

"There is a strong relationship between genetic ancestry and facial traits," said Shriver. "Using individuals of combined ancestry, European and African, we can see how the target genes alter facial traits," he told attendees at the 2009 Annual Meeting of the American Association for the Advancement of Science.

The researchers looked at a combined sample of African Americans with West African and European ancestry whose genetic makeup was known through DNA testing. To make it simpler, anyone with Native American ancestry was eliminated so that only two genetic pools were represented -- West African and European. The researchers reported on a sample of 254 individuals using three-dimensional imaging and measured the distances between specific portions of the face. Each individual had provided a DNA sample.

"We started with 22 landmarks on the faces that could be accurately located in all the images," said Shriver.

These landmarks might be the tip of the nose, the tip of the chin, the outer corner of the eye or other repeatable locations. They then recorded

the distances between all the points in all directions, so they had a distance map of each of the faces.

From their DNA profiles, Shriver could determine the admixture percentages of each individual, how much of their genetic make up came from each group. He could then compare the genetically determined admixture to the facial feature differences and determine the relative differences from the parental populations.

"This type of study, done on admixed populations shows that each person is a composite of their ancestors and that the range of facial features is a continuum," says Shriver.

Shriver found that there was a very strong statistical correlation between the amounts of admixture and the facial traits.

"We chose to look at African Americans because they were a large enough and available admixed population," said Shriver. "We are trying to solidify our understanding of the origins of humans and the evolutionary processes. Looking at admixed populations shows us the influence genes have and how they relate to physical features."

Source: Penn State