Cameron Heck (left) doesn’t have Usher syndrome but is a carrier for the disease. Mom Darcie Heck says cochlear implants have helped her son Blake (right), who has Usher syndrome, perform above average in a mainstream first-grade class.
The Sound of Hope

New research helps the fight against a rare disease that afflicts descendants of the original Acadians.

BY DAVID WINKLER-SCHMIT

Even on an overcast muggy January day, Cory and Darci Heck's home in rural Albany, La., is paradise for the Hecks' sons, Blake and Cameron. The house that sits in the middle of the property is a large one-story Acadian ranch where each boy has his own room filled with toys. Outside a rambunctious dark brown and white border collie, Belle, patrols the grounds and eagerly waits to fetch tennis balls. Six-year-old Blake and his 5-year-old little brother Cameron have the woods on the back of the land to explore. The family recently dug a pond and plans to stock it with fish. As long as they're wearing helmets, the boys can sometimes motort around the Heck's 5 acres on their four-wheelers.

A fishing hole, mini-bikes and a loyal dog with boundless energy. Blake Heck is living a dream, but it didn't begin that way. It started as a quiet nightmare.

"He fainted the test. He can't hear." A nurse stood by Darcie Heck's hospital bed and matter-of-factly reported that 1-day-old Blake hadn't responded to the test's auditory cues. Darcie, who had gone through more than 20 hours of labor delivering Blake, tried to get out of bed to confront the nurse but could only manage to tell the nurse to get out. The test was repeated, but the result was the same. Blake was deaf.

At first, Darcie searched for answers, but not so much because she needed an explanation. She wanted someone to blame. The doctor that delivered Blake was old, Darcie believed he must have waited too long to remove Blake from the womb, depriving him of oxygen and making him lose his hearing. "I knew that baby should have come out sooner," Darcie's mother said in agreement. Surely it was the doctor's fault.

Decisions had to be made. How would the Hecks raise Blake? Should he learn sign language and become a member of the deaf community, which views deafness not as a handicap and sign language as an alternative way of communicating? Another option was pursuing an auditory and verbal path where the child learns how to lip-read, is fitted with hearing aids and is taught how to speak.

The Hecks decided on auditory and verbal instruction. At the age of three weeks, Blake was wearing his first hearing aids, and soon he was enrolled in the Chinchuba Institute in Marrero, which bases its curriculum on teaching deaf children speech and language skills. At Chinchuba, a therapist taught Blake how to express his emotions; she taught him how to cry.

Darcie Heck cried everyday. Blake's hearing aids didn't work and likely never would, since Blake was severely deaf. It would be a long and hard road for Blake to ever really speak. During the day while Blake was napping in his crib and Cory was away at work, Darcie sometimes felt the need to disturb the silence of her house.

"I would be so angry that I would cling to sides and pains," Heck says. "Blake would sleep right through it."

When Carl Heck Jr., Blake's paternal grandfather, found out about Blake's deafness, he remembered Kevin Talbot, his cousin. Talbot had two daughters who were deaf — the cause a genetic disease called Usher syndrome. Heck visited Talbot and found out his daughters probably weren't the first in the Talbot family line to have Usher syndrome. Although they weren't officially diagnosed, there were two long-deceased men from the Talbot family who were deaf. They were also blind.

Usher syndrome is a rare disease in which children are born deaf and eventually become blind because of retina deterioration. Usher syndrome patients usually begin to lose their sight during adolescence, progressing from night blindness to loss of peripheral vision to complete blindness.

A link between Usher syndrome and the Acadians of Louisiana was first established more than 40 years ago by Dr. H.W. Kleefer. That allowed scientists to narrow down the disease's cause; records from the Catholic Church demonstrated that many Cajuns were unknowingly intermarried, sometimes from several family lines, with couples often being third, fourth or fifth cousins. Carl Heck's mother was Verna Mae Talbot Heck, a descendant of one of the original Acadian families in Louisiana. When he found out that there was going to be a symposium on genetic diseases and the Acadian population at Nicholls State University in Thibodaux, Heck convinced his son Cory that they should attend.

The symposium was led by Dr. Briony Keats, head of the department of genetics at Louisiana State University's Health Sciences Center in New Orleans. Keats is also director of the Molecular and Humur Genetics Center of Excellence, part of the Center for Acadiana Genetics and Hereditary Health Care. Carl and Cory listened as Keats explained why Acadians have a higher probability of genetic disease and how the Acadian population plays a significant role in genetic research.

It's a matter of a relatively small genetic pool growing into a fairly large number of people," Keats explains in a recent interview, also citing probability, gene muta-

ations, and the geographical and cultural isolation of the Acadians. As a result, Acadians became more likely to be born with certain genetic diseases including Tay-Sachs disease, Friedrich's ataxia, and Usher syndrome.

The family names of the original Acadians are familiar surrines in southern Louisiana: Doucet, Carrier, Richard, Labreche, and many others. Since the Acadians were living in the frontier in Nova Scotia and isolated...
from others, marriages were almost exclusively between the offspring of the first 300 pioneers. Keats says the population grew from 300 to about 18,000 by 1765. Keats believes the mutation that causes Usher syndrome likely occurred in the DNA of one of these ancestral Acadians.

From 1765-1785, more than 3,000 people from that population of 18,000 who had lived in Nova Scotia migrated to Louisiana under Lie Grand Département. This reduced population—settling mostly in 22 parishes including Lafayette, Avoyelles, St. Martin, Vermilion, Cameron, and Assumption—kept to themselves and their indigenous culture, with very few marriages outside of their community. Since Usher's syndrome is caused by two recessive genes, the likelihood of both parents having a copy of that recessive gene increases in a smaller gene pool like that of the Acadians.

The Talbots, Blake Heck's ancestors, moved to Assumption Parish shortly after 1765.

Following the symptom, Carl and Cary Heck lobbied Darcie Heck to test Blake for Usher syndrome. The family came to Bronya Keats' office when Blake was 6 months old. A positive test result would mean that Blake would someday lose his sight, but it would also provide the couple with the answer for why Blake was deaf. Plus, the test would let Darcie Heck, who was already pregnant with Cameron by this time, know what the chances were that she would have another child with Usher syndrome.

"When I first started speaking to them, I asked them about ancestry," Keats recalls. "Cary said he was Acadian, but Darcie said no, so I thought it might just be deafness. But there were cousins on the Acadian side that had it, so I felt we should check her off chance Darcie carried the gene. Sure enough when we sequenced Darcie's genes, it turned out she was carrying a form of the mutated gene."

It was confirmed Blake had Usher syndrome. Later, they found out that Cameron doesn't have Usher syndrome but is a carrier for it. Since two recessive genes cause Usher, there is a 25 percent chance that a couple carrying these recessive genes will have offspring with Usher.

Darcie's anger dissipated when she realized Blake's deafness was nobody's fault. The Hecks became determined that their son's chances for a normal life would be as good as any other child.

The next step was cochlear implants — tiny electronic devices that can partially restore hearing for the severely deaf. Having the surgery doesn't guarantee the patient will benefit from the implants. Pre- and post-surgery counseling is vital so that patients can learn to interpret the sounds coming from the devices. The younger the patient, the more likely he is to become accustomed to the implants, which bypass damaged cells and convert sounds into electrical signals that stimulate the hearing nerve. A surgeon would implant the tiny electronic device under the skin behind Blake's ear, and post-surgery he would wear a headpiece microphone to capture incoming sounds.

The implants immediately made a difference. Blake's hearing isn't like an average person's — it's actually amplified louder and sounds such as an air conditioner kicking on or the busy noise of a classroom can distract him. But he does hear and now talks so well that he only goes to speech therapy for a monthly checkup.

The early diagnosis and the cochlear implants made a significant difference. Blake attends a mainstream first grade class and is performing above average.

Linda Hebert's parents thought she was mentally handicapped. She is sitting at a long folding table in a large hall, which serves as a meeting place and a makeshift church, in the Deaf Action Center in Lafayette. Linda is signing to her daughter, Lynn Leblanc, recalling the day she asked her parents if they could have a baby, so Linda, an only child, would have a sibling.

"They said, 'No. One is enough.' They thought they might have another mentally retarded child," Linda recalls. Sixty-four-year-old Linda Hebert still believes she is mentally slow, but she certainly doesn't appear that way. Considering that she received a minimal education and wasn't taught how to communicate until she was 12 years old, Linda, who has Usher syndrome, has accomplished a great deal. She has raised four daughters, worked much of her adult life and smiles and laughs most of the time.

The center is a wooden prefabricated building in the Village De Lac, an apartment complex for the disabled that is owned and operated by the Catholic Diocese of Lafayette. Myron Mouton, the center's director, says the Deaf Action Center serves about 400-500 people with Usher syndrome and other deaf people throughout a seven-parish area. Mouton's worked at the facility for 30 years, and says the majority of those with Usher that use the center are elderly. Joining Linda on this day are about 10 others with the disease; she likes to come here to visit with old friends.

Says Mouton, "When I first got involved with the deaf and the deaf/blind population, people weren't out there as much as they are today."

After her birth in Abbeville, Linda moved with her Acadian parents Gladston and Beulah Hebert to her father's Air Force station in New Mexico. It was there that the Heberts realized their daughter couldn't hear. They tried to get Linda treatment, which amounted to doctors painfully pressing on her ears in order to somehow manipulate them into functioning.

When the treatments proved fruitless, Linda was enrolled in a school for the deaf. Not much of a school, it was more like a prison, she says. The 3-year-old Hebert wasn't really taught anything and spent her days doing little except occasionally being allowed to draw.

When Linda turned 8, her father was transferred to a base in England. She couldn't speak or read but for some reason was enrolled in a speaking school. The only way she could communicate was by elementary hand gestures, which weren't welcomed by the school's staff.

"I was pointing and grunting," Linda says. "The teachers would slap my hands down."

It wasn't until her family moved back to Abbeville when she was 12 that Linda finally learned sign language. She was placed in Baton Rouge's Louisiana School for the Deaf, where she was taught how to sign and read, played sports, and received some vocational training. She graduated from the school in 1961 with what Leblanc describes as the equivalent of an eighth grade education.

Within a year in 1962, she was married to a fellow deaf student, Joseph Lennar Griffith, and pregnant. Griffith's deafness was not caused by Usher syndrome. The marriage lasted 14 years, and the couple produced four daughters, none of whom are deaf. The family continues to live on the Hebert's 20-acre plot that runs along the Vermilion River in Abbeville, with Linda's daughters raising their own families in houses or trailers on the property.

Linda wasn't diagnosed with Usher syndrome until she was 41 years old. Realizing that she would lose her sight, she learned how to sign. Even with her sight failing, she continued to perform physically demanding jobs including dish washing, working in a canning factory, and even farming. Until recently, Linda has been peeling shrimp and cleaning pots and pans at a local bakery two days a week but lost the job after the bakery bought a dishwashing machine. She receives a small monthly allowance from Social Security Disability, and her children and grandchildren help her out. She knows her land very well and visits her extended family daily by using her cane.

Still, Linda gets agitated and visibly frustrated when she's asked about her new daily routine.

"I'm bored, bored," she admits.

Educational outreach is a critical component toward Acadians understanding their genetic heritage. The federally funded Center for Acadiana Genetics and...
Hereditary Health Care performs these outreach services by conducting symposiums like the one at Nicholls State that Carl and Corey Heck attended. Dr. Keats stresses that the point of these events isn’t to scare people.

"It’s focused on disease, but that will come out," Keats explains. "Our biggest goal is to help them understand genetics.

Even with a restricted gene pool, genetic disease within the Cajun population is still rare. About 1 in 70 Acadians carry the recessive gene that causes Usher. For the rest of the United States population, it is approximately 1 in 100,000. Therefore, the person has to couple with another carrier, again very unlikely. One in 20,000 Acadian children are born with this syndrome compared with 1 in 50,000 nationally. Keats reminds people that Cajuns and similarly small populations like them aren’t the only ones with defective genes.

"We all have these," Keats says. "We all have at least a dozen or more of these bad genes, but they’re paired with normal genes, so there isn’t a problem."

The reduced genetic pool within the Cajun population has also led researchers to better understand genetic disease and locate the specific gene and mutation that leads to the disease.

The blood samples from the Cajun Usher patients first identified by Dr. H.W. Kleefer in 1966 were collected and kept at Coriell Institute for Medical Research in New Jersey, a repository for genetic diseases. Instead of trying to analyze blood samples from Usher patients from around the world, which Keats says "will lead you nowhere," researchers could focus only on Cajun Usher patients.

Researchers from around the world could order copies of these samples for study.

By 2000, two studies precisely identified the gene that causes Usher syndrome — USH1C — in Cajuns. Interestingly, Keats was one of the researchers involved in the discovery, but her research paper was about a Lebanese family, which had members with Usher that was caused by the same gene, USH1C. A group of researchers in London using the Acadian samples was also able to identify the USH1C gene. Although it is the same defective gene, the Lebanese family had a different mutation on USH1C than the Acadian sample. Researchers to date have confirmed that eight different genes can cause Usher syndrome, but for Acadians with Usher there is only one possible gene, USH1C.

By identifying the gene, researchers like Keats could start hypothesizing how to treat the disease. In Usher syndrome, blindness slowly occurs through retinal degeneration, because the defective gene doesn’t produce harmonin, a protein that helps maintain the retina.

"Our goal," Keats states, "is to prevent or slow down the retinal degeneration in these kids."

Keats thinks the best approach is cellular therapy. If a cell with a good copy of the gene could be introduced into the patient’s retina, it could stimulate harmonin production. If the therapy works, it would be beneficial to any Usher sufferer, not just Cajun Usher patients.

In order to test out cellular therapy, researchers have to replicate the disease in lab animals. For the past three years, Keats

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Linda Hebert recently began to lose her sight as a result of Usher syndrome.

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Blake Heke is like many 6-year-old boys. When he asks him what he likes to do in school, Blake doesn’t mention reading or mathematics.

"I like playing with Play Dough, playing at recess and watching movies," Blake says.

Darec Heck just shrugs. "He’s normal."

Blake knows that someday he could begin to lose his sight. His mom says he becomes anxious about it now and then, but like his father, he’s even keeled and rarely displays too much emotion. As he gets older, he will realize how far he has already come, how his life is connected to an old woman living along the Vermilion River, and how his future might hold a cure for Usher syndrome.

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