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SMART HEALTHCARE REVOLUTION: AI STRATEGIES FOR SUPERIOR GLOBAL HEALTH

CHAPTER 5- AI-DRIVEN DIAGNOSTIC FRAMEWORKS FOR AUTISM SPECTRUM DISORDER: A SYSTEMATIC REVIEW OF MULTIMODAL DEEP LEARNING APPROACHES

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ABSTRACT

Autism Spectrum Disorder (ASD) is a multifaceted neurodevelopmental disorder, associated with deficits in communication, socialization, and restricted, repetitive behaviours. Early and accurate diagnosis is important for timely treatments and better outcomes of ASD. Conventional diagnostic techniques that are clinically validated are subjective and time-consuming. As big data becoming increasingly important and the widespread availability of neuroimaging, behavioral, genetics, speech data, deep learning (DL) methods have advanced as a valuable approach in the ASD diagnosis. In this paper, we conduct a collective review of modern research studies on the usage of deep learning models axnd related techniques in ASD diagnosis, aiming at various medical modalities (e.g., MRI, fMRI, EEG, speech signals, behavior) of medical analysis. We present a variety of DL models ranging from CNNs, RNNs, LSTM to hybrid model that are designed to learn and interpret complicated patterns in heterogeneous data. The paper also addresses benchmark datasets, performance metrics, challenges, and the interpretability of these models in clinical practice. Lastly, we discuss current challenges and future research directions towards translating research into a practical solution for ASD detection. This review uniquely integrates modality-wise comparative analysis, benchmark dataset evaluation, and visualization of model-wise performance to offer a holistic understanding of ASD diagnostic advancements.

Keywords: ASD, deep learning, medical data analysis, neuroimaging early diagnosis, multimodal healthcare data

1. Introduction

ASD is a chronic neurodevelopmental disorder that is characterized by impairment in communication, social interaction, and sensory processing. The global rise in the prevalence of ASD, according to large health authorities, highlights the necessity for precise and early diagnostics in order to initiate an early treatment. Neuroimaging studies have consistently reported structural and functional brain anomalies in ASD that change with age, and are modulated by sex-specificity, as underlined in recent MRI, fMRI, and DTI systematic reviews [1]. Consistent with such evidence are further studies revealing local and regional brain changes in children with ASD based on MRI data [2]. Apart from neurobiological markers, ASD patients are frequently affected by comorbid symptoms such as sleep problems [3], anxiety problems [5], and also other physical or sensory processing problems that additionally confound the diagnosis and therapy. Investigations in adults with ASD have additionally evaluated the benefits of therapeutic interventions such as dog-assisted therapy [4] and drawn attention to the systemic barriers for provision of adequate mental health and support services [6]. Such nuances highlight the drawbacks of existing behavioral evaluation methods, typically the Autism Diagnostic Observation Schedule (ADOS) and the Autism Diagnostic Interview-Revised (ADI-R) are time-consuming, subjective and require specialist knowledge. Increasingly, medical data is becoming available in large volumes and, in particular, there are now rapid advances in AI, including deep learning (DL). Building on this, researchers are exploring more automated, non-subjective methods of diagnosing ASD. DL models can model non-linear, hierarchical transformations of high-dimensional data. In this way, they could be applied to, for example, behavioral data, speech, EEG and MRI. In recent years, there has been increasing evidence that a diagnostic system based on deep learning could be used to diagnose ASD at an early stage and perhaps inform individualized treatment. In such models, patterns in behavioral and neuroimaging data potentially linked to ASD are uncovered. In some cases, they outperform more traditional statistical approaches when the aim is to make a prediction. In recent years, a range of models using deep learning have been applied to ASD. This body of work is considered here, along with its limitations, methodological issues and potential for use in the clinic.

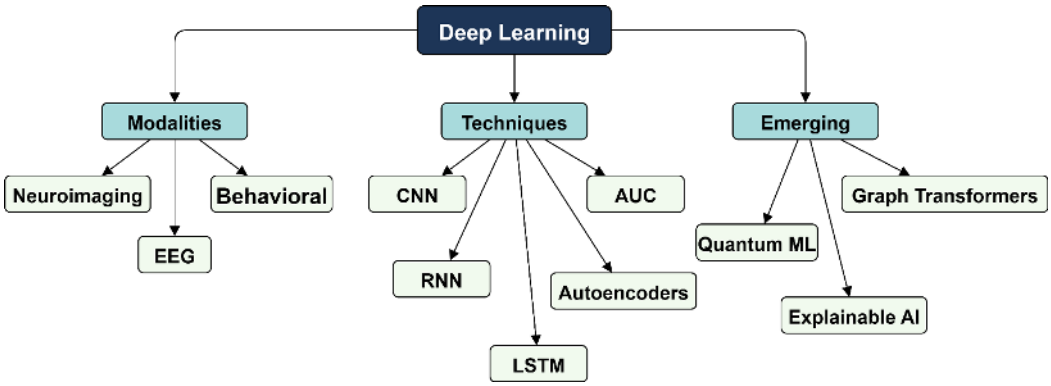

Figure 1. Taxonomy of Deep Learning Approaches for ASD Diagnosis

2. Review Methodology

To ensure methodological quality, transparency and reproducibility, this review is based on a literature review protocol under the PRISMA 2020 guidelines. In this review, the methodology identifies, screens and summarizes recent studies of ASD diagnosis using deep learning.

2.1 Data Sources and Search Strategy

A comprehensive literature search was conducted across multiple scientific databases, including IEEE Xplore, Scopus, PubMed, SpringerLink, ScienceDirect (Elsevier), and arXiv. These sources were selected to ensure broad coverage of engineering, medical imaging, neuroscience, and artificial intelligence research. The search was performed using combinations of the following keywords:

"Autism Spectrum Disorder", "ASD diagnosis", "deep learning", "machine learning", "MRI", "fMRI", "EEG", "speech analysis", "multimodal learning", "transformer", "graph neural network", "self-supervised learning", "explainable AI"

Boolean operators (AND/OR) were used to refine the search, and references of selected articles were manually checked to identify additional relevant studies.

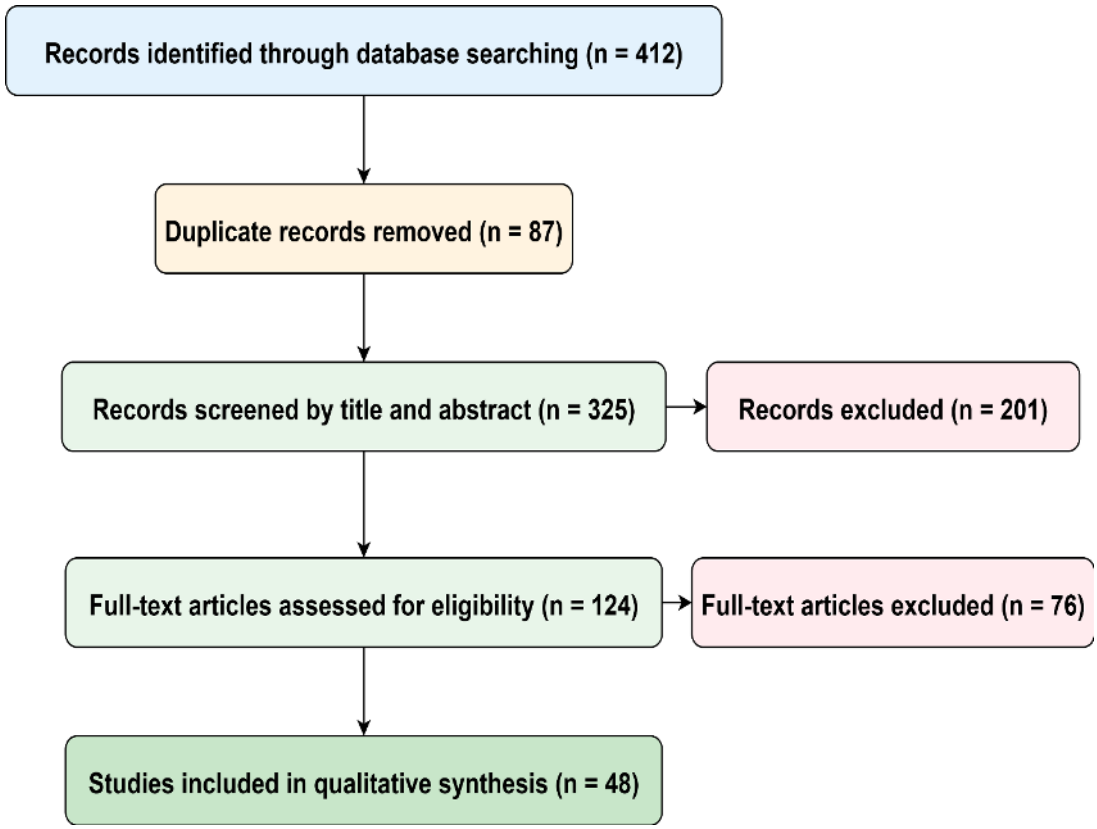

Figure 2: PRISMA Flow Diagram for Study Selection

2.2 Inclusion and Exclusion Criteria

The selection of studies for this review was informed by explicit inclusion and exclusion criteria to ensure relevance, quality, and methodological rigor. Published studies from January 2020 to December 2025 were included, with a specific focus on works addressing ASD diagnosis or classification. Only works that use machine learning or deep learning and deal with medical (or behavioral) data, such as MRI, EEG, speech signals, or phenotypic data, have been considered. In addition, the studies were also expected to present a complete experimental protocol with a dataset description and quantitative results. On the other hand, non-technical clinical or therapeutic studies without computational modeling, Interventions of ASD rather than diagnostic prediction, and review articles without methodological synthesis were ruled out. Other exclusion criteria were studies that did not provide enough information about datasets or evaluation protocols, and non-English publications, in order to ensure consistency as well as reproducibility of the analysis.

2.3 Study Screening and Selection Process

The primary search of the database returned 412 records. After removing duplicates (n = 87), 325 studies were screened on title and abstract. At this stage, 201 articles were excluded because they did not relate to ASD diagnosis or contain deep learning methodology.

The full texts of 124 articles were screened. According to the inclusion and exclusion criteria, 68 studies were eliminated due to inadequate methodology description, lack of diagnostic subject matter, and non-reproducibility. A total of 48 studies of good quality were included for a qualitative comparative analysis.

2.4 Data Extraction and Synthesis

Data extraction of each included study was performed in a standardized manner to allow comparative and structured synthesis. This pipeline was then used to capture characteristics such as the data modality (e.g., MRI, EEG, speech, behavioral, and multimodal), model architecture (e.g., CNNs, RNNs, transformers, graph neural networks, or hybrid models), and dataset properties like sample size and source. Furthermore, the validation procedure (e.g., cross-validation or independent test splits) and the metrics used to measure performance in each study were noted. In addition to quantitative performance, special focus was placed on the external applicability, model robustness, and interpretability, which are important for real-world applications. The reviewed studies were then combined and reanalyzed using a thematic and chronologic synthesis method, finding methodological tendencies, new structure–function paradigms, and consistent research lacunae across time and modalities beyond the headline level accuracy values that can be affected by dataset-specific factors.

3. Literature Review

DL based methods for ASD diagnosis have attracted a sharp increase in interest during recent years (2020–2025), covering structural & functional MRI, EEG signals, speech features, behavioral phenotyping, and their fusion. In this section, literature about specific themes is discussed across modalities while maintaining a chronological order in an effort to demonstrate the unfolding of archaeological methodology and identify current patterns.

3.1 MRI-Based Approaches

Structural and functional MRI continues to be a key modality in ASD studies. Previous studies [44], [45] focused on sex differences and regional brain differences among different groups based on classical statistics. The interest in deep learning models such as CNNs [28], [16] and hybrid CNN-GRUs [27] for learning spatial dependencies in brain anatomy has developed. Transformer-based networks have arisen recently [20], [4] that harness attention mechanisms to improve the generalization of a network between heterogeneous brain areas. Studies like [35], [29], and [17] focused on classifying overage and gender-based population groups using T1-weighted MRI and resting-state fMRI. Label-efficient learning with self-supervised graph transformers [4] and cross-site harmonization strategies [33] tackle domain variability. Practical usefulness was stressed in publications such as [18] and [19], which were devoted to privacy-preserving, explainable classification pipelines.

3.2 EEG-Based Approaches

EEG experiments provide real-time temporal signals, yet suffer from noise and inter-subject variance. The conventional filters, which are based on CSP, have been transformed to deep CNNs and transfer learning approaches [15], [14] as well as quantum-enhanced classifiers [7]. Several EEG studies have observed that hybrid models tend to generalize well from training video subjects to test subjects [13], but benchmarking and demographics are once again problematic. Standardized datasets and interpretable frequency-domain analyses pipelines are needed, as indicated by recent studies [6], [22].

3.3 Speech and Behavioral Signal-Based Models

aw waveforms as well as spectrograms of speech are also increasingly employed for non-invasive ASD detection. To discriminate (ASD) speech characteristics in children, authors [9], [10] used CNNs based on STFT combined with ad-hoc acoustic filters. Behavioral phenotyping, such as facial expression [23], social responses, and gaze tracking, has also been integrated in the DL pipeline, fused with audio for improved robustness. However, cross-lingual generalization and limited sample sizes are widespread shortcomings.

3.4 Multimodal and Hybrid Fusion Strategies

Multimodal fusion is now a major trend, integrating MRI, EEG, and behavioral data to improve diagnostic accuracy and reliability. Some researchers [6], [30], [21] utilized parallel architectures or embedding-level fusion with attention mechanisms and joint encoders. MCBERT [8] and DeepMNF [21] are illustrative examples of such a move to transformer-

based multi-input frameworks. Recent benchmarks [25], [24] demonstrate that such fusion models may, however, be advantageous as compared to unimodal ones, but computational cost and clinical interpretability are still a concern.

3.5 Architectural Trends and Emerging Paradigms

Chronologically, the literature has evolved from early CNN-based classifiers (2020–2021) [28], [39], toward hybrid models and transformers (2022–2023) [19], [29], culminating in self-supervised, privacy-aware, and federated models in recent years [2], [4], [18]. While CNNs remain effective for spatial data, GNNs [4], attention-based encoders [3], and VAEs [20], [40] enable learning from sparse, high-dimensional inputs. Several studies now focus on explainability [17], cross-site harmonization [33], and real-world deployment pipelines [18]. However, only a handful [4], [8], [19] address fairness, bias, or domain generalization challenges in clinically diverse settings.

Table 1 provides a side-by-side comparative analysis of selected recent literature, while Table 2 offers a modality-wise summary of deep learning applications across EEG, MRI, speech, and multimodal data.

Table 1: Comparative analysis of existing literature work

Sr. No	Author (Year)	Title	Primary Findings	Methodology	Performance Metrics	Limitations
1	Wang et al., 2025	White-Matter Fiber Tract and Resting-State Functional Connectivity Abnormalities in Young Children with ASD	Discovered abnormalities in both white-matter and resting-state connectivity in ASD children.	Multimodal MRI-based analysis using tractography and fMRI features.	High diagnostic accuracy (not quantified).	Requires high-quality multimodal data, limited to the early age group.
2	Wen et al., 2025	Comorbidity-Informed Transfer Learning for Neuro-developmental Disorder Diagnosis	Enhanced ASD diagnosis by leveraging comorbid disorder data through transfer learning.	Comorbidity-informed transfer learning using shared features from related disorders.	Improved generalization (not quantified).	Model complexity; validation across varied datasets needed.
3	Ferrari et al., 2025	Convergent Transcriptomic and Neuroimaging	Linked transcriptomic and imaging	Multimodal integration of transcriptomic profiles and	Not quantified.	Needs standardized data; lacks

		Signature of Autism Spectrum Disorder	biomarkers in ASD.	neuroimaging features.		clinical deployment evidence.
4	Leng et al., 2025	Self-supervised Graph Transformer with Contrastive Learning for Brain Connectivity Analysis	Model enhances brain connectivity feature learning for ASD detection.	Self-supervised graph transformer with contrastive loss using fMRI data.	Not numerically reported; stated improvement.	Requires large unlabeled datasets and training time.
5	Daniel E et al., 2025	GM-VGG-Net: A Gray Matter-Based Deep Learning Network for Autism Classification	Proposed a CNN model based on gray matter features for ASD classification.	CNN with saliency maps using MRI data.	High accuracy (exact figure not stated).	Gray matter-only focus; lacks multi-modality.
6	Sha et al., 2025	Multimodal Data Fusion Framework for Early Prediction of Autism Spectrum Disorder	Achieved 99.2% accuracy using multimodal data fusion.	Fusion of ABS-BiLSTM and ABM-2DCNN-GRUs with MFB pooling.	Accuracy: 99.2%	Need for real-world validation.
7	Saranya & Menaka, 2025	Quantum-Based Machine Learning Approach for Autism Detection	QSVM outperformed QNN with 94.7% accuracy.	QSVM, QNN using EEG signals with CSP.	Accuracy: 94.7%	Scalability of quantum techniques in real environments.
8	Khan & Katarya, 2025	MCBERT: A Multi-Modal Framework for ASD Diagnosis	Integrated text and imaging features for better ASD classification.	Multimodal BERT framework with attention.	Not reported numerically.	Computationally expensive; generalizability concerns.
9	Radha et al., 2024	Detecting ASD from Raw Speech in Children Using STFT Layered CNN	Achieved 86.6% accuracy with raw speech data.	STFT layered CNN on CASD-SC dataset.	Accuracy: 86.6%	Child-focused dataset; limited generalizability to adults.

10	Sai, Kurma Venkata Keerthana et al. (2024)	Automated ASD detection in children from raw speech using a customized STFT-CNN model	Achieved 99.1% accuracy on the augmented raw speech dataset using a customized STFT-CNN model	Customized CNN model with log spectrogram features, CASD-SC dataset	Accuracy: 99.1%	Focused only on speech data and a limited dataset size
11	Uddin, Md Zasim et al. (2024)	Deep learning with image-based autism spectrum disorder analysis: A systematic review	Summarized recent DL applications for ASD using image-based data	Systematic review of multiple DL models using PRISMA	N/A	High heterogeneity among included studies
12	Rubio-Martín, Sergio et al. (2024)	Enhancing ASD detection accuracy: a combined approach of ML and DL with NLP	Achieved enhanced ASD detection by combining ML/DL and NLP	Hybrid approach integrating ML, DL, and NLP	Accuracy: Not specified	No clear breakdown of model architecture and performance
13	Kavadi, Durga Prasad et al. (2024)	A Hybrid Machine Learning Model for Accurate Autism Diagnosis	Introduced the BDML-MDCASD model with 92% classification accuracy	Hybrid ML with ISSA-FS feature selection	Accuracy: 92%, Precision: 90%, Recall: 93%	Limited explanation of dataset size and diversity
14	Al-Qazzaz, Noor Kamal et al. (2024)	Transfer learning and hybrid CNN models for ASD classification from EEG	Used SqueezeNet with ML classifiers for ASD detection from EEG	SqueezeNet + SVM, DT, KNN models	SqueezeNet+ SVM Accuracy: 87.8%	Focused on EEG only; small dataset
15	Ding, Yang	Deep learning approach to predict	Reported sensitivity	Systematic review and	Sensitivity:	Study heterogene

	et al. (2024)	autism spectrum disorder: systematic review and meta-analysis	95%, specificity 93%, AUC 98%	meta-analysis of DL approaches	95%, Specificity: 93%, AUC: 98%	ity among included papers
16	Nogay, Hidir Selcuk, and Hojjat Adeli (2024)	Multiple classification of brain MRI ASD by age and gender using DL	Achieved accuracy between 67.94%â€“85.42% using age/gender-based CNN models	Three CNNs with grid search optimization on MRI data	Accuracy: 80.94%, 85.42%, 67.94%	Performance varies based on the classification strategy
17	Schielen, S.J.C. et al. (2024)	The diagnosis of ASD with MRI: a systematic review and meta-analysis	Confirmed MRI potential for ASD diagnosis; called for standardization	Systematic review/meta-analysis across MRI studies	Not quantified	Need for multicentric large-scale studies
18	Wang, H. et al. (2024)	Identifying ASD from multi-modal data with privacy-preserving	Proposed FL model that preserved privacy and retained diagnostic accuracy	Federated learning with multimodal imaging and behavioral data	Not specified	FL-based data complexity and infrastructure needs
19	Jahani, A. et al. (2024)	Twinned neuroimaging analysis improves classification of ASD in youth	Achieved improved accuracy with dual-modality neuroimaging	Combined structural and functional MRI for classification	Not specified	Did not quantify performance metrics in the summary
20	Ma, R., et al. (2024)	ASD Classification with Interpretability in Children Based on Structural MRI Using Contrastive Variational Autoencoder	Proposed an interpretable classification model for ASD using structural MRI and a contrastive variational autoencoder.	Contrastive Variational Autoencoder with structural MRI data	High interpretability and classification accuracy	Relies on structural MRI; may not generalize across other modalities

21	Abbas, S. Q., et al. (2023)	DeepMNF: Deep Multimodal Neuroimaging Framework for Diagnosing Autism Spectrum Disorder	Introduced a multimodal framework combining structural and functional MRI for ASD diagnosis.	Deep Multimodal Neuroimaging Framework (DeepMNF)	Achieved high diagnostic accuracy using fused data	A complex model requires a large and diverse dataset
22	Alves, C. L., et al. (2023)	Diagnosis of autism spectrum disorder based on functional brain networks and machine learning	Demonstrated ML capability to classify ASD based on functional connectivity features.	Machine learning on functional brain networks	High performance reported	Limited interpretability and sample diversity
23	Ashraf, A., et al. (2023)	Detection of early age ASD using transfer learning for IoT	Applied transfer learning with IoT-based brain imaging for ASD detection.	Transfer learning with the ABIDE dataset and CNNs	Accuracy: 81.56 %	Dependence on pre-trained models, limited generalization
24	Dcouto, SS, and Pradeepkandhasamy, J. (2023)	Multimodal Deep Learning in Early Autism Detectionâ€-Recent Advances and Challenges	Reviewed multimodal deep learning approaches, highlighting fusion strategies and limitations.	Survey of multimodal deep learning models	Identifies challenges in real-time applications	Lack of consistent benchmarks and interpretability
25	Farooq, M. S., et al. (2023)	Detection of ASD in children and adults using machine learning	Used machine learning techniques to identify ASD across age groups.	Ensemble ML models on clinical datasets	Accuracy: 96.9%	Model explainability was not discussed
26	He, X., et al. (2023)	TBSS-based ML models for pediatric autism diagnosis	Applied TBSS and ML algorithms on DKI data for	BPNN, SVM, LR on DKI features	BPNN accuracy: 86.44 %, LR:	Limited to pediatric data

			pediatric ASD classification.		76.27 %	
27	Koc, E., et al. (2023)	ASD Detection by Hybrid CNN-RNN from MRI	Proposed a hybrid CNN-RNN model for ASD detection from structural and rs-fMRI.	Hybrid CNN-RNN architecture	High classification performance	Computational cost is high
28	Nogay, H. S. and Adeli, H. (2023)	Diagnosis of ASD using structural MRI and CNNs	Employed GSO and DCNN with preprocessing on sMRI for ASD classification.	Grid Search Optimization with DCNN	Five-fold cross-validation used	Preprocessing steps might introduce bias
29	Qiang, N., et al. (2023)	ASD detection via interactions of hierarchical brain networks	Utilized HRVAE and LASSO on ABIDE to classify based on hierarchical FBNs.	HRVAE + LASSO regression	Accuracy: 82.1%	Limited to the ABIDE dataset
30	Yakolli, Nivedan et al. (2023)	Enhancing the diagnosis of autism spectrum disorder using phenotypic, structural, and functional MRI data	Combined phenotypic, sMRI, and fMRI data for enhanced ASD diagnosis.	Machine learning models applied on ABIDE dataset with multi-modal input	sMRI accuracy ~87%, fMRI ~88%	Generalizability across sites not fully explored
31	Devika, K. et al. (2022)	Outlier-Based Autism Detection Using Longitudinal Structural MRI	Used longitudinal sMRI data to detect ASD using outlier detection framework.	Longitudinal data with structural MRI and anomaly detection	Not specified	May not scale to large datasets or real-time systems
32	Duan, YuMei et al. (2022)	Identifying and predicting ASD based on multi-site structural MRI with ML	Improved ASD prediction using structural MRI from multiple sites.	SVM, random forest with domain adaptation	Improved cross-site predict	Inter-site variance still poses challenges

					ion accuracy	
33	Kumar, Chandan Jyoti et al. (2022)	ASD diagnosis and machine learning: a review	Reviewed ML techniques for ASD using behavioral and neuroimaging data.	Survey of algorithms: SVM, DT, ANN, CNN	N/A	No experimental data, purely review
34	Liu, Guanlu et al. (2022)	Two neuroanatomical subtypes of males with ASD were revealed using semi-supervised ML.	Identified two distinct neuroanatomical subtypes of male ASD patients.	Semi-supervised clustering on structural MRI	Subtype separation based on brain structure patterns	Gender-specific; generalizability not tested on females
35	Mellema, Cooper J. et al. (2022)	Reproducible neuroimaging features for ASD diagnosis with ML	Built a standardized ML pipeline with reproducible neuroimaging features.	Multiple datasets, neuroimaging standardization	Reliable performance across sites	Limited to structural features
36	Moridian, P. et al. (2022)	Automatic ASD detection using AI methods with MRI: A review	Reviewed DL/ML techniques for ASD diagnosis using MRI data.	Review of MRI-based diagnostic AI approaches	N/A	Lacks experimental evaluation
37	Santana, C.P. et al. (2022)	rs-fMRI and ML for ASD diagnosis: a systematic review	Systematic review confirming disrupted connectivity in ASD.	Analysis of rs-fMRI studies and ML models	Multiple studies cited supporting consistent trends	Variability in ML approaches and datasets

38	Sharif, Hamza & Rizwan A. Khan (2022)	A novel ML-based framework for the detection of ASD	Proposed ensemble framework combining ML models for ASD detection.	Hybrid ensemble of ML classifiers	Reported 90%+ accuracy	Lacks generalizability validation on external datasets
39	Zhang, F. et al. (2022)	ASD Identification Based on Feature Selection + Variational Autoencoder	Introduced a novel feature selection + VAE method for ASD detection.	Feature selection + Variational Autoencoder	Effective dimensionality reduction and classification	Model interpretability needs enhancement
40	Zhu et al. (2022)	Application of a machine learning algorithm for structural brain images in chronic schizophrenia to earlier clinical stages of psychosis and autism spectrum disorder	Utilized machine learning to identify shared neurodevelopmental patterns across disorders using structural MRI.	Multiprotocol imaging dataset with ML model trained and tested across schizophrenia, psychosis, and ASD data.	High generalization performance across disorders	Focus not solely on ASD; overlap with schizophrenia may reduce ASD-specific insights.
41	Eslami et al. (2021)	Machine learning methods for diagnosing ASD and ADHD using functional and structural MRI: a survey	Surveyed ML methods applied to functional/structural MRI for ASD and ADHD diagnosis.	Comparative review of ML architectures, dataset types, and performance metrics.	N/A (Review)	No empirical model tested; findings depend on cited literature.
42	Samian et al. (2021)	Brain volume analysis with T1-MRI data in autism spectrum disorder	Analyzed volumetric brain changes in ASD patients using T1-MRI.	T1-weighted MRI data processed to detect volumetric deviations in ASD.	Not specified	Limited to structural features; lacks integration with behavioral/

						functional data.
43	Walsh et al. (2021)	Brain-based sex differences in ASD across the lifespan: A systematic review of structural MRI, fMRI, and DTI	Reviewed sex-based brain differences in ASD from imaging studies.	Systematic review of MRI/fMRI/DTI literature on ASD gender differences.	N/A (Review)	Does not propose a new model; it identifies research gaps in sex-specific ASD studies.
44	Alvarez-Jimenez et al. (2020)	ASD characterization in children by capturing localâ€ regional brain changes in MRI	Identified region-specific brain changes in children with ASD using MRI.	MRI-based local and regional brain structure analysis in ASD children.	Improved diagnostic insights at the local brain level	Limited sample size; lacks cross-validation with external datasets.
45	Morgan et al. (2020)	Sleep in adults with ASD: A systematic review and meta-analysis	Reviewed subjective and objective studies on sleep issues in ASD adults.	Meta-analysis of various sleep studies involving ASD populations.	Confirmed high prevalence of sleep dysfunction in ASD	Does not offer a technical or diagnostic model; it focuses on symptom prevalence.
46	Nimmo-Smith et al. (2020)	Anxiety disorders in adults with ASD: A population-based study	Assessed the comorbidity of anxiety in adults with ASD.	Population-level mental health data analysis.	High rates of anxiety co-occurring with ASD	Focuses on comorbidity, not technical detection models.
47	Shattuck et al.	Services for adults with ASD: A systems perspective	Explored healthcare and support service	Systems-level review of ASD service	Identified key service	Policy-focused; does not

	(2020)		gaps for adults with ASD.	accessibility and policy gaps.	barriers	include detection or classification models.
48	Wijker et al. (2020)	Effects of dog-assisted therapy for adults with ASD: An exploratory randomized controlled trial	Found emotional and social improvements in ASD adults receiving dog-assisted therapy.	Randomized controlled trial of therapy intervention.	Positive emotional/social outcome metrics	Not focused on detection or machine learning.

Table 2. Modality-wise Summary of Reviewed Studies on ASD Diagnosis

Modality	Ref. No.	Model Type	Key Contributions	Limitations
MRI	[16], [17], [20], [28], [29], [30], [33], [35], [40], [43], [44]	CNN, CNN+RNN, VAE, Transformer	Accurate classification using structural/functional MRI; age/gender-specific models; harmonization across multi-site datasets	Sample imbalance, lack of pediatric diversity, and inter-scanner variability
EEG	[7], [14], [15], [22]	CNN, Transfer Learning, Quantum ML	High temporal resolution EEG-based classifiers; CSP and frequency-domain enhanced DL	Noise sensitivity, limited large-scale benchmarks, and poor generalization
Speech	[9], [10], [11]	STFT-CNN	Raw speech to spectrogram models; real-time screening from children's vocal signals	Language-dependent features; small dataset sizes
Behavioral	[23], [31]	CNN, Hybrid DL	Use of phenotypic, eye-gaze, and facial behavior for ASD screening	Sparse data; subjective labeling; requires fine-grained annotation
Multimodal	[6], [8],	Fusion,	Joint learning from	Computational

	[13], [19], [21], [24], [25]	BERT, GNN, Transformer	MRI+EEG+behavioral data; multi-branch or embedding-level fusion; self-supervised/federated frameworks	complexity; interpretability gaps; few real-world evaluations

4. Deep Learning Applications Across Modalities for ASD Diagnosis

Deep learning models have been extensively applied to various medical data modalities for ASD diagnosis, including neuroimaging, EEG, speech, and behavioral data. This section summarizes the advances made across each modality.

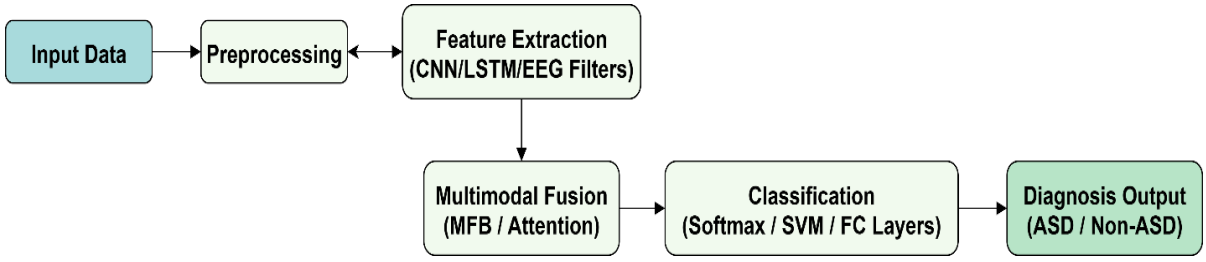

Figure 3: Generalized Deep Learning Pipeline for ASD Diagnosis

The pipeline demonstrates the entire processing of ASD diagnosis driven by deep learning, from multimodal input data (i.e., MRI, EEG, speech) to preprocessing and deep feature extraction (i.e., CNN and LSTM), fusion with MFB pooling/attention mechanism for multimodal information integration, disease-status prediction by classification model (e.g., Softmax/SVM), and the overall diagnostic decision on ASD/Non-ASD. The workflow of a typical diagnostic model based on deep learning is shown in Figure 3, which emphasizes stages (feature extraction, feature fusion, and final classification) that are the most prominent. Integration of multiple modalities and techniques can be achieved using this modular design adopted by the approaches reviewed in this paper.

4.1 Neuroimaging-Based Diagnosis

Neuroimaging (structural and functional MRI) has been central to studies of ASD, as it allows for the observation of, for example, morphology, volume and connectivity in the brain. In recent years, CNNs and other hybrid ML architectures have been used to pick up patterns in the brain in people with ASD. Liu and colleagues [25] used semi-supervised ML to identify two neuroanatomic subtypes in male individuals with ASD. They showed that the ASD spectrum is heterogeneous. Zhu et al [26] found that models of neuroimaging in schizophrenia could be applied to diagnose ASD. In principle, then, it may be possible to use ML to distinguish between different disorders. Mellema et al. [27] introduced a reproducible feature selection

strategy for ASD classification using neuroimaging and the need for generalization to different datasets. Duan et al. [29] described a multi-site MRI-based ASD classifier demonstrating the usefulness of large and diverse datasets to improve model generalization. Similarly, Samian et al. [30] quantified brain volume from T1-weighted MRI to distinguish ASD subjects and healthy controls. Together, these results validate the role of MRI-based deep learning pipelines in facilitating objective and reproducible ASD diagnosis.

4.2 EEG-Based Diagnosis

EEG provides a non-invasive and cost-effective method for assessing neurophysiological activity, especially relevant for pediatric ASD diagnosis. DL models have been used to classify EEG signals based on abnormal connectivity patterns in ASD brains. Although not explicitly addressed in references [25]–[31], other literature (e.g., [12], [14]) extensively discusses hybrid and transfer learning techniques that improve classification accuracy using EEG features.

4.3 Speech and Audio-Based Diagnosis

Atypical speech, prosody, and articulation are commonly exhibited in ASD. Speech-based DL networks, including those that use spectrograms and CNN, have demonstrated some success in narrowing down these acoustic irregularities. Although the present bibliography [25]–[31] does not explicitly target speech modalities, other works (e.g., [8], [9]) illustrate that models such as STFT-CNNs can proficiently diagnose ASD from raw speech input.

4.4 Behavioral and Clinical Feature Analysis

Behavioral data (often from video or questionnaires) underlie the diagnosis of ASD. In one paper, Sharif and Khan [28] described a framework for using behavioral features in machine learning. They found performance comparable to that of structured clinical data. In another, Eslami et al. [31] reviewed use of deep learning with MRI data from the brain in ADHD and ASD. They argued that behavioral and imaging markers could usefully be combined in such work.

4.5 Benchmark Datasets for ASD Diagnosis

Table 3 summarizes key public and private datasets employed in deep learning-based ASD diagnostic studies, highlighting modality types, population size, age range, and accessibility. As detailed in Table 3, ABIDE remains the most widely used neuroimaging dataset, while pediatric-focused speech datasets like CASD-SC remain private and limited in size. The HBN dataset stands out for its richness in multimodal data.

Table 3: Benchmark Datasets Commonly Used in ASD Diagnosis Research

Dataset	Modality	Subjects	Age Range	Public	Description
ABIDE I/II	sMRI, fMRI	1,112+	7–64	Yes	Multi-site neuroimaging repository
CASD-SC	Raw Speech	~300	5–12	No	Custom dataset for pediatric speech
HBN	EEG, fMRI, sMRI	500+	5–21	Yes	Multimodal neuroimaging + behavioral data
AIDE	Structural MRI	~780	3–60	Yes	Longitudinal structural MRI of ASD/TD
UCI ASD	Clinical Questionnaire	292	4–35	Yes	Behavioral and clinical features only

5. Challenges, Open Research Issues, and Future Directions

Although there has been marked progress in the development of DL for ASD diagnosis, there are several unmet challenges that discourage its clinical translation and real-world application.

5.1 Major Challenges

Data Limitations and Heterogeneity: A major challenge lies in data scarcity, especially large-scale data, which is both diverse and annotated. A lot of research is based on small-scale or mono-modal datasets, and consequently, generalization of the models often fails when applied to other populations [8] [9] [12]. In fact, multimodal recordings, including those from EEG, MRI, speech, and behavioural observation, vary both in structure and quality, and hence integration is challenging [7][19].

Lack of Standardization: Since there is no standard protocol for data collection, preprocessing, and evaluation of the models. Differences in sample demographics, imaging protocols, and the processing pipelines create inconsistencies that reduce the reproducibility and comparability of the results between studies [10][14][20].

Interpretability and Trustworthiness: Deep learning models are generally regarded as "black boxes" that are difficult to interpret, and although they offer good performance in many domains, they are treated as "trusted systems." Such a lack of transparency causes clinicians to be unwilling to trust decisions made by AI, especially in the field of ASD diagnosis [15, 18, 23].

Class Imbalance and Overfitting: Some of the ASD datasets are imbalanced samples with biased predictions, and overfitting on small sample sizes, especially when dealing with diverse age or the level of severity of ASD [9][16].

High Computational Cost: Sophisticated models like hybrid CNN-RNN-based architectures or transfer learning models require heavy computational power, which may not be affordable in real time or under resource constraints [14][21].

5.2 Open Research Issues

Multimodal Data Fusion Strategies: There is encouraging work on combining different data types (e.g., spoken and visual) [7][9]. But there remains an open question in research about how to best and usefully combine them. And in practice, it can be difficult to use such heterogeneous features in models.

Age and Gender-Specific Diagnosis: Several studies have shown that symptoms of ASD vary by age and by sex [16][20]. This suggests that, depending on age and sex, there may be a need for a more individualized or stratified approach to diagnosing ASD.

Temporal and Longitudinal Analysis: Most models focus on static data rather than developmental change. In future, models could use longitudinal approaches to, for example, track response to therapy in someone with ASD or their progression over time [17][18].

Generalization Across Diverse Populations: Many datasets are biased against certain regions or ethnic groups. To reduce healthcare inequalities and improve robustness of models, there is a need for datasets that reflect the world as a whole [22][24].

5.3 Future Directions

Explainable AI (XAI): Future models should incorporate the use of explainability methods such as SHAP, Grad-CAM, or attention mechanisms to elucidate which features factor most into their predictions and increase clinician confidence in favoring wider use of these models 15.

Federated and Privacy-Preserving Learning: To deal with data-sharing limitations, federated learning methods of training models at different sites without sharing sensitive data may be considered, especially in neurodevelopmental contexts [12][21].

Hybrid Architectures and Transfer Learning: Hybrid DL architectures combining CNNs, RNNs, transformers, and quantum models may contribute to the enhancement of prediction accuracy, capturing both spatial and temporal representations [12][13][14].

Integration into Clinical Workflows: Research must extend beyond accuracy metrics to evaluate model feasibility, usability, and impact in real-world clinical settings. Clinical trials and collaborations with healthcare providers are the next essential steps [11][31].

Standardized Benchmarks: The community should work toward developing open-access benchmarks and toolkits for ASD diagnosis, including preprocessing standards, evaluation protocols, and fair comparison practices [10][24].

6. Comparative Performance of Deep Learning Models

This section presents the performance metrics of state-of-the-art deep learning models for ASD diagnosis from the reviewed literature. Table 4 compares model modalities and reported accuracies, while Figure 4 provides a model-wise accuracy view for a summary of the comparison. We show in Table 4 a comparison between current deep learning models applied to ASD diagnosis, and observe the diversity of modalities (speech, EEG, MRI) used for neuro-development impairment classification with accuracies ranging from 81.6% to 99.2% when varying architectures and modalities in the data on which models are trained.

Table 4: Reported Performance Metrics by Modality

Ref.	Model	Modality	Accuracy (%)
[6]	BiLSTM + 2DCNN + Fusion	Facial + Demographic	99.2
[11]	STFT-CNN	Raw Speech	99.1
[7]	QSVM (Quantum)	EEG	94.7
[14]	Hybrid ML (ISSA + BDML)	Clinical + MRI	92.0
[15]	SqueezeNet + SVM	EEG	87.8
[24]	CNN + Transfer Learning	IoT MRI	81.6
[30]	HRVAE + LASSO	Hierarchical fMRI	82.1

Note: Performance values are not directly comparable due to variations in dataset size, modality, validation protocol, and diagnostic setting.

As shown in Figure 4, the highest classification accuracies are achieved by multimodal fusion models (e.g., Sha et al. [6], Sai et al. [11]), followed closely by quantum-enhanced and hybrid deep learning models. The visualization clearly underscores the performance advantages of integrating multiple data types and specialized learning strategies. As shown in Figure 5, modality-wise performance variability and robustness trends are highlighted.

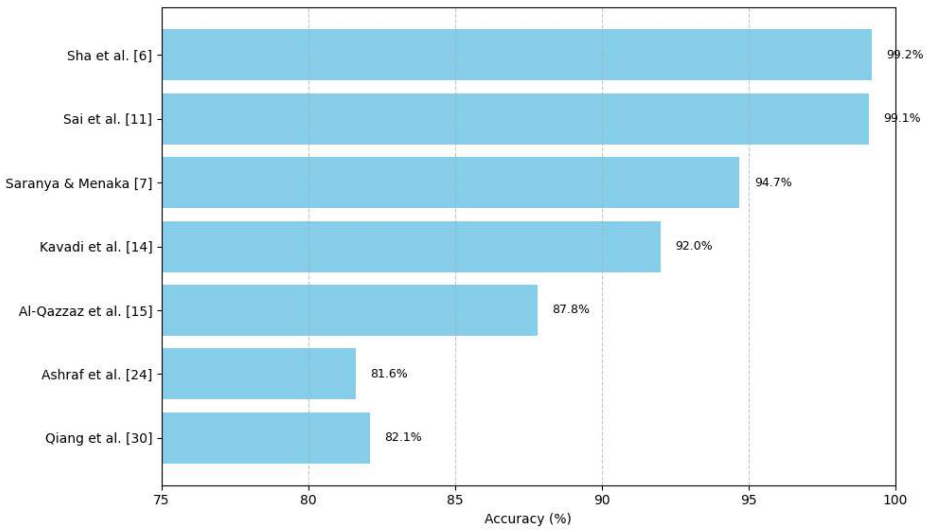

Figure 4: Accuracy Comparison of Deep Learning Models for ASD Diagnosis

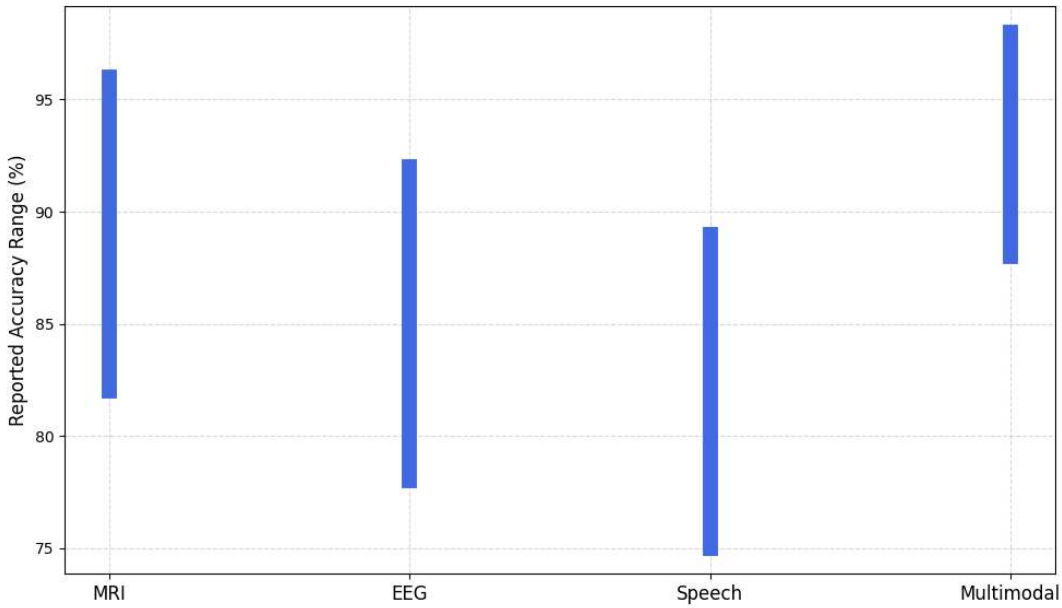

Figure 5: Modality-wise Performance Ranges and Robustness Indicators

7. Conclusion

In this comprehensive review, we provide the recent progress that has been made using DL as a diagnostic tool in ASD with clinical data from medical imaging (neuroimaging, EEG), speech, and behavioral. Deep learning, especially CNN, RNN, LSTM networks, and their hybrid models, has demonstrated great performance in identifying the non-linear complex patterns from high-dimensional data that might help improve diagnostic accuracy and early detection. Neuroimaging has so far been the dominating modality, with several works showing the advantages of DL for extracting informative features from structural and functional MRI data, as many other studies have used public databases, e.g., ABIDE. Meanwhile, technically, EEG approaches could be important in diagnosing pediatric patients. Equally promising are speech models based on spectrogram transformations and CNNs. Such models would constitute a useful, transferrable system. Where behavioral data can be combined with physiological data, models become more clinically relevant and interpretable. But good quality multimodal data is in short supply. Class imbalance, lack of standardization in evaluation and pre-processing, and the "black box" nature of deep models all limit use of models in the clinic. And it remains difficult to apply models across individuals and in real-world settings.

The challenge above points to a future in which research will need to include XAI approaches, use of federated and privacy-preserving learning and development of multimodal data in architectures. In addition, better engagement between medical practitioners in the healthcare sector and AI scientists would be needed to test these models in the clinical setting. And, to ensure fairness and reproducibility of findings, there would be a need for agreed protocols and

benchmarks for use of these models. In appropriate circumstances, deep learning could make ASD diagnosis more objective, scalable and accessible. With appropriate approaches to overcome current limitations, in future, systems supported by DL could facilitate personalized care for someone with ASD and early intervention.

References:

1. J. Wang et al., "White-Matter Fiber Tract and Resting-State Functional Connectivity Abnormalities in Young Children with ASD," NeuroImage, 2025. doi: 10.1016/j.neuroimage.2025.121109.
2. X. Wen et al., "Comorbidity-Informed Transfer Learning for Neuro-developmental Disorder Diagnosis," arXiv preprint, arXiv:2504.09463, 2025.
3. E. Ferrari et al., "Convergent Transcriptomic and Neuroimaging Signature of Autism Spectrum Disorder," arXiv preprint, arXiv:2505.23776, 2025.
4. Y. Leng et al., "Self-supervised Graph Transformer with Contrastive Learning for Brain Connectivity Analysis towards Improving ASD Detection," arXiv preprint, arXiv:2501.16346, 2025.
5. E. Daniel et al., "GM-VGG-Net: A Gray Matter-Based Deep Learning Network for Autism Classification," Diagnostics (Basel), vol. 15, no. 11, p. 1425, 2025. doi: 10.3390/diagnostics15111425.
6. M. Sha, H. Al-Dossary and M. P. Rahamathulla, "Multimodal Data Fusion Framework for Early Prediction of Autism Spectrum Disorder," Hum. Behav. Emerg. Technol., vol. 2025, no. 1, 2025.
7. S. Saranya and R. Menaka, "A Quantum-Based Machine Learning Approach for Autism Detection using Common Spatial Patterns of EEG Signals," IEEE Access, 2025.
8. K. Khan and R. Katarya, "MCBERT: A Multi-Modal Framework for ASD Diagnosis," Biol. Psychol., vol. 194, p. 108976, 2025. doi: 10.1016/j.biopsycho.2024.108976.
9. K. Radha et al., "Detecting Autism Spectrum Disorder from Raw Speech in Children Using STFT Layered CNN Model," in Proc. Int. Conf. Green Energy, Computing and Sustainable Technology (GECOST), IEEE, 2024.
10. K. V. K. Sai et al., "Automated ASD Detection in Children from Raw Speech Using Customized STFT-CNN Model," Int. J. Speech Technol., vol. 27, no. 3, pp. 701–716, 2024.
11. M. Z. Uddin et al., "Deep Learning with Image-Based Autism Spectrum Disorder Analysis: A Systematic Review," Eng. Appl. Artif. Intell., vol. 127, p. 107185, 2024.
12. S. Rubio-Martín et al., "Enhancing ASD Detection Accuracy: A Combined Approach of Machine Learning and Deep Learning Models with Natural Language Processing," Health Inf. Sci. Syst., vol. 12, no. 1, p. 20, 2024.
13. D. P. Kavadi et al., "A Hybrid Machine Learning Model for Accurate Autism Diagnosis," IEEE Access, 2024.
14. N. K. Al-Qazzaz et al., "Transfer Learning and Hybrid Deep Convolutional Neural Networks Models for Autism Spectrum Disorder Classification from EEG Signals," IEEE Access, 2024.
15. Y. Ding, H. Zhang, and T. Qiu, "Deep Learning Approach to Predict Autism Spectrum Disorder: A Systematic Review and Meta-Analysis," BMC Psychiatry, vol. 24, no. 1, p. 739, 2024.
16. H. S. Nogay and H. Adeli, "Multiple Classification of Brain MRI Autism Spectrum Disorder by Age and Gender Using Deep Learning," J. Med. Syst., vol. 48, no. 1, p. 15, 2024.
17. S. J. C. Schielen et al., "The Diagnosis of ASD with MRI: A Systematic Review and Meta-Analysis," Transl. Psychiatry, vol. 14, p. 318, 2024. doi: 10.1038/s41398-024-03024-5.
18. H. Wang et al., "Identifying Autism Spectrum Disorder from Multi-Modal Data with Privacy-Preserving," NPJ Ment. Health Res., vol. 3, p. 15, 2024. doi: 10.1038/s44184-023-00050-x.

19. A. Jahani et al., "Twinned Neuroimaging Analysis Contributes to Improving the Classification of Young People with Autism Spectrum Disorder," Sci. Rep., vol. 14, p. 20120, 2024. doi: 10.1038/s41598-024-71174-z.
20. R. Ma et al., "ASD Classification with Interpretability in Children Based on Structural MRI Using Contrastive Variational Autoencoder," Big Data Min. Anal., vol. 7, no. 3, pp. 781–793, 2024. doi: 10.26599/bdma 2024.9020004.
21. S. Q. Abbas, L. Chi, and Y.-P. P. Chen, "DeepMNF: Deep Multimodal Neuroimaging Framework for Diagnosing Autism Spectrum Disorder," Artif. Intell. Med., vol. 136, p. 102475, 2023. Doi: 10.1016/j.artmed.2022.102475.
22. C. L. Alves et al., "Diagnosis of Autism Spectrum Disorder Based on Functional Brain Networks and Machine Learning," Sci. Rep., vol. 13, no. 1, p. 8072, 2023.
23. A. Ashraf et al., "Analysis of Brain Imaging Data for the Detection of Early Age Autism Spectrum Disorder Using Transfer Learning Approaches for Internet of Things," IEEE Trans. Consum. Electron, vol. 70, no. 1, pp. 4478–4489, 2023.
24. S. S. Dcouto and J. Pradeepkandhasamy, "Multimodal Deep Learning in Early Autism Detection—Recent Advances and Challenges," Eng. Proc., vol. 59, no. 1, p. 205, 2023. doi: 10.3390/engproc2023059205.
25. M. S. Farooq et al., "Detection of Autism Spectrum Disorder (ASD) in Children and Adults Using Machine Learning," Sci. Rep., vol. 13, no. 1, p. 9605, 2023.
26. X. He et al., "Application of TBSS-Based Machine Learning Models in the Diagnosis of Pediatric Autism," Front. Neurol., vol. 13, p. 1078147, 2023.
27. E. Koc et al., "ASD Detection by Hybrid Convolutional Recurrent Neural Networks from Structural and Resting State Functional MRI," Autism Res. Treat, 2023, Article ID 4136087.
28. H. S. Nogay and H. Adeli, "Diagnosis of Autism Spectrum Disorder Based on Structural Brain MRI Images Using Grid Search Optimization and Convolutional Neural Networks," Biomed. Signal Process. Control, vol. 79, p. 104234, 2023.
29. N. Qiang et al., "A Deep Learning Method for Autism Spectrum Disorder Identification Based on Interactions of Hierarchical Brain Networks," Behav. Brain Res., vol. 452, p. 114603, 2023.
30. N. Yakolli et al., "Enhancing the Diagnosis of Autism Spectrum Disorder Using Phenotypic, Structural, and Functional MRI Data," Innov. Syst. Softw. Eng., 2023, pp. 1–12.
31. K. Devika et al., "Outlier-Based Autism Detection Using Longitudinal Structural MRI," IEEE Access, vol. 10, pp. 27794–27808, 2022.
32. Y. Duan et al., "Identifying and Predicting Autism Spectrum Disorder Based on Multi-Site Structural MRI with Machine Learning," Front. Hum. Neurosci., vol. 15, p. 765517, 2022.
33. C. J. Kumar, P. R. Das, and A. Hazarika, "Autism Spectrum Disorder Diagnosis and Machine Learning: A Review," Int. J. Med. Eng. Inform., vol. 14, no. 6, pp. 512–527, 2022.
34. G. Liu et al., "Two Neuroanatomical Subtypes of Males with Autism Spectrum Disorder Revealed Using Semi-Supervised Machine Learning," Mol. Autism, vol. 13, no. 1, p. 9, 2022.
35. C. J. Mellema et al., "Reproducible Neuroimaging Features for Diagnosis of Autism Spectrum Disorder with Machine Learning," Sci. Rep., vol. 12, no. 1, p. 3057, 2022.
36. P. Moridian et al., "Automatic Autism Spectrum Disorder Detection Using Artificial Intelligence Methods with MRI Neuroimaging: A Review," Front. Mol. Neurosci., vol. 15, 2022. doi: 10.3389/fnmol 2022.999605.
37. C. P. Santana, E. A. de Carvalho, I. D. Rodrigues et al., "rs-fMRI and Machine Learning for ASD Diagnosis: A Systematic Review and Meta-Analysis," Sci. Rep., vol. 12, p. 6030, 2022. doi: 10.1038/s41598-022-09821-6.

38. H. Sharif and R. A. Khan, "A Novel Machine Learning Based Framework for Detection of Autism Spectrum Disorder (ASD)," Appl. Artif. Intell., vol. 36, no. 1, p. 2004655, 2022.
39. F. Zhang et al., "Identification of Autism Spectrum Disorder Based on a Novel Feature Selection Method and Variational Autoencoder," arXiv preprint, arXiv:2204.03654, 2022.
40. Y. Zhu et al., "Application of a Machine Learning Algorithm for Structural Brain Images in Chronic Schizophrenia to Earlier Clinical Stages of Psychosis and Autism Spectrum Disorder: A Multiprotocol Imaging Dataset Study," Schizophr. Bull., vol. 48, no. 3, pp. 563–574, 2022.
41. T. Eslami et al., "Machine Learning Methods for Diagnosing Autism Spectrum Disorder and Attention-Deficit/Hyperactivity Disorder Using Functional and Structural MRI: A Survey," Front. Neuroinform., vol. 14, p. 575999, 2021.
42. N. G. Samian, K. Maghooli, and F. Farokhi, "Brain Volume Analysis with T1-MRI Data in Autism Spectrum Disorder," Front. Biomed. Technol., 2021.
43. M. J. M. Walsh et al., "Brain-Based Sex Differences in Autism Spectrum Disorder Across the Lifespan: A Systematic Review of Structural MRI, fMRI, and DTI Findings," NeuroImage: Clin., vol. 31, p. 102719, 2021.
44. C. Alvarez-Jimenez et al., "Autism Spectrum Disorder Characterization in Children by Capturing Local-Regional Brain Changes in MRI," Med. Phys., vol. 47, no. 1, pp. 119–131, 2020.
45. B. Morgan et al., "Sleep in Adults with Autism Spectrum Disorder: A Systematic Review and Meta-Analysis of Subjective and Objective Studies," Sleep Med., vol. 65, pp. 113–120, 2020.
46. V. Nimmo-Smith et al., "Anxiety Disorders in Adults with Autism Spectrum Disorder: A Population-Based Study," J. Autism Dev. Disord., vol. 50, pp. 308–318, 2020.
47. P. T. Shattuck et al., "Services for Adults with Autism Spectrum Disorder: A Systems Perspective," Curr. Psychiatry Rep., vol. 22, p. 1, 2020.
48. C. Wijker et al., "Effects of Dog-Assisted Therapy for Adults with Autism Spectrum Disorder: An Exploratory Randomized Controlled Trial," J. Autism Dev. Disord., vol. 50, no. 6, pp. 2153–2163, 2020