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ML-DRIVEN APPROACH FOR IMPROVED DIAGNOSIS OF AUTISM SPECTRUM DISORDER

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Abstract

Autism Spectrum Disorder (ASD), commonly referred to as "autism," is a psychiatric condition that affects a person's linguistic, cognitive, and social abilities. It's a prevalent disorder, with approximately 1 in every 54 births being diagnosed with ASD, and about 1% of the global population living with it. Unfortunately, despite its prevalence, the cause and cure for autism remain unknown, posing significant challenges to parents who suspect their child might have ASD. Early diagnosis of autism is crucial for a child's development, but it can be incredibly tough since symptoms manifest as the child grows. Typically, diagnostic tests conducted on children between the ages of 2 to 3 years are less reliable than those performed on children aged 4 to 5 years. This creates a worrying situation because early diagnosis is vital for autistic individuals to reach their developmental milestones successfully. Autism is often characterized by difficulties in social interaction and communication, making it challenging to diagnose accurately even with advanced tools like the ADOS and ADI. This work addresses the concerns surrounding autism diagnosis by focusing on improving the diagnostic pipeline. It involves training and testing machine learning models i.e., Random Forest with Standard scaler using an autism spectrum disorder dataset to identify the most significant indicators of autism in toddlers. The goal is to develop a quantitative approach to aid in early screening and subsequent treatment, as timely intervention can help mitigate long-term symptoms associated with autism. By leveraging machine learning, this work aims to provide valuable insights into diagnosing autism effectively and facilitating better support for individuals with ASD and their families.

Keywords: Autism spectrum disorder, Predictive analytics, Data analysis, Supervised learning.

1. Introduction

Autism spectrum disorder (ASD) is a developmental disorder that involves persistent challenges in social interaction, speech and nonverbal communication, and restricted and repetitive behaviours. In the USA, the prevalence of ASD has increased substantially in the past two decades, with an estimate of every 1 in 44 children to be identified with ASD by age 8 in 2016 [1]. Although there exist evidence-based interventions which improve core symptoms in children with ASD, many children with ASD still experience long-term challenges with daily life, education and employment [2]. Early diagnosis is the key to early intervention for improving the long-term outcomes of children with ASD. However, despite the growing evidence shows that accurate and stable diagnoses can be made by 2 years, in real-world settings, the median age of ASD diagnosis is 50 months. To improve early diagnosis, the American Academy of Paediatrics (AAP) has recommended universal screening among all children at 18-month and 24-month well-child visits in the primary care settings using the Modified Checklist for Autism in Toddlers (M-CHAT) [3], a questionnaire that assesses children's behaviour for toddlers. However, growing evidence has shown that using M-CHAT alone may not yield sufficient accuracy in detecting ASD cases, with a sensitivity below 40% and a positive predictive value (PPV) under 20% [4, 5]. In addition to ASD-specific behavioural questionnaires, general clinical and healthcare records may also

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contain meaningful signals to differentiate the ASD risks among very young children. Studies have found that children with ASD are oftentimes accompanied by certain symptoms and medical issues such as gastrointestinal problems, infections and feeding problems. This implies that past diagnosis and healthcare encounter information, commonly available from health insurance claims or Electronic Healthcare Record (EHR), could potentially be used for ASD risk prediction. In fact, medical claims and EHR data have been widely used in the health informatics literature for identifying disease-specific early phenotypes even before the hallmark symptoms start to manifest, such as for chronic diseases like heart failures, diabetes and Alzheimer's disease [6].

2. Literature Survey

This section explains previous studies that use machine learning-based approaches to detect and predict the autism spectrum disorder. The main motive is to analyze and find some limitations to propose a new, better, and improved machine-learning based approach for autism spectrum disorder prediction. Automated algorithms for disease detection are being deeply studied for usage in healthcare. Graph theory and machine learning algorithms were used. For each age range being examined, the pipeline automatically selected 10 biomarkers. In discriminating between ASD and HC, measures of centrality are the most operational [8]. The study [9] used a neural network-based feature selection method from teacher-student which was suggested to have the most discriminating features and applied different classification algorithms. The results are compared with the already presented methods at the overall and site level. The authors in [10] also utilize the neural network to acquire the distributions of PCD for the classification of ASD as it has far more hyper parameters that make the model extra versatile. Payabvash et al. [11] used computer leaning algorithms to classify children with autism based on tissue connectivity metrics, hence, observed decreased connectome edge density in the longitudinal white matter tracts. It illustrated the viability of it in identifying children with ASD, connectome-based machine-learning algorithms.

The authors in [12] conclude that the data may be used to establish diagnostic biomarkers for the progression of autism spectrum disorders and to distinguish those with the condition in the general population. Wang et al. [13] proposed an ASD identification approach which focuses on multi-atlas deep feature representation and ensemble learning technique. In study [14], the multimodal automated disease classification system uses two types of activation maps to predict whether the person is healthy or has autism. It was able to achieve 74% accuracy. Rakić et al. [15] suggested a technique which is based on a system composed of autoencoders and multilayer perceptron. Because of a multimodal approach that included a set of structural and functional data classification classifiers, the highest classification precision was 85.06%. In study [16], advanced deep-learning algorithms are proposed where HPC solutions can increase the accuracy and time of broad fMRI data analysis significantly. Thomas et al. [17] introduced a novel analysis technique to identify changes in population dynamics in functional networks under ASD. They have also introduced machine learning algorithms to predict the class of patients with ASD and normal controls by using only population trend quality metrics as functions. The limitation of this approach is that the outcomes of the classification are highly dependent on the threshold parameter T. Another problem is that despite age variations in the experimental samples, the same spatial normalization design was used for all subjects. The authors in ref [18] proposed a collection of new features based on MRI images using machine learning algorithms to diagnose ASD which achieved 77.7% accuracy using the LDA approach.

3. Proposed Methodology

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Activity diagrams are graphical representations of Workflows of stepwise activities and actions with support for choice, iteration, and concurrency. In the Unified Modeling Language, activity diagrams can be used to describe the business and operational step-by-step workflows of components in a system. An activity diagram shows the overall flow of control.

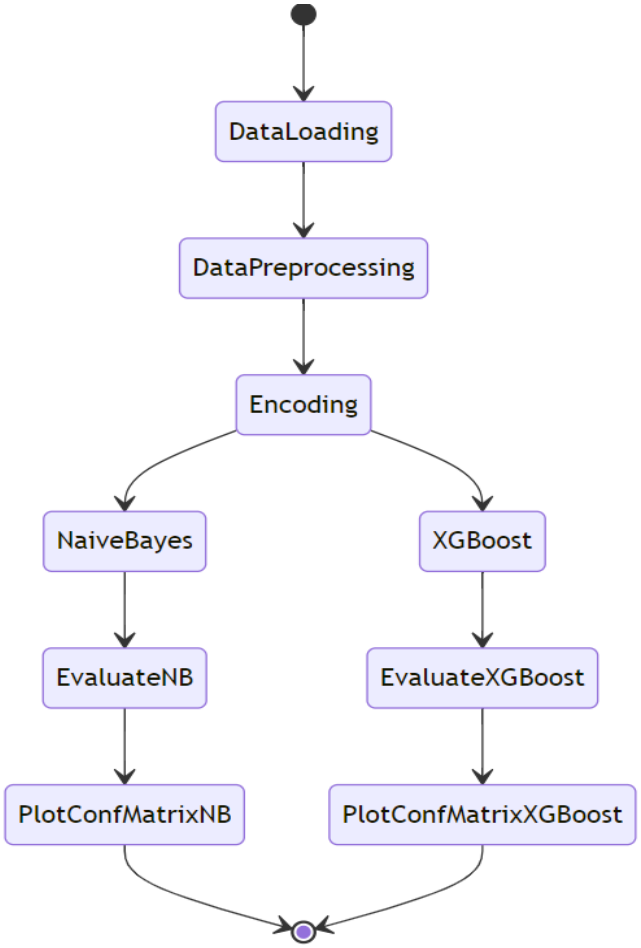

Figure 1. Proposed system design.

3.1 XGBoost Classifier

XGBoost is an optimized distributed gradient boosting library designed for efficient and scalable training of machine learning models. It is an ensemble learning method that combines the predictions of multiple weak models to produce a stronger prediction. XGBoost stands for "Extreme Gradient Boosting" and it has become one of the most popular and widely used machine learning algorithms due to its ability to handle large datasets and its ability to achieve state-of-the-art performance in many machine learning tasks such as classification and regression. One of the key features of XGBoost is its efficient handling of missing values, which allows it to handle real-world data with missing values without requiring significant pre-processing. Additionally, XGBoost has built-in support for parallel processing, making it possible to train models on large datasets in a reasonable amount of time. XGBoost can be used in a variety of applications, including recommendation systems, and click-through rate prediction, among others. It is also highly customizable and allows for fine-tuning of various model parameters to optimize performance.

4. Results and Disussion

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This dataset contains information about various attributes of individuals, including demographic information (age, gender, ethnicity), medical history (jaundice), family history of ASD, and assessment scores (Q-CHAT-10) used to predict the presence of ASD traits. The "Class/ASD Traits" column appears to be the target variable used for classification purposes, indicating whether the individual exhibits ASD traits or not.

	Case_No	A1	A2	A3	A4	A5	A6	A7	A8	A9	A10	Age_Mons	Qchat-10-Score	Sex	Ethnicity	Jaundice	Family_mem_with_ASD	Who completed the test	Class/ASD Traits
0	1	0	0	0	0	0	0	1	1	0	1	28	3	f	middle eastern	yes	no	family member	No
1	2	1	1	0	0	0	1	1	0	0	0	36	4	m	White European	yes	no	family member	Yes
2	3	1	0	0	0	0	0	1	1	0	1	36	4	m	middle eastern	yes	no	family member	Yes
3	4	1	1	1	1	1	1	1	1	1	1	24	10	m	Hispanic	no	no	family member	Yes
4	5	1	1	0	1	1	1	1	1	1	1	20	9	f	White European	no	yes	family member	Yes
...	...	...	...	...	...	...	...	...	...	...	...	...	...	...	...	...	...	...	...
1049	1050	0	0	0	0	0	0	0	0	0	1	24	1	f	White European	no	yes	family member	No
1050	1051	0	0	1	1	1	0	1	0	1	0	12	5	m	black	yes	no	family member	Yes
1051	1052	1	0	1	1	1	1	1	1	1	1	18	9	m	middle eastern	yes	no	family member	Yes
1052	1053	1	0	0	0	0	0	0	1	0	1	19	3	m	White European	no	yes	family member	No
1053	1054	1	1	0	0	1	1	0	1	1	0	24	6	m	asian	yes	yes	family member	Yes

1054 rows × 19 columns

Figure 2: Sample dataset used for classification of ASD.

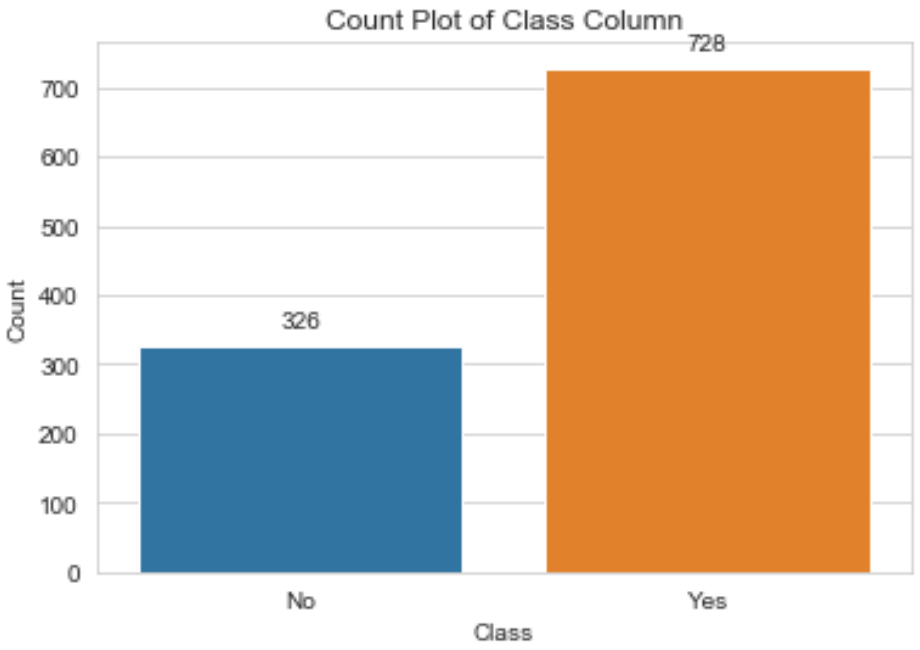

Figure 3: Count plot of class column i.e., Autism or Normal.

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```
Accuracy: 100.00
Classification Report:
              precision    recall  f1-score   support

           0       1.00      1.00      1.00        68
           1       1.00      1.00      1.00       143

    accuracy                           1.00       211
   macro avg       1.00      1.00      1.00       211
weighted avg       1.00      1.00      1.00       211
```

Figure 4: Obtained accuracy and classification report using XGBoost model.

5. Conclusion

In the analysis of autism prediction in toddlers, the XGBoost model emerges as the superior choice compared to the Naive Bayes model. This assessment is based on various performance metrics and evaluations. Firstly, in terms of accuracy, the XGBoost model demonstrates a notably higher accuracy score when compared to the Naive Bayes model. This suggests that XGBoost excels at correctly classifying cases, a crucial aspect of any predictive model. Secondly, a closer examination of the classification report reinforces the superiority of the XGBoost model. The classification report provides insights into precision, recall, and F1-score values for both classes—likely autism and not autism. The XGBoost model consistently showcases higher values across these metrics, indicating a more balanced and accurate classification of positive and negative cases. Furthermore, the confusion matrix, which offers a detailed breakdown of true positives, true negatives, false positives, and false negatives, reflects better performance by the XGBoost model.

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