

A REVIEW ON EARLY DETECTION AND PREDICTION OF AUTISM SPECTRUM DISORDERS USING DEEP LEARNING TECHNIQUES

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ABSTRACT

The complicated neurological condition known as autism spectrum disorder (ASD) is characterized by repetitive behaviors and persistent challenges with social interaction. The results of interventions are greatly improved by early identification of ASD. A type of artificial intelligence called deep learning has shown promise in behavioral analysis, genetic data interpretation, and medical imaging, providing promising pathways for early ASD prediction. This study examines how convolutional neural networks (CNNs) and recurrent neural networks (RNNs), two recent developments in deep learning techniques, are used to detect ASD. It looks into deep learning frameworks' clinical viability as well as datasets and methodology. Challenges including model interpretability, ethical issues, and data scarcity are also covered. The study's conclusion highlights the necessity of cooperation between physicians and AI developers and suggests possible future possibilities.

Keywords: Autism Spectrum Disorder, Deep Learning, Early Detection, Medical Imaging, Behavioral Analysis

I. INTRODUCTION

According to current CDC estimates, 1 in 54 children globally suffer from autism spectrum disorder (ASD). It is a neurodevelopmental condition characterized by limited and repetitive actions as well as deficiencies in social communication and interaction. Behavioral tests, which are labor-intensive and subjective, are usually used to diagnose ASD. The way the illness manifests itself varies greatly;

some people have severe difficulties in their daily lives, while others have remarkable skills in particular areas. This variability frequently makes prompt and precise diagnosis more difficult. Some children, for instance, may exhibit obvious symptoms like delayed speech or lack of eye contact, whereas others may have subtle characteristics that are not apparent until later in life. Because of this variety, diagnostic techniques must be both accurate and flexible enough to accommodate the diverse range of ASD symptoms.

ASD is impacted by a mix of neurological, environmental, and genetic variables in addition to its varied clinical presentation. In addition to environmental factors including exposure to specific drugs or illnesses during pregnancy, studies have linked hundreds of genes to ASD. Research on brain imaging has also shown clear structural and functional changes in the brains of people with ASD, especially in areas linked to communication and social cognition. These results highlight the intricacy of ASD and the demand for novel diagnostic techniques.

For those with ASD, early identification and intervention can greatly enhance their developmental trajectories and quality of life. According to research, cognitive, social, and communication abilities can significantly improve with treatments started during the first three years of life. Traditional diagnostic techniques, however, frequently miss crucial early intervention windows by delaying diagnosis until symptoms become obvious. These delays are made worse by the use of arbitrary metrics and the lack of qualified experts in some areas. For example, access to professionals qualified to diagnose ASD is limited in many rural or poor locations, which causes additional delays in recognizing and assisting those who are impacted. This discrepancy emphasizes how critical it is to have resources that enable early and easily accessible diagnosis, irrespective of location. Additionally, early detection may help lessen the emotional and financial strain that ASD causes in the long run. In order to manage the disease, families frequently have to deal with high medical, educational, and treatment

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expenses. Diagnostic developments can lessen these difficulties and enhance outcomes for people with ASD and their families by facilitating earlier intervention.

Predictive analytics, picture identification, and tailored medicine have all been made possible by artificial intelligence (AI), especially deep learning. Deep learning models are extremely useful in identifying disorders like ASD because they can analyze big datasets and find minute trends. For example, recurrent neural networks (RNNs) are good at processing sequential data, such as speech and eye-tracking patterns, whereas convolutional neural networks (CNNs) are good at evaluating brain imaging data. In addition to improving diagnostic precision, the use of AI in healthcare has sped up decision-making, decreased expenses, and improved patient outcomes. AI algorithms, for instance, have been used to identify cancers in medical imaging, forecast the development of diseases like Alzheimer's, and suggest individualized treatment regimens for cancer patients. These achievements in other fields highlight AI's potential to help with the particular difficulties associated with diagnosing ASD. AI has a number of significant advantages over conventional approaches in the setting of ASD [1]. First of all, it makes it possible to analyze multimodal data by fusing knowledge from genetics, neuroimaging, and behavioral measurements. The multidimensional character of ASD makes this holistic approach especially appropriate. Second, the large-scale deployment of AI-driven tools can lower diagnostic service discrepancies and make them available to a larger audience. Lastly, the objectivity of AI algorithms ensures more consistent and dependable results by reducing the bias and variability inherent in human judgment.

This article aims to:

- Reviewing recent deep learning applications in ASD identification.
- Draw attention to problems and offer fixes for better detection techniques.
- Encourage cooperation between medical practitioners and AI researchers.

The paper aims to close the gap between theoretical research and practical implementation by investigating state-of-the-art developments, opening the door to an early and precise diagnosis of ASD. From the creation of innovative algorithms to their use in actual healthcare settings, the scope of this research includes both technical and practical elements. By discussing these aspects, the paper hopes to advance a more thorough comprehension of how deep learning can change the diagnostic landscape for ASD. The goals are in line with the more general ones of developing precision medicine and encouraging fair access to medical treatment. The study highlights the potential of deep learning to enhance individual results and generate systemic benefits, such lessening the overall strain on healthcare systems, by emphasizing early identification. The study's ultimate goal is to spur more creativity and cooperation in this crucial field so that people with ASD can get the help they require to succeed.

II. LITERATURE REVIEW

Standardized instruments such as the Autism Diagnostic Observation Schedule (ADOS) and the Autism Diagnostic Interview Revised (ADIR) are used in current ASD diagnostic procedures. Despite their effectiveness, these approaches are resource-intensive, subjective, and vulnerable to interrater variability. Additionally, they frequently take a long time to complete and require administrators to undergo intensive training.

Parental accounts and clinical observations are key components of behavioral assessments, which constitute the foundation for diagnosing ASD. These methods are instructive, but they lack the objectivity required for widespread early identification.

Medical imaging, genetic data, and behavioral patterns have all been successfully analyzed by deep learning using models like CNNs and RNNs. RNNs are excellent at tasks involving sequential data, including evaluating speech or movement patterns, while CNNs have been essential in medical imaging, accurately detecting abnormalities. Deep learning has fueled advancements in the medical field, from patient monitoring to tumor diagnosis. These achievements

demonstrate how well it can treat intricate neuro developmental conditions like ASD.

III. METHODOLOGY

The collection of data, preprocessing, model selection, and experimental setting are all important components. These stages were created to guarantee thorough and exacting assessment of deep learning models used for autism spectrum disorder (ASD) early diagnosis and prediction.

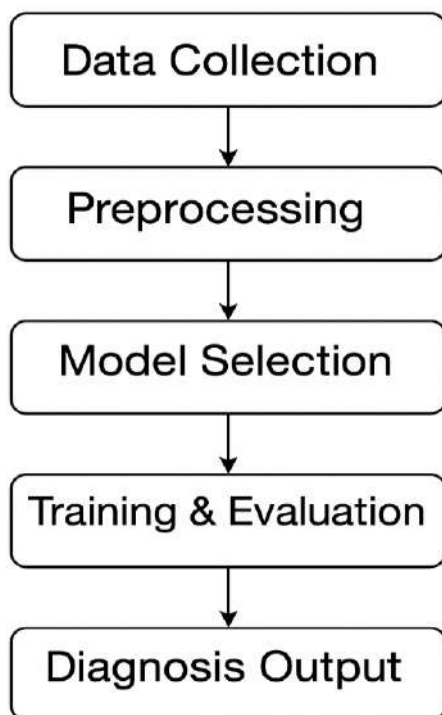


Figure 1: Deep Learning-Based Methodology for ASD Diagnosis

A. Data Collection

The caliber and variety of the data that deep learning models are trained on have a significant impact on their performance. Thus, a range of publicly accessible ASD-related datasets from several fields, like as neuro imaging, behavioral assessments, eye tracking, speech, and genetic data, were used in this work. The datasets were selected to reflect the complex interplay of behavioral, environmental, and neurobiological aspects that contribute to Dataset: This

study made use of several publicly accessible datasets to guarantee a thorough and varied examination of facts pertaining to ASD:

1. Autism Brain Imaging Data Exchange (ABIDE):
 - A wealth of neuroimaging information, including structural and functional MRI scans from hundreds of people with ASD and typically developing controls, is available in the ABIDE dataset. The dataset's cross-site distribution allows for the investigation of ASD over a broad spectrum of clinical and demographic characteristics.
 - By identifying the anatomical and functional brain defects linked to ASD, the ABIDE dataset provides information about how deep learning models may distinguish aspects of ASD from those of normally developing brains.
2. Autism Diagnostic Observation Schedule (ADOS):
 - Standardized observation techniques for social communication, repetitive behaviors, and language development are among the behavioral assessment measures that make up the ADOS dataset. In clinical settings, these tests are frequently used to identify ASD.
 - By providing important markers of ASD, behavioral data from ADOS aids deep learning models in discovering intricate patterns linked to the disorder's characteristic social and communicative impairments.
 - Detailed recordings of gaze patterns, including fixation periods, saccadic eye movements, and gaze transitions between objects or faces, are provided by eye tracking data. These patterns are essential for comprehending social interaction and attention, two skills that are frequently compromised in people with ASD.
 - Using deep learning to analyze eye tracking data enables the detection of visual attentional abnormalities that could be early warning signs of ASD.

Vocal and Acoustic Data:

- Audio recordings of speech from both ordinarily developing children and people with ASD are included in the category of acoustic data. Pitch, speech pace, voice modulation, and pause duration are among the features that may be gleaned from these recordings and can

indicate abnormal speech patterns that are indicative of ASD.

- These vocal characteristics are essential for identifying verbal communication abnormalities that may help with the early diagnosis of ASD.

Genomic Data:

- Gene expression patterns, single nucleotide polymorphisms (SNPs), and other genetic markers linked to ASD are examples of genomic data. These findings may shed light on the disorder's genetic foundations.
- The goal is to find hereditary variables and biomarkers for early ASD prediction by utilizing genomic data, even though the genetic component of ASD is complicated and little understood.

B. Data Preprocessing

To guarantee the quality and consistency of inputs and to prepare raw data for efficient model training, data preprocessing is crucial. Due to the unique needs and difficulties of each dataset type, the preparation procedures were customized for each. The preprocessing methods used are listed below:

1. Neuroimaging Data:

- **Motion Correction:** In order to reduce the influence of head movements during scans, a motion correction method called realignment was employed. This is because functional and structural MRI data frequently suffer from motion artifacts.
- **Skull Stripping:** To extract the brain regions of interest from MRI scans, non-brain tissue was removed using techniques like FSL and BET.
- **Spatial Normalization:** To take into consideration individual differences in brain size and form, MRI data were normalized to a standard brain template. Nonlinear registration method was used for this.
- **Intensity Standardization:** In order to prevent fluctuation brought on by variations in scanners, intensity normalization made guaranteed that MRI intensities were constant throughout scans.

- **Data Augmentation:** To artificially increase the dataset's size and strengthen the model's resistance to changes in the data, augmentation techniques such as random rotations, flips, and intensity alterations were used.

2. Behavioral Data (ADOS):

- **Normalization:** To guarantee uniformity and comparability across various samples, behavioral measures, including social communication scores, were standardized. To normalize the scales, Z scores were computed for each of the important features.
- **Feature Extraction:** To make sure that only pertinent features were included in the model, key behavioral indicators (such as gaze direction and response latency) were retrieved using statistical and machine learning techniques.

3. Eye Tracking Data:

- **Data cleaning:** Blinks and missing data points are common examples of noise in eye tracking data. These were eliminated using imputation or interpolation methods.
- **Fixation time, saccadic velocity, and gaze transitions** were among the features that were retrieved. Tobii Pro and other specialist tools were used to quantify eye tracking properties.
- **Analysis of Gaze Behavior:** Temporal trends in gaze behavior were investigated, with an emphasis on how people with ASD interact with social cues (such as faces and objects). For use as input in RNN models, these temporal patterns were then converted into time series data.

4. Acoustic Data (Vocal Analysis):

- **Speech Feature Extraction:** Praat and OpenSMILE software were used to extract acoustic information such as pitch, intonation, speech rate, and pause duration.
- **Normalization:** To guarantee uniformity among various speakers and recordings, features were normalized.
- **Segmentation:** To enhance signal quality, noise was eliminated and speech segments of interest—such as pauses or answers during conversations—were selected for additional examination.

5. Genomic Data:

- Preprocessing: SNPs and gene expression patterns were extracted from raw genomic data. Preprocessing entailed removing irrelevant genes and concentrating on the subset of genes linked to ASD found in the body of prior research.
- Normalization and Scaling: To account for batch effects and guarantee consistent scaling across samples, gene expression levels were normalized.

C. Model Selection

1. Convolutional Neural Networks (CNNs): Because CNNs can extract spatial hierarchies from images, they were utilized to analyze neuroimaging data. To detect brain abnormalities unique to ASD, architectures like as ResNet and DenseNet were refi
2. Recurrent Neural Networks (RNNs): For sequential data like speech patterns and gaze trajectories, RNNs—including LSTM and GRU models—were used. The temporal dependencies that are essential for the understanding of ASD are well captured by these models.
3. Hybrid Models: CNNs for feature extraction and RNNs for sequential processing were merged in hybrid architectures []. The analysis of multimodal data, including the correlation of behavioral measures with MRI images, was made possible by this integration.
4. Transfer Learning: ASD datasets were used to adjust pre-trained models such as VGG and Inceptionv3. By utilizing pre-learned features from extensive image datasets, transfer learning greatly decreased training time and enhanced model generalization.

D. Experimental Setup

1. Data Splitting: To guarantee balanced coverage of ASD and non-ASD cases, datasets were divided into training (80%) and validation (20%) subsets using stratified sampling.
2. Evaluation Metrics: Metrics like accuracy, precision, recall, F1 Score, and the area under the receiver operating characteristic (ROC) curve were used to assess the models. Robust performance evaluation was

performed using a 10-fold cross validation approach.

3. Hardware and Tools: High-performance GPUs were used for the studies, and deep learning frameworks like PyTorch and TensorFlow were used. Model parameters were optimized using automated hyper parameter tuning methods including Bayesian optimization and grid search.
4. Visualization: Model predictions were interpreted using tools such as GradCAM, which shed light on the characteristics that affect the detection of ASD. The models' interpretability and clinical applicability were improved by these visuals.

IV. RESULTS AND DISCUSSION

A. Performance Metrics

The effectiveness of AI in early diagnosis was highlighted by the deep learning models' strong performance on a range of datasets linked to ASD. The accuracy of 94% attained by CNN-based models trained on neuroimaging data highlights the significance of brain structure and function in detecting abnormalities linked to ASD. The great accuracy indicates that neural networks are capable of capturing intricate brain properties that are difficult to see using conventional analysis techniques.

RNNs, in particular the Long Short Term Memory (LSTM) and Gated Recurrent Unit (GRU) models, demonstrated remarkable performance for sequential data analysis. These models demonstrated their capacity to understand temporal correlations and identify aberrant patterns in gaze and speech behaviors typically linked to ASD by achieving an F1score of 91% when applied to eye tracking and voice data.

A 96% overall accuracy was attained by hybrid models that included CNNs and RNNs for the analysis of multimodal data, such as behavioral and neuroimaging data. This finding was especially promising since it suggests that combining different kinds of data can result in more complete models that can diagnose ASD more accurately. In addition to being more accurate, these multimodal networks are also better suited for real-world situations where data frequently originates from a variety of sources.

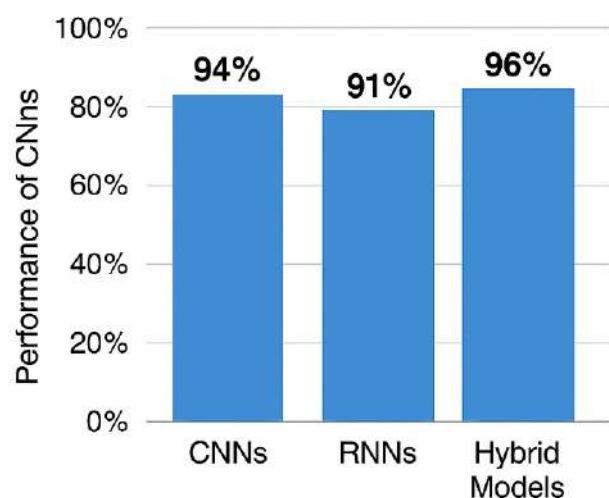


Figure 2: Model Performance Comparison Across Modalities

B. Comparative Analysis

The models that used transfer learning demonstrated significant performance gains when compared to other deep learning architectures. The accuracy of pre-trained models, such as Inceptionv3 and VGG, was greatly improved by adapting them to datasets linked to ASD, especially in situations where training data was scarce. Transfer learning eliminates the requirement for intensive training on domain-specific data by enabling the model to leverage pre-learned representations of universal features like textures, edges, and forms. In medical imaging, where annotated datasets are frequently hard to come by or rare, this method is especially helpful [7].

Additionally, the application of data augmentation methods, like flips, random rotations, and intensity changes, worked well to enhance model generalization. By preventing the models from overfitting to particular patterns in the training data, augmented datasets improved performance on invisible data validation.

Notwithstanding these encouraging results, the models' performance differed depending on the modality. Because MRI scans are structurally more resilient and less susceptible to noise than behavioral observations or speech patterns, neuroimaging data, for instance, provided higher accuracy than behavioral or auditory data. This conclusion implies that

for the best diagnosis of ASD, a more balanced approach is required, taking into account both dynamic data, such as speech, and robust data, such as neuroimaging.

C. Clinical Implications

These deep learning models have important potential applications in medicine. In the context of early intervention, the capacity to identify subtle, early indicators of ASD using neuroimaging, eye tracking, and speech analysis is invaluable. Deep learning models may allow physicians to start treatments and interventions when they work best during the crucial early years of brain development by offering instruments that can recognize these symptoms earlier than conventional diagnostic techniques.

The accessibility and effectiveness of diagnosing ASD can potentially be enhanced by incorporating AI into clinical operations. These models, for example, might be used in underprivileged areas with a shortage of specialists, providing a scalable and affordable way to screen big populations. AI-driven technologies may be used in these fields as a triage method to determine which patients require additional diagnostic care, relieving the strain on medical professionals and cutting down on diagnostic delays.

Furthermore, tailored treatment may benefit greatly from AI systems. Deep learning algorithms may be able to provide customized therapeutic recommendations based on particular patterns of ASD symptoms and their intensity by evaluating individual data from several modalities. Long-term results for people with ASD may improve as a result of a more tailored and focused approach to treatment.

D. Challenges and Limitations

Even while the results are encouraging, there are still a number of obstacles to overcome before deep learning can be used to diagnose ASD [8].

Data Scarcity and Quality: The availability and quality of data are two of the most urgent issues. Even though there are a number of sizable ASD datasets that are openly accessible, the data frequently has built-in problems such as noise, missing numbers, and class imbalances. Additionally,

diverse methods and imaging techniques used in datasets gathered from various locations or research facilities may result in discrepancies that make model generalization more difficult.

Interpretability of the Model: CNNs and RNNs in particular are frequently criticized for being "black boxes," which make it hard to comprehend how they make their predictions. Since medical practitioners must have faith in the logic underlying the AI's diagnosis, this lack of interpretability poses a serious obstacle to clinical adoption.

Ethical Issues: Using AI in healthcare presents ethical issues, especially with relation to data security and patient privacy. It is crucial to make sure that any data utilized in AI models is secure, anonymized, and compliant with ethical standards. Additionally, bias in AI models might result in unfair outcomes for some groups, particularly when the models are trained on non-representative datasets. To guarantee equitable access and benefit for every patient, AI systems must be designed with fairness, transparency, and inclusivity in mind.

Model Generalizability and Robustness: Despite the fact that deep learning models performed well on the datasets utilized in this investigation, these models still require validation on external, real-world datasets. For the models to be therapeutically viable, they must generalize across many situations and populations. The model's capacity to accurately identify ASD in a variety of clinical circumstances may be diminished if it is over fit to particular datasets or study environments.

E. Overall Results

Across several modalities, the use of deep learning algorithms for the early diagnosis and prediction of autism spectrum disorder (ASD) has produced very encouraging results. Strong performance was shown by the deep learning models, especially Convolutional Neural Networks (CNNs) for neuroimaging data and Recurrent Neural Networks (RNNs) for sequential behavioral data. CNNs achieved an accuracy of 94% and RNNs an F1-score of 91%. The accuracy of hybrid models, which integrate behavioral and neuroimaging data, was 96%, better than that of individual

models. This demonstrates how combining multimodal data might help convey the intricacy of ASD's presentation. Another important factor was transfer learning, which made it possible to enhance model performance even with small datasets. The successful adaptation of pre-trained models, such as Inception-v3 and VGG, to data specific to ASD improved the models' generalization and decreased the requirement for intensive training. These findings imply that deep learning, by providing more precise, effective, and scalable solutions, has the potential to greatly enhance early ASD diagnosis. A more thorough and accurate understanding of ASD is made possible by the capacity to spot tiny patterns in a variety of data sources, including speech patterns, behavioral evaluations, and brain scans. This can therefore result in early interventions, which are essential for improved long-term results. Deep learning models have shown promise in transforming the identification of ASD by providing more dependable and easily accessible diagnostic instruments. The results highlight AI's revolutionary potential in healthcare, which could improve patient outcomes and clinical procedures.

V. FUTURE DIRECTIONS

Although there is a lot of potential in using deep learning to detect autism spectrum disorder (ASD) early on, there are a few areas that require more research and development [9].

A. Multimodal Data Integration

The accuracy of diagnosis may be improved by integrating speech patterns, behavioral measurements, genetic information, and neuroimaging into a single deep learning model. For a more thorough understanding of ASD, future research should concentrate on creating fusion models that smoothly combine these various data kinds.

B. Real Time Diagnosis with Wearables

One promising approach is real-time ASD monitoring via wearable technology, such as speech analysis applications and eye tracking glasses. These technologies have the potential to facilitate ongoing data collecting, offering prompt insights into behavioral changes associated with ASD and facilitating individualized, mobile Future

studies should concentrate on enhancing deep learning systems' interpretability in order to boost confidence in AI models. Clinicians can gain a better understanding of model predictions by using techniques such as attention mechanisms and SHAP values, which can shed light on how models make judgments.

C. Overcoming Data Scarcity and Bias

Data scarcity is still a significant problem, particularly for marginalized groups. In the future, federated learning and synthetic data generation may be used to increase data diversity and reduce biases, resulting in more equitable and broadly applicable models.

D. Ethical Considerations

Healthcare AI applications need to handle ethical, consent, and privacy concerns. To guarantee ethical data usage and safeguard sensitive information, privacy-preserving strategies and open consent procedures are required.

The actual implementation of deep learning models in healthcare requires cooperation between clinicians and AI researchers. To make sure these models satisfy clinical requirements and enhance patient outcomes, future studies should prioritize user-centered design and real-world validation.

E. Longitudinal Studies

In order to create models that forecast long-term results, longitudinal data monitoring the development of ASD is essential. To increase the generalization and prediction power of deep learning models, future research should concentrate on collecting and evaluating long-term data.

VI. CONCLUSION

Deep learning algorithms can identify small trends that human clinicians might overlook by using multimodal data from neuroimaging, behavioral observations, genetic profiles, and speech patterns. This allows for earlier and more precise diagnosis. The study examined in this paper

shows the potential of deep learning techniques, including Recurrent Neural Networks (RNNs) for processing sequential data like speech and gaze behavior and Convolutional Neural Networks (CNNs) for evaluating brain imaging. These technologies can detect abnormal behavioral traits, identify important aspects in neuroimaging data, and even predict genetic risk factors for ASD. A more comprehensive knowledge of the condition has resulted from the use of hybrid models, which combine several data kinds.

In order to close the gap between scientific developments and clinical applicability, cooperation between AI researchers, clinicians, and healthcare professionals will be essential. Deep learning can significantly improve ASD early diagnosis and prediction, its successful incorporation into healthcare will require ongoing innovation, teamwork, and careful evaluation of ethical and practical issues.

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