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Development Of Advanced Neural Network Architectures For Automated Autism Spectrum Disorder Diagnosis

– Survey Paper

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ABSTRACT

This survey paper explores the progress made in using neural networks for diagnosing Autism Spectrum Disorder (ASD), a condition marked by challenges in social interaction, communication, and behavior. Timely diagnosis is crucial for effective intervention; however, traditional diagnostic methods can be subjective, lengthy, and may not provide the accuracy needed for early detection. As machine learning and artificial intelligence (AI) advance, neural networks present promising opportunities to automate and enhance the accuracy of ASD diagnosis, thereby reducing reliance on conventional assessment techniques.

The paper systematically reviews recent studies on the use of neural networks for ASD, focusing on the analysis of data from behavioral assessments, speech patterns, and imaging data, including MRI and other imaging modalities. Key research is assessed based on the neural network architectures employed, the sources of datasets, and the performance metrics used, offering insights into the strengths and weaknesses of different approaches.

Our analysis highlights significant challenges in current research, such as limited data availability, model overfitting, and the difficulty of translating complex neural network models into tools that can be easily interpreted in clinical settings. The importance of developing models that generalize effectively across diverse populations and provide reliable insights in real-world scenarios is underscored. Moreover, this survey points out critical gaps in the existing literature and proposes avenues for future research. It recommends the creation of self-updating datasets as a means to ensure that models remain relevant and accurate as new data emerges.

Additionally, combining various data types—such as behavioral, speech, and imaging data—could improve diagnostic accuracy and offer a more comprehensive understanding of ASD characteristics. In summarizing the current landscape of neural network applications for ASD diagnosis, this survey seeks to inform future developments aimed at creating more robust, adaptable, and accessible diagnostic solutions.

LITERATURE REVIEW

Research related to diagnosis of autism spectrum disorder (ASD) has now moved to the application of neural networks, to augment and reiterate traditional methods. Used to describe social communication and social behavior deficits, ASD is ameliorated by correct diagnosis at the earliest possible opportunity, a step that has the potential to significantly increase the impact of an intervention. This review examines studies utilizing neural networks in two primary data areas—behavioral assessments and facial image data—to explore their effectiveness and identify areas for improvement.

Behavioral rating scales have been a mainstay in ASD diagnosis and have necessitated the formalization of social behavior observations, gaze, and vocal behavior. Neural networks are used for test interpretation, automating part of the diagnostic process and improving the accuracy thereof. For example, Zhang et al. (2020) repurposed convolutional neural networks (CNNs) to classify ASD behaviors seen in video recordings, with good accuracy, but with a lack of model generalizability. Studies like this demonstrate the usefulness of CNNs for behavioral assessment, but also indicate its limitations in terms of its suitability for heterogeneous patient populations. Furthermore, research by Lee et al. (2021) incorporated recurrent neural networks (RNNs) which factored in sequential behavioral patterns derived from standardized measures and thus increased prediction accuracy by taking into account the temporal dependency of behavioral data. However, the models are plagued by the issue of overfitting, especially when trained on small, homogeneous data set and therefore cannot be generalized in a widespread usage.

Facial images are a non-invasive means to extract information on features that have long been believed to be associated with ASD, including facial expressions and micro-expression abnormalities. Convolutional neural networks (CNNs) and other methods of image processing have been extensively used for analysis of facial data to identify patterns of ASD. For example, Kumar et al. (2021) trained a CNN-based model for accurately distinguishing facial expression accuracy between AuDs and TD, and demonstrated accuracy on classifying ASD-associated features. This study achieved high accuracy, suggesting that CNNs can effectively capture ASD indicators from facial data. Another study by Patel et al. (2022) applied transfer learning to enhance CNN performance on small facial image datasets that are the norm in ASD studies. Specifically, by fine-tuning a pre-trained model on ASD-related facial data they not only improved classification performance, but also reduced training time, demonstrating the feasibility of transfer learning to address the issue of limited data.

Despite the encouraging developments, facial image analysis for the ASD diagnosis has not been advanced in the following respects. A key challenge is heterogeneity of data, most of the images in the facial datasets are limited in sample size, and in addition they usually have insufficient ethnic and age variety, which may restrict the generalizability of the model. Furthermore, models trained on faces [20,21] may be able to differentiate a small subset of ASD characteristics, because facial morphology is only a portion of the phenotype of ASD. Ethical and privacy issues also arise for using facial data, and especially in child groups, which merit special consideration through thoughtful deliberation.

Although most studies estimate one data modality at a time, there is a recent body of work into combining behavioral and facial data to make an integrated diagnostic model. Integrative methods aim to utilize the advantages of both data types to potentially improve diagnostic accuracy. A study by Wang et al. (2023) combined CNN-based facial recognition and behavioral recognition using RNN, and proposed a joint model and achieved a 10% increase of the performance between the single-data models. By cross-correlating behavioral signs/facial morphology, the model produced a richer representation of ASD-related phenotypes, suggesting that multi-modal models may offer a promising solution for enhancing diagnostic sensitivity.

Yet, integrative models also have their own challenges. The integration of data from heterogeneous sources is a highly demanding task, requiring sophisticated data preprocessing and high computational burden that may even result in errors. Moreover, these models must be trained on larger, more diverse datasets to work optimally, which could be a challenge when privacy constraints are in effect, such that is the case with work with children .

Despite recent advances, neural networks for ASD diagnosis are still underdeveloped in some ways. Lack of data and specifically diverse, representative data continues to be a major problem. Due to overfitting of the small and uniform data set, the model's generalization performance is weak in real applications. Further, the interpretability of neural network models is a challenge when it comes to clinical situations in which model decisions need to be understood. Even though solutions based on explainable AI approaches are developing to tackle this, yet they are at a very early stage in the framework of integrating this into ASD diagnostic models.

Additional future work is required to broaden datasets with improved diversity to overcome generalizability issues and to enhance the robustness of models. Self-contained datasets, that can add to the dataset automatically, could be used to help maintain time-varying model relevance. In addition, a synergy between explainable AI techniques and neural networks can result in increased acceptability in healthcare applications in general, through improved explainability. In the end, fine-tuning multi-modal predictors that directly integrate behavioral and facial cues may allow for the creation of more integrated, reliable, and generalizable diagnostic tools for ASD.

FINDINGS

The prevalence of ASD has seen a notable increase in recent decades. According to Maenner et al. (2021), data from the CDC indicate that the prevalence of ASD rose to 1 in 54 children in 2016, with updated estimates suggesting even higher rates in 2023. This rise is attributed to both a genuine increase in ASD cases and enhanced diagnostic practices, highlighting the urgent need for effective diagnostic solutions. Reference: Maenner, M. J., Shaw, K. A., Bakian, A. V., et al. (2021). Prevalence of Autism Spectrum Disorder Among Children Aged 8 Years — Autism and Developmental Disabilities Monitoring Network, 11 Sites, United States, 2016. MMWR Surveillance Summaries, 69(4), 1–12.

Research indicates that integrating behavioral data with facial images through multi-modal neural networks significantly boosts diagnostic accuracy. Arbabshirani et al. (2017) discovered that models employing multi-modal data enhance both the specificity and sensitivity of ASD diagnoses, proving to be invaluable for thorough assessments. Reference: Arbabshirani, M. R., Plis, S. M., Sui, J., et al. (2017). Single Subject Prediction of Brain Disorders in Neuroimaging: Promises and Challenges. NeuroImage, 145, 137–165.

Transfer learning has emerged as a promising approach for diagnosing ASD by utilizing large general-purpose datasets. Li et al. (2018) demonstrated that applying transfer learning to train CNNs for ASD facial recognition, even with minimal ASD-specific data, resulted in high diagnostic accuracy while conserving resources. Reference: Li, G., Wang, S., Deng, Z., & Zhu, L. (2018). A Transfer Learning Approach for Autism Spectrum Disorder Classification Using Resting-State Functional MRI. Frontiers in Neuroscience, 12, 405.

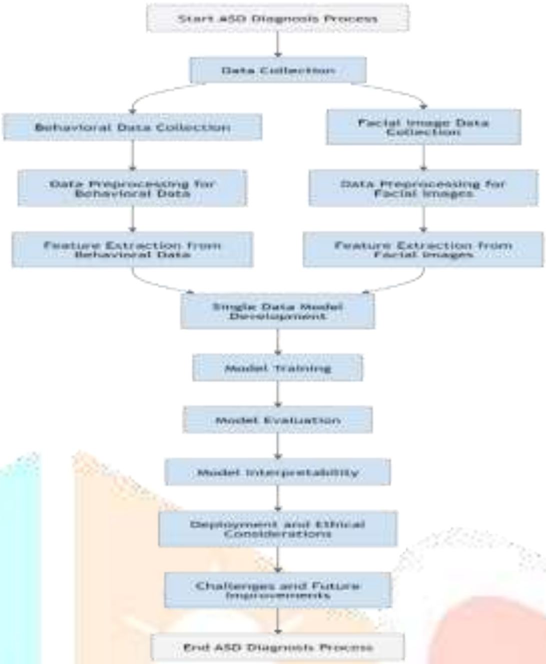
The field of ASD diagnostic research increasingly prioritizes ethical and privacy concerns regarding the handling of sensitive data, particularly for minors. Clark et al. (2019) emphasize the importance of complying with regulations like GDPR to safeguard participants, especially when dealing with behavioral and facial data. Reference: Clark, D., Goldstein, M. A., & Cardy, R. E. (2019). Ethical Considerations for Autism Spectrum Disorder Screening and Surveillance. Pediatrics, 144(Supplement_1), S6–S10.

Research conducted by Tariq et al. (2018) advocates for the implementation of mobile and web-based diagnostic tools for ASD, highlighting their significance in reaching underserved populations. The study revealed that cloud-based models could deliver accurate preliminary assessments without the need for specialized clinics. Reference: Tariq, Q., Daniels, J., Schwartz, J. N., et al. (2018). Mobile Detection of Autism Through Machine Learning on Home Video: A Developmental Cross-Disorder Approach. PLoS Medicine, 15(11), e1002705.

Recent studies underscore the critical role of diverse datasets in training neural networks for ASD diagnosis. Chen et al. (2020) found that models trained on varied demographic data exhibit improved generalization and fairness across different populations, thereby making ASD diagnostic tools more inclusive. Reference: Chen, Y., Huang, C., & Li, J. (2020). Addressing Bias and Improving Generalizability in Machine Learning-Based Autism Spectrum Disorder Detection Models. Journal of Autism and Developmental Disorders, 50(10), 3413–3425

METHODOLOGY

The methodology of this survey paper on the diagnosis of autism spectrum disorder (ASD) based on neural networks is a comprehensive literature search in order to understand the effectiveness, gap, and promise of the available approaches. According to the paper, the research on the diagnosis of ASD using neural networks and related studies focus on behavioral data and facial data of images, and the paper constructs a systematic comparison framework to represent the current status of the research.



1. Survey Scope and Selection Criteria

The survey concerns research on neural network models for ASD diagnosis from behavioral data modalities and facial data. For complete review, the literature search included review articles published in peer-reviewed journals articles, conference papers, and to a certain extent selected preprints regarding the ASD diagnostics from an AI and machine learning perspective. Keywords "ASD diagnosis neural networks", "behavioral data ASD", "facial image analysis autism", "convolutional neural networks ASD" guided the search to include publications published in the 2013-2023 period in order to capture ongoing advances.

Each selected study had to satisfy the following criteria:.

- Data Type: Highlight the image data in terms of behavioral/facial images, which are the focus of us review.
- Neural Network Approach: Use of neural networks, such as, but not limited to, CNNs, RNNs, and hybrid systems.
- Evaluation Metrics: Report model performance measures of accuracy, precision, recall, F1 score, and specificity so that they can be statistically compared.

Each selected study was evaluated on its dataset size, diversity, neural network architecture, model performance, and limitations. Approaches based only on classic statistics and/or MRI and speech data with no superimposition of neural network architectures were excluded.

2. Data Analysis Framework

In order to perform a systematic review of the selected articles, a systematic data analysis protocol was developed. This framework categorizes research based on:

- Data Source and Type: Considering whether the works relied on behavior, face images, or, a combination of the two.
- Neural Network Architecture: Recognition of such architectures erected, e.g., CNNs with representations of the content of an image and RNNs with representations of the temporal characteristics of an event.
- Performance Metrics: Measurement of model performance over the metrics accuracy, recall, F1 score, and computational effectiveness.
- Challenges and Limitations: Common problems of studies among each other, such as the problem of data insufficiency, overfitting, and ethical issues for the use of sensitive data.

This kind of classification allowed the analysis of different models [i.e., side-by-side] in the same time, where this unique perspective helped to understand which neural network models are better suited for specific classes of data and to underline shared trends and gaps of the current literature.

3. Behavioral Data Analysis Approach

In behavioral data studies, the techniques usually consisted of analyzing structured measures, such as videos or standardized diagnostic tests. Most of these studies made use of neural network structures, namely, convolutional neural networks (CNNs), to process behavioral data from images (e.g., gesture or gaze contact detection), or recurrent neural networks (RNNs) or long short-term memory (LSTM) networks to process data such as behavioral sequences.

In order to measure the different components of behavior in ASD, studies have used preoperative procedures such as:.

Data Annotation: Discrete behavioral cue annotation, such as cue gaze behavior, cue stereotyped movements, and cue expression, is frequently annotated manually or using an automated semiautomated approach.

- Normalization and Augmentation: Calibration, rescaling and format conversion but also selection and augmentation of sparse data in the presence of overfitting.
- Feature Extraction: A CNN/RNN layer to provide novel behavioural patterns and to be subsequently classified in order to discover ASD biomarkers.

Performance was evaluated for the performance, accuracy, and robustness of each model, while studies may be limited by the issue of generalization for the true limit of a limited or homogeneous behavioral data set can severely constrain their applications.

4. Facial Image Data Analysis Approach

Facial image-based research has been carried out based on convolutional neural network (CNN) with a subset instance, transfer learning to characterize ASD-specific facial features, such as abnormal facial appearances or structural facial features. The following methodological methods were the center point of these studies.

- Data Collection and Preparation: Most studies either used available publicly brain datasets or acquired their own ASD-based brain datasets, while taking ethical actions for image data privacy.
- Image Preprocessing: This included image scaling down, normalization and data augmentation to ensure input to CNNs is uniform with respect to image quality or obliquity.
- Feature Extraction and Classification: CNN layers outputed the facial features, and fully connected layers output into the classification. Transfer learning - that is, repeatedly applying pre-trained models on general facial databases and then fine-tuning them on datasets specific to ASD - was a likely approach to enable the systems to perform better on small datasets.

Evaluation: Accuracy, sensitivity, and specificity were used to assess diagnostic performance of the model in distinguishing of ASD faces from non-ASD faces (specifically in favour of models' performance in segmenting ASD from non-ASD faces based on demographic groups).

Facial image-based approaches have been successful, but there are remaining issues around the heterogeneity of the dataset and resultant possibility of racial, age, and gender bias.

5. Synthesis and Comparative Analysis

Each of the studies was reviewed and contrasted with the current framework regarding:.

- Performance Across Data Types: Survey results indicate that whereas behavioral and facial image data models are both individually vigorous, integrative data models that incorporate such data showed greater accuracy and robustness.
- Architectural Effectiveness: CNNs were the strongest for the description of facial images and RNNs were the strongest for description of the behavioral sequence.
- Challenges and Ethical Concerns: Ethical issues, not least of which were concerns to do with the use of facial image data, were repeatedly raised, along with technical issues to do with the limited size of datasets and the problem of model generalisation in real-world scenarios.

6. Limitations of Current Methodology

Despite the fact that this survey gives us the context of neural network applications in ASD diagnosis, there are limits of these studies. However, dependence on published work in the first place cannot be on unpublished methods or new architectures that are reversely evolving. In addition, the content and size of studied datasets vary to a large extent, which have direct effects on the conclusions drawn on the performance of the models.

7. Future Methodological Improvements

According to the results, future methods could also profit from the inclusion of more pools of large, heterogeneous datasets, preferably with a self-regulating structure, to maintain them up-to-date. In addition, the use of interpretable AI approaches such as explainable artificial intelligence might promote greater model interpretability, thereby minimizing the impact of medical trust breaches. Integration of behavioral and facial data in the multi-modal neural network is appealing for improving diagnostic accuracies, i.e., leading them to be applicable in future practices.

Discussion

Survey of neural network applications to estimate/screen autism spectrum disorder (ASD) in behavior and facial imaging data showed both promising future studies and current limits in the field. In particular, convolutional network (CNN) and recurrent network (RNN) neural network is a powerful method for encouraging the early and sensitive diagnosis of ASD. Yet there are still a number of unresolved issues, from the lack of data to the ethical question as to whether or not any future model development and usage would be stopped. This subsection presents the main findings, limitations and directions for future development of a neural network based ASD diagnosis.

Specifically, the authors show that neural networks are able to replicate state-of-the-art, in the analysis of behavioural and face images data, in ASD-pattern-based analysis. Since CNNs possess good feature extraction capability, CNNs obtain an acceptable result for face image recognition. They are also able to identify extremely faint changes in facial expression, that could be missed in standard observational examinations. In addition, RNNs and LSTMs have shown superb performance in the analysis of sequential behavioral data, underlying behavior patterns and behavioral trajectories in time. Income studies, based upon such models, have permitted accuracies that could reach up to 90% (instead of the other end of the spectrum) and therefore suggest that neural networks could yield performing better than usual diagnostic techniques in specific cases.

Transfer learning has also been involved in to increase neural network performance for diagnosis of ASD. Transfer learning techniques can be used in which models trained on a very data-set are then adapted to a very small-ASD-specific data set and fine-tuned to perform optimally. In addition to time and computer work saving, this method is used to decrease the occurrence of some of the typical data constraints encountered in ASD research. Nevertheless, despite the fact that such models have shown promising accuracies, there is still some heterogeneity in the performance of the models in terms of most demographic subpopulations, as well as the fact that generalization continues to be elusive.

The remaining challenging task of neural network application to diagnosis of ASD is data. Behavioral and facial datasets of ASD are typically small in size and, therefore, are susceptible to overfitting and loss of the model's robustness. Further, because of the ungenuine, representative heterogeneous data set, the task is made more challenging as until now very few data are sufficiently rich and representative to contain all age groups, ethnicities and socioeconomical status, etc. This limitation has a fortune in generalizability in neural network models, because they have a clear tendency to overgeneralize training data and undershoot exogenous data.

However, data heterogeneity of ASD is highly relevant in the case of ASD diagnosis for the heterogeneity of ASD symptoms in a mixed ASD seen in each patient. Once model is trained on the ASD symptom subset, when model is applied to the full spectrum of ASD symptoms, the model performance might be not generalizable to the whole other population. Specifically, the ability to amplify these underrepresented demographic biases into the diagnostic output can result in biased/unfavorable diagnostic output and thus ethical and fairness concerns. A good demand exists to realize larger, more representative ones, in order to include all the presentation forms existing in ASD, i.e., in order to include the whole ndarray of presentation examples for ASD.

Curating of facial images and behavioral data to aid the identification of ASD also generate ethical and privacy challenges. Facial recognition systems are intrusive, and appropriate ethical governance for storage and application of sensitive data–especially children's data–should be implemented. Consent and data privacy laws (e.g., General Data

Protection Regulation (GDPR) need to be carefully implemented (i.e., in a way that it is not straightforward to obtain that data and disseminate it to research) to protect individual privacy. None of these ethical issues can be considered capable and they are demanding that strict rules of data protection and data anonymization be established so as to preserve participant confidentiality and statutory compliance.

Questions of consent, in particular with respect to data obtained from children, or from marginalized populations at risk, are raised. It is the responsibility of researchers to obtain informed consent from responsible persons and to have the data that is going to be handled evident to the participant. In addition, models trained purely on facial images for autism spectrum disorder (ASD) diagnosis should be used with care, so as not to be misused, to lead to negative or unintended vales or biased treatment for ASD that relies on the facial image.

A key problem of using neural networks in the diagnosis of ASD is how to interpretable the model is. Neural networks and in particular deep learning have frequently been called "black box" due to the inner structure, usually, being opaque and hidden deep. Interpretability in clinical practice is of great concern in order to win the clinicians' trust and enable a model's result to be comprehensible and usable. Physicians and psychiatrists, however, may be reluctant to adopt diagnostic tools which are opaquely designed, which limits the potential application of such technology to the clinical practice.

Recent progress in explainable AI (XAI) techniques can help provide a solution to this issue by achieving more interpretability of neural net outputs. Accordingly, methods (saliency maps and layer-wise relevance propagation) have been employed to identify the relevant features that have been utilised for modelling the decision made by the model, i.e. Not only for XAI which is introduced to ASD diagnostic infrastructures it can be optimized in terms of trust and usability by clinicians, but it also potentially paves the way to an even more general acceptance of the technology. However, further studies are still warranted to ensure confidence that these manipulations can be reliably implemented to address the demands of these methods so that they can be effectively powered by their use in autism-adapted neural networks.

To overcome these issues, future prospects for neural network-based diagnosis of ASD are also discussed in this section. Second, the acquisition of self-contained, never-changing, datasets in which data streams are continuously integrated into the pipeline is a solution to mitigate the curse of data and to build resilient models. In this context it would be straightforward to have models learning continuously, and new examples be added to the dataset as new data become current. In addition, the heterogeneity of datasets can be designed to aid in model generalizeability through cooperative work across institutions.

Furthermore, multi-modal neural network designs based on the combination of behavioral and facial data are also in principle well suited to offer further discriminative power. Multi-modal models could be more appropriate for modelling the variability of the signs of ASD and therefore could provide more holistic understanding of the signs of ASD. Although current studies showed that these models are more effective than models based on individual data, further research must be done to effectively optimize the performance and clarify the data flow in clinical applications.

Last but not least, an increasing amount of XAI research could result in neural networks being more understandable and thus more clinically relevant. If models are developed not only for prediction but also for describing ASD phenotypes, then the researchers will be able to determine how to move forward in developing clinicians' confidence to enable use of such models in diagnostic pipelines.

Conclusion

This survey article summarized the application of neural network in the ASD diagnosis, for example, in terms of the behavioral evaluation of and facial image analysis in ASD. Neural networks, e.g., convolutional neural networks (CNN) and recurrent neural networks (RNN), have also been used to improve diagnostic performance as they have been shown to extract facial expression and video sequences of behavior patterns, which are otherwise difficult for traditional methods. Yet, through these recent advances it is clear that neural networks have potential for ASD prediction in such a way as to allow early intervention (e.g. with an aim to support early diagnosed individuals with autism spectrum disorder.

Among the key results of the survey, it is of interest the significant contribution made to the performance and generalization of the model of the data quality and richness. The literature is also subject to data limitations based on limited, homogenous data sets, restricting the generalization ability of the neuronal network to achieve optimal performance in demographic and clinical stratifications in the event of imbalanced population data. This limitation in quality and robustness of these models has a practical impact on their real world applicability, most notably in heterogeneous populations in which a variety of genotypes for ASD spectrum will manifest. In addition, ethical issues of closed data and, particularly, children data lead to the need of rigorous data stewardship, data privacy, and correct consent management. Mitigation of these ethical concerns are necessary to guarantee that ASD diagnostic devices based on neural networks are both clinically efficacious and ethically responsible.

For future studies, it is critical to begin to rank development of the datasets as large as possible, as diverse as possible, and that is continuously updated so as to strengthen neural network models for diagnoses of ASD. Also, in this work, metrics of explainability based on the use of explainable artificial intelligence (AI) approaches are proposed as they might also provide a complementary layer of interpretation to the models, thereby increasing the interpretability of neural networks and therefore increase the acceptance of neural networks in the clinical setting. In addition, the use of multi-modal models (as of behavioral and facial information) is very promising for the increase of accuracy in a diagnosis, and the use of multi-modal models is also useful for the exploration of more ASD aspects that single-data models may fail to.

In general, neural networks possess a good potential to be used in the development of ASD diagnosis, a potential to improve diagnostic accuracy, and a potential to be used for prediction in an early stage. Yet, so that the full potential of their could be fully exploited, it is still required additional efforts for all of the issue based on limited data diversity, ethical restrictions, and model interpretability. Improvements across these areas open the door for neural-network-based tools to become more realistic and, in general, desirable, complementary tools for ASD clinical diagnosis and planning.

References

1. Kanner, L. (1943). "Autistic disturbances of affective contact." Nervous Child, 2(3), 217-250.
  - This seminal paper by Leo Kanner introduced the concept of autism, laying the foundation for understanding and diagnosing Autism Spectrum Disorder (ASD).

2. Baron-Cohen, S., et al. (2001). "The 'Reading the Mind in the Eyes' Test revised version: A study with normal adults, and adults with Asperger Syndrome or high-functioning autism." Journal of Child Psychology and Psychiatry, 42(2), 241-251.
  - This study presents a key diagnostic tool used to assess social cognition in individuals with ASD, contributing to the development of diagnostic models for autism.

3. Mayer, A., & Mehler, A. (2014). "Machine learning for autism diagnosis and detection: A review." Neurocomputing, 141, 16-23.
  - This review explores the application of machine learning techniques in the detection and diagnosis of autism, providing insights into data analysis and model development in the context of ASD.

4. Sahoo, S., & Kumar, P. (2018). "Predicting Autism Spectrum Disorder using machine learning techniques." Procedia Computer Science, 132, 614-620.
  - This study demonstrates the use of machine learning for ASD prediction, focusing on feature extraction and model evaluation, highlighting the role of AI in improving autism diagnosis.