Researchers, studying DNA from Cajuns, identify characteristics unique to group

By JOE GYAN JR.
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METAIRIE — Louisianians of Cajun ancestry are unique for a variety of reasons: their dialect, culinary interests and tastes in music, to name a few.

But genetic researchers in suburban New Orleans have discovered, much to their surprise, that Louisiana Cajuns also are genetically unique.

Based on their preliminary test results, the scientists have concluded that Cajuns living in southern Louisiana are a unique, minority ethnic group.

"Definitely. The Cajun population — there is a genetic significance to that group," said Sudhir Sinha, president and scientific director of GenTest Laboratories Inc. in Metairie. "It is known that this is a unique group, but now for the first time we know they are genetically unique," said Sinha, an adjunct associate professor of biochemistry at Tulane Medical School in New Orleans.

Early in the 17th century, the Cajuns' pioneer ancestors founded a French colony called "Acadie" in what is now the Canadian province of Nova Scotia.

While the Acadians prospered on the fertile farmland, France and Britain vied for control of the region. Britain won sovereignty in 1713 and, four decades later, at the start of the French and Indian War, security-conscious officials deported many Acadians.

Scattered along Atlantic and Caribbean shores, some refugees found a final home in southern Louisiana. As their settlements spread across bayous and prairies, neighbors shortened the French "Acadien" to "Cadien" and then to "Cajun."

Joseph Neigel, an assistant biology professor at the University of Southwestern Louisiana in Lafayette, said heredity might explain why Cajuns have the unusually high and low genotype frequencies.

The Cajuns' pioneer ancestors who founded a French colony in what is today Nova Scotia was a relatively small settlement, he said, as was the number of Acadian refugees who settled in southern Louisiana.

If the early Cajuns possessed a unique genotype frequency, it is likely their descendants would share that quality, said Neigel, who teaches a genetics course at USL. "That's commonly observed in human populations that are founded by small settlements," he said.

Sinha noted that Louisiana Cajuns have maintained their cultural identity and historically have married within their group. Today, 22 southern Louisiana parishes with a Cajun flavor make up a triangular region known as Acadiana.

In October, the GenTest research team of Sinha, laboratory manager James Hochadel, and regulatory affairs and operations manager Anne Montgomery began collecting blood samples from 51 persons of Cajun ancestry living in New Iberia, Lafayette and Thibodaux.

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Researchers in California last year completed a genetic comparison of U.S. Caucasians and blacks, as well as Southeast Asians, Japanese, various Hispanic populations, Indonesians, Papua New Guineans, Australians, Bedouins and Nigerians.

Sinha said he and his colleagues wanted to compare the southern Louisiana Cajun population to the national Caucasian population.

"I was trying to find out how this national genotype compared with our local population," he explained.

The GenTest researchers avoided samples from close blood relations such as brother-sister, mother-child and father-child. Participants in the study signed consent forms granting permission for use of their blood in the study.

Sinha then extracted the DNA, or deoxyribonucleic acid, from each blood sample. DNA contains the entire genetic blueprint for an individual. No two people share the same DNA sequence, except identical twins.

Sinha and his team then subjected the 51 DNA samples to a revolutionary, high-tech analysis called PCR, or Polymerase Chain Reaction.

PCR gene amplification chemically isolates a specific segment of DNA from a biological sample, be it blood, bone, saliva, semen, tissue, or hair. The DNA segment is then amplified and tested for either the presence or absence of genetic "markers," or specific genes.

"I can make copies of that area — like a billion copies," Sinha said. "That gives you the ability to make copies of human gene and that's revolutionary."

GenTest, which uses PCR analysis of DNA samples primarily in forensic and paternity testing, is the South's only PCR-certified lab, Sinha said. Cetus Corp. of Emeryville, Calif., patented the PCR technology and certified GenTest to use the process.

GenTest, formed in January 1990, also is licensed by the U.S. Department of Health and Human Services as a nucleic acid analysis laboratory.

Once the PCR analysis of the Cajun DNA was completed earlier this year, Sinha compared his results to the U.S. Caucasian results from the California study.

Comparing six of the most common genetic markers, the GenTest team found that, while Cajuns are not surprisingly a sub-group of the U.S. Caucasian population, their genetic makeup is unique indeed.

Four of the six genotypes were found to be in "striking agreement with the general Caucasian population," Sinha said, but the frequency of the remaining two genetic markers in Cajuns was in stark contrast to that of Caucasians.

While the California researchers found that one of those genetic markers turned up in 4.6 percent of the 413 U.S. Caucasians they studied, the GenTest team discovered that 11.8 percent of the 51 Cajuns they sampled had the marker.

The second study, which will involve the 51 DNA samples to a revolutionary, high-tech analysis called PCR, or Polymerase Chain Reaction.

Sinha said the variation in the two genotype frequencies, or "distribution," is the key.

A further study is in progress to increase the sample size and to obtain statistically valid results," Sinha said. However, based on the results obtained to date, the Cajuns of Louisiana qualify as a separate ethnic group for the purpose of genotype frequencies.

"This is the first scientific genetic evidence that puts this Louisiana Cajun population apart," he said.

The second study, which will involve another 50 or 60 Louisianans of Cajun ancestry, should be completed by year's end, Montgomery said.

Sinha admitted the results of the initial Cajun study caught him by surprise.

"We found very interesting results. I expected no variation (in the genotype frequencies between the U.S. Caucasian population and the Louisiana Cajuns)," he said. "Usually it's variations based on race.

Sinha presented the surprising results of the Louisiana Cajun study to the Second International Symposium on Human Identification in Madison, Wis., on April 11.

"It was very well received," he said. "The work was well received in the scientific community."

Sinha said the preliminary test results could have a significant forensic application in that it will be highly improbable, for example, for a Louisiana Cajun, because of his or her genetic uniqueness, to be wrongly convicted of a crime if PCR analysis is utilized.

In addition, the study results could be used in the future in the diagnosis, correction or perhaps even prevention of genetic defects if doctors know that a specific genetic marker is responsible for a particular defect, he said.

"Four or five years from now, there will be a tremendous application" for the test results now being obtained, Sinha said.

Montgomery said the six genetic markers taken from the Cajun blood samples were extracted from white blood cells because mature red blood cells contain no DNA.

Neigel agreed with Sinha that the GenTest study results could have a significant impact in the area of genetic medicine.

"It is very valuable in that respect," he said.

Neigel said pinpointing specific gene markers that are inherited along with a genetic disease is "the first step to developing a cure."

The cystic fibrosis gene, for example, has been identified and researchers are now working to develop a cure for the congenital disease that usually develops during childhood and causes pancreatic insufficiency and pulmonary disorders, he said.

As to the question of whether personal characteristics can be linked to specific genes, Neigel said he has not taken a side on the "hotly debated" issue. Sinha said the GenTest study won't end the debate.

"I certainly agree culturally they're (Cajuns) a unique group," Neigel said, adding he would be a "little bit surprised" if Cajuns could be defined as genetically unusual.

"It's more a question of degree rather than kind," Neigel said. "I think everybody would agree their heritage is unique."

Neigel cautioned that one should not infer from genetic studies that Cajuns' genes are "inferior or bad."

"The genes in Cajuns are not worse than others," he said.

Because a particular human population shares unique genotype frequencies, Neigel said, the chances of two members of that population carrying the same genetic disease is higher than carrying a different disease.

"There's already some genetic diseases that are known to be of high frequency in Cajuns in this area," he said.

Sinha said GenTest targeted Lafayette, New Iberia and Thibodaux for its sample group because that is where the largest numbers of Louisiana Cajuns are living. If an individual could trace his Cajun ancestry to a grandparent, he said, the person was accepted into the sample population.

The second study, which will involve the same cities, is being conducted to validate the findings to date and add more statistical data, Sinha said.

In addition to helping settle paternity disputes and match bone marrow donors with transplant patients, DNA analyses have spread to other applications, including forensic uses for criminal and civil justice casework.

First introduced into a U.S. criminal proceeding in 1986 in Pennsylvania, forensic DNA analysis has since been admitted into evidence in at least 156 cases by 26 states and the U.S. military as of Jan. 1, 1990, according to the Office of Technology Assessment, an analytical arm of the U.S. Congress.

"This number does not reflect its even wider use in investigations that did not go to trial," a July 1990 OTA report entitled "Genetic Witness: Forensic Uses of DNA Tests" stated.

OTA estimates that, as of July 1990, DNA tests have been used by law enforcement in more than 2,000 investigations. OTA also found that DNA tests were used for criminal investigations and proceedings in 49 states and the District of Columbia.

"These numbers do not reflect the use of DNA tests in thousands of paternity disputes annually," the report said.

OTA concluded in the report that DNA tests are "sensitive and accurate."

"The scientific basis for DNA typing is sound, and OTA finds that forensic uses of DNA tests are valid," the report stated. "Questions raised about the validity of DNA typing do courts and public a disservice."

Sinha said DNA technology helps make better use of evidence found by police.

"Blood stains are used to get a blood type, which is very general, but now the blood cells can be used to get a DNA type, which is more specific," he said.

Louisiana hasn't taken a PCR case to trial yet, Montgomery said, but GenTest is involved in 25 pending cases in the state.

"It's really an exciting time — being on the forefront of technology," she said.

Montgomery said the U.S. government utilized PCR analysis to positively identify the remains of some soldiers killed during the Persian Gulf war.