Group meets to cope with problems of rare disorder

By LAURIE SMITH ANDERSON
Advocate staff writer

Ten-month-old Ethan Walters is just now fitting into newborn clothes.
Weighing less than 4 pounds at birth, Ethan now weighs about 9 1/2 pounds, is eating well and is trying to crawl, although that’s difficult because he’s missing his lower arms and hands.

Ethan has Cornelia de Lange Syndrome, a rare genetic disorder believed to be caused by a defective gene that causes delays in physical and mental development.

“We knew from prenatal testing that his arms were not fully formed, but nothing else showed up on the amniocentesis, so we thought he was going to be a normal baby,” said Ethan’s mother, Candace Walters.

“We were devastated when he was diagnosed with CdLS. I had never heard of the syndrome and the doctors painted a very grim picture for us. In fact, we didn’t expect him to live. I remember thinking ‘our family is so normal; this happens to other families,’ ” said Walters, who is now the state awareness coordinator for the Cornelia de Lange Syndrome Foundation.

The Walters and 20 other families in Louisiana affected by CdLS and listed with the foundation will meet for the first time at 10 a.m. on Sept. 20 at Greenwood Park, with a trip to the Batou Rouge Zoo to follow. Invitations have been sent out to all the families in Louisiana who are registered with the foundation; any others are urged to call Walters in Slidell at (504) 781-1955.

Most children born with CdLS bear a resemblance to each other, according to the foundation. Common characteristics include low birth weight, delayed growth and small stature, as well as small head size. Typical facial features include eyebrows that meet at midline, long eyelashes, upturned nose and thin, downward lips. Other frequent findings are excessive body hair, small hands and feet, seizures, heart defects, cleft palate, gastrointestinal problems and developmental delays. Limb abnormalities, such as Ethan’s, are not uncommon.

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Because the disorder varies greatly in severity from one case to another, it is difficult to make a long-term prognosis for anyone affected.

However, the syndrome usually involves significant delays in speech and communication, some degree of mental retardation and other developmental delays.

CdLS was first identified in 1933 by Dr. Cornelia de Lange, a Dutch pediatrician. The incidence is believed to be about one in 20,000 live births. The syndrome is thought to be caused by a genetic mutation, though it is rarely passed directly from a parent to a child.

“These children have their own growth and development charts,” Walters said. “Their average age to talk is 3; to walk is 5. Ethan has hearing aids in both ears because his ear canals are so small that it’s difficult for sound to penetrate.”

Ethan was born a month early and had to be aspirated to start breathing on his own. He was diagnosed with a heart murmur, had feeding difficulties and was not released from the hospital until he was 1 month old.

He has undergone two operations so far for reflux and has a feeding tube in place; his digestive problems have been complicated by hernias which will also need to be repaired surgically.

A geneticist diagnosed Ethan with CdLS the night after his birth. Walters was given the number for the foundation and she immediately called for information.

“They sent me a packet of information and a list of families I could contact. That was so helpful because, even though I had the support of friends and family, I needed to talk to other parents who had been through what I was going through. Next year, the foundation is holding its annual meeting in Minneapolis and we’re already planning to go.”

Ethan’s father, Patrick Walters, is in the military and the couple, formerly of California, decided to move back to Louisiana this year to be closer to family.

Walters has chosen not to go back to work until Ethan is older. He gets physical therapy, occupational therapy, speech therapy and special education services every week because early stimulation is critical in order to help him reach developmental milestones. Walters supplements his therapy at home.

“I get so excited when he does something new. He smiled for me first. He’s really a happy, sociable baby except when he’s in pain from his reflux problems.”

The toll-free telephone number for the Cornelia de Lange Syndrome Foundation in Connecticut is 1-800-223-8355. The foundation provides information and support services, sponsors an annual international convention and publishes a bimonthly newsletter.