

Should All Children Be Screened for Autism Spectrum Disorders?

Yes: Merging Science, Policy, and Practice

PAUL H. LIPKIN, MD, *Johns Hopkins University School of Medicine, Baltimore, Maryland*

SUSAN L. HYMAN, MD, *University of Rochester School of Medicine, Rochester, New York*

The Centers for Disease Control and Prevention has declared that autism spectrum disorders (ASDs) are “an urgent public health concern,” with recent estimates that one in 110 children has an ASD.¹ Before routine screening was recommended, the median age of diagnosis was 5.7 years,² despite 70 to 95 percent of children having identifiable symptoms by three years of age.¹ The primary care physician is challenged to identify children at risk of ASDs before they enter school, with referral for early diagnosis and effective intervention when developmental delays are detected.

Screening for ASDs in primary care was advanced by the American Academy of Pediatrics (AAP) through its policy statement on developmental surveillance and screening, and its clinical report on the identification and evaluation of children with ASDs.^{3,4} The guidelines aim to identify infants and toddlers at risk of developmental delay or disability, leading to further diagnosis and treatment in federally mandated early intervention programs. The AAP recommends developmental surveillance at every preventive care visit, with additional use of standardized general screening tests at the nine-, 18-, and 30-month visits, and ASD-specific screening tests at the 18- and 24-month visits, because general screening tests may not identify toddlers with ASDs.^{3,5} Continued surveillance for ASD symptoms through school age is also recommended because higher functioning children may not be identified as symptomatic until social demands become more complex.

The recommendations for ASD screening were based on promising results using



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the Modified Checklist for Autism in Toddlers (M-CHAT), a parent questionnaire with a reported sensitivity of 87 percent and positive predictive value of 80 percent.⁶ Subsequent evaluation of the M-CHAT has revealed weakness, with a positive predictive value as low as 11 percent (or a false-positive rate of 89 percent) when used as a questionnaire alone; however, the positive predictive value increases to 65 percent (false-positive rate of 35 percent) with a structured interview follow-up. For example, if the initial evaluation is positive, then a second visit with the physician or office staff would be scheduled to answer additional questions and possibly refer the child for further testing and intervention.⁷

Screening tools that have since been developed may be more specific while also identifying children at risk of other developmental disorders. One example, the Infant-Toddler Checklist, screens for ASDs and language delay in children nine to 24 months of age, with a positive predictive value for language disorders higher than 70 percent and sensitivity for ASDs higher than 90 percent.⁸ With its use in screening more than 10,000 children who were one year of age in 137 pediatric practices, the positive predictive value remained high at 75 percent.⁹ Community nurses achieved sensitivities of approximately 70 to 85 percent by combining ongoing developmental surveillance for ASD symptoms and observation

of behaviors as a direct form of screening.¹⁰ These and other studies now indicate that ASD screening improves with repeated surveillance using screening tests and structured observations aimed at identifying the signs of ASDs in infants and toddlers.¹¹

Should the primary care physician perform routine ASD screening if it overidentifies children requiring further assessment? Although false-positive screening tests may result in stress, anxiety, and the added expense of diagnostic evaluation, other developmental disorders (e.g., language disorders, global developmental delay) are often diagnosed in children who falsely test positive for an ASD.^{8,12} False-positive rates can also be decreased with a follow-up interview visit. Children who continue to screen positive should be referred for evaluation using validated cognitive and language tests; those younger than three years can be referred to a local early intervention program, and those three years or older can be referred to their school district. Specific diagnostic testing for ASDs should be pursued simultaneously through a developmental professional.

Does such screening impose other burdens on the primary care physician? Developmental screening has not added to the length of the preventive care visit.^{13,14} Physician concern about liability associated with such screening has not been borne out, and developmental screening is included as standard of care in the Medicaid Early & Periodic Screening & Diagnostic Treatment program and Bright Futures guidelines. Finally, parents of affected children prefer early diagnosis and treatment. These parents consistently report dissatisfaction with the care provided by their physicians, including lack of expertise using screening tools and delay in acting on their concerns.^{15,16}

Children strongly suspected to have an ASD should also be evaluated for other disabilities and for an underlying cause. In a recent multistate cohort of more than 2,500 children with ASDs, 83 percent also had another developmental disorder, 10 percent had a psychiatric condition, and 16 percent had an underlying neurologic problem.¹⁷ Clinically relevant genetic abnormalities identified by chromosomal microarray are present in up to 7 percent of children with ASDs.¹⁸ Further

testing may be important in finding the cause of an ASD and in the medical monitoring of the affected child and family members.¹⁹

Screening offers the potential for early treatment, which can lessen the effect of a condition. This is true for ASDs and related disorders in which randomized controlled trials have demonstrated improvements in young children with ASDs with regard to parent-child social communication,²⁰ socially engaged imitation,²¹ joint attention,²² and core behavioral symptoms of autism.²³ A recent systematic review of early intensive interventions also found improvements in cognitive performance, language skills, and adaptive behavioral skills in young children with ASDs using behavioral interventions or more comprehensive approaches using developmental and behavioral frameworks.²⁴ Diagnosis of ASDs is reliable in children younger than three years,²⁵ permitting access to early treatment and potentially improving outcomes. Therefore, early and ongoing ASD screening is appropriate and important in primary care. Until science identifies all the causes of ASDs, allowing for prevention or even a cure, health care professionals who provide care to young children must offer the continuum of developmental surveillance, screening, diagnosis, and intervention for these common developmental disorders.

Address correspondence to Paul H. Lipkin, MD, at lipkin@kennedykrieger.org. Reprints are not available from the authors.

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