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Table Of Contents

Journal Cover 1
Author[s] Statement 3
Editorial Team 4
Article information 5
Check this article update (crossmark) 5
Check this article impact 5
Cite this article 5
Title page 6
Article Title 6
Author information 6
Abstract 6
Article content 8

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Early Identification of Autism Spectrum Disorder Role of Artificial Intelligence and Digital Screening Tools

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Abstract

General Background Autism spectrum disorder constitutes a prominent neurodevelopmental condition necessitating early intervention to maximize long-term cognitive and social outcomes for children. **Specific Background** Conventional screening methods, including parent-report questionnaires and specialized clinical observations, frequently cause substantial diagnostic delays due to human error and limited specialist availability. **Knowledge Gap** While automated classification models exist, their cross-cultural generalizability, algorithmic transparency, and seamless integration into real-world clinical workflows remain insufficiently explored and validated. **Aims** This review evaluates the efficacy, methodological integrity, and practical deployment challenges of artificial intelligence and machine learning technologies for early autism detection. **Results** Automated analysis demonstrated high diagnostic precision across various modalities; feed-forward neural networks processing clinical questionnaires achieved a 99.72 percent classification accuracy using fewer than twenty instructional items. Similarly, medical claims-based models attained an AUROC of 0.834, while objective eye-gaze and video analysis methods yielded AUC values ranging between 0.81 and 0.83. **Novelty** Synthesizing multimodal data—ranging from behavioral videos to medical claims—through machine learning provides a scalable, objective framework that bypasses traditional resource bottlenecks in early neurodevelopmental screening. **Implications** Healthcare systems must prioritize independent, multi-site prospective validation studies and establish standardized evaluation protocols to ensure these automated tools reduce rather than exacerbate existing diagnostic disparities.

Highlights:

- Feed-forward neural networks achieved a 99.72 percent classification accuracy analyzing automated clinical screening questionnaires.

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- Medical claims-based prediction models successfully identified high-risk pediatric cases with an AUROC of 0.834.
- Objective eye-gaze and video analysis methods yielded high diagnostic AUC values ranging between 0.81 and 0.83.

Keywords: Artificial Intelligence, Autism Spectrum Disorder, Digital Screening Tools, Machine Learning, Pediatric Neurology

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I. Introduction

Autism spectrum disorder represents a diverse array of neurodevelopmental conditions delineated by enduring impairments in social communication along with constrained or repetitive behavioral patterns, interests, or activities, situated along a spectrum of severity. The manifestation of clinical symptoms generally occurs during early childhood and is associated with a lifelong trajectory. Contemporary epidemiological studies estimate the prevalence of ASD among children in the United States to be approximately 1 in 36, a statistic that has escalated over the preceding decades. Might this observation indicate enhanced awareness, expanded diagnostic criteria, diagnostic substitution, or a genuine rise in incidence? Either way, it underlines the need for good early detection.

The importance of early diagnosis cannot be overstated. Children who receive intervention services before the age of three have considerably better long-term outcomes in language, adaptive behavior, and social skills than children who are diagnosed later in childhood [7]. These interventions use the principle of neuroplasticity within sensitive periods of brain development to target potential and limit functional impairment, but clinical practice is still characterized by diagnostic delays. However, most children are not diagnosed with ASD until after four years of age, meaning a sensitive period for intervention has passed [8].

Current screening methods involve parent-report questionnaires and clinical observation instruments, which are administered during well-child visits. The Modified Checklist for Autism in Toddlers, Revised with Follow Up (M-CHAT-R/F) is the most commonly used Level 1 screener for toddlers aged 16 to 30 months [1]. Despite their psychometric qualities, these tools can be impractical. The M-CHAT-R requires follow-up interviews for positive screens, creating additional burden on families and healthcare systems. Time-consuming and highly susceptible to mistakes made by humans are two elements of assessment. Furthermore, these instruments depend upon parents to recognize and disclose subtle developmental variations, which can be impacted by factors like the parents' education, socioeconomic status, and experience with typical child development [4].

Schedule (ADOS), Autism Diagnostic Interview-Revised (ADI-R)) used to diagnose ASD are the gold-standard. They require specific training and lengthy in-person administration, leading to limited access in many communities [20] and bottlenecks in diagnostic pathways. Families may wait months or years between concern and an autism diagnosis. There is also a shortage of developmental specialists, particularly in rural and medically underserved areas [15].

Due to these difficulties with customary methods, artificial intelligence (AI) and machine learning may help reduce long-standing issues around the early diagnosis of ASD, and overcome issues data scientists have experienced with the analysis of high dimensional data, integration of different data sources, and the identification of subtle behavioral changes. Machine learning algorithms can detect patterns in high-dimensional data, which makes them well-suited to build models of the numerous behavioral profiles seen in ASD [5], [6]. Machine learning can also analyze data derived from many different modalities, including questionnaires, medical claims, eye-tracking, video recordings of naturalistic behavior, speech, and physiology [7].

In the past 5 years, there has been a surge of research into the use of machine learning as a tool for screening and diagnosis of ASD. These systems have classified home video footage for behavioral indicators of ASD [16] and healthcare utilization patterns to determine risk of ASD [8]. Others have classified ASD by using eye-gaze metrics during social stimuli [10], [11]. Deep learning architectures have achieved impressive classification accuracy on retrospective datasets [6], [14]. To determine linguistic indicators of ASD, conversation transcripts can be analyzed with methods of natural language processing [17]. The common objectives of these various methods are to minimize time to diagnosis, improve screening reliability, minimize reliance on specialized expertise, and enable scalable population-level screening.

In my opinion, there is a glaring lack of study on socioeconomic, cultural, and geographic situations. Clinical endorsement and assurance may be restricted by comprehension issues caused by the unclear features of certain models trained with machine learning [19]. Careful consideration must be given to the moral implications of algorithmic bias, data confidentiality, and the proper application of predictive risk scores [4], [7].

However, the enthusiasm for AI-enhanced screening must be restrained by a thorough assessment of methodological integrity, validation standards, and hands-on application challenges. Many published studies provide performance measures based on retrospective evaluations of convenience samples, raising questions with regard to their relevance to real-world clinical populations [2], [5]. Despite attempts to find reliable external validation in various geographic, cultural, and socioeconomic settings, it is still deemed insufficient for addressing the issues at hand. Machine learning models with highly complex properties are frequently untransparent, leading to interpretability issues that could affect clinical validation and confidence [19]. Ethical considerations regarding data confidentiality, algorithmic prejudice, and the appropriate application of predictive risk scores necessitate thorough scrutiny [4], [7], especially in light of the ethical implications.

In this article, we explore the use of AI and digital screening tools to identify individuals with early acquired sensory deficiency (ASD) by utilizing AI and digital screening tools. We synthesise evidence from various technological approaches, analyze their performance characteristics and limitations, and critically assess their readiness for clinical application. The current paper employs cutting-edge research methodologies from the domains of questionnaire automation, claims-based prediction, eye-tracking and computer vision, speech analysis, and multimodal integration strategies, as well as recent scholarly articles. The question presented does not only entail the effectiveness of these technologies in a controlled research context, but also encompasses the practical, ethical, and methodological issues that must be tackled before they

Vol. 3 No. 2 (2026): Oktober
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can prove useful in transforming early ASD detection in real-world healthcare settings.

II. Methodology

This narrative review aims to analyze the most current literature on the use of artificial intelligence and machine learning for the early detection of autism spectrum disorder. The current paper focused on research from 2020 to 2026 on the development, validation, and/or evaluation of AI-based screening tools for autism in children. The paper analyzed several technological approaches, including machine learning and questionnaires, medical claims data mining, eye tracking and computer vision, video analysis, speech and language pattern analysis, and systems with integrated multiple modalities.

The review covers a wide range of study types: retrospective cohort studies, cross-sectional studies, prospective validation studies, and systematic reviews. The authors collected data on the study population and sample population, AI methodologies, modalities of data, study performance, and clinical implications noted by the authors. The paper paid special attention to the study's validation approach, generalizability, and practical use.

To synthesize evidence on performance characteristics, methodological strengths and limitations and clinical deployment readiness, the authors arranged findings thematically based on technological approach and data modality. The critical discussion section examines cross-cutting issues such as ethical considerations related to algorithmic transparency, integration challenges, validation rigor, and other cross-cutting issues. The paper analysis aims to provide a fair evaluation of the accuracy and limitations of AI-based artificial intelligence for early ASD detection in light of recent empirical data.

III. Result and Discussion

Artificial Intelligence in Autism Spectrum Disorder Identification

- **Questionnaire-Based Machine Learning Approaches**

AI has entered the ASD screening process with great maturity by analyzing automated parent-report screening questionnaires through automation. Achenie and co-authors developed a feed-forward artificial neural network (fANN) to analyze M-CHAT-R responses from 14,995 toddlers aged 16-30 months, which were then processed by neural networks. Their model's impressive performance was due to the fact that they correctly classified only 18 of the 20 M-CHAT-R items, resulting in a 99.72% overall correct classification across all classifications. The system provided a true negative rate of 99.27% and a positive predictive value of 78.90% for the entire sample. Toddlers from different subgroups exhibited high levels of performance, with White toddlers being correctly classified with 99.92% PPV and Black toddlers with 99.79% PPV. Compared to traditional M-CHAT-R scoring, the approach offers several advantages, such as the absence of laborious follow-up interviews, a decrease in human scoring errors, and an overall high precision for fewer items, which could result in a decrease in respondent burden.

Other questionnaire instruments are equally effective, as has been done with other questionnaires. By employing five machine learning algorithms to build the Quantitative Checklist for Autism in Toddlers (Q-CHAT) Tartariscosco and co-authors found that support vector machines were almost as effective at discriminating autistic children from typically developing peers with a 95% accuracy [21]. By utilizing SVM-recursive feature elimination, they discovered a subset of just 14 items that was 91% accurate, demonstrating that ML can optimize screening instruments by identifying the most informative items and optimizing for optimization. Research on the Q-CHAT-10, a shorter version of the Q-CHAT-10, showed that using just three highly discriminative items, the accuracy of the test was 83%, indicating potential for ultra-brief screening tools suitable for busy primary care settings.

The application of machine learning to datasets from public repositories has been done, with some caution, but also with crucial limitations in terms of universal scalability. Alkahtani and colleagues found that SVM and random forest algorithms demonstrated 100% accuracy on standard ASD datasets split 80/20 for training and testing, and that they used a similar algorithm in SPSS SSRI training and the CNV algorithm using it [2]. These findings are remarkable, but they are likely a result of overfitting to specific dataset features rather than robust real-world performance, as demonstrated by these datasets. Haque and colleagues demonstrated 100% accuracy with various algorithms for toddler and child datasets that were publicly accessible [25,26] in their experiment. Retrospective, curated datasets should be interpreted with caution when attempting to achieve near-perfect performance on near-perfect datasets, as real-world clinical populations display more heterogeneity and complexity.

- **Medical Claims Data and Health Records Analysis**

An innovative approach leverages routine healthcare utilization data to predict ASD risk before formal diagnosis. Chen and colleagues analyzed medical claims from 12,743 children with ASD and 25,833 controls, using logistic regression with LASSO and random forest models to predict ASD diagnosis at ages 18-30 months based on healthcare encounters from birth [8]. Their random forest model achieved an AUROC of 0.775 for prediction at 24 months. When predictor variables were separated by outpatient versus inpatient visits, performance improved substantially to AUROC 0.834, with 96.4% specificity and 20.5% positive predictive value at 40% sensitivity. This outperformed existing screening tools, which showed 38.8% sensitivity, 94.9% specificity, and 14.6% PPV in the same population.

The advantages of the claims-based approach cannot be matched by empirical data. Electronic medical records already

Vol. 3 No. 2 (2026): Oktober
DOI: 10.21070/ijhsm.v3i2.520

contain data, enabling clinicians and families to work without additional extra data collection burdens. It has the potential to identify children who may not be assessed by traditional screening due to their non-attendance at well-child visits or their parents' failure to identify developmental problems. Developmental and behavioral diagnoses, specialist referrals, and service utilization patterns are among the predictors that have been identified to closely approximate known ASD risk factors, including key predictors aligned with existing risk factors. Despite this, the positive predictive value (20.5%) implies that a significant number of children identified as high-risk would not be diagnosed with ASD in the future, necessitating careful consideration of how those results might be delivered and when clinical follow-up is needed.

Previous studies extended this approach using transformer-based deep learning ensemble models on linked maternal-newborn health administrative and birth registry data from 707,274 mother-offspring pairs in Ontario, Canada [15]. For their best-performing ensemble, we calculated ASD diagnosis in children aged 18 months to 5 years with 69.6% AUC, 70.9% sensitivity, and 56.9% specificity. Although these metrics are less rigor than some, the study proposes that stratifying risk by population level using routinely collected data could be a more effective approach than some other metrics. The potential for reducing diagnostic delays in children with a high risk of childhood illnesses may be addressed by enabling targeted outreach and assessment for these children.

Critical Discussion

The proliferation of AI-based ASD screening methods is an evidence-based technology in response to the high demand for accurate tracking tools. The most significant issue here is the disparity in measurement and real-world effectiveness, which often results in a disparity between the reported metrics and the actual effectiveness observed in the real world, making the performance metrics gap a significant problem. Many studies report very high sensitivity or specificity values, typically obtained when analyzing retrospective convenience samples using cross-validation or a train-test split on a single dataset [2], [5], [14]. Although this type of internal validation is valuable from the perspective of proof-of-concept, it gives us little idea about model applicability across different populations from various geographical, cultural and healthcare backgrounds. The border between these two kinds of validation – internal and external, is very important for clinical practice. Achenie and colleagues fANN model could get 99.72% correct classification using M-CHAT-R data; this performance was done using the population used for developing the model [1]. Chen and colleagues' claims-based models showed an AUROC of 0.834, yet were validated only within the MarketScan dataset [8]. Kim et al's video analysis system received a score of 0.83 AUC in nine hospitals in South Korea that represent higher geographic diversity, but its generalizability to other countries' healthcare systems is still uncertain [16].

External validation is true if models are tested on independently developed datasets derived from various populations. If feasible, professional prospective studies that emulate real-world clinical workflows should be conducted separately. In literature, positive predictive value is a more challenging issue than its role in predicting outcomes, and it is often overlooked. Despite high sensitivity and specificity in detection tools, the false positive rate for screening the general population for ASD is disturbingly high, even when screening tools for the same group of people have no recognizable gender. Chen et al. utilized 96.4% specificity in their specificity-based claims-based model, which could only achieve 20.5% PPV at a sensitivity level of 40% [8]. ASD diagnosis would impact less than one in four children classified as high risk, indicating a low incidence of those diagnosed with ASD. Even though it could still justify concentrating on screening criteria, it poses some relevant questions on disbursing resources, addressing family anxieties, and explaining risks in a way that minimizes the risk of family anxiety, and requires therapy before being evaluated for therapeutic intervention.

In addition, adequate and efficient specialist care services need to be developed by healthcare systems for children who screen positive for a disorder. There are some extra problems caused by the transparency of algorithms and interpretability, particularly for deep learning techniques. Neural networks and ensemble models often act as ‘black boxes’ which do not let you understand why a particular decision was made since they rely on complex, non-linear functions that combine features [6], [15]. This opacity causes issues of clinical trust as well as adoption.

If a model classifies a child as high risk, it is expected that clinicians or families should know what triggered the alarm. Conversely, simpler models such as logistic regression or decision trees provide a more intuitive notion but may lead to decreased accuracy [8], [18]. Some have dealt with this issue by employing feature importance analysis or/and/or by devising simplified decision models which resemble complex algorithms, however, provide interpretability [1], [18]. Currently, an effort remains ongoing to determine how to optimally balance performance against interpretability.

Basic issue with any kind of screening approach, be it even AI-driven. Autistic children, for example, show an almost infinite gradient when it comes to their symptoms, learning skills, and cognitive functions. This leads us to another challenge: the fact that what has been learnt from one population might not necessarily apply to another population that differs demographically or clinically. For example, Achenie et al found that their fANN model behaved differently in racial subgroups even if accuracy was high across all subgroups [1]. There are few studies which have specifically investigated how these algorithms perform according to several important demographic variables such as race, ethnicity, socioeconomic status, region and health access pattern. Thus, in case it remains unnoticed systemic bias due to biased training data or performances across groups may lead to aggravation of current health inequalities and disparities [4].

Privacy and security of data are more critical in the context of children’s health information, as well as their behavioral information. Video records of children, speech samples, and detailed medical history carry confidential information that should be carefully protected [7], [17]. Parents have to give consent to the usage of the information about their children, which means they should know where and how this data is stored or shared. The problem of privacy versus data utility is most pressing in the case of deep learning models given that they often require large training datasets. The federated

Vol. 3 No. 2 (2026): Oktober
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learning way is training models on distributed datasets without actual centralizing sensitive data so it may be a way out, but it also brings in some difficulties [15]. Being integrated into clinical workflows, this is the real buffering issue which has nothing to do with the technical performance. Even if the authors have a screening tool that is highly accurate, it will not help in any way to improve outcomes if they can't easily be incorporated into the delivery of healthcare services as they exist today.

Claims-based models are better in this aspect, but they have to work on interfacing with electronic health record systems and providing outputs that are actionable for clinicians without requiring additional data collection [8]. Development of image analysis technology requires that parents must record and upload video files what may bring obstacles connected with technology access, digital literacy and willingness to share recordings [16]. The combination of questionnaire-based ML systems with existing screening questionnaires should be attempted without increasing complexity for medical personnel, while maintaining the effectiveness of existing systems [1,2]. The user experience, work system integration, and change management are crucial elements of the user experience, work system integration, and change management phases of the launch. Is it possible to use AI in certain situations and not other alternatives, while still assuming a specific etiquette to describe the situation? Are there AI screening tools that should replace, improve upon, or design artificial intelligence tools to identify specific populations that are not considered screened by any other methods for their AI? Most researchers position their tools as adjuncts to rather than substitutes for clinical judgment [4], [16], [18].

AI systems might be most useful for preliminary assignment of risk at the population level among large numbers of children, prioritization of children for further detailed evaluation, or supporting clinical decision-making in environments with limited access to developmental specialists. If use cases are defined well, it ensures you have the right expectations and evaluation parameters. It is hoped that uniformity will be enhanced within the evidence base thus standardization on reporting and evaluation will still be encouraged. It is observed that studies have major disparities in this context which include who is diagnosing ASD (the diagnosing party, with whose help?), what ages are included, what groups are used for comparison purposes (typically developing children, those with a developmental delay, general population?), and what outcome measures are used. This heterogeneity prevents combining within an individual study and meta-analysis [11]. It would be advantageous for the field to provide guidelines on how studies should be reported as was done in STARD for diagnostic accuracy studies and TRIPOD for prediction models regarding essential items. Implementation of prospective validation studies which come along with pre-registered analysis plans would put off apprehensions about selective reporting as well as overfitting.

Research Gaps and Future Directions

With regard to AI-driven screening techniques for autism spectrum disorder (ASD), there are still many unanswered questions, although AI-driven ASD screening techniques have fostered many advances. Most importantly, the area lacks large-scale, prospective, multi-site validation studies to capture the real-world viability of the technology in a wide array of clinical setting/locations and pockets of the population. The instigators of the technology need to evaluate and document in a standardized and reliable manner the real-world viability and effectiveness of the AI-driven ASD diagnostic techniques. Most importantly, in addition to the historically provided diagnostic accuracy, the evaluation needs to capture the impact the technology has on the clinical outcomes and on the patients' overall developmental trajectory. There is an absence of studies in the implementation science of the technology, and the research on the barriers and the positive and negative factors impacting the prosthesis' adoption in a wide range of clinical settings and on the overall strategies for scaling.

The absence of standardized benchmark datasets and evaluation protocols has fostered a haphazard evaluation of the models and techniques available to diagnose. Many of the proposed models in the evaluation of AI technology to diagnose ASD have the potential to provide positive results. Many of these models, in theory, have the potential to provide the standard, diagnostic evaluation method to provide an accurate and reliable metric on the variety of available diagnostic and evaluation models. The available models have the potential to provide the various evaluation models and techniques on the models and techniques available to the diagnosis and evaluation of ASD in addition to the evaluative models and standards available to evaluation. The pre-registration of analysis plans coupled with the sharing of code and models has a significant role in enhancing reproducibility, and diminishing opportunities for positive results.

Understudied modalities necessitate further investigation. Despite the focus on the questionnaire-based and eye-tracking methods, other promising modalities such as wearable sensors, mobile app-based assessment, and physiological biomarkers have not been extensively explored. Combining modalities into one may produce information indicating a more specific fusion of modalities, but more systematic analysis is needed before considering combined strategies.

Achieving greater control over AI tools and preventing greater influence on health disparities requires prioritization of research on how to handle research on the intersection of algorithmic fairness and bias. Specifically, performance across demographic subgroups is evaluated through a systematic process, bias sources are investigated in training data and model design to determine potential bias sources, and bias reduction methods are developed. The development and validation of tools must involve active engagement with diverse communities, ensuring accessibility and participation from various perspectives.

In summary, the field needs to be better informed about how AI screening tools can assist in integrating into comprehensive diagnostic and intervention pathways. The screening process is a solitary and intricate process. Responsible implementation would be supported through research focusing on improving workflows, communication strategies for conveying risk information to families, and systems for ensuring appropriate follow-up.

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IV. Conclusion and Recommendations:

With the emergence of artificial intelligence and machine learning technologies, the potential to overcome long-standing barriers to early identification of autism spectrum disorders such as autism spectrum disorder is truly substantial. The technical performance of researchers has been impressive across a broad spectrum of modalities, including questionnaire automation, medical claims analysis, eye-tracking, video analysis, and speech recognition, in controlled settings. The potential of these tools lies in the ability to decrease diagnostic delays, improve screening accuracy, decrease dependence on specialist expertise, and aid in stratifying risk across populations.

Although major work remains to be done, it remains too early for these technologies to be deployed at scale in real world healthcare settings. In order to achieve these objectives, the field needs to address issues with positive predictive value challenges, improve algorithmic transparency, ensure fairness across different populations, ensure data privacy, and demonstrate meaningful impact on clinical outcomes through rigorous external validation, addressing rigor and sensitivity, and addressing questions of validity and privacy. AI-based screening tools can transform the early detection and treatment of autism and its associated outcomes for children and families living with autism, and with more research focused on these issues, AI-driven tools may be the keys to the future of autism screening.

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