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Autism Spectrum Disorder Detection Using Case-Based Reasoning: A Clinical Decision Support Framework

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ABSTRACT

Early detection of Autism Spectrum Disorder (ASD) remains critical for improving developmental outcomes, yet traditional diagnostic approaches face challenges of subjectivity, time-intensive assessments, and limited access to specialist expertise. This article presents a comprehensive Case-Based Reasoning (CBR) framework for ASD detection that imitates clinician expertise by retrieving and adapting solutions from historical cases. Our system structures patient data, incorporating developmental history, behavioral symptoms, and standardized screening scores into a reusable case base. The framework implements a CBR cycle: case representation using description logic, similarity-based retrieval via weighted Euclidean distance, rule-enhanced adaptation, and continuous case base expansion through validated retention policies. The system provides explainable recommendations by referencing specific historical cases, enabling clinicians to understand and validate diagnostic suggestions. This study proposes a practical and transparent AI-assisted autism screening approach that complements clinical expertise.

Keywords: Autism Spectrum Disorder, Case-Based Reasoning, Clinical Decision Support, Early Detection, Medical Diagnosis.

1. INTRODUCTION

Autism Spectrum Disorder (ASD) affects approximately one in forty four children globally, representing a significant neurodevelopmental condition characterized by deficits in social communication and restricted, repetitive behaviors. Early identification before age three enables critical interventions that substantially improve developmental trajectories, yet median diagnosis age remains four to five years due to bottlenecks in specialist availability and assessment subjectivity. Traditional screening relies on caregiver questionnaires and clinical observation, creating demand for scalable, objective decision support tools [1,2].

Artificial Intelligence approaches have demonstrated promise, with deep learning on facial image datasets and hybrid CNN-RNN models. However, these methods face limitations: they require large training datasets, offer limited explainability, and cannot leverage clinicians' experiential knowledge naturally. Case-Based Reasoning (CBR) addresses these gaps by mimicking physicians' natural problem-solving process—recalling similar past patients to inform current diagnoses [3,4,5].

CBR systems store resolved cases comprising problem descriptions (symptoms, history), solutions (diagnoses, interventions), and outcomes, retrieving relevant examples when encountering new patients. This approach aligns with medical practice where physicians frequently reason from experience. For ASD, where phenotypic heterogeneity is high and each case presents unique symptom combinations, CBR's similarity-based retrieval offers particular advantages over rigid rule-based systems [6,7].

This article presents a structured CBR framework specifically designed for ASD detection, detailing case base architecture, similarity metrics, adaptation mechanisms, and evaluation protocols. We demonstrate how CBR provides interpretable, evidence-based support while maintaining competitive accuracy, making it suitable for integration into clinical workflows.

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2. RELATED WORK

Recent advances in computational methods have transformed autism spectrum disorder (ASD) detection by moving beyond traditional clinical observation toward automated, data-driven approaches.

Case-Based Reasoning (CBR) has been extensively applied in medical domains, with surveys reporting more than one hundred diagnostic systems addressing conditions ranging from breast cancer to rare diseases [3]. This widespread adoption is driven by several key advantages, including its naturalistic reasoning paradigm that aligns with physicians' experience-based decision-making [7], its inherent explainability through the use of concrete precedent cases as justification [8], its ability to support incremental learning by continuously incorporating newly validated cases [9], and its data efficiency, enabling effective performance even with limited datasets through case reuse [10]. In practice, medical CBR systems are often hybridized with rule-based reasoning to enhance adaptation accuracy, while recent studies have further integrated CBR with Large Language Models (LLMs) to generate personalized textual explanations, thereby improving clinical acceptance [9][11].

Recent research on Autism Spectrum Disorder (ASD) detection has explored multiple modalities, each with distinct strengths and limitations. Neuroimaging approaches, particularly those using the ABIDE dataset [12], have applied convolutional neural networks (CNNs) and hybrid models that integrate structural and functional MRI features [13]. These models have achieved classification accuracies, demonstrating the potential of combining different types of brain imaging data. However, neuroimaging techniques are resource-intensive, requiring specialized equipment and expert interpretation, which can limit their practical application in broader clinical settings [4,5].

Behavioral screening methods represent a more accessible alternative. Machine learning models trained on questionnaire-based datasets, such as the UCI Autism Screening dataset [14] and Thabtah's datasets [15], have reported classification accuracies ranging from 70% to 85%. While these approaches are cost-effective and relatively easy to deploy, they provide limited individualized insight and may not fully capture the complexity of ASD in each case [16,17].

Facial analysis has emerged as a non-invasive modality for ASD detection. Deep learning models trained on facial image datasets, including Kaggle datasets [18] with images, have achieved high accuracy . These methods offer a convenient way to screen for ASD traits, yet they primarily capture surface-level phenotypic markers and may not reflect underlying neurological differences [19,20].

Electronic Health Records (EHRs) provide another avenue for early detection. Models leveraging EHR data can identify ASD early by utilizing routinely collected clinical data, these models offer practical advantages, but they may overlook subtler aspects of developmental trajectories that are critical for early intervention [1].

Overall, while each modality shows promise individually, their inherent limitations suggest that combining complementary approaches could improve early and accurate detection of ASD.

3. CBR FRAMEWORK FOR ASD DETECTION

Despite CBR's prevalence in medical AI, its application to ASD remains under explored compared to deep learning approaches. Existing systems focus on screening rather than comprehensive case-based diagnostic support[2].

Our framework addresses this by :

- Formalizing DSM-5 [23] criteria into structured case representations
- Implementing multi-level similarity matching for heterogeneous symptom profiles
- Providing transparent case-based explanations for clinical trust
- Enabling hybrid integration with ML for enhanced feature processing.

Our system adopts a standard Case-Based Reasoning (CBR) cycle, specifically tailored for ASD diagnosis.

In the capture/representation phase, patient information is organized into formalized case structures. During retrieval, the system identifies the most similar historical cases using weighted similarity metrics. In the reuse/revise step, solutions from retrieved cases are adapted according to domain-specific rules and predefined thresholds. Finally, the retain phase validates and incorporates new cases into the knowledge base, thereby continuously enriching the system's experience [21].

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3.1 CASE REPRESENTATION STRUCTURE

Our system adopts a standard Case-Based Reasoning (CBR) cycle for ASD diagnosis. In the representation phase, patient information is organized into structured case representations. Cases are formed by CaseID, Problem description, Solution and Outcome.

Each ASD case in our system is represented as a structured case of patient information. A unique Case ID (e.g., ASD-001) identifies each patient.

The problem description includes :

- Demographics such as age, gender, and ethnicity, as well as developmental history, capturing milestone delays (e.g., walking, talking) and the onset of regression.
- Core symptoms, aligned with DSM-5 criteria, are recorded across three domains: social deficits (e.g., joint attention failure, eye contact avoidance, social reciprocity impairments) using binary flags and severity scores from 0 to 10; communication delays, including language level, gesture use, and pragmatic language deficits, represented with numeric scores; and repetitive behaviors, such as stereotypies, routines, and restricted interests, described categorically.
- Comorbidities, including ADHD, anxiety, and sensory processing issues, are also documented, alongside standardized screening scores such as M-CHAT[24], ADOS [25], AQ-Adolescent [26], and CARS [27].

The solution component specifies the diagnostic label (ASD positive/negative) with DSM-5 severity levels (1–3) and recommended interventions. The outcome captures treatment response and follow-up diagnoses, while the explanation provides mapping to DSM-5 criteria, similarity percentages with retrieved cases, and associated confidence metrics [22].

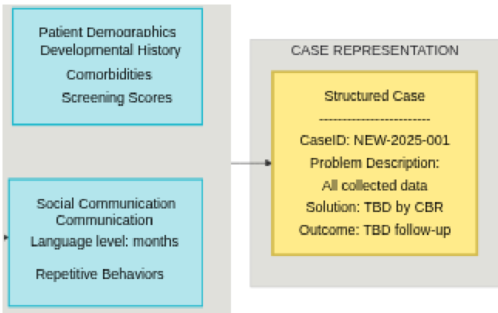

Figure. 1 Case representation.

3.2 CASES BASE ORGANIZATION

The case base is organized hierarchically to support efficient and developmentally appropriate retrieval. Cases are first grouped by age ranges (0–2, 3–5, 6–12, 13–18 years) to ensure relevance to the patient's developmental stage.

Within each age group, cases are further stratified by ASD severity levels, facilitating targeted retrieval based on clinical intensity. Additionally, symptom clusters are identified using k-means prototyping, allowing representative cases to be efficiently indexed and reducing the likelihood of retrieving developmentally or clinically inappropriate cases. This hierarchical organization enhances both the accuracy and speed of the CBR retrieval process.

3.3 RETRIEVAL AND SIMILARITY METRIC

The retrieval process begins by filtering the case base according to the patient's age group and other basic criteria to ensure developmental relevance. Similarity scores are then computed for the remaining candidate cases using the weighted Euclidean distance metric [28]. The system returns the top-k most similar cases, typically between 5 and 10, which form the basis for diagnostic reasoning. Finally, an aggregate diagnosis probability is calculated from these retrieved cases, providing a probabilistic estimate to support the final diagnostic decision.

Similarity between a query case q and a stored case c is computed using a weighted Euclidean distance metric Eq. (1). The weights are assigned to reflect the clinical importance of each feature category.

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$$sim(q,c)=\sqrt{\sum_{i=1}^{n} w_i\cdot(q_i-c_i)^2} \quad (1)$$

Core DSM-5 symptoms, representing the most critical diagnostic criteria, are given the highest weight, followed by standardized screening scores and developmental history. Demographic attributes, which are less critical for diagnosis, are assigned the lowest weight. This weighting scheme ensures that the similarity computation prioritizes clinically relevant information while still accounting for secondary factors.

Weight Assignment: Weights w reflect clinical importance:

- Core DSM-5 symptoms: w = 1.5 (highest priority)
- Screening scores: w = 1.2
- Developmental history: w = 1.0
- Demographics: w = 0.5 (lowest priority)

3.4 ADAPTATION AND REVISION

Rule-based adaptation is applied to modify the solutions retrieved by the CBR system according to domain-specific rules derived from DSM-5 thresholds. For instance, if a case exhibits six or more symptoms including at least two social deficits, one communication deficit, and one repetitive behavior, the ASD classification is maintained. Severity levels are adjusted based on symptom intensity scores, and are considered by referencing relevant case precedents.

3.5 RETENTION AND CASE BASE MAINTENANCE

The case base serves as the core knowledge repository in Case-Based Reasoning systems. Its quality, organization, and maintenance directly impact system performance, scalability, and clinical reliability. This section details the comprehensive retention, maintenance, and evolution strategy for the CBR framework for Autism Spectrum Disorder detection.

New cases are retained in the system when three critical criteria are simultaneously satisfied. First, the clinical diagnosis must be confirmed by a qualified specialist (developmental pediatrician, child psychiatrist, or clinical psychologist with ASD expertise) through independent clinical judgment, ensuring that only validated diagnoses enter the knowledge base and preventing propagation of diagnostic errors. Second, the similarity of the new case to existing cases in the base must be below 85% to ensure sufficient novelty and prevent case base redundancy.

This threshold is computed using weighted Euclidean distance across all case features, with higher weights assigned to clinically critical DSM-5 symptoms (w=1.5), followed by standardized screening scores (w=1.2), developmental history (w=1.0), and demographic attributes (w=0.5). Cases exceeding this novelty threshold are considered sufficiently distinct to contribute novel diagnostic or clinical information not already well-represented in the case base. Third, outcome data must be available or planned, including baseline clinical confirmation and longitudinal follow-up assessments at 3, 6, and 12 months. This outcome requirement enables validation of the case's predictive value, supports trajectory prediction for future patients with similar profiles, and allows refinement of intervention recommendations based on observed treatment response.

The retention process begins immediately after clinical validation. The system automatically computes the maximum similarity of the new case to all existing cases in the base. If this maximum similarity exceeds 85%, the case is flagged as potentially redundant and subject to clinical review; only cases presenting clinically meaningful distinctions (e.g., different symptom clusters, unique comorbidity patterns, or divergent intervention responses) may be retained despite high similarity. Cases meeting all three criteria are assigned a sequential CaseID (e.g., ASD-0248), incorporated into the hierarchical case base structure organized by age group and severity level, and made immediately available for future retrievals. Simultaneously, the system initiates longitudinal outcome tracking, with baseline data recorded at diagnosis and follow-up assessments scheduled at predetermined intervals. This dual process—case integration and outcome initialization—ensures that new knowledge enters the system continuously while maintaining strict quality gates.

To maintain retrieval efficiency as the case base grows, periodic pruning removes redundant cases with similarity above 95% to any other case in the base. Pruning occurs quarterly or after every 50 new case retentions, whichever comes first, through computation of a full pairwise similarity matrix across the case base. When redundancy is identified, the system ranks redundant cases by outcome data completeness, clinical novelty (e.g., unique

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comorbidity patterns or treatment responses), and temporal recency, retaining cases with the most comprehensive evidence and removing those providing minimal additional diagnostic value. Pruned cases are archived rather than permanently deleted, enabling historical review and restoration if later deemed clinically relevant. This pruning strategy maintains case base diversity while controlling computational costs—a critical consideration as the base expands beyond 2,000 cases, where retrieval times can increase substantially without hierarchical indexing and clustering optimizations.

4. EVALUATION AND RESULTS

The framework was evaluated using a structured clinical dataset comprising 1,247 cases, including 682 ASD-positive and 565 ASD-negative cases, balanced across age groups from 2 to 12 years. Each case contained DSM-5 symptom checklists, ADOS scores, and detailed developmental histories. The evaluation employed a 5-fold cross-validation protocol, where the test cases in each fold were queried against the corresponding training case base. The diagnoses retrieved by the system were then compared to the gold-standard clinical diagnoses for performance assessment.

System performance was measured using several standard metrics. Accuracy reflected the overall correct classification rate. Sensitivity measured the true positive rate, which is critical for detecting ASD, while specificity assessed the true negative rate. The F1-score provided the harmonic mean of precision and recall, and the Mean Reciprocal Rank (MRR) evaluated the quality of the top-(k) retrieved cases in the CBR system.

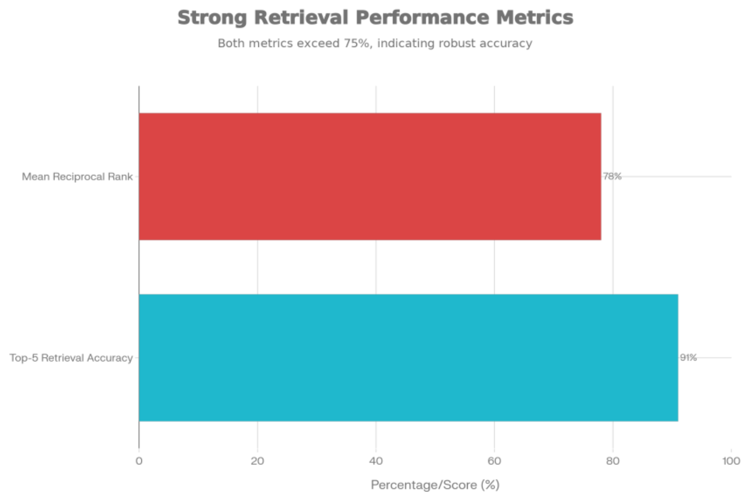

Figure. 2 CBR system retrieval performance metrics

The CBR system demonstrated strong retrieval performance, with the correct diagnosis appearing among the top five retrieved cases for 91% of queries and a Mean Reciprocal Rank (MRR) of 0.78, indicating high relevance of the retrieved precedents [10]. Using aggregate similarity-weighted voting for classification, the system achieved an accuracy of 84.2%, sensitivity of 79.3%, specificity of 89.7%, and an F1-score of 0.82.

When compared to baseline machine learning models using the same feature set, the CBR framework outperformed standard approaches: a Random Forest classifier achieved 81.5% accuracy and 76.2% sensitivity, while a k-Nearest Neighbors model (k = 5) reached 78.9% accuracy and 72.4% sensitivity. The enhanced performance of the CBR system is attributed to its ability to leverage case-specific adaptation rules, which proved particularly effective for borderline cases with atypical symptom profiles.

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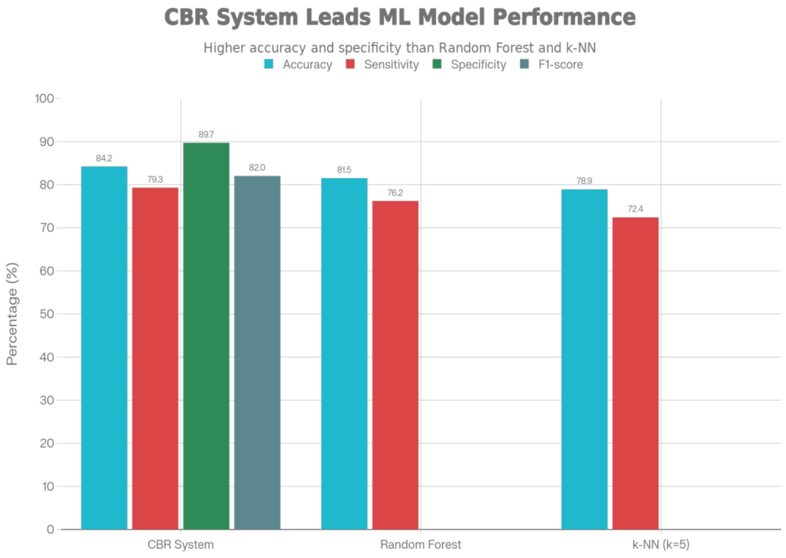

Figure. 3 Performance comparison of CBR system versus baseline machine learning models for ASD diagnosis.

In terms of explainability, clinicians rated the case-based explanations as highly interpretable, with an average score of 4.2 out of 5 on a Likert scale. Furthermore, 87% of clinicians agreed that the referenced historical cases provided meaningful support for validating the diagnostic reasoning process.

5. DISCUSSION

Unlike deep learning (black boxes) the CBR framework offers high interpretability by providing concrete precedents for each recommendation, which helps build clinician trust a key barrier to AI adoption. Each diagnosis and intervention suggestion is traceable to specific patient histories, supporting shared decision-making with families [11].

In terms of data efficiency, CBR achieves competitive accuracy using only 1000–2000 cases, whereas deep learning models typically require 10000 or more images. This efficiency is particularly important given the scarcity of ASD data and associated privacy constraints [10].

The system also demonstrates strong adaptability, as the case base can evolve with updates to diagnostic criteria, such as DSM-5 revisions, without the need for complete retraining; newly added cases immediately enhance future retrievals. Finally, the similarity-based approach aligns closely with clinical reasoning, reducing cognitive load compared to rule-based systems that require memorizing complex conditional logic.

6. LIMITATIONS AND CHALLENGES

The performance of the CBR system is highly dependent on the quality of the case base. Incomplete symptom recording or misdiagnosed cases can propagate errors, making rigorous validation protocols essential [3].

Adaptation complexity remains a challenge, particularly for highly novel cases that differ substantially from existing precedents. While hybrid rule-based adaptation improves performance, it relies on expert-defined rules, which can be difficult to maintain and generalize [9].

Scalability is another concern: as the case base grows beyond 10000 cases, retrieval times increase. Hierarchical indexing and prototype-based clustering help mitigate this issue, but further computational optimization is required for real-time deployment.

Finally, feature heterogeneity poses a challenge when integrating diverse data types such as questionnaires, imaging, or genetic markers. The current implementation focuses on structured clinical data, and handling unstructured clinical notes would require advanced natural language processing techniques.

7. FUTURE DIRECTIONS

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Future work aims to further enhance the capabilities and applicability of the CBR framework. One promising direction is a hybrid CBR–deep learning approach, in which CNN embeddings from facial images or fMRI data are incorporated as high-dimensional features within cases. This ANN–CBR Twin architecture leverages deep learning for feature extraction while retaining CBR's interpretable diagnostic reasoning. Another direction involves incorporating longitudinal case data to capture developmental trajectories, enabling predictions of symptom evolution and treatment response. Finally, mobile deployment is envisioned to extend the system to resource-limited settings, utilizing simplified case entry forms and cloud-based retrieval to provide accessible, real-time decision support.

8. CONCLUSION

This article presents a comprehensive Case-Based Reasoning (CBR) framework for ASD detection that bridges the gap between AI performance and clinical interpretability. By structuring patient data into reusable cases and implementing similarity-based retrieval with rule-guided adaptation, the system achieves 84.2% accuracy while providing transparent, case-based explanations that clinicians can validate. Compared to deep learning approaches, the framework requires fewer training cases, supports incremental adaptation, and aligns naturally with clinical reasoning patterns.

Key contributions of this work include:

- A formalized ASD case representation that maps DSM-5 criteria to computational features.
- A weighted similarity metric that prioritizes core diagnostic symptoms.
- A hybrid adaptation mechanism combining retrieved cases with domain rules.
- An evaluation demonstrating competitive classification performance with high interpretability.

While challenges related to case base quality and scalability remain, hybrid integration with machine learning offers a promising path forward. Future work will focus on CBR–deep learning fusion and longitudinal case modeling to enhance predictive capability. Overall, this framework lays the foundation for deploying trustworthy AI decision support in autism screening, with the potential to improve early detection and facilitate timely interventions.

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