# Decoding Autism Diagnosis: A Journey Towards Transparency with XAI in ML Models

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Abstract: Autism Spectrum Disorder (ASD) is a developmental condition that manifests within the first three years of life. Despite the strides made in developing accurate autism classification models, particularly utilizing datasets like AQ-10, the lack of interpretability in these models poses a significant challenge. In response to this concern, we employ eXplainable Artificial Intelligence (XAI) techniques, specifically Local Interpretable Model-agnostic Explanations (LIME) and Shapley Additive exPlanations (SHAP), to enhance transparency. Our primary aim, following the commendable accuracy achieved with the AQ-10 dataset, is to demystify the black-box nature of machine learning models used for autism classification. LIME provides locally faithful explanations, offering a more nuanced understanding of predictions, while SHAP quantifies the contribution of each feature to the model's output. Through instance-based analyses, we leverage these XAI techniques to delve into the decision-making processes of the model at an individual level. Integrating LIME and SHAP not only elevates the model's trustworthiness but also helps a deeper comprehension of the factors influencing autism classification. Our results underscore the efficacy of these techniques in unraveling the intricacies of the model's decisions, shedding light on relevant features and their impact on classification outcomes. This research contributes to bridging the gap between accuracy and interpretability in machine learning applications, particularly within the realm of autism classification.

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## 1 INTRODUCTION

Autism Spectrum Disorder (ASD) is a behavioral condition that affects how individuals interact with society throughout their lifetime. Symptoms of ASD typically appear during childhood and persist into adolescence and adulthood (Hasin et al., 2013). ASD patients exhibit repetitive behaviors, and analyzing these behaviors can aid in early detection. The diversity of behaviors demonstrated by ASD patients depends on age and ability. Common behavioral disorders in ASD patients include deficient expressive gestures, non-responsiveness to sound, lack of proper eye contact, no sensation of pain, repetition of words, and agitation with changes in daily routines (Hasin et al., 2013). Compared to healthy populations, siblings of individuals with autism are at a fifty times greater risk of developing ASD (Joseph and Tager-Flusberg, 1997). Additionally, males are 4-5 times more likely to be affected than females. The World Health Organization reports that 1 in 160 children worldwide is prone to developing ASD at any given time. Hong Kong, South Korea, and the United States have the highest prevalence rates of ASD. In India, the prevalence rate is 1 in 500, with an incidence rate of 11,914 people yearly. In recent years, ASD prevalence has increased from 15 to 64 per 10,000 in India (Geetha et al., 2019). According to the Autism Society of America, the incidence rate of autism is rising at a rate of 10-17% each year in the USA. In 2020, the CDC (Centers for Disease Control and Prevention) reported a 10% increase in autism incidence rate, with 1 in 54 children in the USA being diagnosed with ASD. These statistics are alarming, considering that ASD is a rare disease (Shaw et al., 2023).

While ASD cannot be fully cured, early detection of symptoms can help reduce the effects of the disease. Machine learning (ML) has been applied to predicting and detecting various diseases with good accuracy, including ASD, based on multiple physical and physiological parameters (Raj and Masood, 2020). However, detecting and analyzing ASD is challenging due to the existence of other mental health problems with typical symptoms, which

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can result in false detection. To address this issue, a machine learning model is proposed in this paper for early prediction of ASD through screening test datasets, with the application of Explainable Artificial Intelligence (XAI) to identify the contribution of features towards accurate prediction. Comparative case studies have been conducted using available features in the dataset and the most contributing features predicted by XAI through performance metrics: accuracy, precision, recall, and F1-score (Das and Rad, 2020).

### 1.1 Contribution

1. This research focuses on detecting ASD using AQ-10 datasets. It provides a comprehensive guide on implementing machine learning algorithms specifically designed for this purpose.
2. Addresses interpretability issue in black box models with XAI techniques (LIME and SHAP).
3. Provides detailed analysis of each prediction and identifies specific attributes with the greatest impact on the decision-making process.

The paper is divided into four main sections. Section 2 explores the related work, providing a context and existing knowledge in the field. Moving forward, Section 3 gives a concise overview of the dataset and describes the pre-processing steps taken. Section 4 presents the proposed work and provides an in-depth examination of the methodology used in our experiments. Section 5 unveils the results of our experiments and engages in a comprehensive discussion. Finally, Section 6 delivers a conclusive summary and findings, encapsulating the paper's key insights and future scope.

## 2 RELATED WORK

Based on the reviewed literature, Machine Learning (ML) techniques have proven highly effective in predicting various diseases based on syndromes. For example, Cruz et al. (Cruz and Wishart, 2006) used ML to diagnose cancer, demonstrating its broad range of applications in predicting diseases. Khan et al. (Khan et al., 2017) also employed ML to predict diabetes, showcasing its versatility in addressing different health concerns. Wall et al. (Wall et al., 2012) utilized the Alternating Decision Tree (ADTree) method to detect ASD traits and achieved high accuracy within a specific age range (5-17 years). However, their approach was limited in its ability to predict ASD across diverse age groups. Bone et al. (Bone et al., 2016) applied a Support Vector Machine (SVM) to predict ASD traits, achieving notable sensitivity and specificity. However, the study faced constraints due to a wide age range (4-55 years). Allison et al. (Allison et al., 2012) achieved over 90% accuracy in ASD screening by employing the 'Red Flags' tool in conjunction with the Autism Spectrum Quotient. Thabtah (Thabtah, 2017) conducted a comprehensive comparative analysis of previous ML algorithms for autism trait prediction. In contrast, Hauck and Kliewer (Hauck and Kliewer, 2017) identified pivotal screening questions for the Autism Diagnostic Observation Schedule (ADOS) and Autism Diagnostic Interview-Revised (ADI-R) methods, emphasizing the efficacy of their combined use. Bekerom (van den Bekerom, 2017) used various ML techniques to discern ASD traits in children, accounting for factors like developmental delay, obesity, and physical activity levels. Wall et al. (Wall et al., 2012) made progress in classifying autism through concise screening tests, with the ADTree and functional tree exhibiting promising results. Heinsfeld (Heinsfeld et al., 2018) explored algorithms and neural networks for identifying ASD patients, achieving classification accuracy ranging from 66% to 71%. Meanwhile, Liu (Liu et al., 2016) investigated the potential of face-scanning patterns to identify children with ASD, achieving exceptional accuracy, specificity, sensitivity, and Area Under the Curve (AUC) metrics. Bone et al. (Bone et al., 2015) conducted a meticulous analysis of prior studies, addressing conceptual and methodological issues and successfully replicating results using their ML approach. Despite the extensive research in this field, a definitive conclusion regarding the universal applicability of ML-based autism screening across age groups remains elusive. Therefore, the existing tools and techniques call for developing a comprehensive app-based solution tailored to different age demographics.

Numerous studies have delved into the application of machine learning techniques to predict ASD from high-dimensional datasets. Archana et al. (Archana et al., 2023) introduced a comprehensive methodology encompassing dimensionality reduction, feature extraction, and classifier selection, achieving notable advancements in ASD prediction. Their approach addresses the challenge posed by large feature sets in high-dimensional data. Additionally, similar efforts have been made by several researchers. They demonstrated promising results in refining classifiers for enhanced ASD prediction. Furthermore, it examined various classifiers, emphasizing the potential of Decision Tree classifiers in achieving high accuracy rates in ASD prediction. Building upon these preceding works, the current study refines and extends these methodologies, achieving a remarkable testing accuracy of 97.47% with a reduced feature set of 254 features and a model training time of 23.508 seconds. This research contributes significantly to the evolving landscape of machine learning-based ASD prediction, offering promising avenues for improving early diagnosis and intervention strategies for individuals with ASD.

# 3 DATASET DESCRIPTION AND PRE-PROCESSING

Data is a crucial component in AI, and to achieve substantial efficiency, a large amount of data must be analyzed. Numerous resources have been dedicated to collecting data, including Dr. Fadi Fayez Thabtah (Thabtah, 2019; Thabtah et al., 2018), who has focused on autism assessment in young children. The dataset used in this study was curated through the ASDTests mobile application, which can be accessed at ASDTest. This dataset represents a significant advancement in screening autism, particularly in infants, and highlights several influential factors that require further exploration. These factors are essential not only for detecting autistic traits but also for refining the categorization of ASD. As part of the diagnosis process for ASD, behavioral traits are recorded using the Q-chat-10 questionnaire.

This dataset has been designed specifically for classification tasks in ASD. It consists of 704 instances, with each instance containing 21 attributes. These attributes include a mix of categorical, continuous, and binary data. The dataset provides demographic details such as age and gender, as well as contextual factors like ethnicity and family history of Pervasive Developmental Disorders (PDD). Additionally, it includes specifics about the screening process, such as who completes the test, the country of residence, and whether a screening app was used previously. The dataset also describes screening methods by age category and captures binary responses (0 or 1) to ten questions as mentioned in table 1 integral to the screening process.

In the process of developing a classification model, the attributes, denoted as [X], serve as independent variables. These features are utilized to construct the model, where the target variable, represented as [Y], is binary. Specifically, [Y] indicates the presence (1) or absence (0) of autistic traits.

# 4 PROPOSED WORK

Our primary objective is to build a model that generates predictions and provides insight into its decision-making process. We aim to understand the reasons behind the model's decisions and the factors contributing to its predictions. This requires us to examine each output and explain the basis of each decision. As we implement machine models, it is crucial to consider interpretability in the model's outcomes.

## 4.1 LIME

Local Interpretable Model-agnostic Explanations (LIME) (Ribeiro et al., 2016) is a technique that aims to explain the predictions of machine learning models, particularly those considered black-box models. The main goal of LIME is to provide interpretable explanations for individual predictions, which can help humans understand why a specific prediction was made. The process involves selecting a particular instance and generating perturbed versions of that instance by slightly and randomly modifying its features. These perturbed instances are then used to obtain predictions from the black-box model. After that, a locally interpretable linear regression model is trained on the perturbed instances and their corresponding black-box model predictions. The optimization problem can represent the working equation of LIME:

$$\text{minimize } L(f,g,\pi_x) = \mathbb{E}_{x'\sim\pi_x}\left[\mathcal{L}\left(f,g,x'\right)\right] + \Omega(g)$$

Here, $f$ represents the black-box model, $g$ is the interpretable model, $\pi_x$ is the probability distribution over perturbed instances, $x'$ is a perturbed instance, $\mathcal{L}$ is a loss function measuring the difference between black-box and interpretable model predictions, and $\Omega(g)$ is a regularisation term discouraging model complexity. This equation encapsulates the optimization problem that LIME addresses, seeking an interpretable model $g$ that effectively captures the behavior of the complex model $f$ for the chosen instance while promoting simplicity through regularisation.

## 4.2 SHAP

SHapley Additive exPlanations (SHAP) (Lundberg and Lee, 2017) is an algorithm that helps explain the output of machine learning models fairly and consistently. It uses the concept of Shapley values from cooperative game theory, where each feature is considered as a "player" in a game, with the prediction being the outcome. The Shapley value provides a

Table 1: Q-chat-10-Toddler Features Corresponding to Variables in Dataset.

| Variable in Dataset | Corresponding Q-chat-10-Toddler Feature |
|---|---|
| A1 | Does your child look at you when you call his/her name? |
| A2 | How easy is it to make eye contact with your child? |
| A3 | Does your child point to indicate that s/he wants something? (e.g., a toy that is out of reach) |
| A4 | Does your child point to share interest with you? (e.g., pointing at an interesting sight) |
| A5 | Does your child pretend? (e.g., care for dolls, talk on a toy phone) |
| A6 | Does your child follow where you're looking? |
| A7 | If you or someone else in the family is visibly upset, does your child show signs of wanting to comfort them? (e.g., stroking hair, hugging them) |
| A8 | Would you describe your child's first words as: |
| A9 | Does your child use simple gestures? (e.g., wave goodbye) |
| A10 | Does your child stare at nothing with no apparent purpose? |

way to distribute the contribution of each feature to the model's prediction fairly.

The algorithm considers all possible feature combinations for each prediction using a permutation-based approach. It calculates the difference in the model output when a feature is included versus when it is excluded, representing the marginal contribution of that feature. The Shapley value for a specific feature is then computed by taking the weighted average of these marginal contributions over all possible combinations. The weights are determined by the number of ways each combination can occur.

Mathematically, the equation for SHAP values for a particular feature $i$ in a specific prediction instance involves a summation of all possible subsets of features. This equation ensures that the Shapley values satisfy essential properties such as consistency and linearity.

$$\phi_i(f) = \frac{1}{N} \sum_{S \subseteq N \setminus \{i\}} \frac{|S|! \cdot (|N| - |S| - 1)!}{|N|!} [f(S \cup \{i\}) - f(S)]$$

In this equation, $N$ represents the set of all features, $S$ is a subset excluding feature $i$, and $f(S)$ and $f(S \cup \{i\})$ denote the model output for subsets $S$ and $S$ with feature $i$ included, respectively. The Shapley value for feature $i$ is determined by considering its marginal contribution to all possible subsets, ensuring a comprehensive and fair explanation of the model's predictions. The workflow of both algorithms is shown in figure 1.

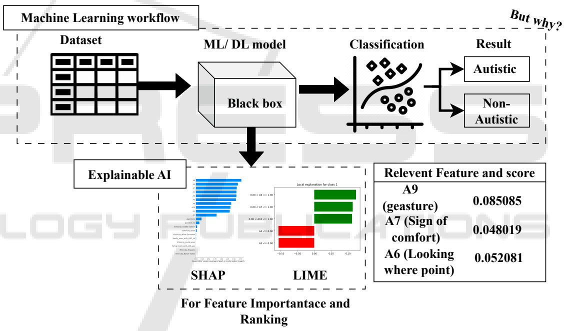


Figure 1: Proposed Methodology for XAI.

## 5 RESULT AND DISCUSSION

We used various machine learning algorithms on the AQ-10 dataset, a questionnaire designed to measure autistic traits. During the development of a model, the data preprocessing stage is carried out on the whole dataset. This stage includes tasks such as handling missing values and scaling features. After this, the dataset is divided into two sets: 80% for the training set and 20% for the testing set. Hyperparameter tuning is then performed. Following this, cross-validation is applied to the training set to assess the model's generalization across different subsets.

The final model is then trained on the entire training set. Its evaluation is carried out on a separate testing set that was not used in the preceding stages. while evaluations are based on metrics such as accuracy, precision, recall, and F1 score. The performance of each model on the AQ-10 dataset is summarized below in the table. 2.

Following the implementation of machine learning algorithms on the dataset, we specifically chose the Random Forest algorithm to interpret the model predictions. The selection was motivated by the ensemble nature of Random Forest, which consists of multiple decision trees. Understanding individual decision trees can be challenging due to their complex structures, prompting our preference for Random Forests as they provide a diverse collection of trees, contributing to a more interpretable and robust model.

Table 2: Autism Screening AQ-10 dataset Accuracy.

| Algorithm | Training Acc | Testing Acc |
|---|---|---|
| Decision Tree | 1.0 | 1.0 |
| SVM | 1.0 | 1.0 |
| Logistic Regression | 1.0 | 1.0 |
| **Random Forest** | **1.0** | **0.995** |
| k-Nearest Neighbors (knn) | 0.962 | 0.947 |
| MLP | 0.93 | 0.92 |

We have chosen two instances to compare. In Instance 1, the class label is 'Yes' or '1', representing the presence of autistic traits. In Instance 2, the class label is 'No' or '0', representing the absence of autistic traits or non-autistic. We applied the random forest algorithm and then used the LIME and SHAP methods to interpret the model's decisions. Below are the results.

## 5.1 LIME: Interpretation

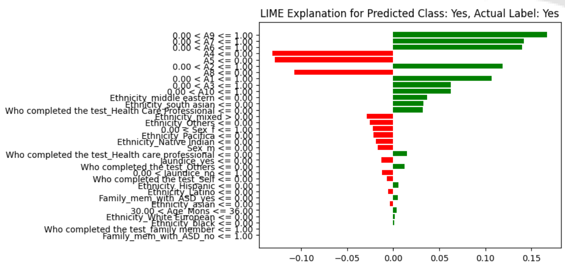


Figure 2: LIME interpretation for Instance 1, where the individual identifies as autistic Class: Yes.

LIME plays a crucial role in making autism classification model predictions more understandable through visualizations that highlight positive and negative associations with specific features. Consider Figure. 2, where the model establishes a positive relation between features A9, A7, and A6 and the likelihood of predicting autism. In this visualization, a green color signifies this positive association. The intensity of the green color indicates that higher levels of these features are linked to an increased probability of autism prediction. Now, consider another scenario, as shown in Figure 3, where features negatively affect autism prediction. In this case, LIME represents the negative correlation with a red color. The increasing intensity of red communicates that as these features increase, there is a decreased likelihood of the model predicting autism. These color-coded visualizations from LIME offer valuable insights into the importance of specific features in predicting outcomes for individual cases. They contribute to a nuanced understanding of how the model makes decisions in autism classification.

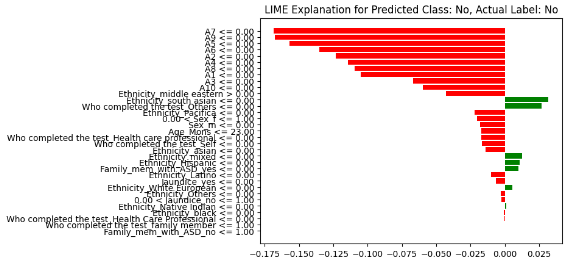


Figure 3: LIME interpretation for Instance 2, where the individual identifies as non-autistic Class: No.

Consider Instance 1, depicted in Figure 4. In this case, the actual label is autistic. The prediction label score is notably high at 0.99 for a "YES" prediction, indicating a 99% probability that the person is autistic. This score is derived from a combination of different features, and the contribution of each feature is visually represented in the figure. Conversely, when the actual label is non-autistic (NO), as illustrated in Figure 5, the prediction score is much lower at 0.071. This implies that there is a mere 0.071% chance that the person could be autistic, leading the model to predict a non-autistic classification. Analyzing such results allows us to interpret the model's predictions and conduct a detailed analysis of the contribution of each feature to the model's decision-making process.

## 5.2 SHAP: Interpretation

SHAP is a global interpretation method that helps understand how a model operates. As shown in Figure 6, the summary plot is an excellent tool for visualizing how each feature contributes to the model's outcome. The plot uses a color-coding system to represent each feature based on its distance from the baseline. Warmer colors are used for features that have a more significant impact on the model's prediction. In contrast, cooler blue colors indicate a negative or lesser impact on the model's outcome. The SHAP summary plot aids users in comprehending the impact of individual features on the model's predictions, enhancing the interpretability of the model by providing

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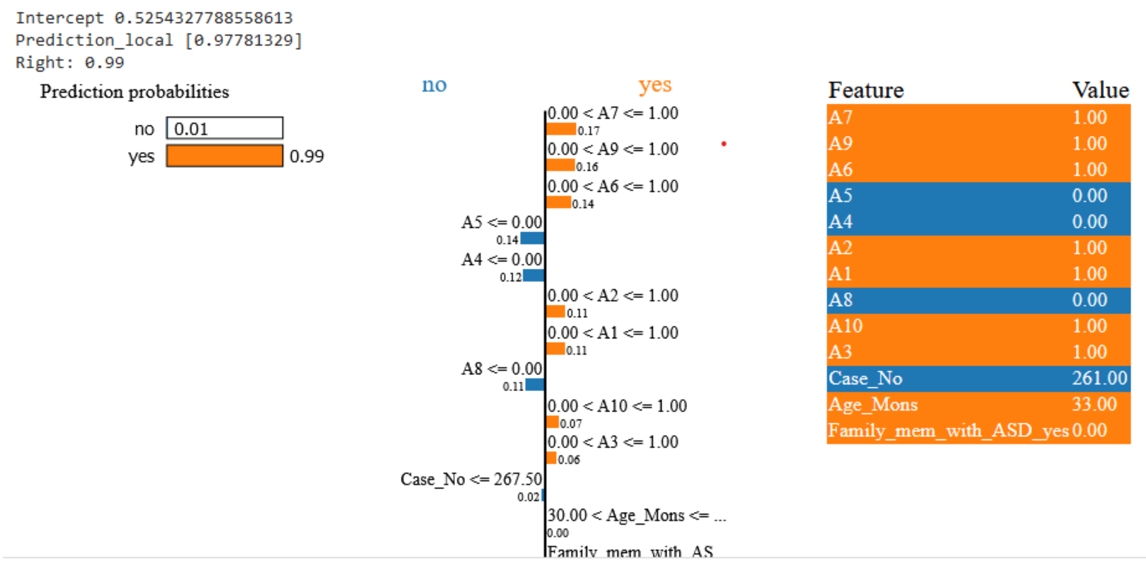


Figure 4: LIME interpretation for autistic class.

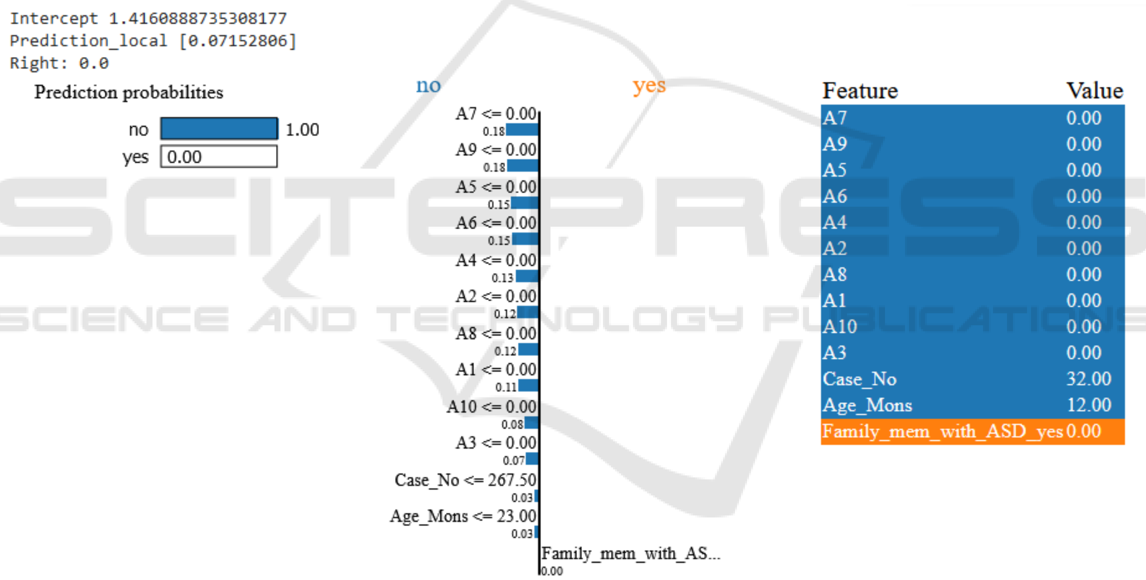


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Figure 5: LIME interpretation for non-autistic class.

insights into the relative importance and directionality of each feature.

In this detailed analysis using SHAP values, we focus on interpreting the predictions of a model for two instances; for instance 1 7, we observe positive SHAP values for attributes A1, A2, A3, A6, A7, A9, and A10. This means that higher values in these attributes positively predict Yes meaning the presence of autistic traits. By this, we can observe that Features such as A9 (with a SHAP value of 0.085085) have a powerful positive impact on the model's affirmative prediction.

On the other hand, for instance, 2, 8, which is labeled as No, we observe negative SHAP values for attributes A1 to A10. This indicates that higher values in these attributes negatively influence the model's prediction of the absence of autistic traits. Features like A9 have a significant impact on predicting the negative outcome. We can clearly understand the interpretation of the SHAP values using the color-coded visualization, where red highlights feature with higher values that strongly impact positive predictions. In comparison, blue indicates features with higher values that have a mitigated impact on

negative predictions. This comprehensive analysis

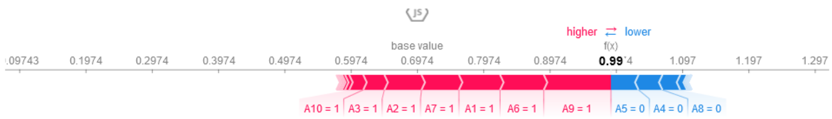


Figure 7: SHAP interpretation for Instance 1, where the individual identifies as autistic Class: Yes.

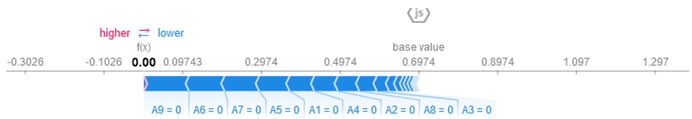


Figure 8: SHAP interpretation for Instance 2, where the individual identifies as non-autistic Class: No.

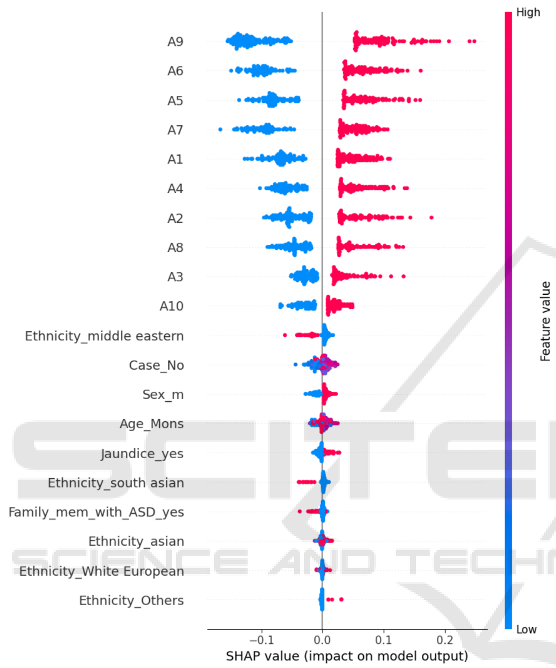


Figure 6: SHAP summary of overall model prediction.

provides a nuanced understanding of the importance of features in individual predictions. It underscores the transparency and interpretability offered by SHAP values in unraveling the intricacies of the model's decision-making process.

Through an examination of LIME and SHAP analyses, it becomes evident that features such as "Does your child use simple gestures? (A9)," "Does your child comfort you? (A7)," and "Does your child follow where you point? (A6)" play a pivotal role in both positive and negative predictions made by the model. Notably, these features exhibit significant prominence in the SHAP values as well. This substantiates the conclusion that gestures, expressions of comfort, and the ability to follow directions are areas where individuals with autism may experience discomfort or challenges. The consistent emphasis on these features in both LIME and SHAP analyses strengthens the inference that they significantly influence the model's predictions. This insight holds valuable implications for tailoring child-oriented therapies, as it underscores the importance of addressing and supporting individuals with autism in the domains of gestural communication, providing comfort, and following cues. Leveraging these findings can contribute to developing more targeted and effective interventions for autistic children, fostering better understanding and support in areas where they may face difficulties.

## 6 CONCLUSION AND FUTURE SCOPE

In our approach to autism classification through various machine learning algorithms, our goal is to achieve consistent accuracy while acknowledging the inherent complexity of these models. We employ Decision Trees, Random Forests, SVM, Logistic Regression, knn, and an MLP. Emphasizing interpretability, we leverage two widely used eXplainable Artificial Intelligence methods—SHAP and LIME. SHAP employs game theory principles to calculate Shapley values, providing insights into how features collectively impact predictions. Meanwhile, LIME offers specific, instance-level explanations. Looking ahead, we plan to enhance explanations by incorporating domain-specific knowledge, exploring advanced visualization techniques, and staying current with emerging interpretability methods in healthcare data. We aim for continuous adaptation, real-time model monitoring, and exploring novel approaches to boost transparency. This includes integrating model-agnostic interpretability methods with domain-specific knowledge and staying informed about state-of-the-art techniques. Through these efforts, we aspire to improve transparency in our autism classification model and contribute to advancing interpretable machine learning in clinical applications.

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