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Autism Spectrum Disorder Screening and Early Action

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A B S T R A C T

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Since its diagnostic inception, autism has garnered stigma and fear, often to the exclusion of action. Early detection and treatment of autism spectrum disorders have become crucial priorities of the National Institutes of Health. The primary care provider is the point of entry to the health care system for infants/children showing risk factors for autism spectrum disorders and/or symptoms of the disorder. Despite the existence of low-cost, well-tested developmental screening tools, many primary care providers rely solely on clinical judgment to ascertain developmental status. Current best screening practices were determined by a review of the literature. Key recommendations are highlighted.

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Autism spectrum disorders (ASDs) comprise several complex neurodevelopmental disorders that coalesce around impairments in communicative abilities, social functioning, and rigid or repetitive behaviors. These pervasive disabilities are described as a spectrum disorder because of the considerable variation in how individuals manifest both symptoms and severity.

On one end of the autism spectrum are individuals who need services resulting from co-occurring minimal verbal and intellectual ability. On the opposite end are individuals living near normal lives with average to superior intellectual ability. The majority of people living with ASDs reside somewhere between these extremes. Regardless of where individuals fall, every person on the autism spectrum has a greater chance to achieve life success with early detection and intervention.¹

Symptoms of ASDs typically emerge in infancy and toddlerhood and lead to individual, family, and community challenges. ASDs have strong established genetic heritability, often clustering in families. Many theories exist for additional ASD contribution, although no unifying cause for ASDs has been found. No current evidence-based data has linked vaccine administration to ASD risk.

Although there is no cure for ASD, timely detection increases the awareness of ASD-associated pathologic behaviors before they progress and become entrenched in a child's behavioral norm. Correcting core ASD deficits when the brain is most pliable to change maximizes the effects of early intervention.² Behavioral redirection is also simpler when physical size is smaller. Evidence-based early interventions build a proactive foundation for learning to facilitate the highest achievable life quality.³

A Growing Health Concern

The prevalence of ASDs has increased across the globe, with an estimated 1% to 2% of the world's population affected by the enigmatic condition. The Centers for Disease Control and Prevention report an average of 1 in 59 children being identified with ASDs.⁴

Primary care providers (PCPs) are essential points of entry to the health care system for infants/children demonstrating potential ASD risks. Every PCP who manages the care of pediatric patients will likely encounter children with ASD over the course of well-child and sick care. As health care "quarterbacks," PCPs have more opportunities to observe and screen infants and toddlers for ASD than any other professional because they should see each child up to 13 times in the first 3 years of life for well-child visits alone.⁵ Unlike many professionals, PCPs are positioned to stay the course for the child with suspected ASD, consistently teaming with the child's caregiver(s) in pursuit of optimal health trajectories.

Although universal use of evidence-based developmental and ASD-specific screening tools in conjunction with developmental surveillance at specific intervals has been recommended for nearly 2 decades, many PCPs continue to rely solely on clinical judgment to discern development.⁶ Researchers have shown an association between the use of clinical judgment alone and missed diagnoses in up to 45% of children eligible for early intervention services.^{7,8}

Review of the Literature

A comprehensive review of the literature was completed to discern current ASD detection strategies and PCP screening

practices. The search used Embase, PubMed, Scopus, PsycINFO, and CINAHL (EBSCO) databases. Initial searches were inclusive and sensitive. Search terms included a combination of keywords, Emtree terms, and Medical Subject Headings.

In PubMed, the Medical Subject Headings terms *autistic disorder*, *childhood developmental disorders-pervasive development disorders*, *Asperger**, *Rett**, *infant*, *child*, and *adolescent* and keywords *autism* and *early* were searched. The initial search yielded 937 articles. Limiters of English language and title/abstract were applied. The revised search provided 379 articles.

In the PsycINFO database, subject headings (focused) of *autism* and *screen**, *rescreen**, *test**, and *detect** and *primary care*, *practitioner**, and *pediatrician** were searched. The initial search returned 555 articles. Limiters included peer reviewed, English, and childhood (birth-age 12 years). The refined search yielded 241 articles.

The Scopus database was searched using the following keywords: *autism* and *screen**, *rescreen**, *test**, and *detect** and *primary care*, *practitioner**, and *pediatrician**. The initial search yielded 319 articles. English language was applied as a limiter. The refined search yielded 300 articles. The same keywords were used in CINAHL (EBSCO). The initial search provided 195 articles. Limiters of peer reviewed, English, and all children (age) were applied. The refined search provided 141 articles.

Studies were selected for complete text review based on their quality, placement in the evidence hierarchy, and clinical practice generalizability. A total of 84 articles were selected for complete text examination.

Synthesis of the Literature

ASD Screening

For almost 20 years, consensus panels and professional groups have advocated for early and regular ASD screening in primary care,^{9,10} yet compliance with these guidelines remains far from ideal.¹¹ Because infant and childhood development is dynamic, any single source of information is likely to be incomplete; therefore, regular and repeated screening, in addition to PCP developmental surveillance at every well-child visit and ascertainment of parental concerns in primary care, is needed¹² (Table 1). For these reasons, since 2006, the American Academy of Pediatrics (AAP) has recommended developmental screening using a validated screening tool at 9-, 18-, and 24- or 30-month well-child visits, with special attention to subtle red flags in infants who have sibling(s) with ASDs as well as conduction of ASD-specific screening using a validated tool at the 18- and 24-month visits.¹⁴ Scheduling a targeted clinic visit when PCP or parent concerns remain after a negative screen and acting quickly on a positive screen or in the presence of 2 or more ASD risk factors (ie, sibling with ASD, older parents, and low birth weight/birth injury) with referral for formal ASD evaluation and/or early intervention is also recommended.¹⁴ Because the presentation and timing of ASD symptoms are widely variable, in addition to universal screening, PCPs should also be aware of absolute indications for ASD diagnostic referral (Table 2).

PCPs may bill for ASD screening using the *Current Procedural Terminology* code 96110. The use of this code requires the following: 1) the screening instrument must be a validated tool, 2) the instrument must be scored, and 3) the PCP indicates that he or she reviewed the screen in the medical record.¹⁵ For practices servicing children with Medicaid, many states now include bonuses for primary care practices that successfully reach developmental screening benchmarks using validated and approved tools.⁵

Table 1

Possible Autism Spectrum Disorder Red Flags

Displays a lack of interest in others and has difficulty relating to them (absence of joint attention)
Makes little or inconsistent eye contact
Tends not to look at or appear to listen to people
Fails to point at objects to convey interest or look at objects when others point at them by 24 months
Prefers solitude
Communication
Has delayed language and speech skills
Fails to respond to name by 12 months
Repeats/echoes words or phrases said or repeats words or phrases in place of normal language
Speaks in a flat, robotlike or singsong voice
Reverses pronouns (eg, says "I" instead of "you")
Gives unrelated answers to questions
Appears not to understand sarcasm, jokes, or teasing
Struggles to talk about their feelings or understand others' perspectives
Has difficulty conveying personal needs and desires
Behavior
Fails to initiate play/interactions
Does not engage in pretend play
Prefers parts of objects (eg, wheels)
Displays obsessive interests and/or repetitive actions (eg, plays with toys the same way each time)
Becomes upset by routine changes
Has obsessive interests
Displays flat or inappropriate facial expressions
Shows hyperactivity and/or impulsivity
Fails to perceive danger
Causes self-injury
Shows aggression and unusual mood/emotional reactions
Sensory
Exhibits unusual responses to sights, smells, tastes, textures, or sounds
Participates in stimulations such as hand flapping, body rocking, or spinning in circles
Avoids physical contact or invades personal space
Displays unusual sleeping or eating habits

Derived from CDC Autism Spectrum Disorder Signs and Symptoms.¹³

ASD Formal Diagnosis

Diagnostic assessment by experienced clinical specialists after positive screening and/or PCP/parental concern for ASDs remains the current standard for formal ASD diagnosis. Parents should be educated that a diagnostic assessment by a clinical specialist is necessary because other diseases and disorders may mimic or coexist with ASD. Specialist evaluation is also important because false-positive screenings are possible. For example, racial minorities and lower maternal education levels have been associated with inflated positive M-CHAT screening rates and lower participation at follow-up. Families of white race and higher educational levels have been cited as more likely to decline formal evaluation participation.¹⁶

PCPs must understand and teach that although diagnostic workup is necessary, if early intervention is needed, it should transcend formal ASD diagnosis.¹⁷ Specialized diagnostic and interventional services are often difficult to access and usually require long wait times.¹⁸ This reality prompted the AAP to

Table 2

Absolute Indications for Autism Spectrum Disorder Diagnostic Evaluation Referral

Behavior Indicator	Referral Time Frame
No gesturing (waving bye-bye and pointing)	By 12 months
No babbling	By 12 months
No single words	By 16 months
No spontaneous (excluding echolalia) 2-word phrases	By 24 months
Any loss of language or social skills (regression)	Any age

Data from Filipek et al¹⁰ and Johnson and Myers.¹⁴

recommend that PCPs make simultaneous referrals to diagnosticians, providers, and audiologists as appropriate to allow necessary intervention to begin as soon as possible.¹⁹

Many families experience significant barriers in obtaining necessary and timely ASD services with access to evidence-based care often being limited or nonexistent. In the study of care barriers, researchers have paid great attention to family characteristics, focusing primarily on sociodemographic factors and their links to age of ASD diagnosis. Later age of diagnosis and underdiagnosis of ASDs have been found among poorer families, rural families, and traditionally underserved minority groups.^{20,21} Firstborn and only children are also more likely to experience delayed ASD detection.²² Streamlining of ASD screening processes helps reduce these health disparities.¹⁶ Although ASD can be reliably diagnosed by 2 years of age, most children are not diagnosed until after age 4.²³

Recommendations

Mounting literature points to the importance and effectiveness of early intervention and supportive services for children with ASD and their families and communities, driving the need for screening and early action practice change.

Recommendations for Clinical Practice

Expedient application of the AAP evidence-based developmental screening recommendations vitally depends on individual PCP practice and performance. Numerous barriers to ASD screening have been cited that reduce PCP willingness to screen. Common barriers include 1) increased demands on provider time occasioned by the necessity to screen for other disorders and 2) challenges posed by families requiring professional support to complete formal questionnaires.¹⁵

The designation of clinic staff to assist with screening may mitigate these challenges. Additionally, shortages of staff and staff turnover complicate the development of consistent screening practices in busy primary care clinics. Efforts to overcome these barriers toward achievement of consistent developmental screening routines will likely increase screening frequency, elicit improved parental cooperation, and provide greater time for direct discussion of potential concerns between PCPs and parents.¹⁵ Primary care providers can access an unexhaustive list of current validated ASD and developmental screening tools at <https://screeningtime.org/star-center/#/screening-tools#top>.

Compelling evidence exists to support the use of health information technology in the facilitation of PCP adherence to ASD screening and developmental surveillance. The use of digital screening has been linked to 1) time/cost savings resulting from automated scoring and report generation and 2) the ability of clinics to monitor and scrutinize their own patterns in ASD screening and detection.²⁴

Primary care practices should ensure that parents completing screenings are able to read (typically a fifth-grade reading level is necessary). Evidence-based screenings should be provided in the parent/caregiver preferred language, and cultural preferences must always be considered. In circumstances in which these challenges are present, effective screening completion should be addressed through clinic support and language translation with attention to possible cultural barriers with ASD screening.

Equally important to the completion of evidence-based ASD screening is the PCP's understanding of appropriate action(s) to take when a screen is positive (Figure). Additionally, PCPs must

address any concerns raised by a parental report and/or personal observation. In any of these cases, PCPs must obtain a careful history and examination. The history should include pregnancy, birth and delivery details. The PCP should also include family history, paying particular attention to a history of ASD or recurrent developmental problems.¹

If the PCP has not previously met the child, he or she should obtain a parental review of the child's early developmental milestones and note any previous evaluations. Was the child described as "too easy" or overly difficult? Recalling specific instances may be easier if the PCP asks parents to remember an occasion (ie, first birthday or Christmas) and describe the child's behavior.¹ The PCP should observe if an appropriate response to the child's parents (and to the PCP) is present.

The PCP should note if there are any symptoms suggestive of seizures or hearing loss (including recurrent ear infections) and other factors that may account for speech delay including 2 spoken languages in the home. Even in these cases, the PCP should not dismiss ASD as a possible cause of delay.¹

Child Find and other applicable agencies may offer evaluation and treatment and possibly assistance in securing treatment. PCPs and/or nurses can connect parents with relevant state agencies and then follow up with the parent(s) to ensure an evaluation was completed. Parents often minimize troubles and/or have obstacles (because of poverty, transportation, language barriers, etc) in accessing services or following through with interventions.¹

PCPs should offer anticipatory guidance to families awaiting diagnostic workup. Accidents are the most common causes of death for children, youth, and adults with ASD.²⁶ Safety is a critical consideration while a PCP awaits a formal ASD diagnostic evaluation, especially for any child whose parents describe a history of departure from safe areas (elopement). Typically developing children may elope, inadvertently, putting themselves at risk, yet children with ASD often continue this behavior long after their same aged peers have ceased to wander away. Documented increased risks are injuries of the face, head, and neck; poisoning; and deaths from suffocation and drowning. Children with ASDs are often fascinated by water. PCPs should recommend early swimming lessons to promote safe water practices. Other actions reviewed with families during office visits may include the following:

1. Encourage the use of identification bracelets, door alarms/locks, and/or fencing as appropriate.
2. Educate on possible warning signs of poisoning (eg, stains on mouth or clothes, sudden onset of vomiting, etc); parents should know how to contact their local poison control.
3. Create a wandering child action plan in case the child becomes lost.
4. Respond to sensory sensitivities (increased or decreased) proactively.
5. Ensure safe opportunities for regular exercise and movement.

Some PCPs are reluctant to share their concerns for ASDs with parents out of fear that premature or tentative *labeling* may contribute to undue familial stress. Yet, concealing concerns in favor of a wait and see approach is rarely appreciated among parents and is associated with poorer outcomes for the child with ASD.¹⁴

Many parents who are suspicious of ASD search the Internet or potentially erroneous sites for more information. It is crucial that PCPs supply and/or teach parents how to seek out peer-reviewed data. Parents should be strongly cautioned regarding information available on the Internet that is not evidence based. Parents should

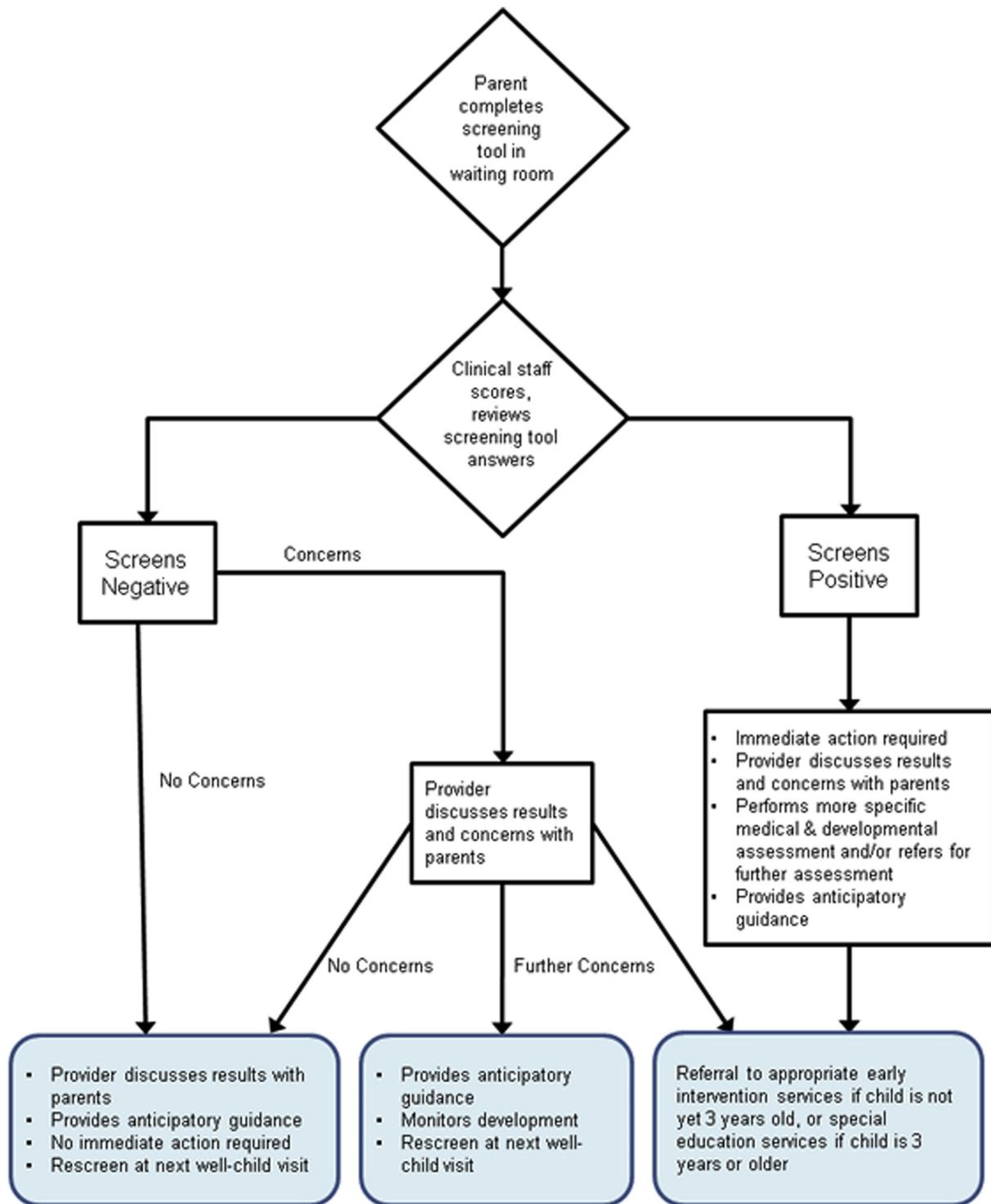


Figure. Centers for Disease Control and Prevention.²⁵ Pediatric developmental screening flowchart.

be advised to bring as much information as possible to their diagnostic specialists' appointment(s) including the following:

1. Video of child behavior(s), particularly in peer settings if possible
2. All paper work from school(s) and/or early intervention providers

Recommendations for Nursing Education

The primary care nurse practitioner (NP) workforce is projected to grow far more rapidly than the physician supply.²⁷ With more NPs filling PCP roles and as part of the largest, most trusted workforce in health care, NPs are uniquely positioned to help ensure timely ASD screening and early action.

Both undergraduate and graduate nursing programs must incorporate the critical necessity of developmental and ASD-specific screening at designated intervals into their curriculums and equip students for practical utilization. Greater practice change may be achieved if NPs of tomorrow consistently partner with practicing PCPs to identify and improve care for those who have or will have ASD.

Recommendations for Research

Critical research considerations must include the strengths and vulnerabilities of the child at risk for or with ASD and their family members. Continued efforts toward improving the process of ASD screening could facilitate rapid, early detection and advance accuracy of future studies. Better efforts to detect and characterize young children with ASD must be sought for a chance to develop and adapt early interventions to most effectively address each child with consideration to their family and community characteristics.¹

Given the continued emergence of promising results, additional work is needed to replicate, expand, and individualize screening tools and available interventions for infants and children at risk for ASDs.²⁸ Improving primary care knowledge, skills, and screening practices may optimize the timing of detection and initiation of treatment, care quality, and outcomes for children with ASD.²⁹

Recommendations for Policy

When PCPs use validated and reliable screening tools consistently at designated intervals, they initiate a process of discussion and the appropriate next steps. These actions position parents to further understand early development and proper procurement of necessary resources to facilitate improved developmental paths for their children with suspected ASD.³⁰

Federal and state policies supporting ASD screening may lead to significant societal cost savings. Care of persons with ASD will most likely be lessened over time, and a parallel increase of overall productivity of many (including many exceptionally bright individuals) may be realized via early intervention. Changes to current policies are needed to ensure significant disparities in access to appropriate health care for children with ASDs do not continue to persist. Urgent advocacy and resource allocation are required.³¹

The importance to our society and its citizens to identify and ensure early evidence-based treatments for the exceptional, vulnerable, and growing ASD population warrants urgent attention. Every child must be screened consistently—a long-standing recommendation that should become a mandate.

Conclusions

Early detection of ASDs and/or developmental concerns is a prerequisite for early intervention. Increasing evidence supports the potential of early intervention to alter adverse development toward more neurotypical trajectories, thereby providing substantial short- and long-term benefits to human capacity across systems. Although not every child who presents with developmental concerns suggestive of ASD may reach the diagnostic threshold, early detection and intervention offer the best outcomes.³²

Developmental and ASD-specific screening must be viewed through the same lens of importance as other screenings recommended or mandated based on age. Developmental and ASD-specific screening can identify children with developmental and behavioral challenges early, when their benefits from treatment

will be highest. For these screenings to be effective, by design they must be applied to all children, not only those who may be exhibiting overt symptoms, those whose caregivers are concerned, or those an individual PCP judges would benefit. Health care teams caring for young children should establish systems and prompts that result in increased automatic developmental and ASD screenings at necessary intervals.

Broad adherence to ASD screening and early action will require health care professionals across clinical areas, academia, policy, and community health to work together in support of PCP practice change. Children who will be diagnosed with ASD cannot afford to endure the research to practice gap, which is currently appraised to average 17 years (the pediatric life span). Positive health transformations for those with ASD may occur if practice is changed now.

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