2012 INTERAGENCY AUTISM COORDINATING COMMITTEE STRATEGIC PLAN UPDATE: QUESTION 1 WHEN SHOULD I BE CONCERNED?

WHAT IS NEW IN THIS RESEARCH AREA, AND WHAT HAVE WE LEARNED IN THE PAST 18 MONTHS?

Prevalence

Several noteworthy new studies from the U.S., South Korea, and England update the recognized prevalence of autism.

In the U.S., the Autism and Developmental Disabilities Monitoring Network (ADDM) released their most recent surveillance data showing a prevalence of 1.1/100 children—an increase of 78% since the first report in 2002—with larger increases among racial/ethnic minority groups (ADDM, 2012). The average age when children were initially diagnosed with ASD remains essentially unchanged—at 4-5 years of age. A second U.S. study found a sharp increase in autism in children (based on parent report) over an 11-year period—an increase that was not found for other neurodevelopmental disorders other than attention deficit hyperactivity disorder (Boyle et al., 2011).

A South Korean study evaluated a large population sample of school-aged children and found an ASD prevalence of 2.6/100 children (Kim et al., 2011). Notably, two-thirds of the children had not been previously diagnosed with autism or received any services.

A British study found a prevalence of 1/100 in adults, most of whom were not previously diagnosed with ASD (Brugha et al., 2011). The large majority of adults with ASD identified in this study were living independently but at lower levels of success than non-ASD peers.

Taken together, these studies suggest that some but not all of the increased prevalence in children is a result of improved identification and that there may still be a sizable population of children and adults with undiagnosed autism.

Diagnosis

The Diagnostic and Statistical Manual of Mental Disorders (DSM) definition of autism will be revised in 2013 when the new version, DSM-5, is released. The goal is to consolidate all current diagnoses under one category (autism spectrum disorder) while making its criteria more appropriate to diagnosis of very young children, adults, and girls. While early studies raised concerns that the new criteria would exclude some people from diagnosis, recent findings suggest this will not happen, and the other goals of the revision will be achieved. Specifically, similar rates of diagnosis were reported in a study based on well-characterized research samples (Huerta et al., 2012) and in preliminary unpublished results of the DSM-5 validation studies presented to the Interagency Autism Coordinating Committee (DATE?). However, there is still a need for prospective studies of the new criteria with larger sample sizes and no data have yet been collected on adults.

Comment [A1]: Discussion Point: Reference needed.

Comment [A2]: Discussion Point: This was just published – Geri has the reference to be added JER

New analyses suggest that the method by which diagnostic information is collected is just as important as criteria. Diagnoses that include information from both clinician observation and parent or caregiver report are more accurate than diagnoses that rely on one approach or the other alone (Huerta et al., 2012; McPartland et al., 2012).

Variations in diagnostic data from the infant sibling studies (Ozonoff et al., 2011), the CDC surveillance network sites (ADDM, 2012) and the Simons Simplex Collection sites (Lord et al., 2011) suggest that clinical diagnoses still remain more variable than they should. This situation may ultimately be resolved by the development of lab tests for autism biomarkers, refined by observation and reporting. Although the criteria for diagnosis are behavioral, the hope is that medical evaluation will identify subtypes and decrease this variability.

Early Screening and Detection

Work on early screening tools has accelerated, with one study accurately identifying some children with ASD as early as 12 months in a community setting (Turner-Brown et al., 2012). However, researchers continue to have difficulty attaining sufficient sensitivity without excessive rates of false positives. Specificity - accurately distinguishing ASD from other developmental disabilities or typically developing children – also remains a challenge.

Simplified ASD assessment tools for use across the lifespan are in development (Allison et al., 2012), but have not yet met the need to appropriately identify adolescents and adults of all ages with ASDs (Pilling et al., 2012).

New research continues to highlight disparities by gender, race, and ethnicity in identification of ASDs (Valicenti-McDermott et al., 2012; Kocovska et al., 2012), while gaps remain in understanding the reasons for these disparities and evidence-based ways to close these gaps.

Early Diagnosis

Symptoms of autism may not be visible in the first year of life, and currently are not measured reliably until the end of the second year. Several groups are looking for performance-based measures to bridge that gap. Pierce et al. (2011) found a very strong correlation between a preference for fixating on geometric images and a later diagnosis of autism in toddlers as young as 14 months. Bedford et al. found an association between atypical eye contact at 6-10 months and a later diagnosis of ASD. Another study correlated differences in vocalization at 6,9, and 12 months with a diagnosis of ASD at 24 months (Paul et al., 2011).

Temperament in the first two years (increased perceptual sensitivity, reduced cuddliness) may also predict a later diagnosis of ASD (Clifford et al., 2012).

Wolff et al. used brain imaging to identify white matter fiber tract development differences in 6-month-old infants who would later be diagnosed with ASD (2012). This suggests that aberrant development of brain pathways may precede the manifestation of autistic behaviors in the first year of life.

Comment [A3]: **Discussion Point:** This is also in Question 2. Do we want this in both chapters?

Other research explored EEG data as a potential biomarker and quantifier of risk in the first year (Bosl et al., 2012; Elsabbagh et al., 2012). Other studies attempt to detect autism through blood

screening – looking at gene expression (Glatt et al., 2012) or gene pathway analysis (Skafidas et al., 2012).

The Bosl study employed a signal processing approach – multiscale entropy – to determine if a neural "signature" of autism risk could be observed. At 9 months of age, they were able to classify high vs. low risk with over 80% accuracy, but it is uncertain whether this is predictive of which infants develop ASD. Elsabbagh et al. found that atypical event related potential (ERP) responses to eye gaze in high risk infants predicts which infants developed ASD. It is too early to tell if this work translates to the general infant population, but it may point the way to very early identification of ASD in children from known at-risk groups.

Comment [A4]: Discussion Point: This is also in Question 2. Do we want this in both chapters?

WHAT GAPS HAVE EMERGED IN THE PAST 18 MONTHS?

The age at which autism is diagnosed in most children has not changed, even though diagnosis is now possible significantly earlier. Better diagnostic tools and skills have been developed, but their existence has not yet translated to earlier detection in the general population.

Data systems – using the existing ADDM infrastructure or elsewhere – to understand how the upcoming DSM-5 changes impact diagnosed prevalence and age at identification are recommended. Autism Speaks has funded a study to prospectively examine the impact of DSM-5 criteria on diagnosis at one ADDM site, but more studies are needed with both younger and older individuals.

The American Psychiatric Association (APA) has proposed a new disorder – A04 Social Communication Disorder – to describe people with communication problems whose severity is not enough to warrant an ASD diagnosis. We recognize the community's concern that there are no therapies or services currently associated with SCD. There is a fear that it will be interpreted as "mild ASD without supports."

The Brugha et al. study suggests that a significant portion of the adult ASD population remains undiagnosed, despite the existence of screening tools and increasingly widespread public awareness of autism. Unrecognized adults with autism have emerged as an overlooked and underserved population.

Some studies show that adults with autism continue to be socially disadvantaged and have significantly lower academic and career attainments as compared to non-ASD adults in similar surroundings (Brugha et al., 2011; Henninger et al., 2012). Autism is a lifelong disability, yet research efforts to date have primarily focused on childhood and adolescent detection and intervention. More emphasis must be placed on adults of all ages.

Comment [A5]: Discussion Point: Should this be moved to Question 5?

As the field shifts to pre-symptomatic diagnosis, a series of new, complex bioethical issues are emerging since diagnosis and treatment could be indicated for an infant without behavioral

symptoms. Many in the autism community have expressed concern that research into early detection will lead to prenatal tests for ASD and that the existence of such tests may have family planning implications. On the other hand, very early detection in infants may be one of the keys to provide the best outcomes for people who will grow up with ASD in the future. Prenatal testing could also have significant quality of life implications if it facilitates effective early intervention. Work leading to prenatal tests for ASD potential should be informed by a full discussion of the bioethical issues cited above.

Word count: 1429

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