

THE DEPARTMENT OF HEALTH AND HUMAN SERVICES  
NATIONAL INSTITUTES OF HEALTH

Examining the Federal Response to Autism Spectrum Disorders

Witness before the

House Oversight and Government Reform  
Subcommittee on Government Operations

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## Introduction

Good morning Chairman Mica, Ranking Member Connolly, and distinguished Members of the Subcommittee. I am Dr. Tom Insel, Director of the National Institute of Mental Health (NIMH) at the National Institutes of Health (NIH). I have served as the Chair of the Interagency Autism Coordinating Committee -established by the Combating Autism Act of 2006 (CAA), and reauthorized by the Combating Autism Reauthorization Act of 2011 (CARA), since my arrival at NIMH in 2002.

progress we have seen since the passage of the CAA and the inception of the IACC. We at NIH are very grateful for the strong support that you in the Congress have always shown for NIH and the thousands of researchers around the country who are working to advance biomedical research in support of people living with a wide array of diseases, disorders and disabilities. As Chair of Federal Agencies represented on the committee for your continued support for our efforts on autism spectrum disorder and related disabilities. I am here to provide an update on the Federal response to autism, including the work that has been done by the IACC to coordinate Federal activities and foster public-private collaboration, and to provide a snapshot of the considerable progress being made in autism research. More details on the specific programs and projects funded by the various Federal Agencies involved in the autism effort can be found in the recently submitted *Report to Congress on Activities Related to Autism Spectrum Disorder and Other Developmental Disabilities Under the Combating Autism Act of 2006 and Combating Autism Reauthorization Act of 2011 (FY2010-FY2012)*.

## Background on Autism Spectrum Disorder (ASD)

Autism spectrum disorder (ASD ) is a neurodevelopmental condition characterized by deficits in social interaction and social communication, along with restricted interests and repetitive behaviors sometimes

accompanied by additional features such as intellectual disability or language impairments. With varying degrees of severity in these symptoms, autism is a heterogeneous condition that affects some with only mild impairments and others with profound disabilities. Like many other neurodevelopmental disorders, autism is thought to be caused by a combination of genetic and environmental factors in this case, by genes and environmental factors that influence the development of the brain. Currently, there are only a small number of proven causes of ASD, including genetic mutations associated with several well-characterized genetic disorders such as Rett Syndrome, tuberous sclerosis and Fragile X, and other rare genetic syndromes. While each of these causative mutations is rare, the discovery of different rare mutations associated with ASD is increasing so quickly that in a recent report, the American College of Medical Genetics and Genomics (ACMG) strongly expressed their support for genetic testing to be routinely provided for individuals who have autism without a known cause, because currently available tests are likely to be able to identify a specific genetic mutation underlying autism symptoms in an estimated 30-40 percent of individuals. Identification of contributing gene mutations could result in benefits for the individual, including better

Overall, does the upward trend in CDC prevalence estimates over the last several years represent a true increase in the number of children with



children, but higher than for Black and Hispanic children in Minneapolis. Overall, the combined prevalence for the Minneapolis children was one in 48, which is higher than most recently published ASD prevalence estimate of one in 68. Somali children with ASD were also found to be more likely to have intellectual disability than children with ASD in all other racial and ethnic groups in Minneapolis (100 percent of the Somali-American children with ASD who had IQ records on file showed an intellectual disability in comparison to only 20-

## Research Highlights

With the coordination provided by the IACC and its *Strategic Plan*, NIH and other agencies within HHS and other Federal departments have been working collaboratively to tackle the challenges of supporting research on this profoundly complex condition. Investment in ASD research over the last decade has increased 90 percent to \$190 million in Fiscal Year 2014.

Some of the most important research investments related to ASD have not been specific to ASD but have created tools or resources for studying brain development, new insights about the immune system, or research on the microbiome that may transform ASD research.

share just a few examples of the scientific progress that has been made toward understanding autism and developing new clinical approaches over the past five years. This brid

The technology advances that are revolutionizing neuroscience are rapidly being incorporated into the autism field, with the promise of greatly deepening our understanding of autism.

One ground-breaking new method developed by NIH-funded researchers, called CLARITY, takes intact postmortem human brain samples, donated with appropriate consent,



University of California at San Diego that found scattered patches of disorganized brain cells in the deep layers of the brain cortex in samples from children with autism also converge on the second trimester of fetal development as a critical time-point in the development of autism, indicating that the origins of autism are present before birth.

If autism begins before birth, why are we making the diagnosis after age 3? A number of new scientific findings from prospective longitudinal studies are helping us make significant progress in the area of early identification. For example, NIH-funded researchers using eye-tracking technology determined that children who later go on to develop autism exhibit a distinct pattern of decline in eye contact with caregivers that is detectable between the ages of two to six months of age. In another study, in slightly older infants and toddlers with autism, from 14-42 months of age, the use of eye-tracking technology revealed pronounced differences in attention to social cues; when given a choice between watching a video of a friendly human face interacting with them versus one showing a moving geometric pattern, the children who eventually developed autism preferred to focus on the geometric pattern. Other studies have demonstrated that children who later develop autism show measurable differences in repetitive behaviors (such as

develops new social behaviors indicates that the brain is or able to adapt and remodel itself to learn new skills and that behavioral therapy is actually brain processes social information. In another study, researchers evaluated children receiving the Early Start Denver Model therapy approach, or ESDM. ESDM focuses on social exchange, social attention, social engagement, and positive affect. Randomized trials have shown that this intervention results in significant improvement in IQ, language, and adaptive behaviors. Very compelling new evidence has also shown that this technique results in normalized patterns of brain activity, as measured by electroencephalography, and that these patterns correlate with improvement in behavioral outcomes. This suggests that ESDM is actually remodeling the brain to respond to social stimuli in a different way perhaps by strengthening existing neuronal circuits or building new ones to compensate in areas where function is reduced and this adaptation results in improved social behaviors.

Pharmacological treatments for autism are in earlier stages of development, but work has intensified in this area. In 2008, only six drug treatment clinical trials were underway. That number is now around 100. ASD is a relatively new area for clinical trials research. Working out the design and the appropriate outcome measures has been an ongoing discussion between NIH and FDA as well as with colleagues at Autism Speaks and scientists involved in ASD clinical trials in Europe. These discussions are helping us to improve trial design and ensure the highest rigor of science along with the best protection of participants. As the dialogue continues and ongoing clinical trials proceed, we expect to have more rapid progress on medications and other interventions for autism in the near future.

In order for a medical treatment to be effective, it must address the problem at hand. What we from person to person. The ultimate goal for autism treatments is that they will soon be defined by biological indicators of the underlying cause, or biomarkers, which will help with both diagnosis and the development of effective treatments.

symptoms, several large national and international efforts to accelerate the discovery of autism biomarkers have been launched. The Foundation for the NIH manages the Biomarkers Consortium, which is a private-public partnership to identify and develop biomarkers to help prevent, diagnose, and treat a variety of conditions such as autism. The Biomarkers Consortium has organized a targeted search for and refinement of biomarkers for ASD, which will unite funding agencies, academic researchers, and pharmaceutical companies. The Consortium is also working with international partners from European Autism Interventions - A Multicentre Study for Developing New Medications, which is the largest single grant for autism in the world at over \$38 million, to focus discovery of autism biomarkers with the ultimate goal of creating effective and personalized treatments for autism.

NIH continues to support its Autism Centers of Excellence

Research has suggested that people with ASD are often under

the implementation of the *IACC Strategic Plan*. In addition, Coordinating Committee has played an important role in helping NIH institutes coordinate their efforts to ensure that areas of the *IACC Strategic Plan* that fall within the NIH mission are being covered, and to foster cross-institute collaborations and prevent duplicative efforts.

As a result of this investment in autism research and our intensive efforts to coordinate and to foster collaboration, over the past few years we have seen remarkable progress in autism research. We have made tremendous advances in our understanding of how autism unfolds during the course of early development, in the identification of factors that may be contributing to increased or decreased risk for autism, and in developing and testing new screening/diagnostic tools, treatments and interventions, and services approaches that can be used in a variety of populations and community settings.

With the availability of unprecedented tools and technologies, we are poised to make significant scientific discoveries that can be translated into the next generation of tools and services to improve the quality of life for people on the autism spectrum. With several promising early results, there is also a need for more replication to validate research findings. Continued focus on coordination and collaboration with external partners will be essential to help us achieve the objectives in the *IACC Strategic Plan*. With sustained support and continued public-private collaboration, the IACC and its members can continue to work steadily toward the eventual collective community goal.

