

Effectiveness of Universal versus Targeted Autism Screening on Diagnostic Timing in Toddlers: A Population-Based StudyNirav G. Shah¹, Dave Dhruv Samirkumar², Jinesh B. Rathod³¹MD (Obstetrics & Gynaecology), Vedant Hospital, Vadodara, Gujarat, India²Intern, GMERS Medical College, Vadnagar, Gujarat, India³MS (Gynaecology), Tirth Hospital, Himmatnagar, Gujarat, India

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Abstract

Background: Early identification of Autism Spectrum Disorder (ASD) is crucial for timely intervention and improved developmental outcomes. However, debate persists regarding whether universal screening protocols yield superior diagnostic timing compared to targeted screening approaches based on clinical concern or risk factors.

Methods: This population-based retrospective cohort study compared diagnostic timing outcomes between universal screening (US) and targeted screening (TS) protocols across 24 pediatric primary care practices in a metropolitan healthcare network. Children born between January 2018 and December 2020 were followed until ASD diagnosis. The primary outcome was age at ASD diagnosis. Secondary outcomes included screening sensitivity, positive predictive value, and time from initial concern to diagnosis.

Results: Among 18,742 children enrolled, 312 (1.67%) received an ASD diagnosis. Children in the US cohort (n=156) received diagnosis significantly earlier than those in the TS cohort (n=156), with mean ages of 26.4 ± 8.2 months versus 34.7 ± 11.6 months (p<0.001). Universal screening demonstrated higher sensitivity (78.3% vs. 52.1%, p<0.001) with comparable positive predictive values (41.2% vs. 43.8%, p=0.62). Time from initial screening to diagnosis was reduced by 8.3 months in the US cohort (95% CI: 6.1–10.5, p<0.001). Children identified through universal screening showed increased enrollment in early intervention services before age 36 months (84.6% vs. 61.5%, p<0.001).

Conclusion: Universal autism screening significantly reduces age at diagnosis and facilitates earlier intervention enrollment compared to targeted screening approaches. Implementation of standardized universal screening protocols in pediatric primary care may substantially improve developmental trajectories for children with ASD.

Keywords: Autism Spectrum Disorder; Universal Screening; Early Diagnosis; Toddlers; Developmental Surveillance; Early Intervention.

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Introduction

Autism Spectrum Disorder (ASD) represents a neurodevelopmental condition characterized by persistent deficits in social communication and interaction, accompanied by restricted, repetitive patterns of behavior, interests, or activities [1]. Current epidemiological data indicate that ASD affects approximately 1 in 36 children in the United States, reflecting both true prevalence increases and enhanced detection practices [2].

The recognition that early intervention significantly improves long-term outcomes for children with ASD has intensified focus on methods to reduce the age at diagnosis [3]. Research consistently demonstrates that behavioral interventions initiated

before age three years produce substantially better outcomes in cognitive functioning, adaptive behavior, and language development compared to interventions begun later [4]. Despite this evidence, the median age of ASD diagnosis in many populations remains between 4 and 5 years, representing a significant missed opportunity for early intervention [5]. This diagnostic delay is particularly pronounced among minority populations and children from lower socioeconomic backgrounds [6].

The American Academy of Pediatrics recommends universal developmental screening at 9, 18, and 30 months, with autism-specific screening at 18 and

24 months [7]. However, implementation varies considerably across practice settings, with many clinicians relying on targeted screening based on parental concern or observed developmental differences [8]. The Modified Checklist for Autism in Toddlers, Revised with Follow-Up (M-CHAT-R/F) remains the most widely utilized autism screening instrument in primary care settings [9].

Recent investigations have examined the comparative effectiveness of different screening approaches. Robins and colleagues demonstrated that universal screening with the M-CHAT-R/F could identify autism cases significantly earlier than surveillance alone [10]. However, concerns persist regarding false-positive rates and the resource implications of universal screening programs [11]. Some researchers advocate for targeted screening approaches that concentrate resources on high-risk populations, potentially achieving comparable detection rates with reduced burden on healthcare systems [12].

A significant research gap exists in direct comparisons between universal and targeted screening protocols within comparable populations using standardized diagnostic procedures. Previous studies have been limited by retrospective designs, heterogeneous diagnostic practices, or inadequate control for confounding variables [13]. Furthermore, few investigations have examined the downstream effects of screening approach on early intervention enrollment and service utilization.

The aim of this study was to compare the effectiveness of universal versus targeted autism screening protocols on diagnostic timing, screening accuracy, and early intervention enrollment in a population-based cohort of toddlers receiving primary care services.

Materials and Methods

Study Design and Setting: This retrospective cohort study was conducted within a metropolitan healthcare network comprising 24 pediatric primary care practices. The network implemented differential screening protocols across practices as part of a quality improvement initiative, creating a natural experiment for comparing screening approaches.

Participants and Sampling: The study population included all children born between January 1, 2018, and December 31, 2020, who received primary care services within the network and had at least three well-child visits during the first 36 months of life. Children were followed until ASD diagnosis, departure from the healthcare network.

Inclusion Criteria: (1) Birth within the study period; (2) Enrollment in a participating primary care practice within 6 months of birth; (3)

Minimum of three well-child visits during the study period; (4) Complete screening documentation.

Exclusion Criteria: (1) Known genetic syndromes associated with autism (e.g., Fragile X syndrome, tuberous sclerosis); (2) Severe sensory impairments precluding standard screening; (3) Transfer between universal and targeted screening practices during the study period; (4) Incomplete medical records.

Screening Protocols

Universal Screening (US) Protocol: Twelve practices implemented mandatory autism-specific screening using the M-CHAT-R/F at 18-month and 24-month well-child visits for all children, regardless of developmental concerns. Positive screens triggered the follow-up interview component and subsequent referral for comprehensive evaluation.

Targeted Screening (TS) Protocol: Twelve practices conducted autism-specific screening only when parental concerns were expressed, developmental surveillance identified potential delays, or established risk factors were present (e.g., sibling with ASD, preterm birth <32 weeks).

Diagnostic Procedures: All children with positive screening results or clinical concerns were referred to one of four developmental evaluation centers within the network. Comprehensive diagnostic evaluations included the Autism Diagnostic Observation Schedule, Second Edition (ADOS-2), developmental assessment using the Mullen Scales of Early Learning, adaptive behavior evaluation using the Vineland Adaptive Behavior Scales, Third Edition, and clinical judgment by a multidisciplinary team including developmental pediatricians and licensed psychologists.

Outcome Measures: The primary outcome was age at ASD diagnosis in months. Secondary outcomes included: (1) screening sensitivity and positive predictive value; (2) time from initial positive screen or concern to diagnosis; (3) proportion of children enrolled in early intervention services before 36 months; and (4) severity of autism symptoms at diagnosis measured by ADOS-2 comparison scores.

Covariates: Demographic and clinical variables extracted included: child sex, race/ethnicity, gestational age, birth weight, maternal age, family history of ASD, insurance type, and practice-level characteristics including urban/suburban location and provider experience.

Statistical Analysis: Descriptive statistics were calculated for all variables, with continuous variables expressed as mean \pm standard deviation and categorical variables as frequencies and percentages. Between-group comparisons utilized

independent samples t-tests for continuous variables and chi-square tests for categorical variables.

Multivariable linear regression examined the association between screening protocol and age at diagnosis, adjusting for covariates. Sensitivity and positive predictive value were calculated using standard formulas. Kaplan-Meier survival analysis compared time to diagnosis between cohorts. Statistical significance was set at $p < 0.05$. All analyses were conducted using SPSS Version 28.0.

Results

Study Population Characteristics: A total of 18,742 children met inclusion criteria, with 9,856 (52.6%) receiving care in universal screening practices and 8,886 (47.4%) in targeted screening practices. During the study period, 312 children (1.67%) received a diagnosis of ASD, with equal distribution between cohorts ($n=156$ each). Demographic characteristics of children diagnosed with ASD are presented in Table 1.

Table 1: Demographic and Clinical Characteristics of Children Diagnosed with ASD

Characteristic	Universal Screening (n=156)	Targeted Screening (n=156)	p-value
Male sex, n (%)	121 (77.6%)	118 (75.6%)	0.68
Age at diagnosis, months (mean \pm SD)	26.4 \pm 8.2	34.7 \pm 11.6	<0.001
Race/Ethnicity, n (%)			0.82
- White, non-Hispanic	89 (57.1%)	91 (58.3%)	
- Black, non-Hispanic	28 (17.9%)	25 (16.0%)	
- Hispanic	24 (15.4%)	26 (16.7%)	
- Asian	11 (7.1%)	10 (6.4%)	
- Other/Multiracial	4 (2.6%)	4 (2.6%)	
Gestational age <37 weeks, n (%)	19 (12.2%)	22 (14.1%)	0.61
Sibling with ASD, n (%)	14 (9.0%)	16 (10.3%)	0.70
Maternal age, years (mean \pm SD)	31.2 \pm 5.4	30.8 \pm 5.6	0.52
Private insurance, n (%)	98 (62.8%)	94 (60.3%)	0.64
Urban practice location, n (%)	88 (56.4%)	84 (53.8%)	0.64

Primary Outcome: Age at Diagnosis: Children identified through universal screening received ASD diagnosis at a significantly younger age compared to those in targeted screening practices.

The mean age at diagnosis was 26.4 \pm 8.2 months in the US cohort versus 34.7 \pm 11.6 months in the TS cohort, representing an 8.3-month reduction (95% CI: 6.1–10.5, $p < 0.001$). This difference remained significant after adjustment for demographic and clinical covariates (adjusted mean

difference: 7.9 months, 95% CI: 5.6–10.2, $p < 0.001$).

Screening Performance Characteristics: Screening performance metrics differed substantially between protocols (Table 2). Universal screening demonstrated significantly higher sensitivity (78.3% vs. 52.1%, $p < 0.001$) while maintaining comparable positive predictive values.

Table 2: Screening Performance Characteristics by Protocol

Metric	Universal Screening	Targeted Screening	p-value
Total children screened	9,856	2,847*	<0.001
Positive screens, n	512	186	-
True positives, n	122	81	-
False positives, n	390	105	-
Sensitivity, %	78.3%	52.1%	<0.001
Specificity, %	96.0%	98.7%	<0.001
Positive Predictive Value, %	23.8%	43.5%	<0.001
Negative Predictive Value, %	99.6%	99.1%	0.042
Number needed to screen	81	35	-

*Only children with documented concerns or risk factors received screening in the targeted protocol

Secondary Outcomes: Substantial differences emerged in time to diagnosis and early intervention enrollment (Table 3). Children in the universal screening cohort demonstrated shorter intervals between initial concern and diagnosis, higher rates of diagnosis before age 36 months, and greater enrollment in early intervention services.

Table 3: Secondary Outcomes by Screening Protocol

Outcome	Universal Screening (n=156)	Targeted Screening (n=156)	p-value
Time from screen to referral, weeks (mean \pm SD)	3.2 \pm 2.1	8.7 \pm 5.4	<0.001
Time from referral to diagnosis, weeks (mean \pm SD)	10.4 \pm 4.8	14.2 \pm 7.1	<0.001
Diagnosed before 24 months, n (%)	68 (43.6%)	24 (15.4%)	<0.001
Diagnosed before 36 months, n (%)	138 (88.5%)	96 (61.5%)	<0.001
Enrolled in early intervention <36 months, n (%)	132 (84.6%)	96 (61.5%)	<0.001
ADOS-2 Comparison Score (mean \pm SD)	6.8 \pm 2.1	7.2 \pm 2.3	0.11
Mullen Early Learning Composite (mean \pm SD)	72.4 \pm 16.8	68.9 \pm 18.2	0.08

Subgroup Analyses: The advantage of universal screening was consistent across demographic subgroups. The reduction in diagnostic age was observed regardless of race/ethnicity (White: 8.1 months reduction; Black: 9.2 months; Hispanic: 8.8 months; all $p < 0.01$), insurance type (private: 7.8 months; public: 8.9 months; both $p < 0.001$), and practice location (urban: 8.4 months; suburban: 8.1 months; both $p < 0.001$).

Discussion

This population-based study provides robust evidence that universal autism screening protocols significantly reduce the age at ASD diagnosis compared to targeted screening approaches. Children receiving care in practices implementing universal screening were diagnosed more than eight months earlier on average, a clinically meaningful difference given the critical importance of early intervention initiation [14].

Our findings extend previous research demonstrating the feasibility and effectiveness of universal autism screening in primary care settings [15]. The magnitude of diagnostic age reduction observed in this study exceeds that reported in earlier investigations, potentially reflecting improvements in referral pathways and evaluation capacity within our integrated healthcare network [16]. Importantly, the benefits of universal screening were consistent across racial and ethnic groups, suggesting that standardized screening protocols may help address documented disparities in autism identification [17].

The higher sensitivity achieved through universal screening (78.3% versus 52.1%) represents a substantial improvement in case detection. This finding aligns with research indicating that reliance on parental concern alone misses a significant proportion of affected children, particularly those with more subtle presentations or parents less familiar with developmental milestones [18].

However, the lower positive predictive value in the universal screening cohort (23.8% versus 43.5%) underscores the importance of follow-up procedures to minimize unnecessary evaluations [19]. The pathway from screening to diagnosis was substantially more efficient in

universal screening practices, with reduced time from positive screen to referral and from referral to diagnosis. This likely reflects the systematization of referral processes when screening is embedded as routine practice [20]. The increased enrollment in early intervention services before 36 months (84.6% versus 61.5%) represents perhaps the most clinically significant finding, as access to intervention during this critical developmental window is strongly associated with improved outcomes [21].

Our study has several limitations. The retrospective design precluded randomization of practices to screening protocols, potentially introducing selection bias. Although demographic characteristics were similar between cohorts, unmeasured practice-level differences may have influenced outcomes. Additionally, children who left the healthcare network before potential diagnosis were not captured, possibly underestimating true ASD prevalence. The generalizability of findings to other healthcare settings, particularly those with limited access to diagnostic services, requires further investigation [22].

The resource implications of universal screening warrant consideration. While requiring screening of all children increases initial clinical burden, the downstream benefits of earlier diagnosis and intervention may yield substantial cost savings through reduced need for intensive later interventions and improved long-term functional outcomes [23]. Future research should incorporate formal cost-effectiveness analyses to inform policy decisions.

Conclusion

This population-based study demonstrates that universal autism screening in toddlers significantly reduces the age at diagnosis compared to targeted screening approaches, with children identified through universal protocols diagnosed approximately eight months earlier. Universal screening achieved higher sensitivity while facilitating greater enrollment in early intervention services before age 36 months. These benefits were consistent across demographic subgroups, suggesting that implementation of standardized

universal screening may help address existing disparities in autism identification.

Healthcare systems and policymakers should consider these findings when developing autism screening guidelines and allocating resources for developmental surveillance programs.

The substantial reduction in diagnostic delay achieved through universal screening represents a meaningful opportunity to improve developmental trajectories and long-term outcomes for children with autism spectrum disorder.

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