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Improving Screening for Autism Spectrum Disorder: Is It Time for Something New?

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Although autism spectrum disorder (ASD) can be reliably detected by age 2, the average age of diagnosis ranges from age 4 to 6, depending on the population.¹ Because earlier access to treatment is thought to improve outcomes,2 delays in diagnosis may lead to avoidable morbidity. In an effort to lower the average age of diagnosis, considerable efforts are being made to promote early ASD screening.^{3–5} Two predominant strategies exist: broad-based developmental surveillance and targeted ASD screening.6,7 Although there are data to support each strategy, targeted ASD screening has recently gained popularity. The American Academy of Pediatrics, for example, recommends all children receive an ASD-specific screener at their 18and 24-month well-child visit, and it proactively supports these guidelines with resources and trainings.3

A number of studies support ASDspecific screeners (the Modified Checklist for Autism in Toddlers [MCHAT] and the Modified Checklist for Autism in Toddlers-Revised [MCHAT-R]) for detecting early ASD risk.⁷ Yet, few longitudinal cohort studies of ASD screening exist, and none of the studies, to our knowledge, has reported on the longitudinal follow-up of screen-negative children until now. In the current study by Øien et al,8 the authors managed 68 197 children who screened negative for ASD using the MCHAT. These children were also screened with the broadbased Ages and Stages Questionnaire (ASQ)⁹ and the Emotionality, Activity,

and Sociability Temperament Survey (EAS).¹⁰ The authors found that children who screened negative on the MCHAT at 18 months but went on to receive a later ASD diagnosis (falsenegatives) demonstrated significantly greater deficits in the developmental and temperamental domains of the ASQ and EAS compared with those with a negative MCHAT screen who did not receive a subsequent ASD diagnosis (true-negatives). The authors assert that even among children who screen negative on the MCHAT, those with ASD frequently display early signs and symptoms. On the basis of this assertion, they conclude that improved screening methods (specifically with more sensitive instruments) may enhance early ASD detection.

Given the publication of this well-designed, population-based study, what does it ultimately mean for ASD screening? Does it lend further support to the American Academy of Pediatrics guidelines recommending ASD-specific screening at 18 and 24 months? Is the MCHAT the appropriate screening tool, or are more sensitive tools needed? Finally, will these data help fill the knowledge gaps cited in the 2016 United States Preventive Services Task Force (USPSTF) statement of insufficient evidence relative to ASD screening?

Unfortunately, the answers may not be straightforward. The current study reveals that young children with ASD, even with a negative MCHAT screen, display some signs and symptoms consistent with ASD that can be detected by

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existing, non-MCHAT screening tools. The authors conclude, "To maximize opportunities for early ascertainment...screening instruments should be refined to improve their capacity for identifying the patterns of deficits...in early life." Or, more succinctly, more sensitive screening instruments are needed.

In light of the findings in the study, such a conclusion is logical. However, the data presented reveal that the MCHAT has a sensitivity of 23% in this population, dramatically lower than that reported in other studies. For example, in the 2016 USPSTF review on ASD screening,7 the 2 included studies in which MCHAT or MCHAT-R sensitivities were reported revealed them to be 75% and 91%, respectively. If the sensitivity of the MCHAT-R is truly 91%, 11 it is hard to argue that more sensitive tools are needed. Of note, the MCHAT was recently replaced by the MCHAT-R.¹¹ The MCHAT-R was developed to improve usability and decrease the false-positive rate of the MCHAT. Therefore, the difference in tools is unlikely to account for the

low sensitivity demonstrated in the current study.

What then accounts for the low sensitivity of the MCHAT in this population? Possibilities include timing (in the current study, MCHATs were evaluated only at 18 months), differential follow-up across falseand true-negative screens, or population differences between the Øien et al⁸ study and previous work. Regardless of cause, the discrepancy between MCHAT sensitivities reveals we may need to temper the conclusions of the investigators that more sensitive ASD screening tools are needed. More importantly, with a sensitivity that is dramatically lower than that reported in other studies, it calls into question the generalizability of the current sample and whether the ASQ or EAS would stand up in other populations.

These criticisms withstanding, the current study does send a clear warning that the MCHAT likely does not identify all manifestations, or clinical phenotypes, of ASD equally. The data begin to address an important gap cited by the USPSTF

(but likely not in the direction many concerned with ASD screening were hoping). The findings lend credence to the USPSTF concern that "clinical and convenience samples do not adequately demonstrate the psychometric properties of screeners in practice." In this study, Øien et al⁸ reinforce the notion that screened and clinical populations may be systematically different and that more research is needed to understand such differences.

ABBREVIATIONS

ASD: autism spectrum disorder

ASQ: Ages and Stages Questionnaire

EAS: Emotionality, Activity, and Sociability Temperament Survey

MCHAT: Modified Checklist for Autism in Toddlers

MCHAT-R: Modified Checklist for Autism in Toddlers-Revised

USPSTF: United States
Preventive Services
Task Force

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