Deaf-blind genetic syndrome stalks Cajun country

By REBECCA ROLWING
Associated Press writer

NEW ORLEANS — Already deaf, Danny Delcambre was 14 years old when he learned he also was going blind.

He was relieved.

Finally, he knew what was wrong. He had Usher syndrome, a genetic disease that generally causes deafness from birth, a loss of balance and a gradual decline of vision beginning in adolescence, the teen years.

He is one of 800 Louisianians with the syndrome — the largest population of deaf-blind people in any state.

Researchers have linked it to the close-knit Cajun world of southwestern Louisiana, where the genetic defect has been passed down through the generations after long-distance intermarriage.

If one person carrying the gene has a child with another carrier, the disease can surface in the child.

"In general they have no idea that they are related. It's only when they have a child with a recessive disease that they will look into it and find that they have a number of ancestors in common," said Bronya Keats, a geneticist at the LSU Medical Center who has identified the gene that causes Usher.

The defect is most prevalent in the Cajun region, renowned for its spicy food, Zydeco music and French heritage. The Cajuns are descendants of Catholics who were expelled from Nova Scotia by British Protestants in the mid-1700s, an exile made famous in Henry Wadsworth Longfellow's epic poem "Evangeline."

Mary Kay Pelias, an LSU genetics professor who has researched Usher for 17 years, counsels possible carriers about the risks.

"Marrying outside the community is a fairly reliable way to avoid passing the disease on to children, she said. But many won't.

"I think it's fair to say most (of the Cajuns) will stay in the community," Pelias said. "Among the families that we have talked to, many regard far away as 20 miles away."

Many of Louisiana's deaf-blind have achieved relatively independent lives. Delcambre, now 38 and living in Seattle, secured a $50,000 loan to open his own Cajun-style cafe there in 1993. Now he delivers motivational speeches around the country. Topics relate to his experiences overcoming obstacles and coping with change.

While Delcambre's achievements have earned him a celebrity-like status among those with Usher, others have mastered tasks that usually rely on eyesight and hearing.

"I love sewing," said Della Mae Childress, a 70-year-old Cajun with Usher. She explains that she uses her hands to replace her eyes and ears.

She also scrubs the house, cooks and applies her own makeup — even though she doesn't remember what her face looks like. It usually goes fairly well except when she mistakes the eye shadow for blush.

But crossing the street unassisted can be life-threatening for those with Usher, said Kristi Mora, deaf-blind program manager at Resources for Independent Living in New Orleans. She said a few have been struck by cars.

Many of the afflicted communicate with tactile sign language, in which the deaf-blind cups his hands over the hands of a person using sign language.

Technology also helps. Childress chats via e-mail with her friends and children.

A special board at the bottom of the keyboard translates the text into Braille. Another device translates phone conversations into Braille.

Gadgets are available that, like a pager, vibrate when there is a phone call, someone at the door or when the house is on fire.

Still, the deaf-blind often face fear and depression, said Ilene Miner, a social worker who specializes in working with people with Usher. "They're ultimately afraid of being abandoned — facing being totally deaf, blind and left in a community that doesn't provide for that."

That's something Childress struggled with.

"I cried a lot. I cried for about six years," she said.

By teaching herself Braille, taking computer classes and memorizing her house, Childress said, she regained her independence and happiness.

Aside from tripping over things, Delcambre says his largest frustration lies with people's attitudes:

"A lot of people don't realize that we're just like everybody else, and we can succeed."
Deaf

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"A year ago I was saying that within a year we might find the gene, but we don't have it yet," said Bronya Keats, an LSU Medical Center geneticist. "It's a very tricky one to find."

Keats is an expert on Usher syndrome, and studies it under a grant from the Foundation Fighting Blindness.

LSU is involved because Usher syndrome is common in Louisianians of Acadian or Cajun descent. Researchers believe one in 70 Cajuns is a carrier of the gene.

"We're trying to find this gene that causes the more severe form in the Acadian population," Keats said.

They have narrowed their search to a small area of genetic material, but have not yet identified the specific Usher syndrome gene.

The work is tedious.

"It's a matter of working through the DNA in the region and looking for a mutation that might be responsible, then checking it out and ensuring it's only in (Usher syndrome) patients," Keats said.

"Once we identify it, we have no idea what kind of protein will be encoded by this gene, so then we need to characterize the protein and see what it's doing," Keats said. "This is likely to be a many more years' research."

Researchers hope that eventually a simple blood test will identify the Usher syndrome gene, and genetic therapies may be developed to control its effects.

Preventing deafness will be extremely difficult, Keats said, but researchers may one day figure out a way to reduce the visual impairment Usher syndrome causes.

Lucy is how educators at Louisiana School for the Deaf first found Usher syndrome among their students in 1977.

Some of the students persisted in describing in sign language — with a symbol they invented — that some of them suffered from tunnel vision. This lack of peripheral vision is a common first phase of blindness in many who suffer from Usher syndrome.

Michelle Eaton, a 1998 graduate of Louisiana School for the Deaf, talks with LSD graduate Jose Perez over lunch at Mulatte's last week. Eaton, who attends community college in New Orleans, spent time with a group of deaf students from Sweden.

Advocate staff photo by Travis Spradling

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Michelle Eaton, a 1998 graduate of Louisiana School for the Deaf, talks with LSD graduate Jose Perez over lunch at Mulatte's last week. Eaton, who attends community college in New Orleans, spent time with a group of deaf students from Sweden.

"None of us knew but it had been going on all these years — people knew they would eventually go blind but they didn't know why," Melancon said.

"We felt so helpless. As a counselor working with the kids I had to explain to them life is going to be rough, and you won't be able to see, able to drive," Melancon said.

"You had to tell them terrible things about their lives — try not to marry someone too close to you."

After a schoolwide screening, they learned that one in 10 suffered from Usher. For the first time, educators knew what was causing the deaf students' blindness.

Since the discovery of Usher syndrome at the Baton Rouge school 22 years ago, educators have been helping the students and their families who carry the gene. A special support group has met since then, helping the students cope with their complex of challenges in daily life.

The 10 percent of the school's students who have Usher today meet regularly. A few years ago, one class dubbed themselves Dice 12 describing the dozen who felt lucky to get out of regular classes for their own gatherings. Later classes have named themselves Lucky 13 and the Bravehearts.

The school also counsels families of those carrying the genes. The incidence of Usher has dropped somewhat, but it is still a threat.

Last week was a rare treat for the Swedish visitors and the Usher students at the School for the Deaf. Between the Cajun food and cultural visits, they hardly spoke about the ugly truth that links them — that eventually they will all probably go blind. Usher syndrome sufferers usually lose their sight by middle age.

Conny Lindfors knows several languages but he has never heard a word in any of them.

He is a 20-year-old Swedish student studying humanities in that country's only high school for the deaf.

"Before I came here, I thought the school here and life for deaf students would not be as good as in Sweden," Lindfors said through interpreters. "Now I have changed my mind."

Thanks to interpreters and translators, the Swedish signing students got along well with their U.S.-signing friends at the Louisiana school. Lindfors made new friends and polished up his American sign language, one of several he is learning.

His sister, Helen Davidsson, especially liked the kitchen at the Louisiana School for the Deaf, and its custom tools and utensils for deaf and blind students. She plans to pursue a career in catering and management.

Maria Nyman of Finland spent a morning at the State Fair looking after a first-grader from Louisiana School for the Deaf. Nyman has a knack for caring for young children, and so the 20-year-old plans to be a first-grade teacher when she finishes college.

The students rarely dwell on or worry about the fact they have Usher.

"I don't think about it today," said Nyman.

Lindfors agreed.

"I don't worry about the future, I worry about what I can do now," Lindfors said.

Perhaps the most important part of the visit was simply seeing the United States.

"When they lose their sight they'll still be able to picture it — it will give them memories," said Catharina Bjur, an adviser to the Swedish students. "It puts a memory in their backpack."