

A Family Faces A Rare Condition - Urea Cycle Disorder Makes A Child's Life A Daily Battle

By JULIE SHAPIRO

Courant Staff Writer

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NEW BRITAIN -- Shortly after C.J. Farr's first birthday, an ambulance rushed him to Connecticut Children's Medical Center. No one knew exactly what was wrong with him - just that he had 10 times the normal amount of ammonia in his blood.

"He was doing bad in the ambulance," said Jennifer Farr, C.J.'s mother. "They didn't think he was going to make it."

The metabolic team at Children's stabilized C.J., and several days later, doctors made a final diagnosis: C.J. has a rare genetic condition called a urea cycle disorder. He is missing an enzyme that helps digest protein. As a result, excess ammonia builds up in his blood and can rapidly cause brain damage, coma and death.

"He should not have survived," Farr said. When she asks doctors why C.J. is alive today, they tell her, "Just call it a miracle."

C.J., now 21/2, is one of only two children in Connecticut living with a UCD. There is no cure, but medication and a low-protein diet keep C.J. stable, most of the time.

Approximately one in 10,000 children is born with a UCD, according to the National Urea Cycle Disorders Foundation. The prognosis is often grim, particularly for young boys. The Foundation estimates that as much as 20 percent of sudden infant death syndrome cases are actually caused by a metabolism disorder such as a UCD.

The rarity of UCDs makes them more dangerous. Farr can't count on emergency room doctors knowing what to do for C.J. - often, they've never even heard of UCDs.

Last year, the Farrs were in Texas at the UCD foundation's annual conference. They had just boarded the plane home when Farr noticed that C.J. was going limp. She rushed him off the plane and to the nearest hospital, where she thrust a packet of information into the hands of the first emergency room nurse she saw.

"[The nurse] took one look at it and handed it to another nurse," Farr said. "She said, 'I'm not touching this.' They don't want to do the wrong thing."

Once, a doctor tried to X-ray C.J.'s chest for pneumonia, misunderstanding Farr's explanation of C.J.'s dangerous ammonia levels.

"They have no idea," Farr said. "Sometimes I feel lost, but I can't get mad, because they just don't understand."

Often, doctors rely on Farr's and her husband Christopher's expertise.

On C.J.'s most recent trip to the emergency room, the doctor flipped through the packet, then handed it back to C.J.'s parents. "Tell me what to do," he said to them.

"My husband and I have saved C.J.'s life many times by being educated," Farr said. "It would be great if the doctors knew more and I could put my trust in them rather than them putting their trust in me."

C.J. also plays an active role in keeping himself healthy. When he sees a Burger King commercial on TV, he points to the close-ups of cheeseburgers and announces, "Too much protein!"

When C.J. vomits, he knows that it means more than just a stomach ache. "Gotta go get a boo-boo now?" he asks his mother, referring to his frequent blood tests. Other times, after he gets sick, he asks his mother whether she's going to call an ambulance.

C.J. isn't always accepting of his disorder. Farr runs a day-care center out of her home, and one child recently brought brownies to share with everyone. Farr had to tell C.J. that he couldn't have one. He got upset and threw a fit, so she sent him to his room for a time out. Once in his room, C.J. kept screaming, "I just want a brownie!"

"I started crying," Farr said. "I feel so bad that he can't be a normal 3-year-old."

Even when C.J. has had months of good blood work, it is impossible for Farr to forget his condition. She analyzes every bite that C.J. eats to make sure that he gets precisely 6 grams of protein each day. She buys special low-protein food for C.J. online. She mixes his powdered medication into a vitamin-rich formula that she puts into an IV and pumps into C.J.'s stomach. She even smells him, because his glands emit a sour odor when his ammonia count is too high.

C.J.'s daily care does not come cheap. One of his medications, Buphenyl, costs \$3,000 a month, and C.J. will need it for the rest of his life. C.J.'s low-protein food costs far more than the normal foods available at a supermarket. A pound of low-protein pasta, for example, costs \$7.58.

After a small battle, C.J.'s insurance company agreed to cover everything.

Despite C.J.'s expensive treatments, his body's careful balance can still get knocked askew. The culprit can be a minor illness, such as a fever; or a bit too much or too little protein; or seemingly nothing at all.

At the beginning of C.J.'s episodes he vomits and is sometimes combative and hyperactive. As the ammonia builds in his brain, he hallucinates and screams for his mother even though she's right beside him. Then, suddenly, he becomes lethargic and goes limp, and nothing will wake him up.

"All the scares are just as scary," Farr said. "I wonder, 'Is his body going to be able to handle this?'... He could die next week, but I don't like to think that way."

Instead, Farr and her family focus on their blessings. The Farris' two other children - Alexis, 6, and Hannah, 5 - each had a 50 percent chance of inheriting a UCD from their mother, who is a carrier. However, both girls are healthy, and neither are carriers.

The Farris are also grateful that, unlike many children with UCDs, C.J. has sustained little or no brain damage during his high-ammonia episodes.

In February, C.J. and his mother will go to Washington, D.C., to participate in a study that Farr hopes will improve the chances for children such as C.J. Doctors will evaluate how C.J. is responding to his medication and his low-protein diet, and they will test his mother for symptoms of a UCD.

Farr looks forward to working with the premier experts in the country. "They're superstars," she said.

Meanwhile, life continues at the Farr residence. C.J. is an energetic boy who loves trucks and anything to do with construction, his father's job. He knows all the words to Rod Stewart's "Have You Ever Seen The Rain" and sings along. He will start preschool next fall.

C.J. helps his parents keep everything in perspective. "We're very thankful because we know things could have been different," Farr said. "When my husband and I are stressing out about something, we just look at each other and say, 'It could be worse.'"

Contact Julie Shapiro at jshapiro@courant.com. Further information about UCDs is available at www.nucdf.org.

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