

ONE FAMILY'S CHALLENGE: CDG

For children with a rare disorder, medical answers don't come easily

by Ellen Mitchell
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When Johnny Lickel was born, his tiny hands did not clench as newborn's hands usually do. The doctors labeled it low muscle tone and kept Johnny in the hospital for tests over the next six days.

"He was our first child. You dream of being wheeled out of the hospital with your baby. We had to leave him there," said Michelle Lickel, of Massapequa.

"We cried for two days," said her husband, Bob.

Over time, Johnny was delayed in rolling over, in crawling and in sitting up; he didn't take a step until 27 months. He had the first of numerous seizures at 15 months and today at age 6, he cannot speak normally, nor is he toilet trained. Throughout his first four years, Johnny underwent test after test administered by mystified doctors. The tests revealed only some slightly abnormal liver enzymes.

"They did EKGs, EEGs, MRIs, blood tests," said Bob Lickel.

"Often they couldn't find the veins and they poked him over and over. It was horrible," said Michelle Lickel.

Always the answer came back, "We have no idea what's wrong with Johnny."

When the Lickels were on the verge of giving up, their general pediatrician, Dr. Stacey Shapiro of Woodbury, urged them on.

"There are some parents who want to keep pushing and others who say they've had enough. They don't want to put themselves through the agony of watching their child go through another test," Shapiro said.

"I remember saying to them, 'He's your first child. I want to keep going on this before you get pregnant again. I want to find out if there is something genetic.'"

When Johnny was 4 and still undiagnosed, the Lickels' daughter, Arianne, was born. During the pregnancy, amniocentesis found no abnormalities and the baby appeared healthy at birth.

But, at 14 months, Arianne suffered two seizures. Tests at North Shore University Hospital in Manhasset revealed she, too, had slightly elevated liver enzymes.



"I had to walk in and tell them," Shapiro said, "that we now knew for sure Arianne had a milder case of what John had, but we still didn't know what it was."

"That changed my life right there," said Michelle Lickel.

But, it also led to an answer. Shapiro and neurologist Ingrid Taff of Great Neck researched on the Internet, now armed with the near-certainty that the mystery illness was genetic.

Identified at last

The diagnosis that seemed to match the children's symptoms, and which was later confirmed with blood tests, was carbohydrate deficient glycoprotein syndrome type 1A (CDG), a disease unknown to the dozen or more specialists the Lickels had consulted. There are only a few hundred recorded cases of CDG. It is a recessive trait and both Michelle and Bob unknowingly carried the recessive gene. They had a 25percent chance of having a child with the disease although no one in their families had ever exhibited signs of CDG.

Unfortunately, the answer provided little consolation. There is no cure for CDG type 1A, and no one can predict what lies ahead for the couple's children.

Meanwhile, Johnny takes medication to control his seizures. He has undergone physical, occupational and speech therapy and attends the Hagedorn Little Village School in Massapequa for children with special needs. He is making slow, but steady progress, has begun to read and on several occasions lately has spoken a few words clearly.

When he walked into her office recently and proudly stated, "Hello, Dr. Shapiro," the pediatrician had tears in her eyes.

Arianne gets physical therapy and seems to be progressing at normal speed, although her speech is not easily understood.

"You appreciate what you get," Michelle Lickel said. "We celebrate the little things."

CDG occurs when sugar molecules fail to attach to proteins in the body, causing the proteins to malfunction. The resulting deficiencies vary greatly and can affect basically any body system. There are many rare metabolic diseases; CDG is only one of them. CDG itself has at least 15 different types, of which 1A is the most common.

Marc C. Patterson, head of the Division of Pediatric Neurology at Columbia- Presbyterian Hospital in Manhattan, is one of the few CDG experts worldwide. He is also one of the Lickels' physicians.

CDG's symptoms

Patterson said some CDG children's muscles are so weak they cannot walk without assistance. Some are in wheelchairs. They are typically subject to seizures and stroke-like episodes and have numerous infections in the early years because their immune systems are impaired. Some are mentally retarded.

Symptoms tend to stabilize in the majority of cases, Patterson said. Extremely severe forms of the disease, however, can result in death, as the Lickels have learned from their own extensive Internet research.

A diagnosis of CDG is confirmed with an obscure blood test known as a carbohydrate deficient transferon test, done primarily in this country at the Mayo Clinic in Rochester, Minn., where the Lickels' tests were done. The disease was first identified in Belgium in 1980. Patterson believes there are numerous cases that no one has ever diagnosed.

Researchers have had some success in controlling the symptoms in CDG type 1B, which affects the gastrointestinal tract, by giving patients mannose or fucose, two forms of sugars. However, that treatment has not been effective with other types of CDG.

Patterson said dietary approaches may one day lead to some treatment, and "ultimately I guess people would like to replace or repair the defective DNA with gene therapy. That's controversial, but I think it's the big hope for the future."

The Lickels and their doctors say public awareness is crucial. The more undiagnosed cases that are found, they say, the greater the research funding will be and the greater the understanding and interest in this virtually unknown disease.

"When a family has a child with something horrible like leukemia or cystic fibrosis, they can talk to other people about it," said Patterson. "But, if they say, 'My kid's got CDG1A,' no one knows what they're talking about. That's a big difficulty for families."