Niemann-Pick Disease

Rare genetic disorder may shed light on other illnesses

By LAURIE SMITH ANDERSON

In the examination room, while waiting for the neurol-
ologist, 15-year-old Michael Landry picked up a small hammer and tested his own reflexes.

"Ow, man. That hurts," he said, putting it down and picking up an otoscope.
Pediatric neurologist Barbara Golden came in, followed by two of her colleagues.

"Hi, Michael. How are you?" she asked.

"Michael is proof of rule number one," she told her fellow physicians. "Rule number one is that the mother is always right."

Michael has Niemann-Pick type C disease — a rare, fatal children's cholesterol disease. Initially, the disease can mimic other illnesses, such as Parkinson's disease or cerebral palsy. Golden, who had never seen a Niemann-Pick case before, is quick to admit that she missed the diagnosis on Michael.

It was only through the persistence of Michael's mother, Maureen Landry, who continued to seek help from specialists, that the diagnosis was made and confirmed.

Niemann-Pick type C disease is an inherited metabolic disorder in which cholesterol builds up in the spleen, liver, lungs, bone marrow and brain, causing progressive deterioration of the nervous system. Symptoms include an enlarged spleen and liver, poor muscle control, slurred speech and dementia.

The disease is so rare that it is only diagnosed in about 300 children in the United States each year. The gene for the disorder has been recently identified; carriers receive two
altered copies of the gene, one from each parent. Those who carry only one copy of the gene are not affected; most do not know they are carriers until they have an affected child. One of the four variants, Type D, is more common in families from Nova Scotia, which includes some Cajun families.

Scientists are hoping that the discovery of the NPC1 gene may also contribute to the understanding of atherosclerosis, as well as a basic understanding of how cells process and transport cholesterol.

The discovery of the gene is an example of how research on rare brain disorders pays off in other ways, said Zach Hall, director of the National Institute of Neurological Disorders and Stroke.

"By identifying this gene, we not only take a crucial step forward in understanding this devastating disorder, but also gain insights into problems (such as heart disease and stroke) that affect every one of us."

At this time, there is no cure or effective treatment for Niemann-Pick disease. However, now that the gene has been discovered, scientists are hopeful that genetic tests will be developed and new therapies can be tested.

In Michael's case, the buildup of cholesterol in his cells is affecting how he walks and talks. His gait is somewhat impaired and his hands draw up into fists.

His speech is slurred and he stutters; his short-term memory is also impaired as he begins to suffer some of the same signs of dementia that Alzheimer's patients have.

His IQ has dropped and he goes through sudden mood changes, that include anger and frustration.

Michael's history shows no particular problems in childhood until he hit adolescence, which is not unusual with Niemann-Pick disease, Golden said. He first saw a neurologist at the age of 12 when he began to experience seizures and other neurological problems: drooling, spasticity and speech deficits.

Three of the hallmark signs of the disease include an inability of the eyes to track up and down, movement disorders, and falling episodes.

Neurologist Steven Zuckerman suspected Niemann-Pick disease when he examined Michael and referred him to a specialist in New York, who confirmed the diagnosis.

Michael recently returned from the National Institutes of Health, where he underwent an extensive evaluation. Diagnostic tests include an MRI, a liver biopsy, bone marrow tests and blood cholesterol studies.

"It was such a relief to finally have an answer," Maureen Landry said, "but it was kind of hard, too. Now, I know what's ahead of us. They told me to expect to see Michael's abilities decrease pretty rapidly. All we can do is address quality of life issues.

"It's hard because he should be playing football, but he can't even tie his own shoes anymore," said Landry, a single parent who works as a nurse at the public health unit in Gonzales.

Maureen gets a lot of support from his teachers, relatives and church, as well as relief from respite workers.

"I want to keep him at home as long as I can, although I realize it's going to get more difficult.

"When we went to the NIH, I knew, in my heart, that we weren't going to get any answers. If our contribution can help another child, or family, in the future, because of the research they're doing, then it will be worthwhile.

And it was reassuring to speak with people who knew this disease and to meet with the two other families who were there with their children who had Niemann-Pick disease. We're all in this boat together."

For now, Michael will stay in school — he attends East Ascension High School — and at home and will continue to be evaluated by Golden and his pediatrician, Charles Daniels.

He loves to sing in church and enjoys watching movies, Maureen said. Dreams Come True Foundation has contacted Maureen about applying to grant his dream.

Anyone wanting more information on Niemann-Pick disease can write to: National Niemann-Pick Disease Foundation Inc., 411 N. Diane Court, Chandler, Ariz. 85226.