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Early Autism Detection: Are We Ready for Routine Screening?

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KEY WORDS

child development disorder, pervasive screening programs, early diagnosis, health care

ABBREVIATIONS

ASD—autism spectrum disorder

ADI-R—Autism Diagnostic Interview, Revised

CHAT—Checklist for Autism Spectrum Disorders in Toddlers

M-CHAT—Modified Checklist for Autism Spectrum Disorders in Toddlers

SCQ—Social Communication Questionnaire

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abstract

FREE

BACKGROUND. Autism is a serious neurodevelopmental disorder that has a reportedly rising prevalence rate. The American Academy of Pediatrics recommends that screening for autism be incorporated into routine practice. It is important to consider the pros and cons of conducting autism screening as part of routine practice and its implications on the community. We have explored this question in the context of screening from a scientific point of view.

METHOD: A literature search was conducted to assess the effectiveness of community screening programs for autism.

RESULTS: Judged against critical questions about autism, screening programs failed to fulfill most criteria. Good screening tools and efficacious treatment are lacking, and there is no evidence yet that such a program would do more good than harm.

CONCLUSIONS: On the basis of the available research, we believe that we do not have enough sound evidence to support the implementation of a routine population-based screening program for autism. Ongoing research in this field is certainly needed, including the development of excellent screening instruments and demonstrating with clinical trials that such programs work and do more good than harm. *Pediatrics* 2011;128:000

Autism (or the autism spectrum disorders [ASDs]) refers to a group of serious neurodevelopmental conditions with major, life-altering implications. The emotional impact can be devastating for people diagnosed with autism and their families. Life is often stressful for the family of a child with autism, because parents and siblings usually have to cope with challenging behavioral and developmental issues. In addition, the financial burden for health care is notably higher for families with children with autism than for children with other special health care needs.¹ Children with autism have difficulty managing in social situations, which isolates many of their families. Autism has considerable impact on current and future participation in society, school, and employment. For example, in a study of the long-term prognosis of 68 adults with autism who had IQs of >50 in childhood, the outcome was described as poor for 46% and very poor for 12%.² Despite these realities, these families and children often experience long waiting periods for therapy or special education services.

A diagnosis of autism may be important to help parents get access to services, including early intervention. The American Academy of Pediatrics recently recommended that screening for autism be incorporated into routine practice.³ Thus, the question that pediatricians must ask themselves is: should we incorporate autism screening as part of routine practice? There are few criteria available in the literature for screening of clinical disorders such as autism. In this article we consider the appropriateness of screening from the perspective of the features and requirements of such programs, guided by the criteria outlined by Cadman et al.⁴

EPIDEMIOLOGY

The reported prevalence of ASDs has risen dramatically over the past few

years. The number of children diagnosed with autism has been estimated to have grown from 0.8 cases per 1000 in 1983 to 4.6 cases per 1000 in 1999⁵ and to a more current prevalence of parent-reported diagnosis of autism at 11 per 1000 in school-aged children in 2007 with a 4:1 male-to-female ratio.⁶ This increase may reflect improved detection and recognition of autism and its variants; it could be attributed to changes in the diagnostic practice, including the need for this diagnosis to access services; or it may reflect an actual increase in the prevalence of autism.⁷

In today's society, the public and physicians are increasingly aware of autism. Information about autism is pervasive in the media and readily available, which almost certainly contributes to considerable alarm among both parents and health professionals. Parents of children with ASDs often recall having developmental concerns by the age of 12 to 18 months, yet confirmation of diagnosis generally does not occur until 3 to 4 years of age or older. In view of the increased awareness of autism, it is important to consider the pros and cons of conducting autism screening as part of regular practice and the implications for the community at large if such screening were to be implemented.

SCREENING PROGRAMS

Screening is a public health service intended to detect a specific medical condition in people who do not necessarily perceive that they are at risk of or already affected by that condition or its complications. A screening questionnaire or test is meant to help identify affected people who are more likely to be helped than harmed by further diagnostic tests or treatment. Established long ago for myriad diseases, childhood screening programs for conditions such as congenital hypothyroidism and phenylketonuria

have proved extremely beneficial for early detection and intervention. The success of these screening programs rests, to a large extent, on the availability of appropriate screening tests, the acceptability of the screening process, known efficacious treatments for these conditions, and the recognized severe consequences for the child of missing early detection and disease-altering intervention.

It is important to note that there are differences between screening and clinical surveillance. With screening, apparently healthy people are targeted to help them make better-informed health care choices. However, screening itself might involve some health risks, so one must weigh the benefits against risks to give the public realistic expectations of what a screening program can deliver. Clinical surveillance, on the other hand, involves the targeted use of diagnostic tests and questionnaires to either rule out or rule in a diagnosis among people who have a relatively high probability of having that condition by virtue of having already been identified in some way. In the specific context of autism, surveillance would involve children seen in pediatric clinics with symptoms or specific parental concerns.

For a community screening program to be effective, certain criteria must be met. Cadman et al⁴ have outlined specific questions that must be addressed and answered thoughtfully to determine if screening is appropriate, feasible, and valuable for a specific condition. These questions are posed and discussed in order with reference to screening for autism.

Question 1: Has the Effectiveness of the Screening Program (for That Condition) Been Demonstrated in a Randomized Trial?

Unlike breast cancer screening, which has well-documented benefits,⁸ no au-

tism screening programs have been studied in randomized controlled trials. There is no solid research evidence on which to base the recommendations of the American Academy of Pediatrics in "Identification and Evaluation of Children With Autism Spectrum Disorders"³. This clinical guideline built on the previously published policy statement "Identifying Infants and Young Children With Developmental Disorders in the Medical Home: An Algorithm for Developmental Surveillance and Screening."⁹ Heightened public awareness, screening tools, and evaluation instruments becoming more reliable probably all contributed to this recommendation.

Question 2: Are There Efficacious Treatments for the Primary Disorder and/or Efficacious Preventive Maneuvers?

After a diagnosis of autism, parents, caregivers, and professionals want effective interventions to be available. Intensive, sustained special education programs and behavioral therapy early in life can apparently help some children with autism acquire self-care, social, and job skills that improve daily functioning, decrease symptom severity, and reduce maladaptive behaviors.¹⁰ However, early intensive behavioral intervention has, at best, produced modest results in selected subgroups of children; the findings are judged to be based on research of variable but almost always poor methodological quality.^{11–13}

There are many available therapeutic approaches to childhood autism,^{14,15} including educational interventions, applied behavior analysis,¹⁶ structured teaching,¹⁷ parent-mediated intervention,¹⁸ speech and language therapy,¹⁰ social skills therapy,¹⁹ and pharmacologic therapy.²⁰ Most children with autism are treated with combinations of these therapies.

Unfortunately, studies of the effectiveness of most interventions have major methodologic problems, which results in lack of evidence to support recommendations in clinical guidelines on autism.¹⁵ These problems include few participants and short-term follow-up that prevent definitive conclusions about efficacy.¹³ Autism is a difficult diagnosis to make because it is, similar to cerebral palsy, a neurodevelopmental disability or phenomenological disorder, not a specific disease. The diagnostic process may differ from one clinician to another depending on which diagnostic tools are being used, which makes comparison across studies somewhat difficult.

Because autism is a heterogeneous condition, further classification of its types and severity is fundamental for research and clinical practice. Unlike the Gross Motor Function Classification System for Cerebral Palsy,^{21,22} there is no valid and reliable functional stratification system with which to assess which children might benefit in what ways from specific interventions.^{23,24} Different therapies may target different symptoms of autism. An ideal therapy would target social, behavioral, and communication defects. In addition, measuring change in social, behavioral, or communicative function requires evaluative tools that have been validated for their capacity to detect change.²⁵ The changes that have been observed in those studies that have reported "positive" outcomes in a child's capacities have rarely been shown to have generalizability to the performance in daily lives of children with ASDs.

Applied behavior analysis, a therapy widely used to treat autism, consists of up to 30 to 40 hours/week of 1-on-1 intervention. The Lovaas program and other well-known behavioral therapy programs often seek to enhance daily living, community living, academic,

and social skills and limit aberrant behaviors such as self-injury and aggression. Systematic reviews of clinical trials of the effectiveness of applied behavior intervention programs on cognitive, adaptive behavior, and language development in preschool-aged children with autism revealed that compared with standard care, applied behavior intervention did not significantly improve the cognitive outcomes of children.^{12,13,15} Results of a recent systematic review suggest that cognitive behavior therapy is an effective treatment for anxiety in people with Asperger syndrome but not for those with other ASD subtypes.²⁶ Thus, more clinical trials are needed, but at present, claims regarding the efficacy of applied behavior analysis have not been substantiated.

A program called Treatment and Education of Autistic and Related Communication Handicapped Children (TEACCH) has been running for many years.²⁷ TEACCH was recently studied over a 3-year period with a group of 34 male children with autism and severe mental retardation in a residential center, at home, and at mainstream schools. The results showed statistically significant changes in all Vineland Adaptive Behavioral Scale (VABS) scores in the home and residential settings groups but not in the mainstream schools group. Although promising, the results of this single study may have been a biased estimate of effect, because it was undertaken in a specific setting, and controlled clinical trials would be needed to validate its apparent benefits.

A systematic research review that focused on parent-implemented interventions for children with ASDs aged 1 to 6 years revealed that there is sufficient evidence that parent training can work in terms of observed improvements in children's social communication skills.²⁸ Moreover, the authors of

this review concluded that parent training does work to produce a positive effect on children's social communication behavior, parental performance, and parent-child interactions. Results of other trials of parent-mediated interventions, including those from a recently published randomized controlled trial that compared the efficacy of the addition of a Preschool Autism Communication Trial (PACT)) intervention to treatment as usual, have shown that these programs might help families interact with their children with autism and increase parental satisfaction.^{15,29,30} None of the early-intervention programs yet reported in the autism literature is an adequately designed study of effectiveness, so no conclusions can be drawn on the question of whether parent-implemented intervention works in broad community practice.

There is little support for the effectiveness of speech and language therapy for people with autism. Most communication support is provided through augmentative and alternative communication methods, which do not seem to impede speech. From their randomized controlled trial, Kasari et al³¹ reported the benefits of joint attention intervention and symbolic play intervention in a sample of 58 autistic children diagnosed according to the Autism Diagnostic Interview, Revised (ADI-R) and Autism Diagnostic Observation Schedule (ADOS) criteria. Howlin et al³² investigated the effectiveness of the use of the Picture Exchange Communication System (PECS) in 84 elementary school children with ASDs in a randomized controlled trial. Future studies will need to determine the long-term effects of such intervention and the carry-over effect in daily activities of the children's lives.

Pharmacologic therapy is available as a treatment but not for the core features of autism, such as language and

social impairment. Pharmaceuticals are used in ASD therapy to treat attention-deficit/hyperactivity disorder, aggression, irritability, self-injurious behavior, and stereotyped, repetitive behavior.¹⁵

In conclusion, at the present time, neither proven therapies nor preventive measures exist for the universal treatment of children and youth with autism, and further intervention research is needed. The best evidence of the benefits of therapy would come from the results of randomized controlled trials, ideally in clinically homogenous groups, and would include evidence of the carry-over of therapeutic effects beyond the specific skills that are being targeted. These conclusions raise a fundamental question: what do we aim to achieve when we screen for a nonpreventable condition for which there are currently no well-validated intervention strategies?

Question 3: Does the Current Burden of Suffering Warrant Screening?

The severity of autism can vary widely, although almost all families of children with autism face major challenges. Children with autism can have an impaired ability to use and interpret nonverbal behaviors such as eye-to-eye gaze, facial expressions, gestures, and body postures. Consequently, autistic children have difficulty reading nonverbal emotional cues. In addition, children with autism fail to develop peer relationships appropriate to their developmental level. In fact, younger children may have little or no interest in developing friendships, which might change as they become older and more interested in social interaction. This lack of understanding of the social boundaries and the needs of others leads to awkward interactions and inappropriate behavior. Children with autism also have sig-

nificant impairment in reciprocal social interaction. Adding to that, their delayed or deviant language development, including both receptive and expressive aspects, makes social connections challenging for them and others around them.

Higher-functioning people with autism do communicate and have relatively normal understanding and use of phonetics, grammar, and some aspects of the meaning of language. However, they might have difficulty recognizing their listener's perspective, realizing meaningful implications of what is said, and understanding and conveying shades of meaning. They might also have difficulty appreciating humor.

Adults with autism continue to have problems with language, social skills, and self-sufficiency. Although authors of follow-up studies often report selected subsamples of people with ASDs, they come to the same conclusion: few people with autism can live as fully self-supporting adults. Approximately 25% of people with autism function fairly well as adults, but the majority of them function poorly. Over the years, the proportion of people with autism finding employment has increased. However, the majority of those jobs are poorly paid.³³ In addition, people with autism often develop psychiatric disturbances in adulthood, including affective disorders and obsessive-compulsive disorder.

All of these factors make it apparent that there is a considerable burden of suffering associated with the diagnosis of autism.

Question 4: Is a Good Screening Test Available?

A successful population-based screening program requires not only that the condition have characteristics identifiable with screening but also that a valid screening test be available. Ideally, the test should be widely acces-

sible, simple to administer, inexpensive, and associated with minimal discomfort and morbidity to the population screened. Moreover, the screening test results must be valid and reproducible.

Of particular concern in autism, the sensitivity of the autism screening tools (correctly identifying persons who actually have the disorder) and the specificity (correctly identifying persons who do not have the disorder) vary depending on the age of the child and the severity of symptoms. Screening tools that are appropriate for toddlers may be less sensitive when used for preschoolers or school-aged children.³⁴

The Checklist for Autism Spectrum Disorders in Toddlers (CHAT) was developed for primary screening of 18-month-olds in the United Kingdom.³⁵ CHAT includes 14 items related to joint attention and pretend play, 9 of which are reported by the parents and 5 of which are directly observed. CHAT has a high specificity of 98% (ie, it successfully excludes the diagnosis of autism in almost everyone who does not have it), but a low sensitivity of 20% to 38% (ie, it does not effectively identify children who do have autism). The results depend on whether a 1- or 2-stage screening protocol is followed. When administered to a population of 16 235 children at 18 months, the CHAT failed to detect children with mild symptoms and those with a regressive pattern of autism. In addition, the CHAT did not discriminate well between children with global developmental delays and ASDs. Sensitivity increased when the CHAT was administered at 24 months. However, low sensitivity overall and the need to observe 5 behaviors limit the CHAT's usefulness as a screening tool.

The Modified CHAT (M-CHAT) was developed as a primary screening tool for autism at health supervision visits for children between 16 and 30 months of

age. The M-CHAT is a 23-item, yes/no parent questionnaire that takes ~5 minutes to administer. The M-CHAT has a high specificity of 99% and a moderate sensitivity of ~85%.³⁶ Note, however, that these data came from the screening of 1293 children: 1122 were nonselected at a regular 18-month screen, whereas 171 children were from a high-risk sample, namely children in an early-intervention screen who did not receive a diagnosis of a disorder as described in the *Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition* (DSM-IV). Children in this latter high-risk group may well have skewed the reported sensitivity to a considerable extent. The M-CHAT continues to be a promising device for screening with the caveat that it is unable to recognize 15 of 100 children with autism. The CHAT and M-CHAT are both considered level 1 autism screening tools.³ They are administered to all children to differentiate children who are at risk of autism from the general population.

The Social Communication Questionnaire (SCQ), formerly known as the Autism Screening Questionnaire, was developed from the ADI-R and is often considered the gold-standard diagnostic interview used in autism research studies.³⁷ The SCQ is a parent-report screen that comprises 40 yes/no questions. There are 2 forms: one for children younger than 6 years and one for children aged 6 years and older. The SCQ can usually be completed by the primary caregiver in less than 10 minutes and takes less than 5 minutes to score. The SCQ was validated in a sample of 200 patients, aged 4 to 40 years, whose parents had previously completed the ADI-R; 160 patients had ASD according to the ADI-R. A cutoff score of 15 on the SCQ had a sensitivity and specificity of 85% and 75%, respectively, for ASD according to the ADI-R. However, given the huge influence of

the prevalence of a condition on the measurement properties of a screening tool (sensitivity, specificity, and positive and negative predictive values), these findings cannot automatically be generalized to the community population.³⁸ The SCQ is a level 2 screening tool to be used (as discussed earlier) for clinical surveillance in early-intervention programs or developmental clinics.³

There are many other autism screening tests available for use by the primary caregiver or the developmental screener, including the Pervasive Developmental Disorders Screening Test-2, the Child Development Inventory, the Infant Development Inventory, and language and cognitive screening tools. However, none of the autism screening tests currently available has been shown to be able to fulfill the properties of accuracy, namely high sensitivity, high specificity, and high predictive value (proportion of patients with positive test results who are diagnosed correctly) in a population-wide screening program.

Question 5: Will the Screening Program Reach a High Proportion of the Persons for Whom It Was Intended?

The screening program would reach a high number of children if it were included as part of an overall wellness program for a given age. For example, most parents take their children to visit their family doctor or pediatrician at 18 and 24 months for vaccinations and routine checkups. If autism screening were incorporated into one of these routine visits, nearly all children who receive regular health care would be reached.

There have been no studies in the literature that addressed the actual impact of the autism screening on parents or children. As discussed earlier, none of the available screening tests

had high sensitivity (the most important feature of a screening test for detecting a condition such as autism). One must then ask: what is the impact of giving the wrong diagnosis of autism on parents, children, and the health care system? The potential burdens on families of receiving a misdiagnosis (either a false-positive or a false-negative) may be enormous, and there might be labeling effects that can be hard to remove. The costs to the health care system of managing these misdiagnoses must also be factored into the calculations for whether these costs could be better directed at case-finding or other surveillance services for children with autism and their families.

Question 6: Can the Health Care System Cope With the Screening Program?

After an autism diagnosis is confirmed, health care professionals have numerous other steps to take, including detailed assessment of the many aspects of a child's development that might be impaired; initiating effective therapy; monitoring compliance with treatment recommendations; and providing a lifetime of follow-up care. Screening is pointless, and almost certainly unethical, without the ability of the health and development services communities to provide the health care the condition requires. Unfortu-

nately, there are barriers that might limit the successful completion of these steps.

Unlike interventions for phenylketonuria or congenital thyroid disease, there is no strong evidence of the effectiveness of the various autism therapies currently provided. In addition, the availability of these therapies is limited (there are waitlists often as long as ≥ 1 year in many therapy centers), and the cost is often prohibitive. Therefore, initiating effective therapy might prove difficult in our current health care systems.

Question 7: Will Those With Positive Screen Results Comply With Subsequent Advice and Interventions?

Treatment compliance might be hard to achieve, because intensive behavior intervention programs require strict compliance on a daily basis, and even they have shown variable results. To date, there have been no published results from studies addressing parents' and children's compliance with interventions.

CONCLUSIONS

Autism is becoming increasingly recognized and understood, but great gaps in knowledge exist. Screening and diagnostic tools are still in progress of ongoing development and revi-

sion. Many therapies are available, but none has curative outcome or even well-established efficacy to change the course of the condition. The financial burden of the treatment can be extremely high, and cost-effectiveness has not been demonstrated.

At this stage, on the basis of scientific principles and the available evidence, we believe that we do not have enough sound evidence to support the implementation of a routine community-wide screening program for ASDs. Ongoing research is certainly needed to assess the effectiveness and acceptability of screening programs for ASDs. It might be thought to be difficult from an ethical perspective to justify a randomized controlled trial to study the benefits of a screening program. Nonetheless, because the implications of instituting a major program without sound research-based evidence are so enormous, we believe that the child health community has an important responsibility to undertake such clinical trials as a matter of priority. At this time we recommend careful surveillance and assessment of all preschoolers who present with impairments in their development of language, social function, or cognitive skills that result in activity limitations,³⁹ but we believe that community screening of all preschoolers is premature.

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