

Combining Information From Multiple Sources in the Diagnosis of Autism Spectrum Disorders

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ABSTRACT

Background: Standard case criteria are proposed for combined use of the Autism Diagnostic Interview-Revised and Autism Diagnostic Observation Schedule to diagnose autism and to define the broader category of autism spectrum disorders. **Method:** Single and combined Autism Diagnostic Interview-Revised and Autism Diagnostic Observation Schedule algorithms were compared to best estimate diagnoses in four samples: U.S. ($n = 960$) and Canadian ($n = 232$) participants 3 years and older, U.S. participants younger than 36 months ($n = 270$), and U.S. participants older than 36 months with profound mental retardation ($n = 67$). **Results:** Sensitivities and specificities of 80% and higher were obtained when strict criteria for an autism diagnosis using both instruments were applied in the U.S. samples, and 75% or greater in the Canadian sample. Single-instrument criteria resulted in significant loss of specificity. Specificity was poor in the sample with profound mental retardation. Lower sensitivity and specificity were also obtained when proposed criteria for broader spectrum disorders were applied. **Conclusions:** The Autism Diagnostic Interview-Revised and Autism Diagnostic Observation Schedule make independent, additive contributions to the judgment of clinicians that result in a more consistent and rigorous application of diagnostic criteria. *J. Am. Acad. Child Adolesc. Psychiatry*, 2006;45(9):1094–1103. **Key Words:** autism diagnosis, Autism Diagnostic Interview-Revised, Autism Diagnostic Observation Schedule.

Autism research has benefited from opportunities to define samples by diagnostic instruments such as the Autism Diagnostic Interview-Revised (ADI-R; Rutter et al., 2003)

and the Autism Diagnostic Observation Schedule (ADOS; Lord et al., 1999). Standardized methods of collecting, coding, and summarizing information result in categorical diagnoses of autism or not autism on the ADI-R, a caregiver interview, and in classifications of autism, broader autism spectrum disorders (ASDs) or nonspectrum on the ADOS, a semistructured observation. These two instruments were intended to be used together, yet there has been no systematic attempt to evaluate how information from the instruments should be combined for diagnosis.

The core characteristics of autism are deficits in communication and social reciprocity accompanied by behavior that is restricted or repetitive (*DSM-IV-TR*; American Psychiatric Association, 2000). The ADI-R and ADOS were developed to operationalize these criteria to identify characteristics that differentiated autism from cases without autism that were equivalent in chronological age and language level. Individual items were not selected for the ADI-R and the ADOS

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algorithms because of their ability to discriminate autism from other ASDs or to discriminate more broadly defined ASD from other disorders. Genetics research has indicated that the boundaries of what is transmitted familiarly, however, extend beyond autism as operationally defined on the ADI-R and ADOS (International Molecular Genetics Study of Autism Consortium [IMGSAC], 2001; Le Couteur et al., 1996). This has led to increasing interest in including in research individuals who do not meet criteria for autism, but who share many of the same characteristics (Constantino et al., 2003). Several different operational definitions of “almost autism,” ASD, and pervasive developmental disorder not otherwise specified exist (Buitelaar et al., 1999; International Molecular Genetics Study of Autism Consortium [IMGSAC], 2001). Unfortunately, studies have indicated that interrater reliability in distinguishing nonautism ASD from autism and in distinguishing ASD from nonspectrum disorders is often poor (Lord et al., 1999; Szatmari et al., 2002).

The purpose of this article is to propose standard criteria for the combined use of the ADI-R and ADOS to diagnose cases of autism and to identify a broader category of ASD cases that may have less pervasive or milder symptoms. Data are also presented about the instruments’ performance with very young children and for individuals with profound mental retardation. In a clinical setting, wrongly denying a child access to services may be the greatest concern, whereas in genetic analyses, wrongly designating unaffected individuals as affected may have more negative consequences. Consequently, data for alternative methods are presented so that these considerations can be taken into account when selecting criteria.

An inherent difficulty in establishing caseness criteria is determining the gold standard to which classifications derived from the diagnostic instruments are compared. Because reporting clinical diagnoses based on information other than the ADI-R or ADOS when those instruments were used is impractical, our solution was to seek replication across different sites. In this study, data are reported from U.S. and Canadian centers that used different strategies for determining consensus best estimate (BE) diagnoses. Standardized administrations of the diagnostic instruments were performed at both sites, but how the information was used to determine diagnoses differed. In the U.S. samples, consensus BE diagnoses were not independent of the diagnostic

instruments. In almost all cases, a psychologist conducted or observed both the ADI-R and the ADOS and summary information from the ADI-R and ADOS was available to physicians who participated in the diagnoses. In the Canadian sample, consensus BE diagnoses were made by physicians and psychologists who had not been directly involved in the ADI-R or ADOS administration but who had access to the clinical information from these instruments.

METHOD: STUDY 1

Participants

Data were collected from 1,039 participants who completed a diagnostic evaluation at the University of Chicago Developmental Disorders Clinic ($N = 627$; 497 males, 130 females), the University of Michigan Autism and Communication Disorders Center ($N = 150$; 115 males, 35 females), as part of a longitudinal study conducted through TEACCH Centers at the University of North Carolina, Chapel Hill ($N = 129$; 100 males, 29 females) and the University of Chicago ($N = 80$; 69 males, 11 females), or in a study of individuals with disorders other than ASD ($N = 53$; 37 males, 16 females). Only participants with known developmental, cognitive, or behavioral diagnoses were included. One hundred twenty-six participants (12%) were affected siblings. The sample was 82% white, 13% African American, 4% Asian American, and 1% other or multiracial. Participants with visual, hearing, or motor impairments that precluded standard administration of an instrument were excluded. Parents signed an institutional review board–approved informed consent form to participate in research before actual participation.

The majority of the 1,039 participants received ASD diagnoses. However, 158 (15%) of the participants had diagnoses other than ASD (41% nonspecific mental retardation, 25% language disorder, 14% oppositional defiant disorder and/or attention-deficit/hyperactivity disorder, 12% Down syndrome, 7% mood and/or anxiety disorder, and 1% Tourette’s syndrome). About 60% of nonspectrum participants had been referred for possible ASD; the remaining 40% were recruited into research comparison groups.

For 182 participants (18%), more than one full assessment was available. No differences were found for separate analyses of all data compared with analyses with only the most recent assessment for each participant. Thus, all 1,297 assessments with contemporaneous ADI-R and ADOS administrations were included. Age at assessment ranged from 14 months to 18 years, with a median of 58 months of age. All sites in this study primarily evaluate individuals referred for possible ASD.

The largest data set consisted of 960 assessments of participants at least 36 months old who had a nonverbal mental age of at least 18 months (Table 1). This data set included cases with clinical diagnoses of autism (540 assessments), pervasive developmental disorder not otherwise specified (PDD-NOS; 252 assessments), Asperger disorder (5 assessments; most cases with a diagnosis of Asperger disorder had a research diagnosis of autism, which was given precedence), or a nonspectrum disorder (163 assessments). In addition, performance of diagnostic criteria for autism and ASD compared with nonspectrum disorders was examined separately for children younger than 36 months’ chronological age (270 assessments) and for participants older than 3 years of age with profound mental

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