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All-out effort on autism

As diagnoses have risen sharply, researchers are pulling together to find the cure

BY JAMIE TALAN
STAFF WRITER

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Ten-year-old Connor Gould and his brother, Bryan, 13, both have been given a diagnosis of autism. Their conditions are not identical: Connor doesn't speak. Bryan does, but in a manner that has prevented him from having a shot at a normal childhood.

The boys' parents, Laurie Murdock and David Gould, are far from alone in wondering why their sons have the condition. The number of cases of autism has grown by 172 percent since 1990, according to the Autism Society of America, and scientists are trying to determine why.

The National Institute of Child Health and Human Development is calling together scientists from various fields - toxicologists, immunologists, neurologists, psychiatrists and public health specialists - to track down the triggers for this serious brain illness. And physicians from six leading medical centers, including Columbia University Medical Center in Manhattan, have launched an Autism Treatment Network, a cooperative effort to develop and test treatments being partially funded by a national organization called Cure Autism Now.

In addition, last month the U.S. Centers for Disease Control and Prevention announced a campaign to diagnose autistic children earlier, hoping that early interventions could help alter the condition's trajectory. Most children are not diagnosed until age 3, a time frame the CDC campaign wants to cut in half.

Looking back to pregnancy

All these efforts come as parents are pressing for an answer: What caused my child's autism?

Children with autism share extreme language disturbances, self-absorption and inability to relate, highly repetitive play, rage reactions, and a predilection for rhythmic movements.

At the Murdock-Gould household in Port Washington, there is a thread that, when pulled, unravels what the couple believes is a possible cause: mercury.

During Laurie's pregnancy with Bryan, doctors learned that mother and child had incompatible blood types. They gave her a shot of Rh immunoglobulin, which prevents the mother's immune system antibodies from cross-reacting with the baby's blood. The medicine contains mercury.

But the debate over whether mercury plays a role is heated and unsettled. It has focused on thimerosal, the mercury-based preservative used in childhood vaccines, which was phased out starting in 1999. The national Institute of Medicine has concluded that evidence doesn't support a connection between vaccination and autism, but

acknowledged "they could not rule out" such a link.

Clues from mercury studies

Two recent studies lend support to the mercury theory. The "Baby Haircut" study, conducted at Arizona State University, studied hair samples from children exposed to thimerosal during development and found evidence that their bodies were not eliminating mercury.

And in a study of animals exposed to low doses of mercury, as well as PCBs and other environmental toxins, Columbia University's Dr. Mady Hornig found that those with a compromised immune system were far more susceptible to developing autistic-like behavior.

The immune system is a prime target for a new generation of autism researchers. S. Jill James, a professor of pediatrics at the University of Arkansas for Medical Sciences, has been studying an antioxidant pathway in the body that connects directly to immune function. Studying blood plasma from 95 autistic children and 75 healthy volunteers, James and her colleagues found a two-fold reduction in the active form of glutathione, a cellular antioxidant critical for the immune system, the brain and stomach lining. (One third of children with autism have gastrointestinal problems, which have gone unexplained.)

Natural body chemicals that help make glutathione were also abnormal, "as if the cells are working hard to get rid of the oxidized form of glutathione," James said. The result of this imbalance: The immune system responds as if it's attacking itself, she theorizes.

Preliminary results indicate this glutathione abnormality may be reversed when patients are given certain vitamins - including methyl B12, trimethyl glycine, and an active form of folic acid, she said.

Wide-ranging research

Columbia's Dr. Ian Lipkin is leading a study to identify markers for risk. Working in Norway, he and colleagues have recruited 100,000 pregnant women, collected blood and spinal samples and will follow the children for the first four years of life. They will have genetic information, infection histories during pregnancy, blood from the umbilical cord, plus repeated examinations and parent surveys.

"I am confident we will find biomarkers for autism," said Lipkin, who is seeking funding to expand the trials.

Researchers at Vanderbilt University in Nashville, Tenn., are studying baby siblings of autistic children for risk factors. Other Vanderbilt scientists are hunting for genetic factors.

Researchers at the Institute for Basic Research on Staten Island have been collecting tissue samples at autopsy from the brains of people with autism, and are trying to identify pathological clues. This research is funded by the National Alliance for Autism Research, a group founded by parents of autistic children.

Quest for a "magic pill"

The mystery of the disease itself has prompted an array of "treatments," from specialized diets to chelation therapy, designed to leach mercury from the body. Scientists worry that the latter approach can have dangerous side effects. Columbia's Hornig said some of these children have lost use of their kidneys and ended up on dialysis, and some have died.

The Autism Treatment Network was formed, in part, to point out that untested treatments may not be worth the risks. "Collaboration through the Autism Treatment Network should improve the quality of treatment and increase awareness of treatment options," said Dr. Thomas Insel, director of the National Institute of Mental Health.

"If your kid has autism, you'll try anything," said Laurie Murdock. "I just want the magic pill."

Murdock and her husband, David, whose father was Pulitzer-prize winning composer Morton Gould, said dozens of fringe therapies are commonly tried, yet not backed up by the rigors of scientific testing.

There is a treatment that most people agree works. Applied behavioral analysis has been shown to help children learn how to respond to their environment with words. The technique is time-consuming and tedious, calling for hours of daily instruction by teachers who break learning into very basic components and reward students for each positive step. Experts say the process could actually help rewire the brain.

The Mosaic School - co-founded by Murdock and Robin Havens, whose child Jake also is autistic - opened in September. The Port Washington school serves a handful of students, including Connor. Applied behavioral analysis is a big component of their work. "We have to help these children get along in the world without us," Murdock said.