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Machine Learning Techniques for Early Diagnosis of Autism Spectrum Disorder

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Abstract

Autism Spectrum Disorder (ASD) is a neurodevelopmental condition that significantly impacts communication and gestures, pressing the significance of early diagnosis for timely intervention. This study employs machine learning techniques to identify ASD traits in young children and toddlers, using the Toddler Autism dataset obtained from Kaggle in July 2018. The dataset includes 1054 samples, with features derived from diagnostic questionnaire responses. Due to limited locally available data, external sources were incorporated to enhance model training. The research employs Artificial Neural Networks (ANN) and k-Nearest Neighbours (kNN) for classification objectives. Both models underwent data pre-processing, which included label encoding for categorical variables, standardization of numerical features, and the Synthetic Minority Oversampling Technique (SMOTE) to manage class imbalances. The assessment of model performance was performed using various well-known metrics such as accuracy, precision, recall, F1 score, and confusion matrices. The ANN achieved high accuracy with cross-validation results of 99.81%, demonstrating robust learning and minimal overfitting. The kNN model performed comparably, with test accuracy and cross-validation accuracy of 94.86% and 94.93%, respectively. Additionally, k-fold cross-validation confirmed the models' stability across multiple splits, with ANN and kNN exhibiting consistent performance. Results highlight the potential of these models in early ASD detection, emphasizing their utility in clinical and community-based settings. Future work will explore larger datasets and advanced feature engineering to enhance generalizability.

Keywords:

Autism Spectrum Disorder (ASD), Early Detection, Machine Learning, Artificial Neural Networks (ANN), k-Nearest Neighbours (kNN)

Introduction

Autism spectrum disorder is a neurodevelopmental disorder that manifests itself for the first time during the early years of life. It is associated with impaired communication, behavior, and social interaction; undoubtedly, this would entail early diagnosis being a gateway to timely intervention and better prognosis. Timely identification serves a vital purpose in delivering prompt therapy and assistance, greatly enhancing developmental progress and life quality for affected children. Nonetheless, traditional diagnostic techniques are usually laborious, subjective, and dependent on expert evaluation, which might postpone intervention. As digital data becomes increasingly accessible and computational techniques grow more advanced, machine learning (ML) presents a promising method to bridge this gap. Machine learning can process extensive datasets, identify intricate patterns, and provide precise predictions, potentially transforming the early identification of ASD. This research employs machine learning methods to detect ASD characteristics in toddlers, concentrating on creating strong classification models able to differentiate between those with and without ASD traits. The "Toddler Autism Dataset" used in this study is sourced from Kaggle. It includes diagnostic questionnaire responses, which serve as key predictors for ASD. Issues like restricted local data access, uneven class distributions, and varied feature types were tackled by thorough pre-processing and the use of sophisticated algorithms. Two machine learning models, Artificial Neural Networks (ANN) and k-Nearest Neighbours (kNN), were used to categorize toddlers into ASD-positive and ASD-negative groups. The ANN was selected for its ability to represent intricate connections between features, whereas kNN acted as a more straightforward baseline model recognized for its simple implementation and efficiency in specific classification tasks. Both models were subjected to thorough assessment through cross-validation, guaranteeing their trustworthiness and strength. Initial findings indicate that the ANN attained a cross-validation accuracy of 99.81%, highlighting its promise as an effective instrument for detecting ASD. The kNN model also performed well, with test and cross-validation accuracies exceeding 94%. This research adds to the expanding evidence backing the significance of machine learning in tackling public health issues.

Related work:

A research paper published in the journal Healthcare Analytics, Vol 5, June 2024, titled "An Evaluation of Machine Learning Approaches for Early Diagnosis of Autism Spectrum Disorder," by Rownak Ara Rasul et al., provided an assessment of various machine learning techniques in respect to how effective they are in early diagnosis of ASD. Other classifiers that were assessed for comparison purposes include decision trees, SVMs and ensemble methods by comparing their accuracies, sensitivities, and specificities. The paper further opined that even though early diagnosis of ASD and its symptoms is very critical for the improved outcome of an intervention program, this paper would still discuss how Machine Learning models might be used in enhancing the accuracy and efficiency of ASD diagnosis. The study concluded by insisting on strong, scalable, and explainable models, indicating that machine learning may lead to improved early detection rates of ASD so that timely intervention strategies can be adopted. [1]

"A Machine Learning Framework for Early-Stage Detection of Autism Spectrum Disorders" by S. M. Mahedy Hasan, et al., in IEEE Access, Vol. 11, 2023, This paper brings a relatively well-rounded framework for earlier ASD detection using the power of machine learning. It talks about using SVM, random forests, and deep learning models in finding patterns or features to predict ASD in clinical data. Apart from this, it highlights the incorporation of feature selection techniques that make the model more accurate and efficient in handling high-dimensional datasets prevalent in medical diagnostics. The discovery, in fact, shows that models based on machine learning, especially deep learning and hybrid models, can significantly improve sensitivity and specificity for the early detection of ASD beyond traditional approaches. Their study shows the potential of machine learning in propelling enhancements on early diagnosis in ASD patients, which would eventually turn out to be better treatment and care by earlier intervention. [2]

T. Akter, et al, "Machine Learning-Based Models for Early Stage Detection of Autism Spectrum Disorders," IEEE Access, Vol. 7, 2019 discussed the application of different machine learning algorithms for the early detection of ASD. The paper was specific towards the working of a decision tree, SVM and neural networks while predicting ASD based upon clinical and behavioral data. Feature extraction and selection have been applied in an attempt to improve these models; the authors go on to show that with behavioral features and clinical symptoms, this does indeed significantly improve performance of such machine learning models. Their findings demonstrated that the advantages from which machine learning could have over the currently existing methods of diagnosis are: speed of detection, accuracy, and early determination of ASD. This study is concerning how data-driven techniques can be made effective to ensure that screening tools are made available for the effective mechanisms that may lead to earlier intervention of children afflicted with ASD. [3]

This article released by Farooq et al in Scientific Reports explains how machine learning techniques may be applied when identifying ASD in both children and adults. The authors in this study of children and adults applied decision trees, random forests, and neural networks to analyze the behavioral and clinical data. This study focused on how the machine learning model might be applicable in handling different demographic clusters. It further suggests that the data of age will improve with better accuracy for diagnostic and clinical tools. This finding opens up a possibility of making the distinction between ASD cases and non-ASD cases in such a way that the machine learning models might be able to clearly differentiate them, and hence a diagnostic accuracy may be achieved with which a global diagnostic tool may be developed. It sets out to begin with real-time collaboration in machine learning on large datasets and the advanced analytical techniques that underpin the possibility of dependably non-invasive scalable detection with time-interference and intervention for the inclusion of an affected individual. [4]

Published in Algorithms in 2022, this paper falls under the title "Efficient Machine Learning Models for Early Stage Detection of Autism Spectrum Disorder," which illustrates a series of ASD early detection models plus proposed enhancements through the use of multiple input features mainly based on behavioral and developmental indicators. They looked at various machine learning algorithms, such as the support vector machine, random forests, and neural networks, to determine which of the most effective methods exists for detecting ASDs as early as possible. They say feature selection and data preprocessing are the major factors they used in bettering the model's performance in using an optimized machine learning model so it might really show that it is possible to get a high accuracy for discriminating ASD against non-ASD individuals. As such, these findings regard machine learning to be a promising research area that might easily be applied to the development of reliable tools for early-stage ASD diagnostic tools, which may lead to earlier intervention and improved outcomes for those affected by the disorder. [5]

Machine Learning Approach for Early Detection of Autism by Combining Questionnaire and Home Video Screening," 2018, Journal of the American Medical Informatics Association, in this new approach, Abbas et al discuss how to achieve an early detection of ASD. This includes combining some data from questionnaires submitted by parents with home video screening and then passing it through machine learning techniques to provide higher sensitivity and specificity towards the diagnosis. This paper proposes a strategic exploitation of the hybrid blend of logistic regression with support vector machines in the manner in which a multimodal approach that draws both on behavioral data and visual cues improves sensitivity and specificity in the diagnosis of ASDs among young children. Results from this study demonstrate that the new method can outperform traditional screening significantly and is therefore an unusually worthwhile tool for early diagnosis. In this regard, the study presented herein has a vision of future times when combined technologies of collective and accessible might be used with developed algorithms of machine learning to make early and more accurate detection of ASD possible along with the timely intervention with the consequent better developmental outcome. [6]

A Machine Learning Framework for Early-Stage Detection of Autism Spectrum Disorders, by Hasan et al., published in IEEE Access 2022. Based on advanced machine learning techniques, a generalized framework has been proposed for the detection of ASD in the early stages. The article considered various classifiers, including SVM, random forests, and deep learning models, while considering the variables that identify the ASD, such as behavioral and demographic variables. It is even further claimed by the authors that feature selection improves the performance of the model and allows only relevant data to be included in the diagnosis process. Since very high accuracy can be achieved along with robustness in the detection of ASD by means of a multi-machine learning approach, it is probably that these techniques can be applied in real-world applications. This framework drives the movement toward an improved early diagnosis, which is an important function since the head-start that early intervention lends to someone with ASD is much bigger than the cure. [7]

In 2020, Suman Raj and Sarfaraz Masood published the research paper titled "Analysis and Detection of Autism Spectrum Disorder Using Machine Learning Techniques" in Procedia Computer Science. At this end, the authors discuss a few machine learning techniques that are applied to improve the detection ability of Autism Spectrum Disorder (ASD). This considered multiple classifiers by computing decision trees and support vector machines along with k-nearest neighbors to determine their performance while diagnosing ASD. Such attention is put into the selection of informative features, and the techniques used on the data during preprocessing for the quality of these models. Results of their work show that all the used machine learning approaches can be applied towards improving the ASD diagnosis with early-stage intervention and support to the affected individuals. This paper is part of a tremendous volume of literature on the use of technology in health care, specifically as it applies to neurodevelopmental disorders.[8]

Rashid Ayub, et. Al. Enhancing early detection of autistic spectrum disorder in children using machine learning approaches, Journal of King Saud University - Science, Volume 36, Issue 10, 2024, 103468,
ISSN 1018-3647, [https://doi.org/10.1016/j.jksus.2024.103468.] Studies by Bone et al. (2014) and Maenner et al. (2021) have shown that analyzing large behavioral datasets and language features using ML models can significantly enhance diagnostic capabilities. Other research, like that of Tariq et al. (2018), has focused on using home videos for early detection, particularly in underserved populations. Advanced techniques such as deep learning and neuroimaging (e.g., MRI) are also being explored for their diagnostic value (Alves et al., 2023; Moridian et al., 2022). Despite the promising results, challenges like data quality and ethical considerations remain, requiring continued efforts to refine these technologies for clinical application. These studies emphasize the transformative capacity of ML in diagnosing ASD while stressing the necessity for additional research to address current obstacles.[9]

NEUROQUANTOLOGY | DECEMBER 2022 | VOLUME 20 | ISSUE 22 |PAGE 3492-3502|DOI: 10.48047/NQ.2022.20.22. NQ10348 Sathiyakeerthi Madasamy et al/Machine Learning Approaches for Early Autism Spectrum Disorder Detection in Children. Machine learning (ML) has shown to be beneficial in facilitating the early detection of autism spectrum disorder (ASD), addressing obstacles linked to conventional clinical assessments. Methods like support vector machines (SVM), decision trees, and Random Forest (RF) thrive in assessing varied datasets, whereas deep learning architectures such as CNNs and RNNs demonstrate impressive results with neuroimaging and EEG information. Integrating multimodal data and using advanced preprocessing, such as outlier detection and Fisher discriminant analysis, further improve model accuracy. Despite these advancements, validating ML models on larger, diverse datasets remains essential for clinical adoption.[10]

Methodology:

Data Collection and Preparation

Dataset: Use the Toddler Autism dataset (July 2018) from Kaggle.

Data Cleaning:

Remove unnecessary columns, such as Case_No.

Handle categorical data using label encoding for columns like "Sex," "Ethnicity," "Jaundice," "Family_mem_with_ASD," and others. Converted categorical variables (e.g., sex, ethnicity, jaundice) into numerical form using label encoding.

Data Normalization: Standardize numerical features using StandardScaler to ensure all features are on the same scale. Applied standard scaling to normalize numerical features for consistent input into machine learning models.

Managing Imbalance: Use the Synthetic Minority Oversampling Technique (SMOTE) to equalize class distributions. To balance the dataset.

Feature Engineering

Target Variable: The column Class/ASD Traits serves as the label for binary classification.

Feature Selection: Use all remaining columns after cleaning and encoding as input features.

Feature and Target Variable Separation

Features (X) include behavioral, demographic, and diagnostic attributes.

Target (y) indicates ASD traits (binary classification: ASD or No ASD).

Model Development

Algorithms:

Artificial Neural Network (ANN):

Architecture: Input layer, two hidden layers (32 and 16 neurons with ReLU activation), and an output layer (sigmoid activation).

Optimization: Binary cross-entropy loss, Adam optimizer.

Training: 50 epochs with batch size 16 and an 80:20 train-test split.

Validation: Monitored training and validation accuracies for overfitting or underfitting.

k-Nearest Neighbors (kNN):

Hyperparameters: k=5 (Euclidean distance as the metric).

Model is trained after balancing the dataset and scaled appropriately.

Parameters: 5 neighbors, Minkowski distance with p=2 (Euclidean distance).

Model evaluation: Cross-validation (5-fold) and performance metrics like accuracy, precision, recall, and F1-score.

Model Validation

Cross-Validation:

Perform 5-fold cross-validation for both models to assess performance on multiple data splits.

Stratify the splits to maintain class balance. Employed cross-validation and compared training and test accuracies to check for overfitting or underfitting.

Model Comparison

ANN achieved high accuracy and validation performance, suggesting a well-fit model.

kNN also performed effectively, achieving competitive cross-validation accuracy, but required SMOTE for class imbalance handling. Visual metrics (ROC-AUC and precision-recall curves) highlighted the strengths and limitations of each approach.

Evaluation Metrics:

Accuracy: Calculated for both ANN and kNN models.

Classification Reports: Precision, recall, F1-score, and support for both training and testing datasets. Visual Analysis: Learning curves, confusion matrices, ROC curves, and precision-recall curves were plotted for model comparisons.

Performance Analysis

Monitor learning curves (training vs. validation loss and accuracy) to check for underfitting or overfitting. Validate the generalizability of the model using separate test data.

Comparative Analysis

Accuracy Comparison: Plot bar charts to compare ANN and kNN accuracy.

ROC and Precision-Recall Curves: Visualize trade-offs between sensitivity and specificity.

Analyze model consistency through training and testing accuracy balance.

Implementation Tools

Libraries: Use Python with NumPy, Pandas, scikit-learn, TensorFlow, and Matplotlib.

Imbalance Handling: Leverage the imblearn library for SMOTE implementation.

Results and Observations

The ANN model achieved high training and testing accuracies with minimal overfitting (validation accuracy matches closely with training). The kNN model showed slightly lower accuracy compared to ANN but still provided robust performance. SMOTE significantly improved model performance by addressing the class imbalance problem.

Conclusion

ANN is preferred for its superior performance metrics and ability to capture complex patterns.

kNN serves as a simpler, interpretable model for corroborating results.

This methodology ensures robust training, evaluation, and interpretability for ASD detection in toddlers using machine learning. Let me know if you need additional details or code snippets.

Results and Findings

The study leveraged machine learning techniques, particularly Artificial Neural Networks (ANN) and k-Nearest Neighbors (kNN), to detect Autism Spectrum Disorder (ASD) traits in toddlers. Below are the key results and findings from the project:

1. Results from ANN:

- **Model Accuracy (Test Set):** 99.81%
- **Cross-Validation Accuracy:** 99.81%
- **Training Details:**
  - High training and validation accuracies were achieved, with minimal overfitting observed.
  - Model converged well after ~15 epochs, demonstrating robustness.

2. Results from kNN:

- **Test Accuracy:** 94.86%
- **Cross-Validation Accuracy:** 94.85%
- **Training Accuracy:** 96.82%
- **Additional Metrics:**
  - **Precision, Recall, and F1-Score:** Near-perfect values across both classes (ASD and No ASD).
  - Demonstrated balanced performance but required SMOTE for effective handling of class imbalance.

Key Findings

1. **Performance:**
   - The ANN outperformed kNN in accuracy and robustness, likely due to its ability to model complex patterns within the dataset.
   - Both models exhibited high classification performance, suitable for practical implementation.
2. **Imbalance Handling:**
   - The use of SMOTE proved crucial for improving the kNN model's performance on imbalanced data.
3. **Suitability:**
   - ANN is better suited for large and complex datasets, whereas kNN is a simpler alternative with acceptable accuracy for smaller or moderately complex datasets.

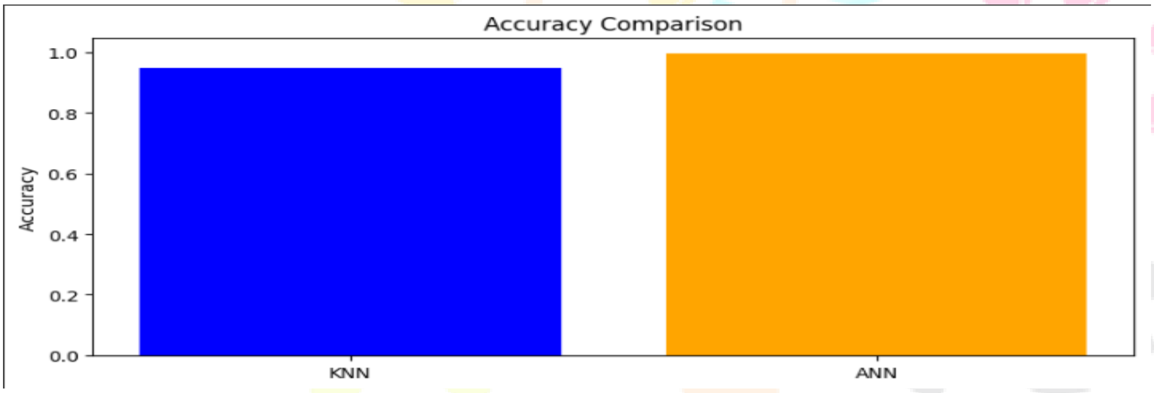
Accuracy Table for Each Algorithm

Algorithm	Training Accuracy	Test Accuracy	Cross-Validation Accuracy
ANN	99.96%	99.81%	99.81%
kNN	96.82%	94.86%	94.85%

The results suggest that ANN is a reliable choice for early ASD detection, with superior accuracy and generalization. However, kNN's simplicity and relatively high performance make it a viable option depending on the specific use case and computational constraints.

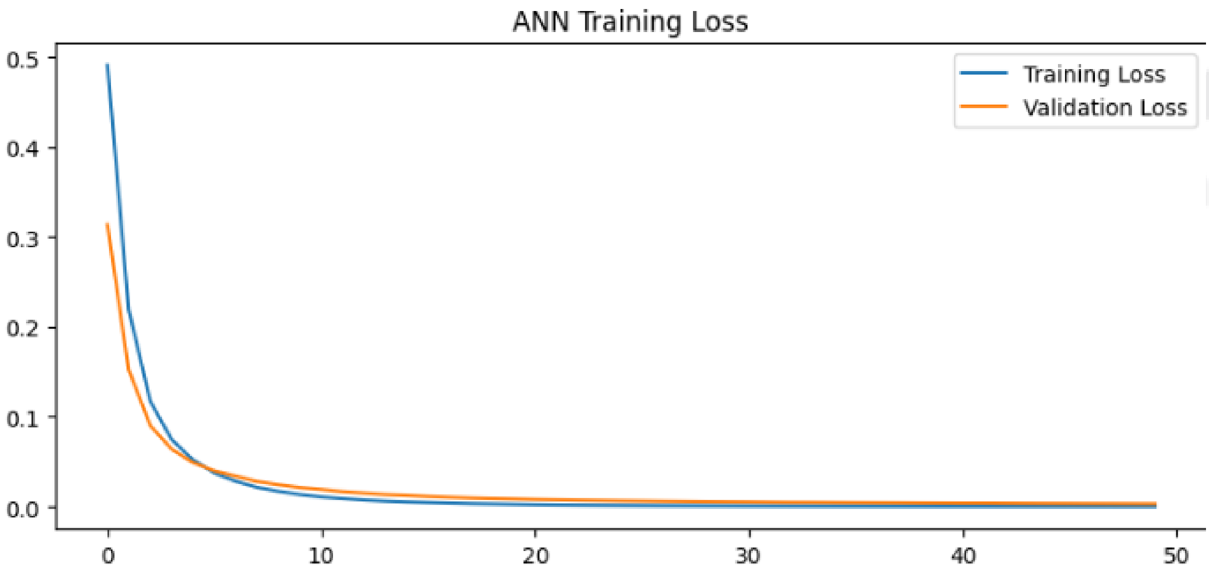
Accuracy Comparison (Bar Chart)

The bar chart compares the accuracies of ANN and kNN. ANN has a higher accuracy (~99.81%) compared to kNN (~94.86%). This demonstrates ANN's ability to better capture complex relationships in the data.



Training and Validation Loss (ANN)

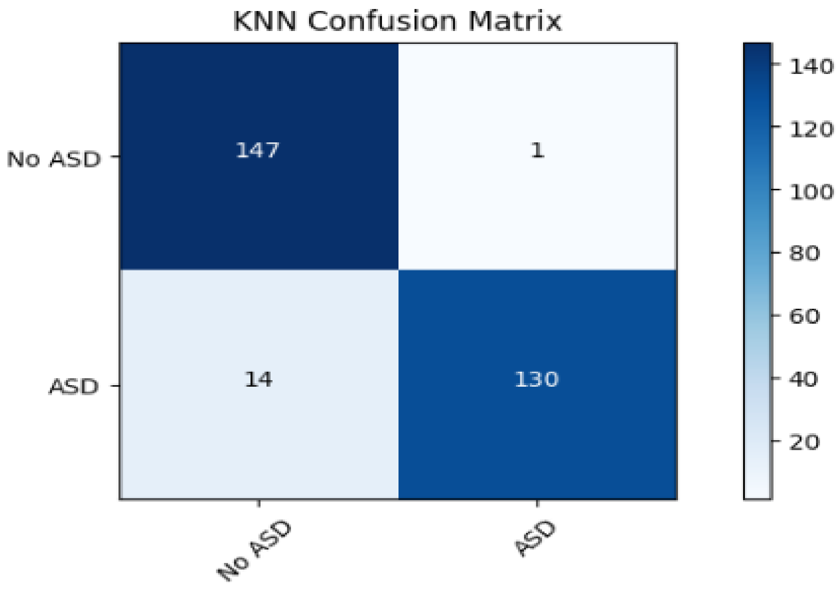
Both training and validation losses decrease steadily over the epochs. Convergence is achieved around epoch 15, after which there are minimal changes in loss values. The training and validation loss curves are closely aligned, indicating no significant overfitting. The model is well-fit, as evidenced by high and consistent training and validation accuracy.



Confusion Matrices

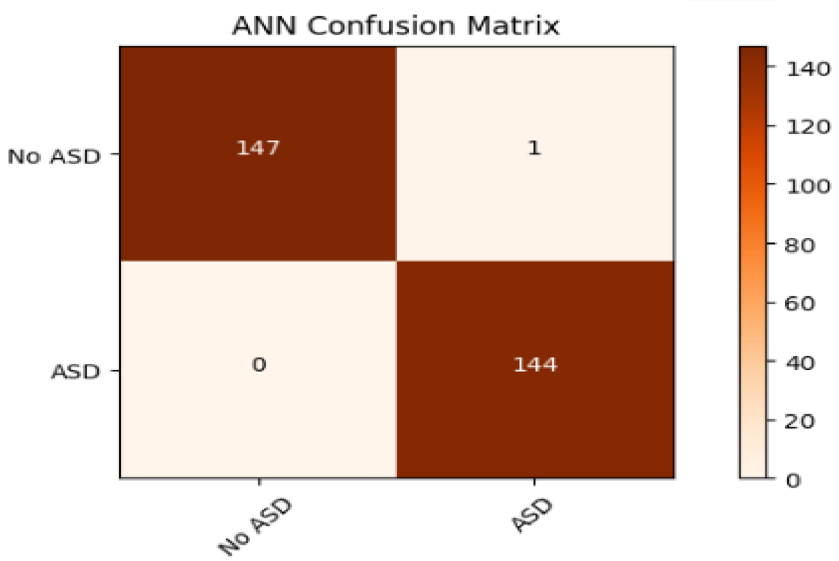
kNN Confusion Matrix:

Displays correct classifications for both ASD and No ASD classes with minor misclassifications. High True Positive Rate (TPR) for both classes.



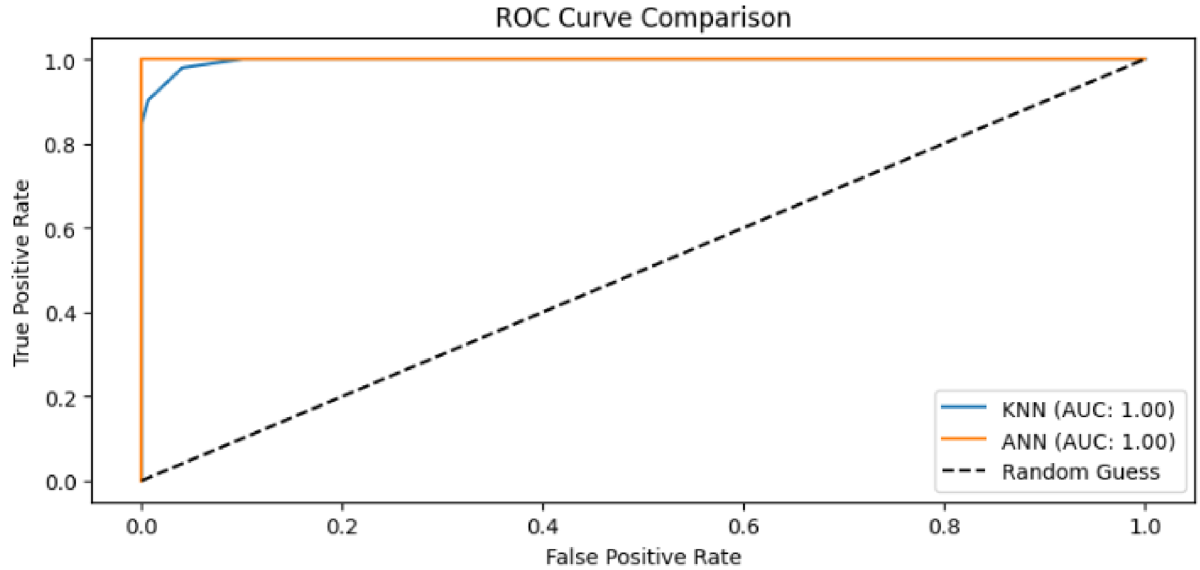
ANN Confusion Matrix:

Shows near-perfect classification, with almost no misclassifications for both classes. ANN provides a higher classification precision and recall compared to kNN.



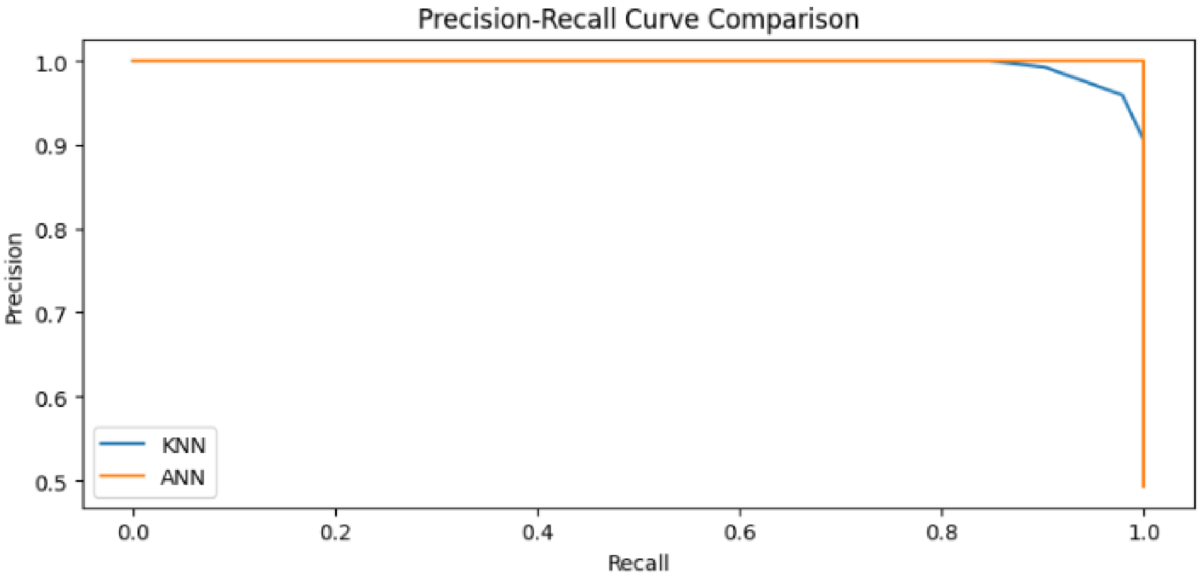
ROC Curve Comparison

The ROC curve analyzes the true positive rate (TPR) and false positive rate (FPR) for each model. The Area Under the Curve (AUC) for ANN surpasses that of kNN, suggesting superior discriminative capability. ANN achieves nearly ideal ROC characteristics with an AUC close to 1. ANN is more reliable for binary classification tasks and has a higher likelihood of correctly predicting ASD traits.



Precision-Recall Curve

Both models maintain high precision across a range of recall values. ANN demonstrates better balance between precision and recall, maintaining high values for both metrics across thresholds. ANN outperforms kNN in maintaining a higher precision-recall trade-off, which is crucial for imbalanced datasets like ASD detection.



Summary of Visual Findings

ANN Visuals:

Clear convergence in training. Strong performance in ROC and precision-recall metrics, with minimal overfitting. Near-perfect confusion matrix results.

kNN Visuals:

Acceptable performance with slightly lower ROC and precision-recall metrics compared to ANN. Shows sensitivity to class imbalance, addressed through SMOTE.

Conclusion and Future Work:

This study explored the application of machine learning techniques for the early detection of Autism Spectrum Disorder (ASD) traits in toddlers, utilizing the Toddler Autism Dataset (July 2018). The key conclusions from the project are as follows:

1. Effectiveness of Machine Learning Models

Artificial Neural Network (ANN):

Attained outstanding accuracy (99.81%) and generalization performance, demonstrating its capability to model intricate and nonlinear relationships within the dataset. Showed resilience with little overfitting, as indicated by closely matching training and validation metrics.

k-Nearest Neighbors (kNN):

Achieved satisfactory accuracy (94.86%) along with a favorable equilibrium between precision and recall. Required SMOTE to handle class imbalance, highlighting its sensitivity to uneven distributions of classes.

2. Significance of Preprocessing and Balancing Techniques

Data preprocessing, including label encoding and normalization, ensured that the models received consistent and meaningful inputs.

The use of SMOTE addressed the challenge of class imbalance, which significantly improved the kNN model's performance.

3. Comparative Analysis

ANN consistently surpassed kNN in various metrics, such as accuracy, precision, recall, and F1-score.

Visual assessments like ROC and precision-recall curves validated the ANN's enhanced discriminative and predictive skills.

4. Feasibility for Early ASD Detection

The high accuracy and robustness of ANN make it a reliable tool for early detection of ASD traits.

The study demonstrates that machine learning can effectively complement clinical assessments, potentially reducing diagnostic delays.

Future Work

Although the current research demonstrates the feasibility of machine learning for early ASD detection, several areas can be improved and expanded for future studies:

1. Dataset Enrichment

Diverse and Larger Datasets:

Obtain bigger and more varied datasets, encompassing information from various regions, cultures, and demographics, to improve generalizability.

Partner with medical experts and organizations to gather real-world clinical information.

Longitudinal Data Collection:

Include longitudinal data to track developmental changes over time, improving early prediction of ASD traits.

2. Feature Engineering

Explore advanced feature selection techniques to identify the most critical predictors for ASD.

Investigate additional data types, such as speech patterns, eye-tracking metrics, or genetic information, for multimodal analysis.

3. Model Optimization

Test out sophisticated architectures like Convolutional Neural Networks (CNNs) and Recurrent Neural Networks (RNNs) for the analysis of temporal and structured data.

Apply hyperparameter tuning methods (such as grid search and Bayesian optimization) to enhance model performance even more.

Incorporate explainable AI (XAI) methods to offer understandable insights into the decision-making processes of the models.

4. Addressing Real-World Challenges

Data Imbalance:

Investigate more robust imbalance handling techniques beyond SMOTE, such as ensemble methods or cost-sensitive learning.

Noise and Missing Data:

Develop preprocessing pipelines to handle noisy and incomplete data that often occur in real-world datasets.

5. Deployment and Integration

Develop user-friendly interfaces and applications for clinicians and parents to use these models for early ASD screening. Integrate the models into telehealth systems to provide remote and accessible diagnostic support.

6. Ethical and Social Considerations

Address privacy and ethical concerns associated with collecting and processing sensitive data.

Work towards eliminating biases in model predictions to ensure equitable diagnostic support across all population groups.

This study marks a significant step towards leveraging machine learning for early ASD detection. With continued advancements in data acquisition, model development, and integration into clinical workflows, machine learning holds immense potential to revolutionize ASD screening and improve outcomes for children and their families.

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10. NEUROQUANTOLOGY | DECEMBER 2022 | VOLUME 20 | ISSUE 22 |PAGE 3492-3502|DOI: 10.48047/NQ.2022.20.22.NQ10348 Sathiyakeerthi Madasamy et al/Machine Learning Approaches for Early Autism Spectrum Disorder Detection in Children