

Understanding the Complexities of Autism

Approximately 1 in 150 eight-year-olds in the United States has autism or a closely related disorder. The risk for this complex brain disorder is three to four times higher in boys than in girls.

U.S. medical schools and teaching hospitals, together with the National Institutes of Health (NIH), are working to better understand autism and the milder autism spectrum disorders (ASDs) that affect so many children.

The following examples of recent NIH-funded research advances achieved at these institutions hold promise for children struggling with autism, as well as their parents and loved ones.

2008

Children's Hospital Boston

Premature infants born weighing less than 1,500 grams (approximately 3 pounds, 4 ounces) have an increased risk for showing early signs of autistic features. Researchers suggest that early autistic behaviors may be under-recognized in infants with very low birth weights, and early screening for signs of autism could be warranted for all preterm infants.

Cincinnati Children's Hospital Medical Center

Unconventional diets could put boys with autism and ASDs at a higher risk of having thinner, less-dense bones than boys of the same age who do not have these disorders. Researchers believe that boys with autism and ASDs are at risk for poor bone development for several reasons, including lack of exercise, reluctance to eat a varied diet, digestive problems, and diets that omit casein, a protein found in dairy products. Casein-free (nondairy) diets are thought by some to lessen the symptoms of autism.

Johns Hopkins University School of Medicine

A team of investigators found that a common genetic alteration appears to be associated with autism only when passed down to sons from their mothers. The alteration, which occurs in a gene called CNTNAP2, is one of the strongest and most common known genetic links to autism susceptibility. This discovery will help guide future studies on understanding autism.

University of California, Davis, School of Medicine

Researchers have identified endocrine dysfunction in children with early onset autism. Examining molecular biomarkers may prove valuable as early diagnostic tools, and provide insights into molecular characteristics in autistic children.

2007

University of Texas Southwestern Medical Center

After discovering two proteins involved in autism, researchers engineered mice with mutations in a gene for these proteins. These mice, the closest animal model to autism, exhibit poor social skills but increased intelligence, as do people with some forms of autism. The proteins, called neuroligins, link brain nerves together.

Vanderbilt University Medical Center

Few rapid screening tools are available that encompass the range of symptoms commonly occurring in autism. Researchers have developed and evaluated a screening checklist—the Parental Concerns Questionnaire (PCQ)—that can accurately identify symptoms of autism.

Johns Hopkins University School of Medicine

A landmark analysis of genomic data, released in 2007, provided the most detailed look thus far at genetic variation patterns in families with autism. The study included 1,250 autistic individuals and their siblings and parents from across the country.

University of Pennsylvania School of Medicine

Researchers analyzed different treatments prescribed for children with autism spectrum disorders who were enrolled in Medicaid in 2001, and examined factors associated with psychotropic, or mind-affecting, medication use. Study results determined that children with ASDs who were male, older, white, in foster care, or diagnosed with more than one psychiatric condition were more likely to have used psychotropic drugs; children in geographic areas with fewer white residents or greater urban density were less likely to use such medications.

2006

Vanderbilt University School of Medicine

Researchers have identified a genetic mutation that increases the risk of autism; the mutated gene is common in children with autism.

Johns Hopkins University School of Medicine

University of Illinois at Chicago College of Medicine

There is a common genetic alteration that leads to autism only when inherited by sons from their mothers. Findings from this research discovery were later replicated in one of the largest-ever group of autism samples ever studied.

Arkansas Children's Hospital

University of Arkansas for Medical Sciences

Scientists at the Autism Metabolic Genomics Laboratory discovered that many children with autism have low levels of glutathione, the major intracellular antioxidant and mechanism used to detoxify environmental contaminants.

2005

**University of North Carolina at Chapel Hill School of Medicine
Duke University School of Medicine**

Results of MRI scans show that, by age 2, children with autism show a generalized enlargement of their brains. The temporal lobe, where language is controlled, was enlarged, along with other parts of the brain. It is not yet clear whether the enlargement is a primary cause of autism or a downstream effect of another process occurring in the brain.

University of California, Davis, School of Medicine

Children with autism have different immune system responses than nonautistic children. This discovery is important evidence that autism, currently defined primarily by distinct behaviors, may potentially be defined by distinct biologic changes as well.

Vanderbilt University Medical Center

The results of two research studies imply that mutations in the gene that regulates brain levels of serotonin may be a risk factor for autism. In individuals with autism, regulatory problems within the gene may disrupt serotonin signaling.

University of Louisville School of Medicine

Researchers found that individuals with autism have smaller and more numerous cortical minicolumns (structures that serve as the brain's microprocessors) than do individuals without autism.

2004

Johns Hopkins University School of Medicine

Scientists found new evidence that the brains of some autistic individuals show clear signs of inflammation, suggesting that autism may be associated with activation of the brain's immune system.

For more information about how medical schools and teaching hospitals are fulfilling the promise of medical research, go to: www.aamc.org/ftp