Systematic Review

ARTIFICIAL INTELLIGENCE ASSISTED EARLY DIAGNOSIS OF AUTISM SPECTRUM DISORDER- A SYSTEMATIC REVIEW

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ABSTRACT

Background: Autism Spectrum Disorder (ASD) is a neurodevelopmental condition marked by social communication challenges, restricted interests, and repetitive behaviours, typically appearing in early childhood. Early diagnosis is essential for timely intervention and improved developmental outcomes in children with ASD. Conventional diagnostic tools, such as the Autism Diagnostic Observation Schedule (ADOS) and Childhood Autism Rating Scale (CARS), rely on behavioural observation and clinical expertise but can be time-consuming and resource-intensive. **Objective:** This review investigates the potential of Artificial Intelligence (AI) to improve the early diagnosis of ASD in children aged 6 to 24 months by analysing various AI methodologies, data sources, and their diagnostic performance compared to traditional methods.

Materials and Methods: A comprehensive literature review was conducted, focusing on AI applications, such as machine learning (ML) and deep learning (DL) models, that utilise behavioural data, eye-tracking metrics, neuroimaging techniques, and video analyses for ASD detection.

Results: The AI models demonstrated superior diagnostic accuracy and efficiency. The fusion of transfer learning with a nature-inspired dandelion algorithm for ASD detection (FTLDA-AASDDC) technique achieved a classification accuracy of 97.5%. Convolutional neural networks (CNNs) that analyse eye movement dynamics have shown promise as objective screening tools. Commercial systems, such as the FDA-approved Early Point, utilise AI and eye-tracking to detect atypical visual attention in toddlers. Emerging models, such as AutMedAI, provide scalable screening using minimal clinical features, which is beneficial in low-resource settings. AI-based tools offer faster, more objective, and highly accurate diagnoses than traditional methods.

Conclusions: AI-assisted early diagnosis of ASD represents a significant advancement, enabling early, precise, and accessible detection. These tools support timely interventions and improve the developmental trajectories and outcomes of children with ASD. Challenges remain in terms of dataset diversity, model transparency, and ethical concerns, which require further research and clinical integration.

Keywords: Autism Spectrum Disorder, Early Diagnosis, Artificial Intelligence, Machine Learning, Eye-Tracking

INTRODUCTION

Autism Spectrum Disorder (ASD) is a developmental condition that can be identified by restricted interests, repetitive behaviours, and challenges with social communication. Its intensity varies greatly, and symptoms typically first appear in early childhood. Although the precise origin of ASD is undetermined, environmental and genetic factors are thought to be involved.[1] Delayed diagnosis limits the effectiveness of early interventions, especially during sensitive developmental windows. As developed nations have greater access to diagnostic resources, the overall prevalence rate of autism is more widespread than in underdeveloped nations. In developed countries, the incidence is 85/10,000, while in underdeveloped countries, it is 155/10,000.[2] Developed countries have a higher pooled prevalence of ASD (0.79%) than underdeveloped countries (0.32%). Male-to-female ratios between 3:1 and 4:1 have also been documented in studies that emphasise particular developing nations. A 4.2:1 ratio is often mentioned in data from the US Centers for Disease Control and Prevention (CDC).[3]

Social interaction, communication, learning, and behaviour are profoundly affected in individuals with autism. Understanding these differences is essential for promoting inclusion and providing adequate support. Families often face challenges that disrupt daily routines, emotional well-being, and financial stability.[4] Common co-occurring psychiatric disorders include depression, Attention Deficit Hyperactivity Disorder (ADHD), and anxiety, along with medical issues such as sleep disturbances, gastrointestinal problems, and epilepsy.[5,6] Distress and behavioural difficulties may arise from hyper- or hypo-sensitivity to sensory stimuli like light, sound, touch, or textures.[7]

A core feature of autism is difficulty with verbal and nonverbal communication, leading to frustration, social isolation, and obstacles in education and employment. Difficulty interpreting social cues and forming relationships may further contribute to isolation and mental health concerns.[6] Conventional tools used to diagnose ASD, such as the Autism Diagnostic Observation Schedule (ADOS) and the CARS, depend mostly on observing behaviour and the expertise of trained professionals, which makes the process slow and requires a lot of resources.[8] These limitations highlight the need for more accessible, accurate, and scalable diagnostic approaches.

Early identification of autism and timely intervention services have become increasingly common in developing countries, especially through integration into routine primary healthcare systems.[9] These early efforts have shown significant benefits in enhancing the language, social interaction, and cognitive abilities of affected children. One such approach, the Early Start Denver Model (ESDM), is recommended for preschool-aged children with autism between 12 and 48 months.[10] This model is based on the understanding that early intervention during sensitive developmental periods can positively influence a child's developmental path. Additionally, the use of Artificial Intelligence (AI), leveraging machine learning (ML), deep learning (DL), and natural language processing (NLP), offers a promising solution for analysing complex datasets to improve early ASD detection and support clinical decision-making.[11]

AI can analyse complex datasets, such as behavioural patterns, eye-tracking data, facial expressions, and neuroimaging scans.[12] These tools can potentially enhance early ASD detection, even in low-resource settings, and support clinical decision-making with high accuracy and speed. Diagnostic screening for autism in very young children is increasingly important due to growing awareness and guidelines recommending checks at 18 and 24 months, though challenges remain in diagnosing those children under two.[13]

This literature review explores the current landscape of AI-assisted early diagnosis of ASD in children. It examines the methodologies employed, evaluates their diagnostic performance and limitations, and compares them with traditional approaches. The goal was to assess whether AI can meaningfully contribute to improving early ASD diagnosis and identify areas for future research and clinical integration.

Literature Review

Etiological conjecture

The following factors were associated with autism risk in the meta-analysis: low birth weight, small for gestational age, congenital malformation, low 5-minute APGAR score, feeding difficulties, meconium aspiration, neonatal anaemia, ABO or Rh incompatibility, foetal distress, multiple births, maternal haemorrhage, summer birth, abnormal presentation, and meconium aspiration. The following factors were not associated with the likelihood of autism: head circumference, high birth weight, post-term birth, anaesthesia, and aided vaginal delivery. Although research indicates that difficulties during pregnancy and infancy could enhance the chance of autism, the precise complications and extent of the impact have not always been consistent.[12]

Certain environmental exposures, metals from the air, occupation (lead, manganese, and mercury), and an array of pesticides are suspected to trigger autism. There were also suggestive patterns for phthalates and some volatile organic compounds, such as methylene chloride, trichloroethylene, and styrene.[13]

Several perinatal and neonatal factors have been linked to an increased risk of autism, including abnormal presentation, low birth weight, foetal distress, congenital malformations, and neonatal complications such as anaemia and meconium aspiration. Caesarean delivery showed a nonsignificant association.[14] In a study conducted with twins, it was revealed that for monozygotic twins, the meta-analysis correlations were practically flawless at 0.98. However, when the ASD prevalence rate was put at 5%, the dizygotic correlation was 0.53, and when a 1% prevalence rate was applied, it rose to 0.67.[15]

According to certain statistics, families may have varying genetic susceptibilities to autism. Families with two or more children diagnosed with ASD are potentially more likely to experience this. In certain families, other siblings have more noticeable broader autism phenotype (BAP) characteristics, and in first-degree relatives of individuals with ASD, research on these characteristics was referred to as BAP.[16]

Early identification and diagnostic red flags of autism

Children with autism often show early signs of social interaction and communication abilities. These include failing to maintain eye contact, no response to their name for nine months, and a loss of facial expressions such as joy, sorrow and surprise. By 12 months, they may stop engaging in simple interactive games, such as pat-a-cake, or show few or no gestures, such as waving goodbye. By 15 months, they might not share interests with others, for example, by showing objects they enjoy. At 18 months, they may not point to indicate interest, and by twenty-four months, they may struggle to recognise others' emotions. As they grow, some may stop interacting with other children by age three, show no pretend play by age four, and not participate in creative activities like singing or dancing by age five.

In terms of behaviour, children with autism may engage in repetitious and restrictive actions, such as stacking toys in a specific order and becoming upset if disrupted, repeating words or phrases (echolalia), having intense interests, needing fixed routines, or showing movements such as hand-flapping or body-rocking. They may also have unusual reactions to sound, smell, taste, or texture.

Additional characteristics can include delayed language skills, slow motor abilities, learning or cognitive delays, inattentiveness, impulsiveness, hyperactivity, seizures or epilepsy, unusual sleep or eating habits, and digestive issues such as constipation. Emotional responses may be unusual, including stress, anxiety, or being more fearful than expected; thus, recognising these signs early is very important. In a 2007 policy statement, the American Academy of Paediatrics (AAP) recommended autism screening for all children between 18 and 24 months to help ensure they receive timely, tailored treatment.[17,18]

MATERIALS AND METHODS

Study Selection

A total of 977 records were identified through database searches (n = 215) and other online sources (n = 762). During the initial screening, 330 publications, including 122 reviews and 208 meta-analyses focusing on randomised controlled trials (RCTs) and clinical trials (CTs), were identified. Among these, 474 studies were categorised as RCTs and CTs (44 RCTs and 430 CTs). A total of 211 studies were excluded for not being related to autism or autism spectrum disorder. Furthermore, 632 studies were excluded because they did not focus on children within the 6–24-month age group. Of the remaining studies, 130 specifically targeted children aged 6–24 months. Among these, eight studies focused on parent-delivered interventions, 15 explored the use of AI and ML, and another 15 utilised neuroimaging techniques. After the removal of 28 duplicate records, 28 studies met the inclusion criteria and were included in the final review.

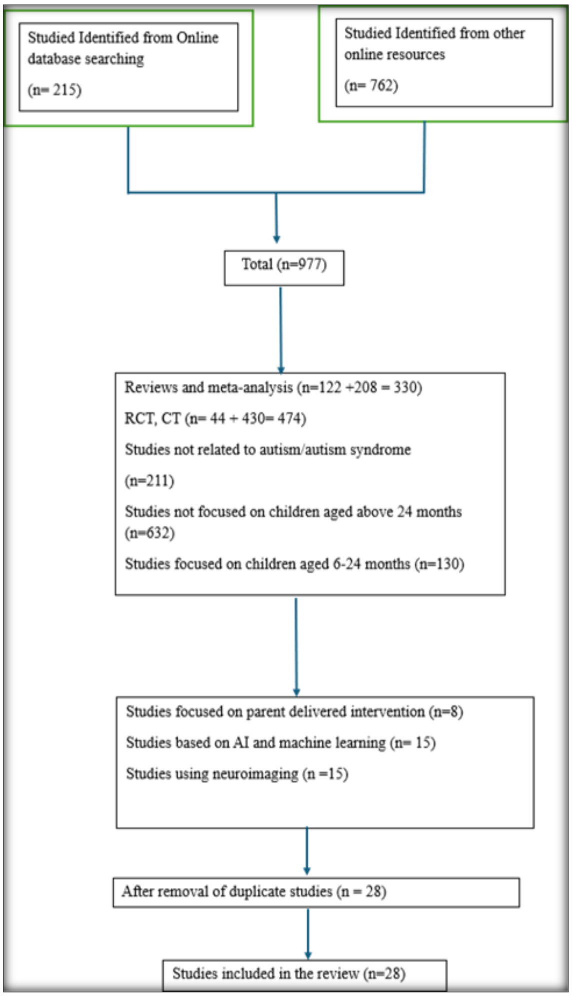

Figure 1: PRISMA 2020 Flow Diagram

Research objectives and guiding questions

The primary objective of this literature review was to assess and synthesise the current research on the application of AI in the early diagnosis of ASD.

Studies published within the last 10 years (2015–2025) will be included to ensure relevance to recent developments in AI technology and ASD research. The key questions to explore are how AI is being utilised to assist in the early diagnosis of ASD and what different AI techniques are being applied (for example, ML, DL, and NLP), what outcomes have been reported regarding the accuracy, efficiency, and effectiveness of AI systems in diagnosing ASD, and what challenges or limitations exist in using AI for early ASD diagnosis.

Literature search strategy

A comprehensive search strategy was developed to identify relevant studies. The following steps will be used: the academic databases PubMed and Google Scholar (for grey literature) will be searched to identify relevant studies. Keywords will be used in various combinations to identify the relevant studies. Examples of search terms include "AI", "ML", "DL", "ASD", "ASD Diagnosis", "Early Diagnosis", "AI in healthcare", " autistic screening", " neural networks for ASD", and "AI-assisted diagnostic tools". Search queries will include the use of Boolean operators such as AND, OR, and NOT to refine the results (for example, "AI AND ASD AND Early Diagnosis"). Only articles written in English will be included.

Inclusion Criteria

The studies included in the review consisted of peer-reviewed original research articles, such as empirical studies, clinical trials, and case studies, as well as reviews or meta-analyses focusing on AI applications for ASD diagnosis. These studies involved individuals diagnosed with ASD, preferably in the early stages. They specifically use AI or ML models, including neural networks and DL, to assist in diagnosing ASD using screening tools, behavioural analysis, or biomarker identification. Additionally, the studies report on the accuracy, sensitivity, specificity, and diagnostic effectiveness of AI models in early ASD detection and include comparisons between AI-based diagnostics and traditional methods, such as clinical diagnosis and psychological assessments.

Exclusion Criteria

Articles were excluded if they did not involve primary research, such as editorials, opinion pieces, or theoretical articles. Articles that focused on AI applications outside ASD diagnosis, including mental health conditions unrelated to ASD, were also excluded. Studies that concentrated on populations outside the ASD spectrum, such as those related to general mental health diagnoses or other neurodevelopmental disorders, were excluded. Studies that did not evaluate the diagnostic performance of AI tools or lacked empirical data, studies not published in English, and studies with incomplete or insufficient data regarding AI models or diagnostic outcomes were excluded.

Data extraction

For each selected article, the following data were extracted: study details, including author(s), year of publication, journal name, and study design; AI methods used, specifying the type of AI or ML algorithms, such as supervised learning, decision trees, or neural networks; sample size, including the number of participants and the inclusion/exclusion criteria; diagnostic tools and methods, detailing the type of AI tool used for diagnoses, such as software, apps, imaging techniques, or behavioural analysis; outcomes, such as diagnostic accuracy, sensitivity, specificity, and Area Under Curve (AUC); and the conclusion, highlighting the main findings of the study, particularly the efficacy of AI in early ASD diagnosis.

RESULTS

Diagnostic models and accuracy in ASD detection

ASD is characterised by deficits in social interaction, language, object utilisation and comprehension, IQ and learning, and verbal and nonverbal communication. Using chi-square-derived characteristics, the experimental results showed a 94% classification accuracy for diagnosing ASD.[19,20] AI integration into ASD diagnosis shows significant promise in healthcare. AI analyses a variety of data sources, including genetics, neuroimaging, behavioural patterns, and electronic medical records, to enable the early detection and individualised evaluation of ASD. Timely therapies are made possible by ML algorithms that demonstrate excellent accuracy in discerning ASD from neurotypical development and other developmental problems.[21]

The classification accuracy of the convolutional neural network model was promising. This implies that the information on gaze motion and its underlying dynamics can be successfully encoded by visuals. Based on the maximal information coefficient, potential relationships between eye movement dynamics and autism severity were investigated. The results demonstrate that eye-tracking, visualisation, and ML together have great potential for creating an impartial tool to help with ASD screening.

The FTLDA - AASDDC (Fusion of transfer learning with nature-inspired dandelion algorithm for ASD detection) technique demonstrated a higher accuracy value of 97.50% compared to other methods in the experimental validation, this method is carried out using face traits.[22]

Types of data used (behavioural, imaging, etc.)

Multiple strategies for detecting ASD have been developed, including psychological testing and neuroimaging modalities such as structural MRI and functional fMRI neuroimaging techniques are non-invasive MRI modalities. Several computer-aided design systems (CADS) based on AI have been created to assist specialised physicians in diagnosing ASD using fMRI and MRI.[23]

Comparison with traditional methods

Traditional ASD diagnostic methods, such as the ADOS, ADI-R, CARS, and M-CHAT-R, rely on behavioural observation and clinician judgment, which can be time-consuming and subjective. In comparison, AI-based approaches using ML and DL offer a faster, data-driven diagnosis with high accuracy, especially when CADS.[24]

Commercial applications of AI in ASD diagnosis

The Early Point Diagnostic System is an FDA-authorised tool designed to aid in the diagnosis of ASD in children aged 16–30 months. It uses AI and eye-tracking technology to assess visual attention patterns. Specifically, it measures how a child looks at social scenes displayed on a screen, capturing eye-movement data at a high frequency. This objective measurement assists clinicians in identifying atypical visual attention patterns associated with ASD.[25]

Research-based applications of AI in ASD diagnosis

Eye movement data analysed through ML models can accurately distinguish children with ASD from typically developing peers. This study confirms that AI-driven eye-tracking analysis offers significant potential for improving early autism diagnosis.[26]

AI analysis of behavioural data from videos is being actively explored to support the early detection of autism. An ML model that combines parent-completed questionnaires with the analysis of short home video clips. The system evaluates behavioural cues, such as eye contact, facial expressions, and motor behaviours, captured in natural settings. Their model demonstrated high accuracy in identifying children with autism, highlighting the effectiveness of integrating video-based behavioural data with AI-driven screening tools.[27]

AI analysis of questionnaire data enhances autism screening accuracy by applying advanced ML techniques to the phenotypic features. A model combining Recursive Feature Elimination with a Graph Neural Network (RFE-GNN) and a Phenotypic Feature Extractor (PFE) to diagnose ASD, this approach analysed data from screening tools such as the Autism Quotient (AQ) and achieved high diagnostic performance by identifying key behavioural traits. This demonstrates the potential of AI to refine traditional questionnaire-based assessments.[28]

Emerging predictive models

A recent model under development, AutMedAI, is an ML-based tool designed to predict autism in children under two years of age with nearly 80% accuracy. The model utilises a minimal set of easily accessible clinical and background features, such as developmental milestones, family history, and demographic data. This approach enables early, scalable ASD screening without requiring complex behavioural or imaging data, making it especially valuable in low-resource or primary care settings.[29]

DISCUSSION

The literature has repeatedly emphasised the importance of early ASD identification, mainly because it can greatly enhance developmental outcomes and provide prompt access to critical interventions.[30] For children with autism, early intervention can result in improved developmental trajectories and increased adaptive skills.[31] Notably, cognitive skills and behaviour have been shown to improve with early intervention, which frequently involves applied behaviour analysis. These sources mostly consist of in-depth reports of a child's developmental history from parents or other caregivers, as well as firsthand behavioural observations made by qualified medical professionals.[32]

The AAP suggests a multi-tiered strategy that includes standardised autism screening tests given at 18 and 24 months of age, in addition to developmental monitoring at each health supervision visit.[33] The Modified Checklist for Autism in Toddlers (M-CHAT), a first step in identifying toddlers who may be at increased risk for ASD, is one instrument that is frequently used in these screenings.[34] For more thorough assessments, professionals use standardised tests such as the ADOS™-2, which evaluates social interaction, play, communication, and repetitive and limited behaviours by interacting directly with the child. A systematic interview with parents called the Autism Diagnostic Interview™ Revised (ADI™-R) explores a child's behaviour in several important developmental categories.[35] CARS is a diagnostic tool used to diagnose autism in children and assess its severity. The CARS behaviour rating scale helps clinicians differentiate between children with autism and those with other developmental disorders. The scale consists of 15 items, each of which focuses on a different characteristic that is commonly associated with autism.[36]

Although screening can begin as early as 18 months, a trustworthy diagnosis by a qualified specialist is usually possible by the time the child is 2 years old.[37] This multifaceted approach emphasises how difficult it is to diagnose ASD and how a variety of data must be interpreted using expert clinical judgment. ASD can present itself in a variety of ways, making it challenging to depend only on conventional behavioural checklists to identify it. There are several reasons for this delay, such as the fact that some parents and even some medical professionals are unaware of the early warning signs that an ASD diagnosis carries a social stigma. Furthermore, access to screening and diagnostic services is severely hampered, especially in impoverished or rural areas, which adds to the delays in diagnosis.[8] These drawbacks highlight the necessity of creative strategies that can improve the precision, effectiveness, and accessibility of early ASD diagnoses.

Numerous AI approaches are being investigated, such as DL designs, CNNs, and ML algorithms, including support vector machines (SVMs), random forests, and ensemble methods. AI approaches analyse a wide variety of data types, from behavioural data gathered from standardised instruments and surveys to more innovative data sources, such as eye-tracking patterns, facial expressions recorded in home videos, speech traits, motor movements, physiological data from neuroimaging methods (e.g. fMRI, EEG), and even genetic information. The main goal of these AI apps is to automate or enhance current diagnostic procedures, which could result in a quicker and more accurate diagnosis of ASD.[11] While AI enhances diagnostic precision, it is intended to support not replace clinical assessment.

Using characteristics such as the age at which a child smiles for the first time, utters a short sentence, and exhibits eating issues, ML models have demonstrated a remarkable capacity to predict autism in very young infants. Because it coincides with the best time for early intervention, which can have more profoundly beneficial effects on a child's development, this early identification window is crucial. AI helps identify functional connectivity patterns, differences in cortical thickness, and increased grey matter volume.[38] This could result in a better understanding of important developmental periods and the effects of early interventions.

AI-assisted early diagnosis has broad practical implications for clinical practice and has great potential to improve outcomes for individuals with ASD and their families. AI can help provide more focused and efficient interventions by identifying subgroups within the autistic spectrum, advancing the management of ASD towards a better future.

Limitations

Most studies have been conducted in high-resource settings with curated datasets, limiting their generalisability to diverse or low-resource populations. The diagnostic accuracy varied widely owing to differences in algorithms and data types, with little standardisation. Small sample sizes and narrow age ranges reduced the robustness of the findings, and few studies offered longitudinal validation to confirm diagnostic stability over time. Additionally, limited model transparency and ethical concerns regarding data privacy and bias remain significant challenges.

CONCLUSION

AI has advanced ASD diagnosis by utilising data from behavioural observations, eye-tracking metrics, neuroimaging results, and questionnaire responses. Compared to traditional tools such as the ADOS, ADI-R, and M-CHAT-R, which require extended evaluation periods and clinical interpretation, AI-based approaches offer faster, more objective, and more accurate diagnostic outcomes. ML and DL models have shown exceptional performance, with techniques such as FTLDA-AASDDC achieving classification accuracies of 97.5%. The integration of eye-tracking and video-based analyses enhances the early and reliable identification of ASD symptoms.

Research-driven and commercial AI applications, such as the FDA-approved Early Point system and predictive AutMedAI model, demonstrate AI's growing role in early ASD detection. These systems enable accurate screening without complex procedures or advanced imaging technologies in low-resource settings. By recognising key behavioural signs, such as patterns in visual attention, facial expressions, and developmental milestones, AI tools help clinicians make precise diagnoses and support personalised interventions. This progress represents a transformative shift in ASD diagnosis, promoting early identification and improved outcomes.

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