MAY 2006

brain BRIEFINGS

NO LONGER CONSIDERED RARE, THE LATEST ESTIMATES INDICATE THAT THOUSANDS OF CHILDREN ARE BORN EACH YEAR WITH THE ISOLATING BRAIN DISORDER AUTISM. UNFORTUNATELY, MANY FAMILIES MAY REMAIN IN THE DARK FOR YEARS BEFORE AUTISM IS PROPERLY DIAGNOSED AND TREATED. NOW, HOWEVER, NEW RESEARCH IS LEADING TO A GREATER UNDERSTANDING OF THE CAUSES AND NATURE OF THE DISORDER, WHICH SHOULD LEAD TO IMPROVED TREATMENT. NEW INFORMATION INDICATES THAT EARLY DIAGNOSIS IS KEY, AND THAT SEVERAL GENES MAY BE INVOLVED. GENETIC STUDIES ALONG WITH IMAGING STUDIES MAY LEAD TO BIOLOGICALLY BASED DIAGNOSTIC TECHNIQUES THAT COULD HELP SPEED DETECTION AND ALLOW EARLY, MORE EFFECTIVE INTERVENTION.

SOCIETY FOR NEUROSCIENCE

AUTISM IN CHILDREN

He acts aloof and withdrawn, and doesn't understand social cues. An introvert, his parents suspect. He also seems oddly obsessed with highways and recites road-related facts. A quirky little guy, they think.

Later, however, a doctor diagnoses the child with autism.

Approximately one out of every 250 to 166 babies will develop autism or a related disorder, which can impair a child's ability to interact socially and communicate. The brain disorder affects individuals differently, but all show abnormal responses to other people. Babies with autism also tend to start speaking later than normal, if at all. Those who can communicate may talk a lot about something they really like instead of having a back-and-forth conversation.

Key to helping these children is the early detection of their autism, followed by specialized assistance. Once confirmed, several methods may be employed to treat and control the disorder. New research is leading to:

- A better understanding of the nature of autism and the major role of genetic factors.
- The development of biologically based diagnostic techniques that could help identify autism earlier, and thus improve treatment.

▼ **POST-MORTEM STUDIES AND RECENTLY DEVELOPED IMAGING TECHNIQUES HAVE HELPED TO IDENTIFY SOME OF THE MAJOR BRAIN AREAS IMPLICATED IN AUTISM. THE AREAS INCLUDE THE CEREBELLUM, CEREBRAL CORTEX, AND TEMPORAL LOBE—PARTICULARLY THE AMYGDALA. THE DISORDER MAY RESULT FROM THE FAILURE OF VARIOUS PARTS OF THE BRAIN TO WORK TOGETHER.**

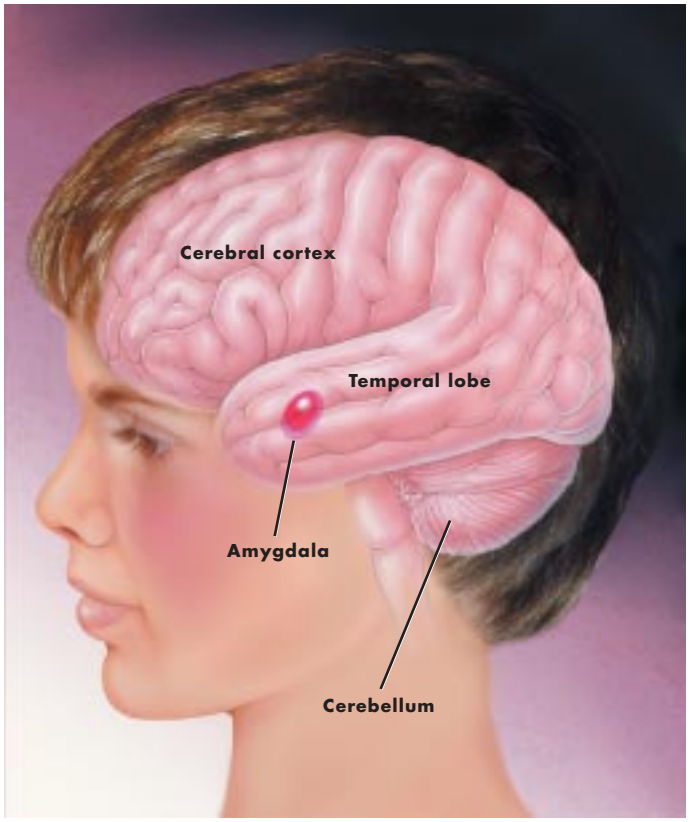

IMAGE BY LYDIA KIBIUK.

Today, autism cannot be identified biologically with a simple medical test like a brain scan or a blood test. Instead, a diagnosis is made on the basis of behavioral symptoms observed or discussed in the doctor's office. Many children spend their first years of life undiagnosed and untreated. Forty percent of children with autism wait more than three years for a clear diagnosis, according to one survey. Other research indicates that most children are accurately diagnosed by 2 to 4 years of age, but in some cases not until age 6. Researchers believe that reducing the time lag in diagnosis may help children gain the full benefit of available interventions.

Although there is no cure,

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research indicates that many with autism can respond well, especially early in life, to highly structured, specialized education programs designed to correct behaviors, teach social skills, and aid language. The young brain is thought to be particularly adept at modifying its connections and function. Researchers believe that starting interventions early may take advantage of this malleability and improve a person's function.

To positively diagnose autism, two components are recommended. One is a "well child" check-up that includes a developmental screening test for a range of behaviors involving speech, social skills, and unusual movements. Then, if indicators of autism are found following this screening, a team of experts conducts a comprehensive evaluation.

In order to help speed detection and start treatment earlier, scientists recently began to scrutinize the brains of those with autism and uncover specific biological signs of the disorder. Converging evidence from multiple groups of scientists suggests that portions of the autistic brain are actually enlarged in early life. Research continues on the use of medications to treat behavioral disturbances in autism.

In other work, scientists hope to gain insight into the roots of the brain changes by studying genes and brain tissue. Our genes control brain development and function. Research suggests that several abnormal genes, including some that may alter brain architecture, likely play a role in autism. While no single gene for autism exists, about five genes recently have been identified as contributing to some cases, suggesting that multiple genetic differences work together to promote the disorder. Genes may interact with environmental factors, which also could play a role.

Some of these genes appear to exert their effect at the synapse—the junction where nerve cells communicate with each other. Other work indicates that a gene may be responsible for controlling the number of nerve cells in a structure called the cerebellum that sits at the back of the brain (see illustration) and may contribute to abnormal brain function in autism. Once the genes that increase a person's risk of developing autism are clearly determined, scientists should be able to develop a simple blood test that establishes whether a newborn harbors any of these abnormal genes. Eventually, researchers also may be able to design methods that counter the genes' actions and treat the disorder, further breaking children free from the world of autism.

As research moves forward, children with autism will be diagnosed sooner, treated with a greater array of options, and start to connect with peers, parents, and loved ones.

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