Linda Hebert Will Not Give Up

By Judy LaBorde

The sun has not yet risen as Linda Hebert sits in her dark and quiet home. She’s dressed and ready for her daughter to pick her up. Today is Monday and that means Linda is going to work.

As soon as she senses the heat from her daughter’s car, she walks out the front door and taps, taps her cane along the gravel path leading to the driveway. Mother and daughter will now travel the 17 miles from their homes in Abbeville, Louisiana, to Southside Bakery in Lafayette, the place where Linda has a job.

There, for the next ten hours she will clean mounds of shrimp, sometimes as many as 50 pounds, that the cooks at Southside Bakery will use to make lunches. Linda will also scald and dry the huge pots and pans used for mixing the cake dough. If there’s enough time, she will slice the tomatoes.

She will do these chores using only her sense of touch — no sight, no hearing. She will work hard and not complain. Linda is not the type to complain — not when she lost her hearing as a child, not when she lost her eyesight as a young adult, and certainly not today, Monday, the one day of the week when she is paid to work.

If there’s one thing Linda loves to do, it’s to work. She has graduated from just about every job training program offered in Lafayette.

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Blake Heck Gets Ready for Take-Off

By Judy LaBorde

This story is about seven-year old Blake Heck who, like Linda Hebert, has Usher syndrome. It is also about the ongoing struggle of his family in making the tough decisions that are fundamentally shaping the future of their young son.

Like Linda Hebert, Blake was born in south Louisiana and his family is of Acadian ancestry.

Also like the family of Linda Hebert, the Heck family has learned to cope on a daily basis with Usher syndrome and the effects it’s had on every member of the family. Every aspect of daily living, including speech, hearing, language, education, and health, is a constant stream of decision making. No matter how hard they try to make the right decisions, Blake’s parents know what they choose will have elements of good and bad.

What the Blake Family didn’t know, what they could not have anticipated, were the dramatic ups and downs their lives would take, starting with the day Blake was born.

continued on page 6
Linda Hebert

For months, she worked in the laundry room of a nursing home in Lafayette. That job was a trainer position she hoped would become a paid job. When it did not, she was disappointed but she knew from a lifetime of disappointments that it's best to accept life as it is and move on. For now she is happy, even excited. She feels fortunate to have this job at Southside Bakery.

Indeed, only 32% of people with disabilities between the ages of 18 to 64 have a full- or part-time job, according to a Harris poll which also found that more than two-thirds of the unemployed with disabilities would prefer to be working.

Back in Lafayette, Linda's boss had this to say: "I have people working at the bakery who can see, hear and can't do half the things that Linda can," says Denny Guibbeau. "I never thought anyone could work being blind and deaf, never thought you could do that. Seeing her work like she does is amazing."

Usher Syndrome

Linda's disability, the reason she cannot see or hear, is known as Usher syndrome, the leading cause of deaf/blindness in America. The condition was first identified in Europe: in 1858 and was named for Dr. Charles Usher, a British eye doctor who believed the disease was inherited. There are three main clinical types of Usher syndrome and cases are found around the world. One form is caused by a mutation specific to the Cajun population of south Louisiana, people like Linda Hebert (see "Acadians, Cajuns & Genealogies" on page 5).

Usher syndrome is known as a recessive genetic disease. Recesive means that it takes two mutated copies of the gene for the disease to be expressed. Usually both parents are carriers which means they do not have the disease but carry one mutated gene and one normal gene. Statistically, each child born to parents who are both carriers has a 50 percent chance of being a carrier with no disease, a 25 percent chance of being a carrier with no disease and 25 percent chance of having two mutated genes and therefore the disease. Linda Hebert is among this last group.

THE EARLY YEARS

She was two years old when her hearing loss was diagnosed. Her father was in the military and the family lived on a base in New Mexico, a place they would call home for the next six years. During that time, in the aftermath of World War II, there were few services for children with special needs. Often, children were kept inside and lived a lonely life, which was especially hard on children like Linda, who was an only child. Linda remembers leaving the house without her parents permission and finding her way to a playground where she spent hours and hours swinging and swinging, all by herself.

Then she started school in Santa Fe where she learned the rudiments of sign language and for the first time was able to communicate with others who also knew sign language. This did not include her parents.

When she was eight, her father was transferred to England and the schools were very different. For the next four years Linda was required to lip read and, in fact, half her hands were slapped when she attempted to use sign language. It was a confusing and difficult time for her but nonetheless she learned the basics of writing and speaking.

RETURN TO LOUISIANA

At age 12, Linda's life took a major turn for the better when the family moved back to Louisiana and she became a student at the Louisiana School for the Deaf in Baton Rouge. Prior to this, Linda's parents thought she might be mentally retarded. Nothing could have been further from the truth.

In this new environment, she thrived. Each student was expected to live independently and Linda quickly learned how to clean her room, do the laundry, cook and communicate using sign language. When she went home for a visit, she impressed her parents while they were away by cleaning the whole house, including laundry and dishes. With new confidence, she insisted that her parents learn to sign. In time her mother did learn some sign language but her father never did.

By communicating with her mother, Linda now had so many questions about her extended Cajun family. It took a while but in time she learned the concept of cousin, aunt, uncle, son and daughter. She also learned that she is the only one in her family who is deaf and blind.

Meanwhile at school, her knowledge of sign language accelerated and with it her knowledge of academics and life in general. Thanks to Ray Max, her favorite teacher, she learned to love math. Even today, Linda enjoys playing math games on her Braille computer.

In 1961, Linda graduated from the Louisiana School for the Deaf with the equivalent of an eighth grade education and the ability to sew, cook, do laundry and live a full life. Soon afterwards, she attended a football game in Shreveport where she met the man who would become her husband and father of her children. He also is deaf. All four of their children are hearing.

Today Linda and her husband are divorced but remain close. He lives nearby and visits frequently with the family. As the years went by, Linda slowly lost her sight. For more than 30 years, she has been blind. In order to communicate with her family, friends and co-workers, Linda now uses a form of tactile communication (photo on page 4).

Two of her daughters are training to become tactile interpreters. Linda has also learned Braille.

LINDA TODAY

Twice a week Linda travels to the Deaf Action Center in Lafayette to do crafts like quilting (see above) and to continue her Braille studies. By using the Braille machine she is able to do her banking, write in her journal, make a grocery list, work with puzzles and play math games. On Wednesdays, she visits neighbors, including her ex-husband, four children, 12 grandchildren and three great-grandchildren.

"She's not scared of anything," says daughter Lyn LeBlanc. "She loves an adventure, to go new places, meet new people and do new things."

In 1993, Linda was determined to visit friends in Chicago who are also deaf and developed a plan: she would travel by train, packing enough food for the long trip so she wouldn't have to get off the train. She enjoyed a three-month visit, then turned around and traveled back home all by herself.

"My Mother is very independent and does not lean on us," Lyn continues. "She's happy doing things and gets bored when she's not active. What my Mother does not want is to be a burden and if she thinks she will be a burden at a family function then she won't come."

At Southside Bakery, Denny Guibbeau has admiration for his productive worker. "It's been a good experience to work with Linda, to see her with so much energy and desire to work. Because of her, I understand how a person's attitude limits what they think they can do. Lots of people feel sorry for themselves. Linda does not. She just wants to better herself with her gifts and talents."

When she's asked how she wants the world to see her, Linda says with a big smile, "She that won't ever give up."

Although she can't see them, everyone around her has a big smile, too.

(Editors' Note: Since this story was written, Linda was laid off from her job at the bakery and is determined to find a new one.)

Linda Hebert at the Deaf Action Center

LADY OF LOVE

Linda is shown with the quilt she made using only her sense of touch. She donated the quilt to the Deaf Action Center to use as a fundraiser.

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Deaf Action Center and Village du Lac Provide Safe Harbor in Acadia

By Judy LaBorde

It's been said that the test of any culture is how the sick, the needy, and the handicapped are cared for. If so, the Deaf Action Center in Lafayette has been fitting tribute to the Cajun tradition of caring for each other.

Indeed, to go to the Deaf Action Center is to go to the heart of Acadia, both geographically and emotionally. Head west on the Interstate, turn at the Breaux Bridge exit, go two miles past Lake Martin, and there it is on the left—another world.

What you notice first are the trees. With trunks so wide they defy the centuries, the trees sway back and forth with moss-laden branches that seem to 'come home and let's play.'

Myra Mouton, right, and Linda Hebert express their thoughts using tactile communication.

Next, you notice the quiet. It's exceptionally quiet even though the Center is on a busy highway that goes from Breaux Bridge to Lafayette. The land seems to dip in from the main road, creating a vacuum.

Then, you see it—the street signs that serve to announce the special people who live here: Blind/Deaf Crossing. These signs are posted all over and are profound reminders of the protective environment that exists.

MEET MYRA

Myra Mouton does not look old enough to be the mother of a 10-year-old son and five other children.

She is the epitome of a gracious Cajun lady with a quick smile, easy laughter, and the soothing way of someone who has solved life's countless problems.

Since 1993, Myra has been the Director of the Deaf Action Center, her labor of love. Her involvement with the Deaf Action Center goes back to the 1960s as a result of her daughter, Monique, who is deaf.

The Center is part of the Archdiocese of Lafayette and has been offering services for the deaf since 1955. In the 1980s, 200 apartment units (called Village du Lac) were built on adjoining property. These units are designed for independent living for individuals with disabilities and are open to anyone with a disability.

Priority is given to the deaf and the deaf/blind and currently 20 individuals in those two categories live there.

Indeed, the Deaf Action Center is an oasis for the deaf and deaf/blind who choose to live in a protected environment. Someone is always on call to help. Each Sunday there are religious services. The Center offers classes in Braille, sign language, and crafts like quilting (see Linda Hebert's photo on page three). Each July there is also a two-week camp for children who are deaf and hard of hearing, as well as their siblings.

One of the Center's most vital services is interpreting. Myra has worked hard to develop a corps of 40 freelance interpreters to help the deaf and deaf/blind communicate with their doctors, hospitals, children's teachers, and the world at large. These interpreters are available for job interviews, the birth of babies, funerals, award ceremonies, Special Olympics, court trials, and even to get someone out of jail.

Interpreters provided services at all three public forums sponsored by Acadians but were forced on the Region of Genetics in 1999, 2001, and 2005 (see pages 17-18).

"Being able to communicate with the deaf and the deaf/blind is its own special reward," says Myra. "To see their smiles of understanding, to read their body language and know that they understand the message is all very gratifying. We get so much more than we can give them because they don't ask for very much. All they want is someone to acknowledge them. They live in a world that is so limited. When they have someone in that world who can communicate with them, they love it and show their appreciation."

"Being able to communicate with the deaf and the deaf/blind is its own special reward."

— Myra

Cajun culture is known around the world. Every year millions of travelers visit southern Louisiana to savor Cajun food, music and lifestyle. That is why when one considers the huge tragedy that brought the ancestors of the Cajans to Louisiana in the first place.

The Cajans of today are descended from a distinct group of about 3,000 French Canadians who came to Louisiana from 1765 to 1785 after being forced from their farms and villages along the Atlantic coast by the English who had taken over the region was called Acadia, thus the people were originally called Acadians.

Prior to their expulsion, the Acadians lived peacefully in Canada for over a century (early 1600s to 1755). They worked together in their communities to transform the salty marshland into fertile farmland. Isolated from Frenchmen living in Quebec and other parts of Canada, they created large, independent families that grew into self-sufficient communities for six generations. They also created a unique way of living based on country customs, French culture, their Catholic faith, and a provincial French dialect.

They knew that their identity as a people - not to mention their very survival - required neutrality from the two superpowers that controlled Canada at that time: England and France. For over a century, the Acadian strategy of neutrality did indeed produce peace. For a 42-year period of time there was not a single recorded crime among the Acadians.

All this tranquility came to an abrupt end when a brutal attack by the British military began a massive, chaotic expulsion of Acadians in 1755. The British were preparing for war with France and demanded that the Acadians leave France or an oath of allegiance to the British king and renounce their Catholic religion. The Acadians refused.

ACADIANA DIAPORE

Thus began the Acadian diaspora of 1755-1809, a sad chapter in the history of mankind in which thousands of innocent Acadians were forced onto ships that took them to places in North America, South America, and Europe. Half of the population was deported. Many died of disease, starvation, and exposure.

Those who survived the deportation found themselves stranded and unwarted in Massachusetts, Connecticut, New York, Pennsylvania, Maryland, Virginia, South Carolina, and Georgia. Families were separated; children were taken from their parents and sold as indentured servants. In Maryland, they were shot on the spot by British soldiers.

Other Acadians were shipped across the ocean to Haiti, Martinique, French Guiana, Argentina, Uruguay, and New- foundland. Still others landed in France and England. Many were jailed or concentrated in coastal camps.

This violent deportation is a seminal event in the history of the Acadians/Cajuns and the State of Louisiana. It is basic to understanding one of the most remarkable populations for the study of genetics in the 21st century.

STORY OF SURVIVAL

Wherever the Acadians were forced to go, they were not wanted, with one important exception: Louisiana. At that time, Louisiana was not a part of America. It was a colony ruled by Spain, a long-time enemy of England. Spanish Governor Bernardo de Galvez knew Louisiana had a shortage of settlers and saw the Acadians as a way to offset the growth of English settlements in central Louisiana.

So, Galvez settled the traumatized Acadians along the isolated bayous and wetlands of the Atchafalaya River in 1763. Areas dominated by mosquitoes and humidity...areas where few people wanted to live. As in Canada, the Acadians lived apart, in geographic and cultural isolation. Adding to their separation were the old and colonial French dialect they spoke. Frenchmen looked down on the Acadians, thus intensifying the isolation that began in Canada.

PRICELESS RESOURCES

According to Dr. Carl Brasseaux, an expert on Acadian history and professor at the University of Louisiana at Lafayette, there were 12,000-18,000 Acadians in Canada before the expulsion. Of this number, about 3,000 arrived in Louisiana. Such a large decrease in population results in fluctuations in the occurrence of genes in a population such as the Acadians. For example, the frequency of a disease gene may be, by chance, be much higher in the smaller population than in the previous one, and this is likely to increase the number of people with the disease in future generations.

Throughout history, there are many examples of ethnic groups leaving their homelands and starting over. A few examples are Ashkenazi Jews from the Jewish Diaspora, the Old Order Amish (originally from Europe and long associated with Pennsylvania), the Memnonites who are a subgroup of the Amish, and the Cajuns.

Genetic studies among these groups have advanced our understanding of the causes of many disorders, including Alzheimer's disease. Because of this, the Cajuns and populations like them are a priceless resource to medical research and the improvement of healthcare for all. That is why geneticists are grateful to the many Acadians who have participated in genetic studies for more than 40 years.

There are several reasons why people participate in medical research...currency among generations yet to be born is one of the most powerful. This type of empathy is something that the Cajuns of Louisiana have in abundance - the deep connections among families and generations.
Blake gets ready for takeoff

HE CAN'T HEAR
That day was Sep-
tember 17, 2000, a
Sunday, and the
place was Houma,
Louisiana, a town of
about 33,000 that's
steeped in Cajun cul-
ture and tradition,
about an hour's drive
from New Orleans.
Dwci, her mother,
remembers Blake's
birth like it was
yesterday. There she
was, lying on a
hospital bed, exhausted. Just 18 hours earlier she had
given birth to her first child, a healthy baby boy, and
now she was told, "He can't hear. He failed the test. He's
dead!"

"What?" Dwci shouted. "What?" she screamed even
louder. "I started coming out of my bed, I was so upset. I
told the nurses to leave and not come back."

How could her nine-pound baby boy with the huge
blue eyes, velvety skin, and ten perfect fingers not hear
the sounds of love all around him? Anger, fear and denial
took over.

Blake's father-in-law told her that two teenage cousins
on his side of the family had recently been diagnosed with
a disease called Usher syndrome, a condition that causes
both deafness and blindness. That was way too painful for
Blake to think about, so she wiped away his words like
she was flicking away a fly.

She didn't want to know that Usher babies are born
profoundly hearing impaired and then begin to lose
eyesight in early adulthood. She didn't know that
cases of Usher have been identified around the world, but
that a specific mutation is unique to those of Acadian
ancestry—the heritage of her husband.

She didn't know Usher is recessive, meaning that
both parents contribute a gene of the disease at conception.
She didn't know that, just 60 miles away, Dr. Britnya Kears,
a geneticist at LSU Health Sciences Center, was studying the
specific genetic mutation that causes Usher in the Acadian
population. Blake didn't know any of these things, nor did
she want to know them. Her anger was too great and she
was a long, long way from acceptance.

When her husband, Cory, brought her and the baby
home from the hospital, Blake decided to do her own
testing. She banged pots and pans, but her son did not
respond. She slapped her hands and she slapped
him. She ran the noisy vacuum all around his tiny body.
He didn't move. "It broke my heart," she says. "I wanted
him to wake up, to see the surprise on his face." But he
never did.

Still reeling in disbelief, she somehow found the
strength to move on, even if it was just barely. "For months,
it seemed that all I did was cry."

At the age of three months, Dwci started Blake in
speech therapy. Five times a week she drove the 45 miles
from Houma to the Chinchuba Institute in Marrero for his
one-hour sessions. Chinchuba Institute has a long record of
Teaching deaf children to talk and was actively involved in
Blake's early training.

He started speech therapy while seated in an infant
car seat and eventually moved to the chair for toddlers.
He was attentive, bright, stronger for more. The therapists
taught him how to cry, how to express happiness,
and how to use his hearing aids. He learned to jump. He
learned his ABCs.

"GENETICS OF THE ACADIAN PEOPLE"

When Blake was six months old, his father and
grandfather heard that the Center for Molecular and
Human Genes at LSU Health Sciences Center in New
Orleans was staging an unusual educational event for the
public called "Genetics of the Acadian People." The event
was open to anyone who wished to attend and the purpose
was to provide education about genetics, especially to the families of Acadian descent.

In addition to the three main speakers, there would
be 14 physicians and researchers available to talk to and
hopefuly answer questions.

The event was a major public service initiative of the Center under the leadership of Bronya J.B. Keats, Ph.D. Eleven organizations were sponsors of the event, including South East Louisiana Area Health Education Center, Louisiana State University Health Sciences Center, Nicholls State University and Tulane University Health Sciences Center.

Blake's father and grandfather decided to go. Maybe
Dwci could ignore the cousins but they knew Usher was
a real possibility for young Blake.

There they met Dr. Britnya Kears who shared with
them the latest in research findings on Usher, including
the newly available genetic test for the Acadian mutation.
Weeks later: the Heck's visited Dr. Keats in her office and
laboratories at LSU Health Sciences Center in New
Orleans. After several discussions and hours of genetic
counseling with Dr. Keats, the Hecks decided to have
Blake tested for Usher.

The test result was what they dreaded most. "Your
baby has Usher."

At least this time the Heck Family was prepared.
Because of their discussions with Dr. Keats, they knew
more of what to expect.

Early detection is a major breakthrough for families
with Usher syndrome. Ducj and Cory now knew more
about their young son's future and what the family would
have to face. Since Usher children lose
eyesight in early adulthood, many parents
look for ways to develop the capacity for hearing before that happens.

TOUGH DECISION, TURNING POINT

Dwci and Cory are concerned about cochlear implants.
A computerized device implanted in the inner ear.
Cochlear implants for children were approved by the Food
and Drug Administration in 1990. More than 25,000 people
have received cochlear implants in this country, including
10,000 children, according to the Centers for Disease
Control. (See cochlear implant story on page 15.)

"We've mentioned it, but we're not limited," Darci says. "We were soul searching, confused... and we didn't have a
guide. We spoke to people in Deaf Culture, people who
questioned why we wanted to change our child from deaf
to hearing. We also talked with the mother of the teenage
cousins with Usher. They felt strongly against implants,
pointing out that there were many implants that were not
successful. We met with two or three people with implants
who could not talk.

Indeed, it was during this interval that a turning point
came for Darci, the point when she faced reality.

There was an involved psychological screening process we had to go through before the surgery. There we learned that a
lot of special care goes into maintaining these devices and
that Blake could not learn how to do this on his own. I would have to be his teacher, all day and everday. This
was going to be a commitment like no other: The decision
to get cochlear implants would change my life forever.

After talking and listening, and then talking and
listening some more, Ducj and Cory came to this
conclusion: "Parents must find out what's best for their
child. We decided to go ahead with the cochlear implant."

On November 27, 2001, Blake Heck, age: 14 months,
went through a three-hour surgery. "He came out of it
wanting to play; he had such a good attitude."

After a six-week recovery, Blake's implant was
activated on January 10, 2002. "He did very well. We started
him in speech therapy immediately."

Sometimes his progress has been uneven. As a baby,
he crawled late and walked late. He needs help with running
and jumping. He's still flagellated and has ongoing
problems with balance.

BLAKE TODAY

Otherwise, he's a regular boy. He likes toy trucks and
tractors and playing in the rain and the hirting the sizzle
that bubbles make. He sometimes fights with his younger
brother, Cameron.

He is now in second grade and on the honor roll. His
teacher says he is smart and attentive.

He's reading above grade level and his speech is
better than many kids his age, says Dacj. "What most
amazes me is how well he understands abstract ideas like
outer space. He understands astronauts and why they went
to the moon. We did research on the computer and found
images on the NASA website of the men eating in the
spaceship without gravity. We saw photos of how the
astronauts saw the earth, in hues of blue and green. Blake
was glued to the TV during the shuttle launch and could
explain all about the exterior gas tank and how it falls into
the ocean and big boats come to pick it up."

Dwci will be the first to admit that she's learned a lot,
too. "When Blake was diagnosed, I was seven months
pregnant with my second child. Cameron. I had been
angry for a long time and finally I apologized to my husband
because I had blamed him for Blake's condition when, in
fact, the genes come from both parents."

The whole experience has made me a stronger
person. I have more patience than I thought I would. I
don't treat Blake special. I treat him like he's a hearing
person with special needs. I want him to play sports, throw
a football, kick a soccer ball. He's also learning sign
language so he can communicate with his peers.

"It's been a long road and it's going to get rougher.
As time passes, we'll worry about his eyesight."

For now, she's grateful for how far Blake and the
whole family have come. "Even if we had known about Usher
we would have had this baby."

"As to choices we've made, I think you have to be
willing to try new things and be positive. Change is going
to happen, no matter what. God made us with brains to
make the technology to make the world better.
Without the technology, Blake could not hear, could not
have the experience of leaves rustling, clocks ticking, cars
passing by."

And for Dacj, today, this moment, that is enough. *
Collaboration amid chaos

The silver lining of Hurricane Katrina

By Judy LaBorde

Stars come out

There's a saying, "when the skies are darkest, the stars come out."

Dr. Bronya Keats, chairman of the Department of Genetics, saw those stars in the form of her colleague, Dr. Ed Rubel, professor in the Department of Otolaryngology in the Virginia Merrill Bloedel Hearing Research Center at the University of Washington in Seattle. After the storm, Dr. Rubel called, offering help, support, whatever he could provide. He and his colleagues at the University of Washington became the silver lining amid the raw chaos of Katrina.

They just never thought that one of the worst natural disasters in American history would be the catalyst for such a new venture.

In solid testament to the generosity of human nature, Dr. Keats received more than 50 offers for help, support and collaboration. "I had a legal pad and kept writing names down, one name per line, single space."

The most appealing offer came from Dr. Rubel. Over the years, he and Dr. Keats had talked about collaborating on studies of Usher syndrome in the Acadian population. Now a storm, they thought that one of the worst natural disasters in American history would be the catalyst for such a new venture.

Acadian Usher

The genetic mutation that causes Usher syndrome in the Acadians is in the gene called USH1C. Babies with two copies of this mutation are born with profound deafness, but also have problems with balance. Later, in early adolescence, they develop retinitis pigmentosa, a progressive form of blindness that begins as night blindness.

Dr. Keats has been a leading investigator in Acadian Usher research since 1990. She knew that what was needed to advance the Usher research was an animal model. Shortly before the storm, a special mouse had been bred with the Acadian Usher mutation but with all the upheaval caused by Katrina, no one knew if the mice would survive.

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Enter the zebrafish. And enter Dr. Dave Raible, respected around the world for his research using zebrafish. He also works at the University of Washington where he is a professor in the Department of Biological Structure.

Dr. Rubel and Dr. Raible had been colleagues for several years. It took Katrina to bring Dr. Raible and Dr. Keats together.

She wanted to know if zebrafish with reduced amounts of hearing proteins that's missing in Usher, would exhibit symptoms of Usher, such as problems with vision, hearing and balance. Dr. Raible offered to help.

As a model organism for scientific studies, zebrafish have a lot of advantages. They usually lay about 200 eggs at a time. The embryos are transparent and they have many of the same genes as humans.

They are an older animal in evolution with organs that have a conserved role, meaning that the function of the organs has stood the test of time.

Zebrafish have a lot to teach us about hearing, balance and vision in humans because of parallel functions. For example, the development of hair cells is a critical component of their hearing and balance organs, just as with humans. Both of these organs are easily accessible and can be seen under a microscope.

To answer the question about hearing, Siampath and Jennifer Lentz, also a graduate student with Dr. Keats at the time of the hurricane, headed for Seattle and the University of Washington in September of 2005. Neither Jennifer nor Si had ever been to Seattle. Neither knew Dr. Raible nor anyone else at the University of Washington. Neither had ever worked with fish or with a collaborating team. And with all the disruptions caused by Katrina, neither had access to the scientific literature that could have helped them study and prepare.

"When I arrived in Seattle, I had been living out of a suitcase for weeks," Jennifer said. "I didn't know how much damage had been done to my home or the laboratories at LSU. My world had been turned upside down while at the same time I was facing the biggest challenge of my professional life. I knew I was unprepared and that I had to succeed."

The situation quickly began to work out when Jennifer met Dr. Kelly Owens at the Seattle airport. Kelly is a post doc in the laboratories of Dr. Rubel and Raible. She had a good idea of what Jennifer needed on personal and professional levels and she committed to making the collaboration work.

Throughout the next three months, Jennifer and Si worked in Dr. Raible's lab. Si also enrolled in the University of Washington which had quickly arranged to take in 91 college students from throughout the Gulf Coast areas that were impacted by Katrina. Students attended classes tuition free and were offered housing in the community.

Si spent the Fall, 2005 semester in Seattle. He worked 12 hours a day, seven days a week in the lab, except when attending classes. Initially, Jennifer spent 30 days in Seattle and then over the next year went back for seven subsequent trips to do intense experiments. In this interval, Kelly allowed Jennifer to stay in her home as a guest. They shared meals and rode to work together. In the lab, she trained Jennifer and helped in any way she could.

The result was one of the most productive collaborations ever undertaken during the 31-year professional career of Dr. Bronya Keats.

"We have now developed two animal models which will facilitate many new experiments in studying Acadian Usher," says Dr. Keats. "Our collaboration has grown to involve the University of Oregon in addition to Washington. We are all busy writing our joint papers for publication.

"Because of our collaborations, we are entering a new era in Acadian Usher research," she said.

(Edited by Note Special thanks to the University of Washington for providing tuition, housing, lab space, supplies and priceless support for "life after Katrina." Thanks also to the Foundation Fighting Blindness for their unswerving financial support for Acadian Usher research.)
Consider what it takes to pursue a career in medical research. A typical scenario finds our budding scientist on graduation day from college holding a diploma affirming a bachelor’s degree in science. This achievement is followed by five to seven years of graduate study to obtain a PhD. Three to 10 years of education now evolve into post-doctoral training. Throughout this interval, an experienced mentor plays a key role in guiding the young scientist.

Now comes the crucial juncture towards self-sufficiency: how to develop funding sources that allow the young researcher to pursue his or her own career in research? As before, the role of mentor is vital. With knowledge and experience in abundance, the mentor can provide coaching and guidance. Mentors can show how to avoid wrong turns and dead ends. In short, mentors, who are usually full professors, can show junior faculty where and how to get the funding needed to develop into productive and successful researchers.

Against this backdrop, it’s a huge accomplishment that scientists from several Louisiana academic institutions have attracted over $120,000,000 in competitive funding from the National Institutes of Health (NIH) to do mentoring. The grants are called COBREs (Centers of Biomedical Research Excellence) and are made available to states like Louisiana with historically low aggregate success rates for grant awards from the NIH.

Of this total, $10.7 million has been awarded to a collaboration jointly directed by Dr. Brosnya Keats with LSU Health Sciences Center and Dr. Prescott Deininger with Tulane University Health Sciences Center and called “Mentoring a Cancer Genetics Program.”

“The research landscape since Katrina has changed,” says Dr. Keats who is chairman of the Department of Genetics as well as director of the Center for Molecular and Human Genetics. “We lost priceless momentum because of the storm,” she said. “We cannot afford to waste resources by not cooperating.”

Since the program’s inception, three of the five original young investigators have received RO1 awards from the National Institutes of Health. RO1 funding is provided by the NIH through peer review for investigator-initiated research and is the gold standard for success.

The current junior investigator in the program are Drs. Andrew Hollembach and Tomoko Ikawa with LSU Health Sciences Center. At Tulane, the researchers are Drs. Astrid Engel, Carl Gregory and Aline Scandurro. Dr. Engel recently received the exciting news that her RO1 application will be funded.

Other COBRE mentoring programs at LSUHSC in New Orleans are “Mentoring Neuroscience in Louisiana,” Dr. Nicholas Bazan, Ph.D.; “Mentoring Oral Health in Louisiana,” Dr. Paul Fidel, Ph.D.; “Mentoring Cardiovascular Biology,” Drs. Daniel Kapusta and Pamela Lucchesi, Ph.D.s, and “Mentoring Translational Research in Louisiana,” Dr. Augusto Ochoa, Ph.D. At Tulane, the other mentoring program is called “Hypertension and Renal Biology,” Dr. L. Gabriel Navar, Ph.D.

Photo left top: Drs. Tomoko Ikawa and Andrew Hollembach, LSU Health Sciences Center.
Photo left bottom: From left, Drs. Aline Scandurro, Carl Gregory and Astrid Engel, Tulane University Health Sciences Center.

It’s a major health concern that requires solutions: nearly one in five children in America are obese or overweight. The health consequences for the individual and society are enormous. According to the Journal of Pediatrics, childhood obesity is the leading cause of pediatric hypertension, is associated with Type II diabetes mellitus, increases the risk of coronary heart disease, increases stress on the weight-bearing joints, lowers self-esteem, affects relationships with peers. Some authorities feel that social and psychological problems are the most serious consequences of obesity in children.

Brian Jakes is committed to the health of children in Louisiana.

It’s a significant problem demanding bold action and Brian Jakes is committed to doing his part. As Director of the South East Louisiana Area Health Education Center (SELAHEC), he is involved in a major collaboration known as the “Mississippi Delta Initiative” to focus on childhood obesity in 45 of the 64 Parishes in Louisiana. In addition to SELAHEC, the collaboration involves Pennington Biomedical Research Center, the Agricultural Center at LSU in Baton Rouge, the Multi-Practice Clinic in Independence, LA, The Wellness Center in Oakdale, LA, and the LSUHSC Department of Genetics.

The program is funded by the Health Resources and Services Administration (HRSA) Office of Rural Health Policy of the U.S. Department of Health and Human Services.

The program includes parent participation, exercise and nutrition.
A NEW APPROACH TO EDUCATIONAL OUTREACH

Dr. Gregory Brings Genetics to High Schools

Paula Gregory is an innovator. The highly skilled educator, who is an associate professor in the Department of Genetics, knows that the pull of tradition can sometimes be powerful. She also knows there can be even more power when you find ways to bend the boundaries, rethink the definition of student and overall expand the big tent of learning about science.

In a career that spans two decades, she's developed programs and found funding for diverse "outside the box" programs in education and outreach. As a result, she's taught businessmen and ministers how to extract their own DNA. She's spent weeks at a time training high school science teachers in the latest laboratory techniques they can take home to their students. She's also the brains behind a unique, hands-on program for high schoolers that teaches them about genetics and cancer. Entitled the "High School Cancer Research Partnership," the weeklong program involves students using special kits to extract DNA from their own cheek cells. In between the 22-step process, Dr. Gregory engages the students in lively dialogue about the ethical implications of genetics in the 21st century.

"High school is the last place that students may ever encounter science, particularly genetics," Dr. Gregory said. "Yet these young people will have to make major decisions regarding their health care in the coming years."

This year she brought the program to East Jefferson High School, West Jefferson High School, Mandeville High School, Baton Rouge Magnet High School, St. Mary's Dominican High School and Ben Franklin High School.

The goals of the program are to bring cutting-edge technology to the classroom and excite high school students about the genetics of common diseases and cancer research. Other goals are to reach a segment of the general population with information about the genetic changes that cause cancer and concepts of new areas of research, including stem cells and gene therapy.

"We have also used this program as a way to identify and recruit students who are interested in our summer internships. Through these two programs, we are not only showing students the interesting research happening in our own backyard but also cultivating their interest in research careers."

Funding for the program comes from the National Cancer Institute. ♦

Summer Intern Connects Small Steps to Big Picture

Dr. Oliver Wessely watches over Veronica Alexander as she performs an in vitro fertilization of frog eggs. Once fertilized, the eggs will develop into tadpoles which are analyzed for kidney function.

Veronica Alexander has always been a thinker and this past summer her brain was in overdrive. As a sophomore at Oberlin College assigned to the laboratory of Dr. Oliver Wessely, Veronica studied the function of the kidney in frogs so that in the future there will be better medications for people with diseased kidneys. 'At first, I thought, oh, I don't know if I'm meant to be a researcher,' she said. 'That's because all I could see was my specific project. Now, after several weeks, I'm thinking and making connections. I see the overall goal. I get the big picture.'

And, she's using all of the experiences she's had in life. In the summer of 2006, she worked as a volunteer at Children's Hospital, often playing with a little boy whose diseased kidney required him to undergo dialysis two and three times a week. 'I've been thinking a lot about that child, how difficult the dialysis was on his body,' she said. 'Now that I'm working with Dr. Wessely, I see how genetic research could improve the functioning of his kidneys and totally change his lifestyle.'

Developing new approaches to kidney disease has been the work of Dr. Oliver Wessely for the last four years. "Like many genetic conditions, most kidney diseases do not kill right away but there are major, long-term consequences," he said. "Right now, there are two major therapeutic approaches for kidney diseases either dialysis, which is a lifelong commitment, or organ transplantation. Both are highly invasive and severe. There's a huge need for improvements in treatment, for ways to fight the symptoms in a more targeted way."

That need translates into more and better laboratory workers which is why Dr. Wessely has eagerly participated in the summer intern program for three years. 'In the end, you get good students who can do good sized projects that help you move the research forward,' he said.

The summer intern program is the brainchild of Dr. Paula Gregory, Associate Professor in the Department of Genetics. She has directed and inspired the program since arriving at LSUHSC in 2002. "Every year, we get more and more interest in the summer intern," she said. Even with Katrina's interest never wavered. Now I'm getting calls from researchers several months in advance asking if they can participate. It's one of those programs that is good for everyone involved."

Funding comes from the National Cancer Institute and the Louisiana Gene Therapy Research Consortium. ♦
Vision Loss From Usher Syndrome
Due to Retinitis Pigmentosa

Children born with Usher syndrome will slowly lose their eyesight due to a condition known as Retinitis Pigmentosa (RP) which causes the degeneration of the retina. Think of your eye as similar to the structure of a camera. Your retina is in that space in the back of the eye that’s similar to film in a camera. It’s the job of the film to capture the details of the images we see. In the human eye, the retina is rich in cells that capture the images. When these cells, called cones and rods, begin to deteriorate, so does the person’s vision.

According to the Foundation Fighting Blindness, which has long supported research into RP, people with the disease are first affected by dim lighting. This results in degeneration of peripheral and night vision. Night blindness is one of the earliest and most frequent symptoms of RP.

RP is typically diagnosed in adolescents and young adults. It is a progressive disorder. The rate of progression and degree of visual loss vary from person to person. Most people with RP are legally blind by age 40, with a central visual field of less than 20 degrees in diameter. It is a genetic disorder and, therefore, is inherited.

How is RP inherited?
An estimated 100,000 Americans have RP mainly caused by mutated genes inherited from one or both parents. Mutated genes give the wrong instructions to photoreceptor cells, telling them to make an incorrect protein, or too little or too much protein. (Cells need the proper amount of particular proteins in order to function properly.) Many different gene mutations exist in RP.

If a family member is diagnosed with RP, it is strongly advised that other members of the family also have an eye exam by a physician who is specially trained to detect and treat retinal degenerative disorders. Discussing inheritance patterns and family planning with a genetic counselor can also be useful.

For more information, go to the website for the Foundation Fighting Blindness at http://www.blindness.org.

What is a cochlear implant?

A cochlear implant is an electronic device that restores partial hearing to the deaf. The implant bypasses damaged hair cells and converts speech and environmental sounds into electrical signals which are then sent to the hearing nerve.

Cochlear implants were first approved by the Food & Drug Administration in 1984.

The implant is surgically implanted under the skin behind the ear. There is an external speech processor, which is usually worn on a belt or in a pocket. A microphone is also worn outside the body as a headpiece behind the ear to capture incoming sound.

Cochlear implants do not restore normal hearing, and benefits vary from one individual to another. Most users find that cochlear implants help them communicate better through improved lipreading, and over half are able to discriminate speech without the use of visual cues.

Blake Heck with implant

Using special glasses developed by The LightHouse for the Blind, the Harris Family of Baton Rouge learn what it’s like to see with tunnel vision caused by retinitis pigmentosa. This demonstration was part of community outreach by the Center for Acadiana Genetics during Earth Day celebrations. Exhibits focused on connections between the earth’s geography, migrations, genes and disease.

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More than 1,000 Attend Sponsored by the Center

More than 1,000

The Center for Acadiana Genetics has staged three large scale public education forums. The first event was held at McNeese State University in Lake Charles in 1999. The second event was held in 2001 in Thibodaux, Louisiana, in the heart of Acadiana, at Nicholls State University.

The third forum was held in 2003 in New Orleans on the City Park campus of the LSU School of Dentistry. The theme was “Genetics: Your Family and Your Health.”

The facilities at the dental school were a perfect match for the event. Dr. Vincent Liberato and his staff went out of their way to make everything go smoothly,” said Dr. Keats.

More than 1,000 people attended these three public forums.

“It’s very gratifying to know that so many people came to us to learn about genetics and what this means for their health care,” said Dr. Keats. The most dramatic story to come out of these events is that of young Blake Heck and his family who are originally from Lockport, Louisiana. His story is detailed in this newsletter beginning on page one.

2003 Forum

The welcome addresses were given by Dr. Joseph Moerschbacher, LSUSC Vice Chancellor for Academic Affairs and Dean of the Graduate School; Dr. Paul Fidel, Director of the LSUSC Center for Oral and Craniofacial Biology, and Alisha Jeanonne with the Office of Congressman Billy Tauzin in New Iberia, Louisiana.

Raychel’s Story

At all three events, the first keynote speaker was Raychel Bartell whose personal story was the catalyst for the founding of the Center for Acadiana Genetics and Hereditary Health Care.

In 1998, Raychel’s son, Keith, was diagnosed with Friedrich ataxia, a disease she had never heard of. In disease she would soon learn was closely tied to the family’s Acadian ancestry, a disease she discovered was being studied by Dr. Bronya Keats at LSUSC in New Orleans.

Raychel was also a longtime assistant to then Congressman Billy Tauzin whose district lies in the heart of Cajun country.

Raychel’s talk was a compelling account of how her family’s diagnosis has been affected since Keith’s diagnosis. “My other two sons may not be affected but they are affected,” she said. She told the audience how the hopes, dreams and ambitions of the whole family have been permanently altered.

Distinguished Speakers

Dr. Lynn Jorde, Professor in the Department of Human Genetics at the University of Utah School of Medicine, focused on the importance of human genetic variation.

Understanding, with precise, how we differ genetically and how we are alike has been invaluable in evaluating DNA evidence in criminal cases. It’s also played a critical role in advancing our understanding of the genetic contribution to many diseases, such as inherited colon cancer, obesity and alcoholism.

Comprehending genetic variation also sheds light on why individuals respond so differently to the same medications. Because more than 10,000 Americans die each year as a result of adverse reactions to prescription drugs, there are major consequences to understanding those differences.

Dr. Wylie Burke gave the afternoon keynote address. She is Professor and Chair of Medical History and Ethics at the University of Washington in Seattle.

Her talk focused on the rapidly growing and sometimes confusing field of genetic testing. On the one hand, testing offers great hope: it can help identify the cause of rare, serious health problems. In turn, however, can help with further diagnostic testing and insight into drug reactions, all hopefully leading to more effective treatments. On the other hand, patients, providers and researchers need to make sure that genetic testing does not lead to discrimination, unnecessary medical treatments, social stigmatization or inaccessible health care.

20 Sponsors and 20 Exhibits

“It took a massive effort with a lot of grassroots support to stage these events,” said Dr. Keats. “I’m glad we did.”

Twenty companies and organizations were responsible for the 2003 forum. Applied Biosystems Children’s Hospital in New Orleans, Congressman Billy Tauzin’s Office, Louisiana Congressional Delegation, Nicholls State University, LSU Health Science Center in New Orleans, South Louisiana Area Health Education Centers, State of Louisiana Office of Public Health, Tulane University Health Sciences Center and Foundation of LSU, Louisiana Cancer Research Consortium, Louisiana Gene Therapy Research Consortium, and BATON Rouge.

Also, there were 20 entities exhibiting at the event representing LSU Health Sciences Center, Tulane University Health Sciences Center, Children’s Hospital, the State of Louisiana, LSU in Baton Rouge and Neurological Disorders in Acadiana.
Director's Message

The Power of Moving Forward

Dr. Bronya Keats

So much has happened, so much has changed, yet so much has stayed the same or improved since a certain infamous storm turned our lives upside down two years ago.

With deep appreciation for our partner, the South East Louisiana Area Health Education Center (SELAHEC), ably led by Brian Jakes, we have been able to get back on track and continue our community and educational activities.

Bronya is a firm supporter of science education and a firm believer in our young people. For the last seven years, he's enthusiastically underwritten prizes for the junior high and senior high school winners of the annual science fair competition. Prizes are awarded for projects exhibiting an excellence in comprehension of genetics. The top prize is a $1,000 scholarship awarded to the senior division winner at the state level. The scholarship is good for tuition at any college or university in Louisiana.

Educating the public is a mission we continue to take seriously. Please take time to read about our third public forum on pages 16 and 17. I still feel a glow thinking about the great turnout from the public, the 20 sponsors, 20 exhibitors and the outstanding talks given by our keynote speakers, Dr. Lynn Jorde from the University of Utah, and Dr. Wyle Burke from the University of Washington.

As with our first two public forums, the tone of the event was beautifully set by Raychel Barilek whose personal story reminds us of the heavy toll of genetic diseases have on families as well as individuals. Her son, Keigh, was diagnosed with Friedreich ataxia in 1998. That event triggered a series of other events leading to the founding of the Center for Acadiana Genetics and Hereditary Health Care in 1999. In a very real way, Raychel and her family have written an important chapter in the history of genetics in Louisiana. So did Congressman Billy Tauzin and we will be forever grateful to them.

Our educational outreach activities have continued throughout south Louisiana. For this, we greatly appreciate the dedication of Dr. John Doucet, Associate Professor of Biological Sciences at Nicholls State University in Thibodaux. John received his Ph.D. from the LSU/BSC Department of Biochemistry and Molecular Biology. He is also a junior investigator in Dr. Nicolas Bazan's COBRE, "Mentoring Neuroscience in Louisiana" (see page 10). John has been tireless in his travels, giving talks to a wide spectrum of groups like science teachers, Kiwanis Clubs, genealogy groups, businesses, groups, you name it. As time goes by, interest grows in genetics and how it impacts everyday health care decisions. That interest translates to educational opportunities and John has been consistently willing to take advantage of those opportunities.

One example of heightened community interest in genetics is our participation in the Abdominal Aortic Aneurysm (AAA) screening program directed by Sheila Arrington, a community leader based near Baton Rouge. These community screenings use ultrasound to detect aneurysms and regularly attract 200 plus people. As with most disorders, there is a strong genetic component.

Dr. Robin McGoye in the LSU/BSC Department of Pathology sees this link in family pedigrees and in her autopsies. That's why she and Dr. Dana Trosclair, also in the Department of Pathology, recently received funding from the LSU/BSC Translational Research Initiative to study AAA. Dr. McGoye has participated in two of the community screenings, often serving as the genetic counselor; in addition to her duties as medical investigator.

Our Human Genetics graduate program is flourishing thanks to the efforts of Dr. Dipaasri Mandal, assistant professor in the Department of Genetics, who is also our graduate program coordinator. We presently have 17 students in the program who are mentored by faculty in the Department of Genetics and Gene Therapy Program.

Heena Mehta with her award-winning poster.

Congratulations to one of our students, Heena Mehta, who won second place in the poster competition at the 21st Annual Graduate Research Day. Heena is working with Dr. Alistair Ramsay, Director of the Gene Therapy Program, who is also to be congratulated.

He was recently awarded $5,500,000 by the Board of Regents Post-Katrina Support Fund Initiative to establish the Louisiana Vaccine Center in collaboration with Tulane University, the Research Institute for Children, and Xavier University.

The Human Genetics seminar series is thriving. Outstanding speakers from around the nation have come to LSU/BSC to give talks. We've experienced consistently high attendance and increased interaction with researchers from other universities. Much praise for this success goes to Dr. Fern Tsien and Dr. Andrew Hollenbach who are the coordinators of the seminar series.

A new addition to our faculty is Dr. Wanguo Liu, associate professor in the Department of Genetics and the Stanley Scott Cancer Center. His research focuses on cancer gene discovery, with emphasis on prostate cancer. Dr. Liu joins us from the Mayo Clinic and we are delighted to have him.

And I'd like to extend a very special thank you to Judy LaBorde, who's been our "go to" person since 1994. Judy coordinates all aspects of the Center's activities and is responsible for the writing and photography in our newsletter. She suggested the name "Linkage" for our newsletter and she's helped us develop links with the community ever since.

Zoe Nicholas and Mary Victoria were junior division winners of the Baton Rouge Regional Science Fair.
Clinical Services in Genetics

Every year 1 in 33 babies born in America has a birth defect, according to statistics compiled by the March of Dimes. A birth defect is an abnormality of structure or metabolism (body chemistry) present at birth that results in physical or mental disabilities or death. Several thousand different birth defects have been identified. Birth defects are the leading cause of death in the first year of life.

The State of Louisiana requires that the 67,000 babies born each year be screened for 28 genetic disorders, including cystic fibrosis. There is also mandatory screening for hearing.

Listed below are the clinical genetic services provided by doctors from LSU Health Sciences Center New Orleans and Tulane University Health Sciences Center.

Clinical genetic services are provided by LSU at Children's Hospital in New Orleans and also at the Children's Hospital Metairie Center (nine times a year). In Baton Rouge, services are provided three times a month at the Children's Hospital Outpatient Clinic. In Lafayette, clinics are held twice a month at the Children's Hospital Outpatient Clinic. Every other month, genetics clinics are held in Monroe at the St. Francis Community Health Center. Clinics sponsored by the State of Louisiana for those with no insurance are held six times a year at the Lake Charles State Clinic and the Thibodaux State Clinic.

Additionally, in New Orleans, there are specialty clinics for Down syndrome, craniofacial anomalies, neurofibromatosis and metabolic disorders.

Services offered by the Tulane Hayward Genetics Center are clinics at Tulane Hospital for Children in New Orleans (weekly), Women and Children's Hospital in Lafayette (every two weeks), Lafayette State Health Unit (monthly), Hammond Health Unit (monthly) and the Huey P. Long Hospital in Alexandria (every three months).

The Hayward Genetics Center also has cytogenetic, biochemical and molecular diagnostic laboratories. A recent addition to the cytogenetics lab is a microarray-based Comparative Genomic Hybridization (CGH) system that greatly enhances the detection of small chromosomal deletions.

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A newsletter that seeks to connect the LSU Health Sciences Community with all who care about the genetic health of our families.