

Help Isn't Just for Kids at Children's

By John Kelly

Wednesday, December 28, 2005; Page C11

Antonietta Stromberg of Johnston, R.I., is on the phone telling me what it's like to have a brain full of ammonia. It's not good.

"I was doing crazy stuff, like somebody high on drugs," the 56-year-old housewife and bookkeeper says.

She's talking about a day 14 years ago.

"I had to go out and make a deposit at the bank and do some shopping," she says. "I never got there. I never knew to this day what happened to that deposit."

She doesn't know how she got home that day, either, just that her car was back in the driveway and she looked bad and felt awful.

"My husband said, 'Why don't you go lie down?'"

"I said, 'I'm fine. I'm gonna make supper.' He set the table. I started supper."

Nobody had a chance to eat.

"I must have taken everything off the table and put it in the freezer. I put supper in the freezer."

Then she got into bed, backward.

"My head was where my feet should have been."

Her husband, *Joseph*, called 911.

"The police came," says Antonietta. "The rescue came. The fire department came."

They all came. She wouldn't talk to any of them.

"I didn't know who I was."

At the hospital, they asked if she knew where she was. "Was I in an accident?" she answered.

"I got worse. I kept passing out, throwing up. I couldn't sit up. They couldn't figure it out. . . . My head hurt so much I wanted to die."

Then she went into a coma. A doctor took Joseph aside and said, "If she does come out of it, she's gonna be a vegetable. If I were you, I'd go home, rest up and make arrangements for your wife's funeral."

Says Antonietta: "My husband threw him out. He said, 'No, she's too young.' "

Antonietta spent four days in a coma. When she came out, doctors had a rough idea what was wrong with her: She had what's known as a urea cycle disorder.

The body is a little chemical factory, creating chemicals that it needs, neutralizing others that it doesn't. The protein we eat is composed of amino acids, which are used for growth and the production of hormones, among other things.

As the amino acids are broken down by the body, they release ammonia. Enzymes in the liver convert the ammonia to harmless urea, which healthy people eliminate. But a person with a urea cycle disorder can't

pass the ammonia. Because the molecule is very small, it can pass through the blood-brain barrier, bathing the brain in ammonia-rich blood.

Doctors in New England told Antonietta that she had a type of urea cycle disorder called OTC. They eliminated protein from her diet and switched her to calorie-rich foods. Soon she was taking 1,800 pills a month.

But she was never quite right. For more than a decade, she was in and out of the hospital constantly.

"Finally a doctor said, 'If someone doesn't figure out what's going on, you're probably going to die in your sleep. You won't feel any pain, you just won't wake up.'"I said, 'You gotta do something.' "

Urea cycle disorders are normally diagnosed in children because it's rare for those with the illness to make it into their forties, as Antonietta had, without being swamped by symptoms. It's a hereditary problem, the result of a faulty gene that both of her parents carried.

Children's Hospital in Washington is one of 10 centers in the country funded by the National Institutes of Health to study rare diseases. *Dr. Mendel Tuchman* took a look at Antonietta's history and called her husband. "I can save her life," he said.

Tuchman confirmed that Antonietta didn't have OTC after all, but a urea cycle disorder he had just discovered called n-acetyl glutamate synthetase deficiency, or NAGS.

NAGS is what's known as an "orphan disease," one that affects fewer than 200,000 Americans. Tuchman had FDA permission to experiment with a European drug known as Carbaglu that substitutes for the missing enzyme and reduces ammonia levels.

Last December, Antonietta traveled to Children's, where she was one of the hospital's oldest outpatients. Under Tuchman's watchful eye, she swallowed some Carbaglu pills. They had an immediate, and miraculous, effect.

"She was completely normalized," said Tuchman's colleague *Dr. Mark Batshaw*.

"That's what keeps Dr. Tuchman and I going," Batshaw said, "the possibility of seeing these miracles occur. Unfortunately, they don't occur as often as we like. But we savor the moment when it does."

So does Antonietta.

"It'll be a year today that I've been feeling great," she says before we hang up. "A year."