

# AI-Driven Early Diagnosis of Autism Spectrum Disorder: Current Status and Future Perspectives

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**Abstract:** *Autism Spectrum Disorder (ASD) is a complex neurodevelopmental condition characterized by challenges in social communication, restricted interests, and repetitive behaviors. Early diagnosis and intervention are critical for improving long-term outcomes for individuals with ASD. In recent years, artificial intelligence (AI) and machine learning (ML) techniques have emerged as powerful tools for assisting in the early detection and diagnosis of ASD. This review provides a comprehensive analysis of the current state of AI-driven approaches for ASD early diagnosis, examining applications across various data modalities including neuroimaging, eye-tracking, electroencephalography (EEG), clinical questionnaires, and genomic data. We systematically review recent advances in deep learning, traditional machine learning, and multimodal fusion techniques, evaluating their diagnostic performance and clinical applicability. Furthermore, we discuss current challenges including data heterogeneity, model interpretability, algorithmic fairness, and the need for large-scale validation studies. Finally, we outline future research directions and potential pathways for translating AI-based diagnostic tools into clinical practice. Our findings suggest that while significant progress has been made, continued interdisciplinary collaboration between computer scientists, clinicians, and researchers is essential for developing robust, interpretable, and clinically deployable AI systems for the early diagnosis of ASD.*

**Keywords:** *Autism Spectrum Disorder, Machine Learning, Early Diagnosis, Deep Learning, Neuroimaging, Eye-tracking, EEG, Multimodal Analysis, Artificial Intelligence*

## 1. Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition affecting approximately 1-2% of the global population, with prevalence rates continuing to rise[1]. The disorder is characterized by persistent deficits in social communication and interaction, accompanied by restricted, repetitive patterns of behavior, interests, or activities[2]. The heterogeneous nature of ASD, combined with its complex etiology involving genetic, environmental, and epigenetic factors, presents significant challenges for early and accurate diagnosis.

Current diagnostic practices rely primarily on behavioral observations and standardized assessment tools administered by trained clinicians, a process that can be time-consuming, resource-intensive, and subject to diagnostic delays[3]. The average age of ASD diagnosis remains around 4-5 years in many regions, despite the fact that reliable identification is possible as early as 18-24 months[4]. This diagnostic delay represents a critical lost opportunity for early intervention, as research consistently demonstrates that early therapeutic interventions significantly improve developmental outcomes for children with ASD.

The emergence of artificial intelligence (AI) and machine learning (ML) technologies has opened new avenues for addressing these diagnostic challenges. AI-driven approaches offer the potential to identify subtle patterns in diverse data types that may not be apparent to human observers, enabling earlier and more objective detection of ASD-related characteristics[5]. These computational methods can process and integrate information from multiple sources, including neuroimaging data, eye-tracking recordings, electrophysiological signals, behavioral assessments, and genetic information, providing a comprehensive framework for ASD characterization.

This review aims to provide a comprehensive examination of the current landscape of AI-driven

approaches for ASD early diagnosis. We systematically analyze recent advances across different data modalities, evaluate the performance and limitations of existing methods, discuss critical challenges facing the field, and outline promising directions for future research and clinical translation.

## **2. AI Applications in ASD Early Diagnosis**

### ***2.1. Neuroimaging-Based Approaches***

Neuroimaging techniques, particularly structural and functional magnetic resonance imaging (MRI/fMRI), have been extensively employed in ASD research to identify brain-based biomarkers. Machine learning analysis of neuroimaging data has shown considerable promise for distinguishing individuals with ASD from typically developing controls.

Several studies have demonstrated the utility of supervised machine learning for diagnostic classification using large-scale neuroimaging datasets. Lanka et al.[6] addressed concerns about generalizability in neuroimaging-based classification, developing robust approaches that maintain performance across different imaging protocols and sites. Their work highlighted the importance of accounting for confounding variables such as head motion and scanner differences when building predictive models.

Deep learning techniques have emerged as particularly powerful tools for neuroimaging analysis. Qiang et al.[7] proposed a deep learning method for ASD identification based on the interactions of hierarchical brain networks derived from functional MRI data. Their approach leveraged the complex network structure of brain connectivity patterns, achieving improved classification accuracy compared to traditional feature-based methods. Similarly, Yang et al.[8] developed a graph attention network framework that explicitly models spatial-constrained sparse functional brain networks, demonstrating the value of incorporating neuroanatomical constraints into deep learning architectures.

Reproducibility remains a critical concern in neuroimaging-based ASD classification. Mellema et al.[9] focused on identifying reproducible neuroimaging features for ASD diagnosis, emphasizing the need for biomarkers that maintain stability across different datasets and imaging protocols. Their work underscored the importance of rigorous validation procedures to ensure that identified features reflect genuine ASD-related brain differences rather than dataset-specific artifacts.

Cross-site generalization represents another significant challenge. Bhaumik et al.[10] investigated domain-adaptive approaches for cross-site evaluation, demonstrating that models trained on data from one imaging center often experience performance degradation when applied to data from different sites. Their findings highlighted the need for domain adaptation techniques and harmonization methods to improve model generalizability.

### ***2.2. Eye-Tracking and Computer Vision Approaches***

Atypical visual attention patterns represent one of the most consistently reported characteristics in ASD, making eye-tracking a valuable tool for diagnostic assessment. Machine learning analysis of eye-tracking data has shown considerable promise for identifying ASD-related attention patterns.

Wei et al.[11] developed a machine learning framework for early ASD identification using eye-tracking data, demonstrating that computational analysis of gaze patterns can effectively distinguish children with ASD from typically developing peers. Their approach integrated multiple gaze features, including fixation duration, saccade characteristics, and attention allocation patterns, achieving high classification accuracy.

The detection of eye contact represents a particularly important application. Chong et al.[12] showed that deep neural networks can detect eye contact with accuracy comparable to human experts, opening possibilities for automated assessment of this critical social behavior. Their work demonstrated the potential for AI systems to augment clinical assessments by providing objective, quantifiable measures of social attention.

Beyond traditional eye-tracking, computer vision approaches analyzing facial images and videos have shown promise. Atlam et al.[13] developed explainable deep learning models for automated ASD identification from facial images, incorporating attention mechanisms to highlight facial regions contributing to classification decisions. This work addressed the important issue of model interpretability, which is crucial for clinical acceptance of AI-based diagnostic tools.

Home video analysis represents an emerging approach with significant potential for increasing diagnostic accessibility. Tariq et al.[14] demonstrated that machine learning analysis of short home videos can accurately classify ASD, enabling remote screening in underserved areas. Their mobile detection system showed promising results in prospective validation studies, suggesting potential for scalable deployment.

### ***2.3. Electroencephalography (EEG) Based Approaches***

EEG offers a non-invasive, cost-effective method for assessing brain function with high temporal resolution. Machine learning analysis of EEG signals has been extensively explored for ASD detection, with several studies reporting high classification accuracies.

Alotaibi and Maharatna[15] developed classification approaches based on EEG functional brain connectivity analysis, demonstrating that patterns of connectivity between brain regions can effectively distinguish individuals with ASD. Their work highlighted the utility of graph-theoretic measures for characterizing atypical brain network organization in ASD.

Feature extraction plays a critical role in EEG-based classification. Djemal et al.[16] proposed a computer-aided diagnosis system combining wavelet analysis, entropy measures, and artificial neural networks, achieving robust classification performance. The integration of multiple signal processing techniques allowed for comprehensive characterization of EEG characteristics associated with ASD.

Deep learning approaches have also been applied to EEG data. Baygin et al.[17] developed lightweight hybrid deep features for automated ASD detection, combining convolutional neural networks with traditional signal processing methods. Their approach demonstrated the potential for end-to-end learning from raw EEG signals while maintaining computational efficiency suitable for clinical deployment.

Barik et al.[18] explored fusion-based machine learning approaches using magnetoencephalography (MEG) signals, demonstrating that combining features from multiple frequency bands and brain regions can improve classification accuracy. Their work highlighted the value of multimodal signal fusion within the electrophysiological domain.

### ***2.4. Clinical Questionnaire and Electronic Health Record Approaches***

Clinical questionnaires and electronic health records (EHRs) represent rich sources of information for ASD prediction. Machine learning analysis of these structured and unstructured data types has shown considerable promise for risk stratification and early identification.

Rajagopalan et al.[3] developed a machine learning model for predicting ASD using a minimal set of medical screening and background history items available before 24 months of age. Their approach demonstrated that accurate prediction is possible using readily available clinical information, potentially enabling earlier identification than traditional diagnostic pathways.

Screening tool optimization represents another important application. Achenie et al.[19] developed machine learning strategies for autism screening in toddlers using the Modified Checklist for Autism in Toddlers, Revised (M-CHAT-R), demonstrating that computational approaches can improve screening efficiency while maintaining accuracy. Their work addressed practical challenges in pediatric screening, including time constraints and training requirements.

EHR-based prediction models have shown potential for population-level screening. Dick et al.[20] developed a transformer-based deep learning ensemble framework using health administrative and birth registry data, demonstrating the feasibility of identifying children at increased likelihood of developing ASD using routinely collected healthcare data. This approach could enable proactive outreach and early intervention for high-risk children.

Algorithmic fairness represents a critical consideration in clinical prediction models. Angell et al.[21] examined fairness in machine learning prediction of autism using EHR data, highlighting the importance of ensuring that AI systems perform equitably across different demographic groups. Their findings underscored the need for careful evaluation of model performance across subpopulations to prevent disparities in diagnostic access.

### **2.5. Genomic and Biomarker Approaches**

Advances in genomic technologies have enabled the identification of molecular signatures associated with ASD. Machine learning analysis of genetic, transcriptomic, and proteomic data has shown promise for developing biomarker-based diagnostic approaches.

Voinsky et al.[22] developed a machine learning-based blood RNA signature for ASD diagnosis, identifying a panel of differentially expressed genes that could distinguish individuals with ASD from controls. Their work demonstrated the potential for blood-based molecular biomarkers to complement behavioral assessments in the diagnostic process.

Proteomic approaches have also shown promise. Tang et al.[23] investigated plasma proteomics combined with machine learning methods for screening ASD biomarkers, identifying protein panels with diagnostic potential. The integration of proteomic data with clinical information may enhance diagnostic accuracy and provide insights into underlying biological mechanisms.

Ismail et al.[24] proposed a hybrid stacking ensemble model with Synthetic Minority Oversampling Technique (SMOTE) for predicting autistic genes, addressing the challenge of class imbalance in gene expression datasets. Their approach demonstrated improved prediction of disease-causing genes, potentially contributing to better understanding of ASD pathophysiology.

### **2.6. Multimodal Fusion Approaches**

Given the heterogeneous nature of ASD, multimodal approaches that integrate information from multiple data sources have gained increasing attention. These methods leverage the complementary information provided by different assessment modalities to improve diagnostic accuracy and robustness.

Several studies have demonstrated the value of combining neuroimaging with clinical data. Dekhil et al.[25] developed personalized ASD diagnosis systems integrating resting-state fMRI with clinical information, showing that multimodal fusion can achieve higher accuracy than single-modality approaches. Their work highlighted the importance of individualized models that account for inter-subject variability.

Abbas et al.[26] developed machine learning approaches combining questionnaire and home video screening, demonstrating that the integration of parent-reported information with behavioral observations can enhance early detection. Their findings suggested that multimodal screening approaches may be particularly valuable for increasing diagnostic accessibility in underserved communities.

Deep learning architectures have been increasingly applied to multimodal ASD classification. Aarthi and Kannimuthu[27] proposed hybrid deep learning models that integrate multiple data types, achieving state-of-the-art performance on benchmark datasets. The ability of deep networks to learn hierarchical representations from diverse inputs makes them well-suited for multimodal fusion tasks.

## **3. Current Challenges**

### **3.1. Data Heterogeneity and Generalizability**

One of the most significant challenges facing AI-driven ASD diagnosis is the heterogeneity of available datasets. Variations in data collection protocols, participant demographics, and diagnostic criteria across studies can substantially impact model performance when the models are applied to new populations[28]. The development of harmonization techniques and domain adaptation methods remains an active area of research.

### **3.2. Model Interpretability and Explainability**

The "black box" nature of many machine learning models, particularly deep learning approaches, presents challenges for clinical acceptance. Clinicians and patients require understandable explanations for diagnostic decisions[29]. The development of interpretable AI methods that provide transparent, clinically meaningful explanations represents a critical research priority.

### **3.3. Sample Size and Data Quality**

Many studies in this field are limited by relatively small sample sizes, which can lead to overfitting and poor generalization. The collection of large, well-characterized datasets with comprehensive phenotypic information remains essential for advancing the field[29].

### **3.4. Clinical Translation and Deployment**

The translation of research findings into clinically deployable tools faces numerous practical challenges, including regulatory approval, integration with existing clinical workflows, and demonstration of cost-effectiveness[30]. Close collaboration between researchers, clinicians, and healthcare administrators is necessary to facilitate successful clinical implementation.

## **4. Future Perspectives**

### **4.1. Large-Scale Collaborative Studies**

Future progress in AI-driven ASD diagnosis will require large-scale collaborative efforts that aggregate data from multiple sites and populations. Initiatives such as the Autism Brain Imaging Data Exchange (ABIDE) have demonstrated the value of data sharing, and expanded efforts along these lines will be essential for developing robust, generalizable models[31].

### **4.2. Integration with Digital Health Technologies**

The proliferation of digital health technologies, including wearable sensors and mobile applications, offers new opportunities for continuous, unobtrusive monitoring of behaviors relevant to ASD. Integration of these data streams with AI-based analysis could enable more comprehensive phenotyping and earlier detection[32].

### **4.3. Personalized Medicine Approaches**

Moving beyond binary classification toward personalized characterization of individual strengths and challenges represents an important future direction. AI systems that can predict treatment response and guide personalized intervention planning could significantly enhance clinical utility[33].

### **4.4. Ethical and Regulatory Frameworks**

The development of appropriate ethical and regulatory frameworks for AI-based diagnostic tools is essential. Issues including data privacy, informed consent, algorithmic fairness, and clinical liability must be carefully addressed to ensure responsible deployment of these technologies[34].

## **5. Conclusion**

AI-driven approaches for ASD early diagnosis have made significant progress in recent years, with machine learning models demonstrating promising performance across diverse data modalities. Neuroimaging-based methods have identified reproducible brain-based biomarkers, eye-tracking and computer vision approaches have enabled objective assessment of atypical attention patterns, EEG-based methods have provided cost-effective screening tools, and multimodal fusion approaches have shown the potential for enhanced diagnostic accuracy.

However, several challenges remain to be addressed before these technologies can be widely deployed in clinical practice. Issues of generalizability, interpretability, and clinical integration require continued attention. Future research should focus on large-scale validation studies, development of interpretable models, and close collaboration with clinical stakeholders to ensure that AI tools meet real-world healthcare needs.

The integration of AI technologies into ASD diagnostic pathways holds significant promise for reducing diagnostic delays, increasing access to assessment services, and ultimately improving outcomes for individuals with ASD and their families. Continued interdisciplinary collaboration

between computer scientists, clinicians, neuroscientists, and ethicists will be essential for realizing this potential and ensuring that AI-driven diagnostic tools are developed and deployed in a responsible, equitable manner.

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