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Towards Refined Autism Screening: A Fuzzy Logic Approach with a Focus on Subtle Diagnostic Challenges

Philip Smith * and Sarah Greenfield

School of Computer Science and Informatics, De Montfort Univeristy, Gateway House, Leicester LE1 9BH, UK; s.greenfield@dmu.ac.uk
* Correspondence: p2585980@my365.dmu.ac.uk

Abstract: This study explores the creation and testing of a Fuzzy Inferencing System for automating preliminary referrals for autism diagnosis, utilizing membership functions aligned with the Autism Quotient 10-item questionnaire. Validated across three distinct datasets, the system demonstrated perfect accuracy in deterministic settings and an overall accuracy of 92.91% in a broad fuzzy dataset. The use of Fuzzy Logic reflects the complex and variable nature of autism diagnosis, suggesting its potential applicability in this field. While the system effectively categorized clear referral and non-referral scenarios, it faced challenges in accurately identifying cases requiring a second opinion. These results indicate the need for further refinement to enhance the efficiency and accuracy of preliminary autism screenings, pointing to future avenues for improving the system's performance. The motivation behind this study is to address the diagnostic gap for high-functioning adults whose symptoms present in a more neurotypical manner. Many current deep learning approaches for diagnosing autism focus on quantitative datasets like fMRI and facial expressions, often overlooking behavioral traits. However, autism diagnosis still heavily relies on long histories and multi-stakeholder information from parents, teachers, doctors and behavioral experts. This research addresses the challenge of creating an automated system that can handle the nuances and variability inherent in ASD symptoms. The theoretical innovation lies in the novel application of Fuzzy Logic to interpret these subtle diagnostic indicators, providing a more systematic approach compared to traditional methods. By bridging the gap between subjective clinical evaluations and objective computational techniques, this study aims to enhance the preliminary screening process for ASD.

Keywords: autism; autism spectrum disorder; autism diagnosis; Fuzzy Logic; Fuzzy Inferencing System

MSC: 03B52

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1. Introduction

Autism spectrum disorder (ASD) is a lifelong neurological condition that affects at least 1% of the population [1,2]. It is characterized by deficits in social communication and social interaction, restricted, repetitive patterns of behavior, interests or activities, and sensory deficits [1]. ASD diagnosis involves a comprehensive approach including behavioral observation, developmental history and sometimes cognitive and language assessments conducted by teams of clinical professionals along with input from relevant stakeholders such as parents, teachers and advocates. The diagnostic process can be lengthy and costly, with complexities varying widely across the autism spectrum. Many researchers have tried to develop short self-administered tests to aid in making accurate suggestions for full referral by comprehensive diagnostic teams. The AQ-10 (Autism Quotient) is such a tool. It is a subset of 10 items drawn from the full 50 item Autism Quotient (AQ) questionnaire [3].

Individuals with less apparent autistic symptoms, often referred to as “high-functioning”, can present unique diagnostic challenges. Those on the higher end of the spectrum typically present with more subtle symptoms and may have normal IQ levels and have typical communicative abilities. Higher functioning individuals form the majority of those on the autism spectrum. Due to the subtlety of symptoms in some individuals, particularly those with typical IQ levels and communicative abilities, obtaining a diagnosis or referral for a full assessment can be challenging [4–6]. Current research in Autism Spectrum Disorder (ASD) is extensively exploring the potential of biomarkers, brain imaging and other quantitative assessments to provide more objective diagnostic criteria [7]. Despite these advances, the diagnosis of ASD still predominantly relies on subjective evaluations based on behavioral observations and interviews, which inherently involve a degree of uncertainty due to their subjective nature [1,4].

The extensive applications of deep learning in ASD are acknowledged. However, this study’s primary innovation lies in its focus on explainability and alignment with clinical tools like the AQ-10. Deep learning models, despite their accuracy, often lack transparency, making them less suitable for clinical settings where decision-making processes need to be clear. The FIS developed in this research addresses the nuanced behavioral traits of autism and manages the uncertainty in diagnosing higher-functioning adults, prioritizing late diagnosis in adults. By using a rule-based approach, which can be easily understood by clinicians and stakeholders, this system enhances trust and usability. Addressing a significant gap, the study focuses on high-functioning individuals and adults at risk of missed diagnoses. Moreover, the system accurately models the AQ-10, enhancing clinical validity and facilitating greater acceptance among clinicians by mirroring familiar diagnostic tools. In this context, the integration of Fuzzy Logic into the diagnostic process presents a promising avenue. Fuzzy Logic, with its ability to handle imprecision and variability inherent in human judgments, offers a novel approach to interpreting the nuanced and often ambiguous symptoms of ASD. This method could potentially bridge the gap between the subjective nature of current diagnostic practices and the objective clarity sought through quantitative research. To this end, a Fuzzy Inferencing System (FIS) was implemented and extensively tested for making referral decisions for full autism diagnosis. Its primary functionality enables diagnosis in line with AQ-10 criteria, providing a systematic and reliable approach that has already been proven to be clinically valid [3]. Additionally, informed by our research and suggestions, the system’s second functionality offers second opinion decisions for borderline cases, again utilizing AQ-10 criteria but with a new interpretation of the data. The system was rigorously tested across its full input range to evaluate its Fuzzy Logic capabilities. This comprehensive testing is crucial, as the first two functions operate under a more deterministic framework. Our approach ensures the FIS is not only accurate accurate according to a widely used clinical tool but also adept at handling the subtleties and complexities of borderline situations. Lastly, validation tests were conducted.

Research Framework Overview

- **Objective**: The primary objective of our research is to develop a FIS that can automate preliminary referrals for autism diagnosis using the Autism Quotient (AQ-10) questionnaire.
- **Rationale**: Autism diagnosis is complex and often involves subjective evaluations. Our framework aims to address this by incorporating Fuzzy Logic to handle the inherent variability and imprecision in human judgments, thus providing a more systematic and reliable approach to preliminary screenings.
- **Framework Components**:
  - *Data Preparation*: Detailed in Section 6, this step involves creating comprehensive datasets from the AQ-10 questionnaire responses, ensuring that all potential input combinations are accounted for.

– *Fuzzy Inferencing System Design*: A Mamdani-type FIS has been developed with membership functions corresponding to the AQ-10 traits. This design is explicitly aimed at replicating and enhancing the decision-making process of the AQ-10, as outlined in Section 7.
– *Testing and Validation*: The system was rigorously tested across three datasets: the AQ-10 dataset, an expanded AQ-10+ dataset and a fuzzy dataset with continuous input values. This phased approach, detailed in Section 8, ensures that the system performs accurately under various conditions. Furthermmore, 100 validation datasets were generated to further enhance and understand the systems reliability.

An outline of the entire research process is provided in Figure 1.

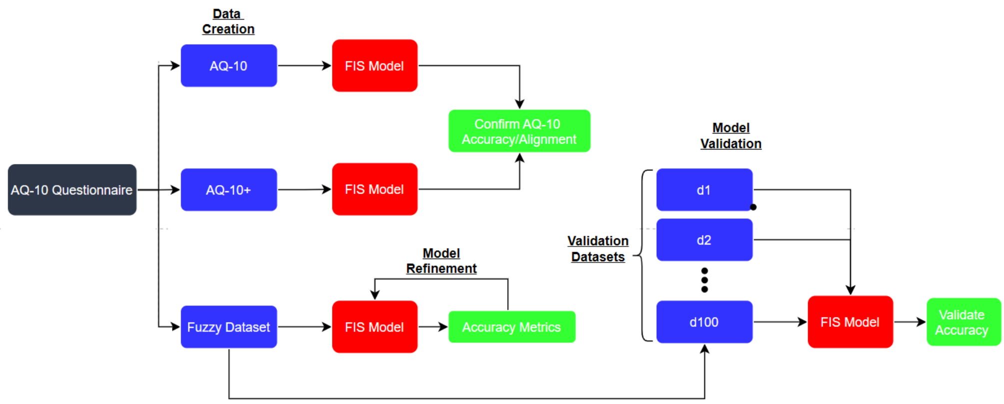


Figure 1. Research flow diagram.

The structure of this paper is outlined as follows: Section 2 gives a concise overview of Fuzzy Logic. Section 3 provides the contextual background, emphasizing the challenges in diagnosing high-functioning adults with ASD and the motivation for using AI and Fuzzy Logic. Section 4 reviews the existing literature, demonstrating how previous research aligns with and informs the current study. Section 5 presents a Fuzzy Logic comparison analysis, comparing the model presented in this paper with those outlined in the literature review. Section 6 details the preparation of the dataset, describing how it was expanded for three different phases of testing the fuzzy system. Section 7 explains the methodology employed in developing the system, including the specific steps and procedures followed. Section 8 reports the findings of the study, showcasing the results obtained from applying the chosen methodology to the prepared datasets, along with validation results. Section 9 discusses these results, offering interpretations of their importance and the impact they may have on the field. Section 10 concludes the paper by summarizing the main findings and outlining the limitations of the work. Finally, Section 11 provides future recommendations.

2. Fuzzy Logic

Fuzzy Logic is a form of many-valued logic that deals with approximate rather than exact reasoning. Unlike traditional binary sets (where variables may take on true or false values) Fuzzy Logic variables may have a truth value that ranges in degree between 0 and 1. This makes it particularly suitable for handling the concept of partial truth, where the truth value may range between completely true and completely false [8].

2.1. Basic Concepts of Fuzzy Logic

2.1.1. Fuzzy Sets

In classical set theory, an element either belongs to a set or does not. In contrast, fuzzy set theory allows for partial membership. A fuzzy set A in a universe of discourse X is characterized by a membership function $\mu_A : X \to [0,1]$ that assigns to each element $x \in X$ a degree of membership in the set A [8].

2.1.2. Membership Functions

The membership function is a crucial component of Fuzzy Logic. It quantifies the degree of truth. Common shapes of membership functions include triangular, trapezoidal and Gaussian. For example, a triangular membership function is defined by three parameters (a, b, c) as follows:

$$\mu_A(x) = \begin{cases} 0 & x \leq a \\ \frac{x-a}{b-a} & a < x \leq b \\ \frac{c-x}{c-b} & b < x \leq c \\ 0 & x > c \end{cases}$$

2.1.3. Fuzzy Operations

Fuzzy Logic extends classical Boolean operations to fuzzy sets. The primary operations include:

- **Fuzzy Complement**: $\mu_{\neg A}(x) = 1 - \mu_A(x)$
- **Fuzzy Union**: $\mu_{A \cup B}(x) = \max(\mu_A(x), \mu_B(x))$
- **Fuzzy Intersection**: $\mu_{A \cap B}(x) = \min(\mu_A(x), \mu_B(x))$

2.2. Fuzzy Inference Systems

Fuzzy inference systems use fuzzy set theory to map inputs to outputs. The two most common types are Mamdani (Figure 2) and Sugeno models. A brief explanation of the Mamdani system is presented as it is the one used in this work. A Mamdani FIS is composed of four steps:

1. **Fuzzification**: Convert crisp inputs into fuzzy sets.
2. **Rule Evaluation**: Apply fuzzy operators to the antecedents of the rules.
3. **Aggregation of the Rule Outputs**: Combine the outputs of all rules.
4. **Defuzzification**: Convert the aggregated fuzzy output back into a crisp value.

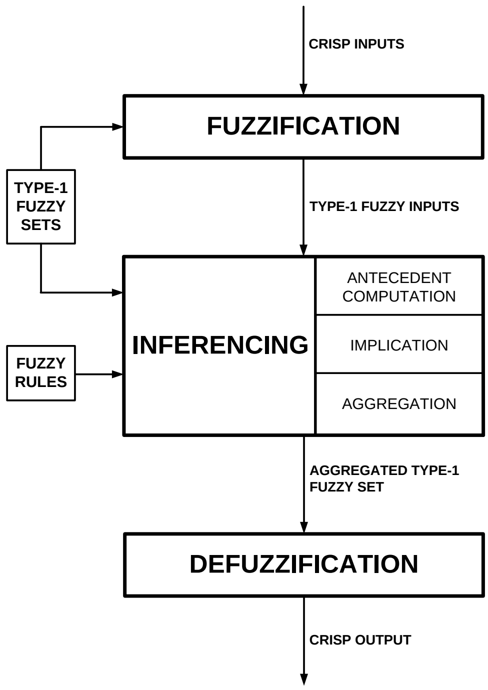


Figure 2. Type-1 Mamdani FIS [9].

2.3. Applications of Fuzzy Logic

Fuzzy Logic has been applied in various fields, including:

- **Control Systems**: Such as in washing machines, cameras and air conditioning systems.

- **Decision Making**: For handling uncertain or imprecise information. See recent usage of Fuzzy Logic in analysing surface regions of Mars for optimal location to land Martian Rovers [10].
- **Pattern Recognition**: For classifying complex patterns in data.

3. Background

This section establishes the pressing need for improved diagnostic tools for adults seeking diagnosis and introduces Fuzzy Logic as a promising solution. The background sets the stage for understanding why traditional diagnostic methods could benefit from an automated aide for this group and the potential benefits of integrating AI-driven approaches. The focus of this research has been placed on high-functioning adults, as these cases are less likely to be detected due to the subtler presentation of their symptoms [6,11]. There exists a greater degree of uncertainty at the higher end of the autism spectrum, which underscores the investigation of Fuzzy Logic as a potential aid in suggesting a diagnosis [12]. There is a clear and urgent need for the healthcare sector to improve the diagnostic process for ASD. Specifically, in the United Kingdom, it has been estimated that the average length of the diagnosis process stretches to five years for adults [13]. This extended duration is attributed to a range of factors, including skepticism towards individuals' symptoms, the logistical challenges of coordinating among multiple stakeholders and the comprehensive data collection required—from self-assessments and contributions from parents and teachers to expert evaluations by healthcare professionals and specialists in autism diagnosis—rendering the process both time-consuming and expensive [1,5,13].

Clinical experience in routine diagnostics often faces the challenge of aligning general guidelines with each unique case. Diagnosticians typically adopt a pragmatic approach due to "institutionally-induced competing demands", requiring thoughtful navigation of these ambiguities [14]. For example, a study of psychiatric diagnoses showed clinicians use "psychiatric workarounds", like negotiating diagnoses with patients or adjusting codes on paperwork, to balance institutional pressures with personalized care [14]. Autism diagnosis exemplifies this tension, as the spectrum disorder blurs into the general population, making precise diagnostic thresholds ambiguous. This uncertainty persists even post-diagnosis in determining the necessary support levels. It is especially pronounced for those on the diagnostic threshold, particularly high-functioning individuals or those seeking diagnosis later in life [1,4]. Rather than eliminating uncertainty, research suggests embracing it as an integral feature of autism diagnosis, acknowledging the challenges of deriving a binary outcome from such a diverse condition [14].

Given these complexities, the adoption of Artificial Intelligence (AI) in the diagnostic procedure presents a promising avenue. This initiative draws inspiration from AI's transformative impact on diagnostic accuracy and efficiency within other medical sectors, such as oncology, radiology and neurology. The exploration of AI to expedite the ASD diagnostic framework, thereby reducing the timeline and associated costs, is imperative. Such advancements align with the broader healthcare trend of leveraging technological innovations to enhance patient care outcomes. The work presented in this paper extends efforts previously detailed by the authors in [6], wherein the efficacy of Fuzzy Logic models for making referrals for patients with high-functioning autism spectrum disorder (ASD) was explored. These initial investigations established the appropriateness of Fuzzy Logic in the ASD referral and diagnostic processes [6]. Despite making strides in elucidating the potential of Fuzzy Logic, the authors' findings emphasized the necessity for a more comprehensive exploration of its application across a broader spectrum of ASD cases and with the utilization of larger datasets. To address this gap, the present paper expands the scope of the original study by incorporating an expanded dataset and developing a new fuzzy system, modeled on the well-established diagnostic tool, the AQ-10 questionnaire.

4. Literature Review

This section delves into existing research on Fuzzy Logic applications in autism diagnosis, providing an overview of previous studies and their outcomes. It also examines related Fuzzy Logic methods and cutting-edge deep learning approaches. The literature review supports the rationale introduced in Section 3 by detailing the current state of research, highlighting gaps and justifying the focus on Fuzzy Logic to address identified challenges. It integrates findings from various studies, demonstrating how Fuzzy Logic has been applied in similar contexts and the potential it holds for improving diagnostic accuracy and efficiency. Additionally, the review places our work into context by highlighting the research gap, particularly in terms of high-functioning adults and the lack of focus on behavioral traits that indicate autism.

Background research indicates that the utilization of Fuzzy Logic to aid in the screening or diagnosis of autism is sparse, yet some research has been identified that attempts to achieve similar goals. This review clarifies the scarcity of studies that examine behavioral characteristics through computational intelligence techniques for referral or diagnostic purposes. The background research identified several studies focusing on the uncertain and vague classification of behavior, as opposed to biomarkers such as vitamin D, serotonin and oxytocin levels, along with brain scans [15]. An overview of other related techniques is provided to illustrate how related technology involving Fuzzy Logic is being used not only for diagnosis but also for the management and treatment of ASD.

In the domain of autism diagnosis, it is paramount to recognize that traditional diagnostic practices predominantly rely on comprehensive, multistakeholder behavioral evaluations. Through the examination of various studies comparing diagnostic accuracies, a nuanced understanding becomes essential. Differences in accuracy should not be interpreted merely as evidence of one system's superiority over another but as a reflection of the divergent diagnostic approaches being employed. While some research, like that involving MRI scans, explores the potential of biomedical markers, and others focus on behavioral assessments, each approach offers unique and critical insights into ASD.

4.1. Fuzzy Logic Approaches

In [16], the researchers employed a fuzzy system to diagnose autism, utilizing a dataset from the UCI Machine Learning Repository. This dataset, however, was not the original but rather derived from the AQ-10 questionnaire. The paper did not specify the formation process of this dataset, which was generated via a mobile application for autism screening that implemented the AQ-10 method [16]. Despite the dataset's derivation from the AQ-10, their system was not compared to other established diagnostic methods, including the AQ-10 itself.

In [17], the authors developed a type-1 Fuzzy Logic expert system to diagnose autism among 96 patients. This system aimed to improve diagnostic accuracy for ASD by leveraging Fuzzy Logic. The results demonstrated a precision rate of 94.4%, a recall precision of 65.4% and an error rate of 3.05%. Diagnostic accuracy, when comparing outcomes from the expert evaluations with those obtained using the fuzzy method, was reported at 90.59% [17]. These findings indicate the system's potential utility in clinical settings by offering quantifiable metrics on precision, recall precision, error rate and comparative diagnostic accuracy, suggesting its effectiveness in diagnosing ASD.

In [18], a Fuzzy Logic-based system was designed to assess the severity of autism in a dataset comprising 100 children aged 2 to 3 years. Utilizing 15 indicators identified by health professionals, the system integrated these into a Fuzzy Logic model to classify autism severity into categories such as *normal, mild, moderate and severe*. The research methodology included the collection of real-world data from these 100 children. In the initial phase of the study, discussions were facilitated with healthcare professionals to gather their insights and expertise. This structured approach aimed at collecting critical information on factors relevant to evaluating autism severity in children. A Takagi–Sugeno–Kang (TSK) type first-order fuzzy inference mechanism was deployed in the decision-making process [18]. Furthermore, the study engaged in a comparative analysis with other soft computing methods and traditional assessment techniques. The results revealed that the fuzzy inference system effectively categorizes children into specific autism severity levels. This classification system aims to support clinicians, especially those at the beginning of their careers or those who might face challenges in determining the autism severity level in children, by offering a structured approach to evaluation [18].

In [19], researchers utilized a neural network classifier and Fuzzy Synchronization Likelihood (FSL) to analyze brain regions from EEG scans to determine the presence of autism. Features selected for the Neural Network classifier were extracted from EEG signals after being subjected to wavelet decomposition, which broke them down into subbands. The calculation of FSL was then performed to measure synchronization likelihood between and within brain regions. The EEG signals were obtained from eyes-closed EEG recordings of two groups: 9 children with autism and 9 without autism (diagnoses were confirmed by behavioral observation from healthcare professionals based on DSM 4 criteria). Despite the small sample size, the classification accuracy of this method is reported to be 95.5% [19].

In the 2019 study by [20], two methods, the FIS and the Adaptive Neuro-Fuzzy Inference System (ANFIS), were applied to determine the severity levels of autism spectrum disorder (ASD) in children [20]. The ANFIS method is characterized by its reduced dependence on expert knowledge compared to traditional FIS. This reduction is considered beneficial in contexts where expert input is limited or where a system that can adapt based on incoming data is preferred. ANFIS's ability to autonomously adjust its reasoning patterns could potentially make the severity assessment process more adaptable and less subjective [20]. The dataset for this study comprised information from 98 children with ASD, with data points collected from questionnaires filled out by their teachers. The findings showed that the FIS method achieved an accuracy of 80%, and the ANFIS method achieved an accuracy of 82% [20]. These results indicate that both methods can be used to estimate the severity of ASD, with ANFIS showing a marginal improvement in accuracy, which could be attributed to its adaptive capabilities and reduced reliance on predefined expert rules.

In [21], an Interpretable Fuzzy Neural Network (IFNN) was developed focusing on enhancing interpretability, aiming to make the model's decision-making process more transparent. This emphasis on interpretability is highlighted as a key aspect that could increase the model's trustworthiness [21]. The IFNN was benchmarked against several comparison models, including k-Nearest Neighbors (k-NN), fuzzy k-Nearest Neighbors (fuzzy k-NN) and traditional neural networks. The study reported that the IFNN achieved an overall accuracy of 91% [21].

In [22], a system named FHGO-DNFN was developed for diagnosing ASD and for feature extraction from brain images. The system employs type-2 Fuzzy Logic for the preprocessing of brain images to remove noise [22]. The preprocessing phase includes Region Of Interest (ROI) extraction, achieved through functional connectivity and the box neighborhood search algorithm. This step focuses on isolating pertinent areas within the brain images for subsequent analysis. The FHGO-DNFN integrates a Deep Neuro-Fuzzy Inference System (DNFN), optimized using a technique named FHGO (Feedback-Henry Gas Optimization). This technique combines the principles of Henry's law regarding gas solubility with a feedback mechanism to enhance the optimization of the DNFN's parameters. According to the study, the FHGO-DNFN system demonstrated an accuracy of 93.3%, a sensitivity of 94.7% and a specificity of 91.4% in classifying ASD from healthy control brain images [22].

In the 2017 study by [23], a system combining a robot and a tablet application was introduced, targeting individuals with ASD. This system utilizes type 1 and type 2 Fuzzy Logic to facilitate and complement social interactions. The robot component is designed to engage in social interactions, while the tablet application provides tasks aimed at enhancing the recognition of facial expressions, a common challenge for individuals with ASD [23]. Fuzzy Logic is employed to enable the robot to handle the variability and uncertainties inherent in interactions with users. Similarly, the tablet application uses Fuzzy Logic to adjust the difficulty of tasks based on the user's previous performance. These adjustments involve membership functions that correlate with the time required to identify facial expressions and modify task difficulty according to the percentage of correct responses on previous tasks [23]. The system is presented as a support and intervention tool for ASD therapy, focusing on social interaction and emotional recognition. The application of Fuzzy Logic aims to adapt the system to the user's interaction style and learning pace. However, the effectiveness of this tool in therapeutic settings is contingent upon further evaluation and practical application [23].

4.2. Deep Learning Approaches

The deep learning approach described by [24] employs Convolutional Neural Networks (CNNs) to analyze facial features for the early detection of ASD. Their study utilizes pre-trained models, including VGG16, VGG19 and EfficientNetB0, achieving accuracies of 84.66%, 80.05% and 87.9%, respectively. Unlike our Fuzzy Logic methodology, which focuses on behavioral traits derived from the Autism Quotient (AQ-10) questionnaire, this deep learning approach leverages physiological markers from facial images.

The study by [25]. employs eye-tracking technology combined with advanced deep learning models, such as CNN, LSTM, GRU and BiLSTM, to detect ASD. This approach analyses gaze patterns and attentional mechanisms, achieving high accuracy rates, with the LSTM model reaching up to 98.33%.

The study by [26]. presents a novel framework for ASD detection that combines Recursive Feature Elimination (RFE) with Graph Neural Networks (GNN) and a Phenotypic Feature Extractor (PFE) to process functional magnetic resonance imaging (fMRI) and phenotypic data. This method achieved accuracies of 78.7% and 80.6% on the ABIDE I and II datasets, respectively. The RFE-GNN approach leverages both imaging and phenotypic data to capture more complex patterns associated with ASD.

The study presented in [27] explores the use of deep learning, specifically deep Convolutional Neural Networks (CNN), to identify diagnostic biomarkers through the facial expressions of children for the early detection of autism. The dataset utilized in this research was sourced from Kaggle. However, significant concerns arise regarding the dataset's integrity, as the children were clinically diagnosed based on typical behavior traits, and the dataset creator admitted to gathering images through internet searches, raising doubts about the authenticity of the 1327 images as solely depicting autistic children. While the application of a quantitative deep model for this purpose is promising, the model's lack of explainability and the questionable source of the dataset's images highlight critical limitations in the study's methodology and reliability.

The study by [28]. employs functional brain networks and machine learning algorithms, specifically focusing on metrics such as Pearson Correlation (PC), Spearman Correlation (SC) and Granger Causality (GC) to differentiate ASD from typical development (TD) using fMRI data. This method achieved a high AUC, with SC emerging as the most effective metric, providing a robust framework for capturing brain changes due to ASD. The functional brain network method leverages advanced neuroimaging data to identify structural and functional anomalies in the brain.

Ref. [29] investigates the use of Graph Neural Networks (GNNs) for identifying Autism Spectrum Disorder (ASD) through MRI data, emphasizing the need for integrating both node and edge features in the graph classification tasks. The proposed model, ALNEGAT, employs adversarial learning techniques to enhance the generalizability of the model across different datasets.

Ref. [30] outlines a data-centric approach using deep learning for diagnosing Autism Spectrum Disorder (ASD) from facial images. Utilizing CNNs and extensive data preprocessing and augmentation, the method achieves a 98.9% accuracy rate. Explainable AI is employed to enhance the interpretability of the diagnostic process, which is beneficial for clinical use. This approach demonstrates significant advancements over previous models in the early and accurate diagnosis of ASD.

Ref. [31] details a study on the use of deep learning for diagnosing ASD. The researchers utilized a CNN to classify brain scans from the ABIDE dataset. The dataset comprised 873 participants, both with and without ASD, ranging in age from 6 to 64 years. The proposed model achieved a classification accuracy of 93.41% and a mean absolute error (MAE) of 0.29.

Ref. [32] explores the use of deep learning to diagnose ASD through multiple classifications considering age and gender factors. Utilizing brain structural MRI (sMRI) scans from the ABIDE dataset, the study applied Canny Edge Detection (CED) for data preprocessing and data-augmentation techniques to expand the dataset. Three convolutional neural network (CNN) models were developed: one for gender-based classification, another for age-based classification and a third for combined age and gender classification. The models achieved accuracy rates of 80.94%, 85.42% and 67.94%, respectively, outperforming several pre-trained models. This research highlights the significant impact of age and gender on the diagnosis of ASD and demonstrates the potential of deep learning in enhancing diagnostic accuracy.

In [33], an autism diagnostic system integrating a Recurrent Neural Network (RNN) and Fuzzy Logic applied to MRI brain scans was developed and assessed on a dataset consisting of 539 ASD patients and 573 controls. The system's performance was evaluated against conventional methodologies, such as RNN, Convolutional Neural Networks (CNN) and Deep Belief Networks (DBN), demonstrating superior performance in terms of diagnostic accuracy. The results indicated an accuracy of 98.21%, precision of 97.56%, recall of 96.74% and an F1 Score of 96.82% [33]. However, when comparing these findings to the work of Wanti et al. (2022) [17], which employed a type-1 Fuzzy Logic expert system based on behavioral traits for diagnosing autism and reported an accuracy of 90.59%, it is crucial to consider the fundamental differences in the diagnostic bases of these two approaches. The method by Nair et al. utilizes MRI brain scans, offering a biomedical diagnostic perspective, whereas the system by Wanti et al. relies on behavioral assessments, aligning with the traditional method of autism diagnosis [33].

Reflecting on the studies reviewed, it becomes clear that the integration of biomarkers and genetic analysis into ASD diagnosis is still in its early stages, particularly when compared to the well-established, interdisciplinary behavioral diagnostic methods involving medical professionals, psychologists and educators, among others [1]. Such an approach promotes a balanced interpretation of research outcomes, recognizing the dynamic interplay between conventional behavioral techniques and the nascent domain of biomedical investigations in the evolving landscape of ASD diagnosis. It is noted that all studies examining biomarkers are only looking at potential combinations of biomarkers that may suggest autism. Given this uncertainty, It is acknowledged that studies of this type are an appropriate subject for Fuzzy Logic research. However, the decision was made not to look at biomarkers, given that all autism diagnoses will require behavioral insight by healthcare professionals; no positive diagnosis comes from biomarkers alone. Thus, the focus is on developing a fuzzy system constructed of membership functions that represent autism behavior traits. This literature review has confirmed that this area is still understudied.

5. Fuzzy Logic Comparison Analysis

The current landscape of ASD diagnostics research is characterized by various machine learning and deep learning approaches leveraging neuroimaging data, such as fMRI and electroencephalogram (EEG) signals. These methods focus on capturing physiological and functional brain differences between ASD and neurotypical individuals. However, a gap remains in addressing the nuanced and uncertain behavioral traits essential for a comprehensive understanding and diagnosis of ASD, particularly in high-functioning adults who are at a greater risk of not receiving timely diagnoses. Most studies do not consider these behavioral aspects, potentially limiting their clinical applicability and acceptance. The

Fuzzy Logic methodology introduced in this study is designed to model the Autism AQ-10 questionnaire, which is widely recognized and trusted in clinical settings. This approach emphasizes interpretability and flexibility, allowing for the incorporation of expert knowledge and adjustments based on clinical feedback. Unlike deep learning models that operate as "black boxes", the Fuzzy Logic system provides transparent decision-making processes through defined rules and membership functions. This transparency may aid in clinical acceptance as it aligns with the AQ-10 screening tool and accounts for the ambiguous and nuanced symptoms of ASD, particularly in high-functioning individuals.

Furthermore, one of the critical concerns with deep learning models is their lack of explainability. Non-machine learning diagnostic experts and stakeholders in ASD diagnosis may struggle to trust systems that are complex and opaque, despite their high accuracies [34,35]. Additionally, ASD diagnosis is not approached through a binary lens; the diagnosis itself captures the inherent uncertainty and the spectrum of autism behaviors [14]. The Fuzzy Logic approach addresses these issues by providing a more interpretable model. Its rule-based structure, which is designed in a human-friendly way, ensures that it aligns with AQ-10 and brings a sense of familiarity to clinicians and stakeholders in the ASD diagnostic process. A recurring theme across various studies is the reliance on the ABIDE dataset for training and validating machine learning models. This dataset, while extensive, presents limitations in terms of demographic representation and balanced data distribution, which can affect the generalizability of the findings. Studies employing deep learning techniques, such as convolutional neural networks (CNNs) and recurrent neural networks (RNNs), demonstrate high accuracy but at the cost of significant computational resources and technical expertise. These approaches often focus on neuroimaging data without integrating behavioral assessments, thus overlooking a critical aspect of ASD diagnostics. Other deep learning studies have employed facial expression analysis for early detection of ASD, utilizing deep CNNs trained on datasets sourced from platforms like Kaggle. These datasets, however, have raised concerns about the reliability and authenticity of the images, as they often rely on images gathered through internet searches without verification. As one dataset creator used in some of the studies outlined in the literature review noted, *"I searched for databases for images of autistic children and found none. Consequently, I had to gather the images via internet searches. In the end, I could only find 1327 images and I am to some extent concerned about them being all of Autistic children"* [36]. Most of the reviewed studies do not explicitly differentiate between high-functioning and low-functioning individuals with ASD. Instead, they predominantly focus on the general ASD population without considering the severity of symptoms or level of functioning. Although these studies are valuable for early detection in children, there is a critical oversight. This study addresses the need for assessing high-functioning adults, who may exhibit subtler symptoms that require nuanced behavioral evaluations rather than relying solely on physiological metrics. The Fuzzy Logic approach, by modelling the AQ-10 adult questionnaire, specifically targets this gap by providing a tool that is sensitive to the behavioral manifestations of ASD across the spectrum.

6. Dataset Preparation

Three separate datasets were created to test the system. The first dataset is designed to assure that the system will output correct decisions according to AQ-10 criteria. The original AQ-10 dataset is bimodal with 70% of the data clustering into a no referral group, with the remaining 30% of the dataset grouped into the referral group (Figure 3).

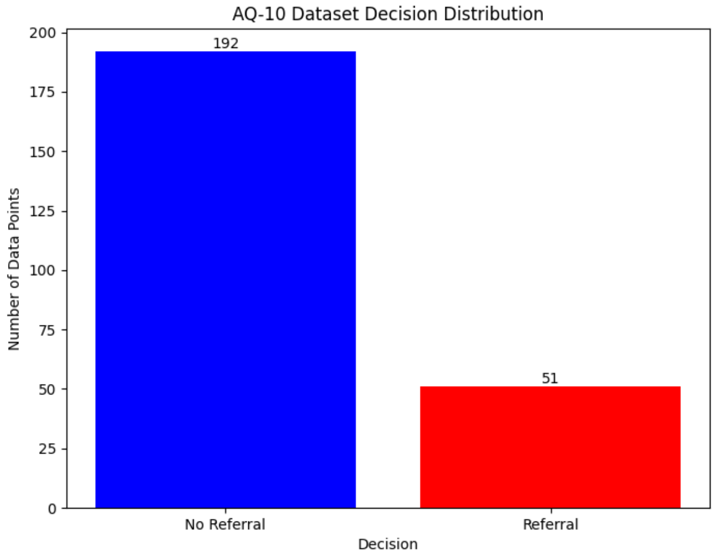


Figure 3. AQ-10 decision distribution.

6.1. AQ-10 Dataset

There are 1024 different combinations that a user can input into the AQ-10 questionnaire, which consists of 10 items, each of which can be answered with a 0 or 1, signifying disagreement or agreement with the statement, respectively. A method was developed to map all potential responses to the AQ-10 into a comprehensive dataset using combinatorics principles. See Table 1 for a description of these 10 items along with their associated autism trait categories.

Table 1. AQ-10 categories and items [3].

AQ-10 Categories	Items
Attention to Detail	1. I often notice small sounds when others do not. 2. I usually concentrate more on the whole picture, rather than the small details.
Attention Switching	1. I find it easy to do more than one thing at once. 2. If there is an interruption, I can switch back to what I was doing very quickly.
Communication	1. I find it easy to 'read between the lines' when someone is talking to me. 2. I know how to tell if someone listening to me is getting bored.
Imagination	1. When I'm reading a story I find it difficult to work out the characters intention. 2. I like to collect information about categories of things (e.g.types of car, types of bird,types of train, types of plant, etc.).
Social	1. I find it easy to work out what someone is thinking or feeling just by looking at their face. 2. I find it difficult to work out people's intentions.

To ensure uniformity in recommendations, these 1024 binary combinations from the AQ-10 were transformed into a fuzzy input dataset. Consequently, this guarantees that any individual completing the AQ-10 will receive a consistent recommendation from the fuzzy system. To create the inputs to the fuzzy system, the two items for each autism trait in the AQ-10 were aggregated in the following manner:

Given an AQ-10 input of the form: $A = [a_1, a_2, a_3, a_4, a_5, a_6, a_7, a_8, a_9, a_{10}]$ where $a_i \in \{0, 1\}$,

the aggregated input is calculated as:

$$B = [a_1 + a_2\,,\ a_3 + a_4\,,\ a_5 + a_6\,,\ a_7 + a_8\,,\ a_9 + a_{10}].$$

After deleting the duplicates the AQ-10 dataset contains 243 input records. Applying a threshold where the total sum exceeds 6, the resulting dataset was concatenated with a single column of the output decisions (Figure 4). Let x_i represent the ith element of the input array, where $i = 1, 2, 3, 4, 5$, and each x_i is a feature mapped to its associated membership function. The five membership functions correspond to the AQ-10 autism trait categories (Section 7).

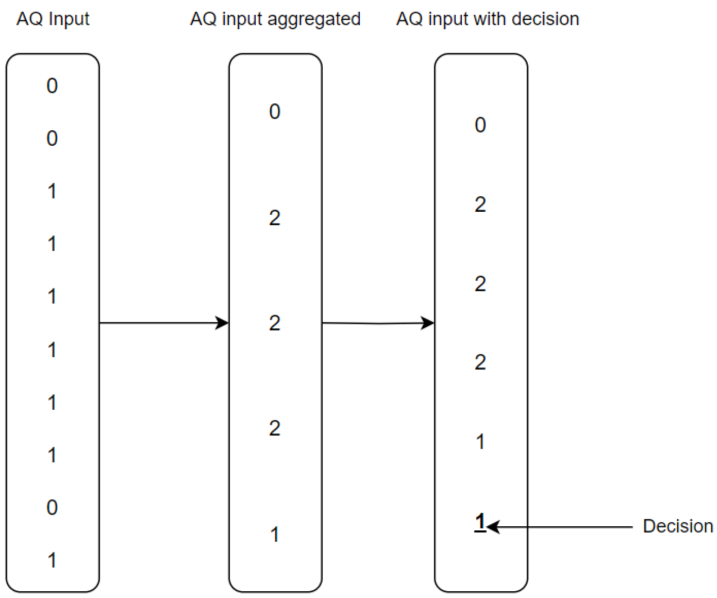


Figure 4. AQ input aggregation flowchart.

6.2. AQ-10+ Dataset

The second dataset is used to assess those who, in the original AQ-10 test, score a total of 6 and are not suggested for referral. These individuals will be taken and the FIS will suggest a second opinion decision instead of no referral. This dataset is identical to the AQ-10 dataset with 243 data points, but it simply has an additional decision variable appended to the data point should the sum of the feature variables add up to 6. Those data points that sum to 6 are appended with the value 0.5 to indicate a second opinion decision.

6.3. Fuzzy Dataset

The fuzzy dataset is designed to evaluate an expanded input range for the input membership functions and for the output range of the FIS. Until now, in alignment with the well-established referral tool AQ-10, the system has exhibited a largely deterministic nature. The inputs to the system have been confined to discrete points on the x-axis, specifically 0, 1 or 2. Similarly, the output range produced the same defuzzified values, with 18.5 indicating a "no referral" decision and 81 signifying a "referral" decision (Section 8.1). The fuzzy dataset involves uniform sampling across the entire potential input spectrum of the fuzzy system, encompassing not only integer values 0, 1, 2, but also the fractional values in between. Feeding the FIS with this continuous range of inputs enables a thorough mapping of its response surface. This process is crucial for visualizing and understanding the variations in FIS output as the inputs change.

The fuzzy dataset is composed of $n = 59,049$ data points, each represented as a 6-dimensional vector. This dataset is organized into a matrix with n rows and $m = 6$ columns. The first $m - 1$ columns, X_1, X_2, X_3, X_4, and X_5, constitute the feature space with values ranging from 0.2 to 1.8 in increments of 0.2. Lowering the step size creates a larger dataset that increases the granularity of the input space, but has the drawback of creating more computational overhead in testing and fine tuning the FIS. A step size of 0.2 resulted in acceptable running speed. A dataset with a step size of 0.1 was tested (Results Section 8).

The last column, denoted as Y, serves as the decision variable, adopting values from the set {0, 0.5, 1}.

Each data point in the fuzzy dataset is an ordered tuple $(x_1, x_2, x_3, x_4, x_5, y)$, where x_i corresponds to the value in column X_i and y is the decision variable's value, determined by the sum of the first five column values. These points are denoted as $d_i = (x_i^1, x_i^2, x_i^3, x_i^4, x_i^5, y_i)$ for $i = 1, 2, \ldots, n$ (superscripts do not indicate exponentiation in the mathematical notation throughout this paper; they are used for indexing only). Specifically, let the sum $S_i = x_i^1 + x_i^2 + x_i^3 + x_i^4 + x_i^5$ for each data point d_i. Then, the value of y (or Y) for each data point is calculated from S_i, akin to the methodology used in the AQ-10+ dataset, allowing for the consideration of borderline cases. The value of y_i is determined as follows:

$$y_i = \begin{cases} 0 & \text{if } S_i < 6; \\ 0.5 & \text{if } |S_i - 6| < \epsilon; \\ 1 & \text{if } S_i > 6, \end{cases}$$

where ϵ is a small tolerance value (like 1×10^{-9}) used to ensure that values extremely close to 6 (due to rounding errors) are still correctly classified.

The fuzzy dataset retains the decision distribution of the AQ-10 dataset (Figure 5).

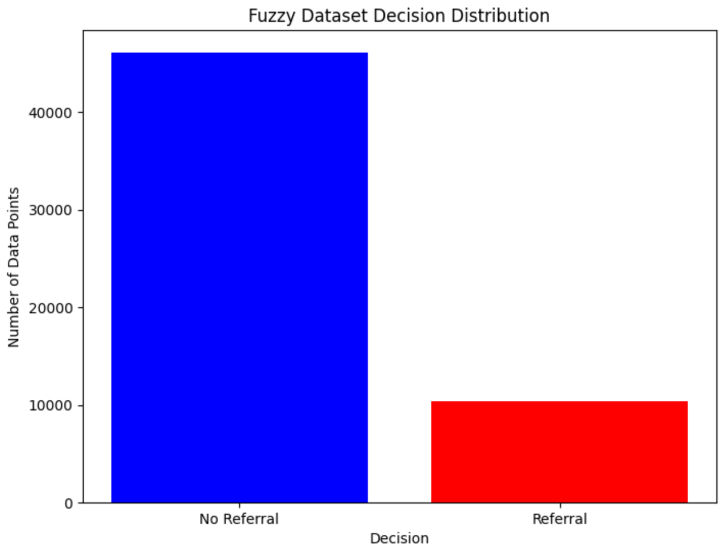


Figure 5. Phase 3 fuzzy dataset decision distribution (not including second opinion).

7. Methodology

7.1. Fuzzy Inference System Design

The Mamdani system operates on the principles of Fuzzy Logic, introduced by Lotfi Zadeh in the 1960s [37]. Unlike traditional binary logic, which is limited to true or false values, Fuzzy Logic accommodates varying degrees of truth. This characteristic aligns closely with human reasoning, which often navigates a similar spectrum of truth values. As discussed, this aspect is a principal reason for suggesting a complementary relationship between the nature of autism diagnosis and its inherent fuzziness.

The next subsection will present the system, focusing on the design decisions made in terms of linguistic variable name choices, defuzzification methods and the rulebase.

7.1.1. Input Membership Functions

The input membership functions are comprised of three fuzzy sets, where the linguistic variables are directly taken from the AQ-10 questionnaire. They are *disagree*, *slightly_agree* and *definitely_agree*. The *slightly_disagree* option was omitted given that the AQ-10 test assigns the same score to all linguistic terms regardless of whether a user expresses slight or complete agreement and slight or complete disagreement with the items in the questionnaire. This presents an advantage, as it allows for the reduction of the system's computational complexity by having fewer fuzzy sets in the membership functions (Figure 6).

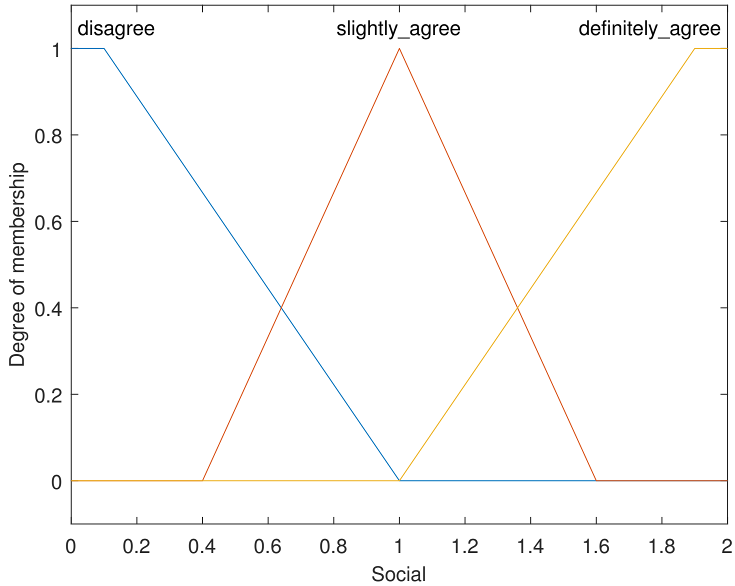


Figure 6. Attention to detail MF.

The five input membership functions are detailed below with their parameters; for clarity, maintainability and overall efficiency, they use consistent fuzzy sets and parameters:

- MF Name: Attention_To_Detail.
  Domain: $X \in [0.0, 2.0]$.
  Fuzzy sets:
  - disagree: $\mu_{\text{disagree}}(x) = \text{trapmf}(x; 0, 0, 0.1, 1.0)$;
  - slightly_agree: $\mu_{\text{slightly\_agree}}(x) = \text{trimf}(x; 0.4, 1.0, 1.6)$;
  - definitely_agree: $\mu_{\text{definitely\_agree}}(x) = \text{trapmf}(x; 1.0, 1.9, 2.0, 2.0)$.
- MF Name: Attention_Switching.
  Domain: $X \in [0.0, 2.0]$.
  Fuzzy sets:
  - disagree: $\mu_{\text{disagree}}(x) = \text{trapmf}(x; 0, 0, 0.1, 1.0)$;
  - slightly_agree: $\mu_{\text{slightly\_agree}}(x) = \text{trimf}(x; 0.4, 1.0, 1.6)$;
  - definitely_agree: $\mu_{\text{definitely\_agree}}(x) = \text{trapmf}(x; 1.0, 1.9, 2.0, 2.0)$.
- MF Name: Communication.
  Domain: $X \in [0.0, 2.0]$.
  Fuzzy sets:
  - disagree: $\mu_{\text{disagree}}(x) = \text{trapmf}(x; 0, 0, 0.1, 1.0)$;
  - slightly_agree: $\mu_{\text{slightly\_agree}}(x) = \text{trimf}(x; 0.4, 1.0, 1.6)$;
  - definitely_agree: $\mu_{\text{definitely\_agree}}(x) = \text{trapmf}(x; 1.0, 1.9, 2.0, 2.0)$.
- MF Name: Imagination.
  Domain: $X \in [0.0, 2.0]$.
  Fuzzy sets:
  - disagree: $\mu_{\text{disagree}}(x) = \text{trapmf}(x; 0, 0, 0.1, 1.0)$;
  - slightly_agree: $\mu_{\text{slightly\_agree}}(x) = \text{trimf}(x; 0.4, 1.0, 1.6)$;
  - definitely_agree: $\mu_{\text{definitely\_agree}}(x) = \text{trapmf}(x; 1.0, 1.9, 2.0, 2.0)$.

- MF Name: Social.
  Domain: $X \in [0.0, 2.0]$.
  Fuzzy sets:
  - disagree: $\mu_{\text{disagree}}(x) = \text{trapmf}(x; 0, 0, 0.1, 1.0)$;
  - slightly_agree: $\mu_{\text{slightly\_agree}}(x) = \text{trimf}(x; 0.4, 1.0, 1.6)$;
  - definitely_agree: $\mu_{\text{definitely\_agree}}(x) = \text{trapmf}(x; 1.0, 1.9, 2.0, 2.0)$.

  Domain: $X \in [0.0, 2.0]$.
  Fuzzy sets:
  - disagree: $\mu_{\text{disagree}}(x) = \text{trapmf}(x; 0, 0, 0.1, 1.0)$;
  - slightly_agree: $\mu_{\text{slightly\_agree}}(x) = \text{trimf}(x; 0.4, 1.0, 1.6)$;
  - definitely_agree: $\mu_{\text{definitely\_agree}}(x) = \text{trapmf}(x; 1.0, 1.9, 2.0, 2.0)$.

7.1.2. Output Membership Function

The output membership function has three Gaussian fuzzy sets labelled "No_Referral", "Second_Opinion" and "Referral" (Figure 7).

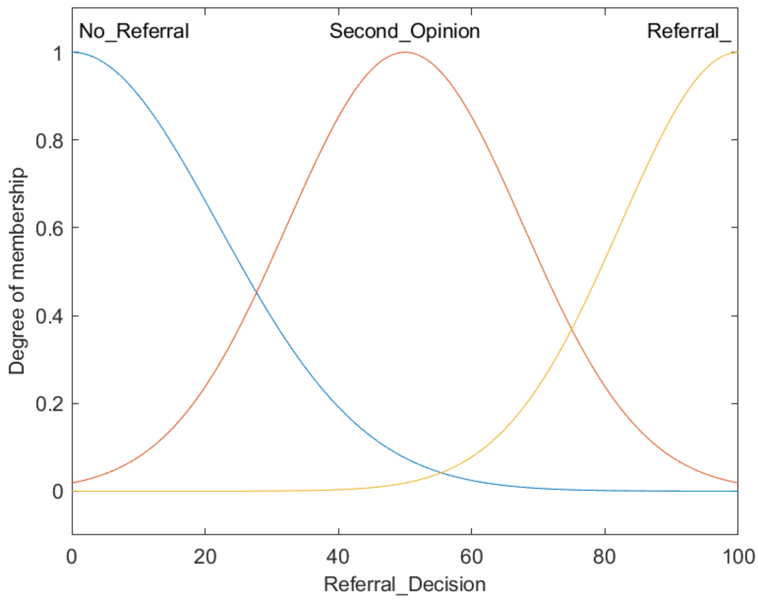


Figure 7. Referral decision.

7.1.3. Defuzzification Methods

During the testing phase of this project, all defuzzification methods available in MATLAB were trialed. Accuracy was maximized by fine-tuning the weights of the rule base, and the impact of each defuzzification method on the system's overall accuracy was observed. The results of this process are displayed in Table 2. The Middle of Maximum defuzzification method yielded the best accuracy.

Table 2. Comparison of defuzzification methods and their accuracy for Phase 3 of testing.

Defuzzification Method	Overall Accuracy (%)
Centroid	84.10
Bisector	88.11
Middle of Maximum	92.91
Smallest of Maximum	89.26
Largest of Maximum	78.99

7.1.4. Rulebase

The development of the rule base was specifically tailored to enhance the FIS with its three key functionalities, focusing on synchronizing the FIS with the AQ-10 decision criteria. To achieve this, a rule base comprising 243 distinct rules was crafted, each corresponding to one of the 243 unique input combinations from the AQ-10 dataset. This alignment ensures that the FIS functions deterministically, mirroring the accuracy of the AQ-10 questionnaire by providing predefined outcomes for each potential scenario.

During the final testing phase, a synthetic dataset was created to comprehensively cover the entire range of inputs for the MFs, resulting in 59,049 data points (with a step size of 0.2). With 243 rules, respectable results were obtained, given the dataset's vastness. These rules were specifically designed to address the 243 AQ-10 inputs, making the fine-tuning of weights unnecessary until the onset of Phase 3 testing with the fuzzy dataset. The challenge of applying a finite rule set to a broader and more complex range of inputs in the fuzzy dataset, which includes 59,049 possible inputs, underscores the difficulty in Fuzzy Logic.

7.2. Example of Fuzzy Rules

The rule base operates on an IF-THEN logic, where each rule specifies that IF a certain combination of inputs (antecedents) is met, THEN a specific output (consequent) is assigned. Structurally, the rule base is organized as an $n \times m$ matrix where $n = 243$ and $m = 8$. The rules are delineated as follows:

- **Five Antecedents:** These correspond to the five inputs, each representing a specific autistic trait.
- **One Consequent:** The output or target ground truth label dictated by the rule.
- **Rule Weight:** Positioned as the seventh element of each rule, indicating the importance or confidence level of the rule.
- **Fuzzy AND Operator:** The eighth element, employing the *min* operator used in Fuzzy Logic to evaluate the intersection of conditions defined by the antecedents.

Below are some examples of the fuzzy rules in both their vector form and textual logic form:

- **No AQ-10 Traits:**
  - **Input Vector:** $[0, 0, 0, 0, 0]$
  - **Rule Vector:** $[1, 1, 1, 1, 1, 1, 0.1, 1]$
  - **Textual Logic:**

    IF Attention_To_Detail is disagree **AND** Attention_Switching is disagree **AND** Communication is disagree **AND** Imagination is disagree **AND** Social is disagree **THEN** decision is No_Referral.
- **All Autistic Traits Present:**
  - **Input Vector:** $[1.8, 1.8, 1.8, 1.8, 1.8]$
  - **Rule Vector:** $[3, 3, 3, 3, 3, 3, 0.1, 1]$
  - **Textual Logic:**

    IF Attention_To_Detail is agree **AND** Attention_Switching is agree **AND** Communication is agree **AND** Imagination is agree **AND** Social is agree **THEN** decision is Referral.
- **Moderate Autistic Traits (where sum of inputs = 6):**
  - **Input Vector:** $[1.0, 1.0, 1.0, 1.0, 2.0]$
  - **Rule Vector:** $[2, 2, 2, 2, 2, 2, 0.5, 1]$
  - **Textual Logic:**

    IF Attention_To_Detail is Slightly_Agree **AND** Attention_Switching is Slightly_Agree **AND** Communication is neutral **AND** Imagination is Slightly_Agree **AND** Social is Slightly_Agree **THEN** decision is Second_Opinion.

Model Complexity

Model complexity for the FIS is defined by several factors. The FIS model comprises 243 fuzzy rules governing the relationships between the input variables and the output decision. Each of the five input variables is associated with three membership functions, while the output variable has three membership functions. The parameters defining these functions include trapezoidal membership functions with four parameters each, triangular membership functions with three parameters each and Gaussian membership functions with two parameters each. The total number of parameters is 56, with 50 parameters for the input membership functions and 6 for the output membership functions. Training the FIS model involved manual adjustments to rule weights over several months, based on identifying fuzzy rules that led to incorrect results. This iterative process required domain knowledge and experimentation to optimize overall accuracy. The inference time was measured, with the single input inference time being approximately 0.0032 s, and the time to evaluate the entire dataset being approximately 32.1801 s. These results indicate that the FIS model can quickly generate predictions, making it suitable for real-time applications.

8. Results and Analysis

The results and interpretation will be presented in four phases, corresponding to the testing stages of the system. Phase 1 involved testing the system's performance with the AQ-10 dataset, Phase 2 used the AQ-10+ dataset, Phase 3 evaluated the system with the fuzzy dataset and the final phase uses perturbed subsets of the fuzzy dataset to validate the system and provide experimental statistics.

8.1. Phase 1 (AQ-10)

It is observed that for Phase 1 and Phase 2, the reported overall accuracy is 100%, which may seem misleading. However, this indicates that the FIS accurately adheres to AQ-10 criteria, meaning any combination of AQ-10 scores inputted into the system will yield the same decision as the AQ-10 itself, demonstrating complete determinism. Figure 8 displays the results for the AQ-10 dataset, and comparison with the original decision distribution in Figure 5 reveals identical distributions, accurately identifying the same number of No_Referral and Referral decisions. The No_Referral decisions have a defuzzified crisp value of 18.5, resulting in a membership degree to the No_Referral output fuzzy set of $\mu_{\text{No\_Referral}}(18.5) = 0.56$, indicating majority membership to the correct output fuzzy set. Similarly, the Referral results show majority membership to the Referral fuzzy set of $\mu_{\text{Referral}}(81) = 0.56$. Despite only having a marginal majority membership within their respective output fuzzy sets, the fuzzy system functions in a binary manner. This binary characteristic aligns with the requirement for the system to operate deterministically, ensuring the accurate categorization of the two distinct outcomes projected by the AQ-10 test.

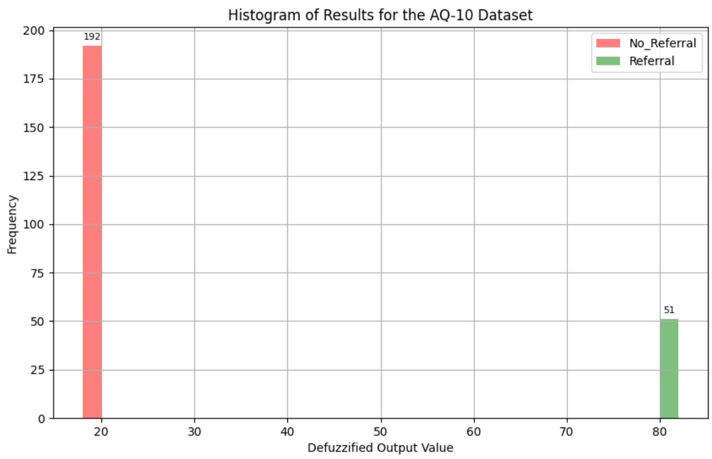


Figure 8. Results distribution compared to the original distribution.

8.2. Phase 2 (AQ-10+)

The results are displayed in the histogram in Figure 9, where the fuzzy system is observed confidently classifying each input. Keeping in mind the appearance of the output membership function (Figure 7), it is noted that no referral inputs are mapped to the No_Referral output fuzzy set with core membership.

There is no gradation in the crisp output values; they are either 0, 50, 81 or 100, which means there is no variability in terms of the degree of membership to the associated fuzzy set. This should not come as a surprise considering the deterministic nature of the AQ-10 and AQ-10+ datasets.

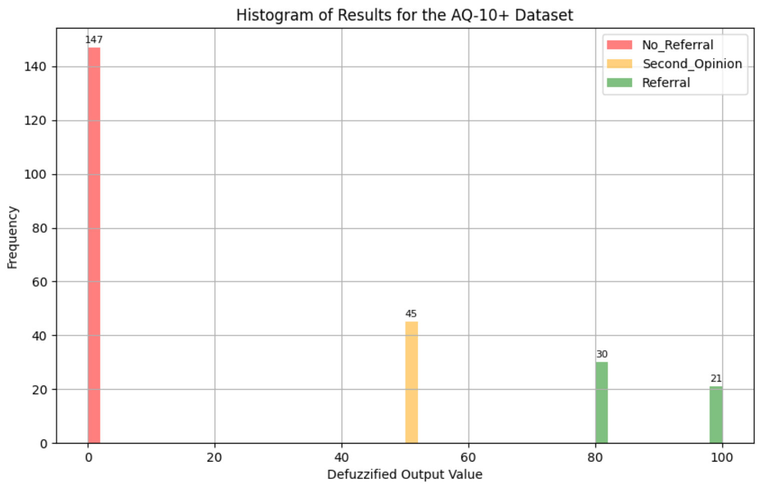


Figure 9. Results distribution compared to the original distribution.

8.3. Phase 3 (Fuzzy Dataset)

To evaluate the overall performance of the FIS, the accuracy was assessed by determining where the crisp output was located within the output membership function on the range of 0 to 100. Overall accuracy was calculated as follows:

$$\text{Overall\_Accuracy} = \frac{\text{TN} + \text{TP} + \text{TP\_SO}}{n}.$$

where TN is the true negative score, TP is the true positive score and TP_SO is the true positive score for second opinion decisions. n is the size of the fuzzy dataset.

The TN, TP and TP_SO were calculated as follows:

$$\text{TP} = \sum_{i=1}^{n} [results_{i,1} > 70 \wedge results_{i,2} = 1],$$

$$\text{TN} = \sum_{i=1}^{n} [results_{i,1} < 30 \wedge results_{i,2} = 0],$$

$$\text{TP\_SO} = \sum_{i=1}^{n} [results_{i,1} \geq 30 \wedge results_{i,1} \leq 70 \wedge results_{i,2} = 0.5],$$

where n = size of the fuzzy dataset, $results_i^1$ is the i-th crisp output value and $results_{i,2}$ is the i-th target value (1 = referral, 0 = no referral and 0.5 = second opinion). TP's were determined to be correct if crisp output was >70, TN's were determined to be correct if the crisp output was <30 and second opinion decisions were assessed to be correct if the crisp output value was between 30 and 70. These values were chosen to ensure that the decisions align with the area where the associated fuzzy output set has significant membership (greater than 0.5). Refer to Figure 7 to visualize these regions within the fuzzy output sets.

Additionally, an investigation is needed to determine the degree of membership each crisp output would result in. It must be ensured that all correct results have a majority membership in their appropriate fuzzy output sets. For instance, all referral decisions resulting in a crisp value of >70 also need to fall within the output fuzzy set, where $\mu_{\text{Referral}}(x) > 0.5$. To this end, a separate membership degree accuracy was calculated as follows:

$$\text{Correct Membership Degree}(i) = \begin{cases} 1, & \text{if Target}(i) = 0 \wedge \mu_{\text{No\_Referral}}(i) \geq 0.5; \\ 1, & \text{if Target}(i) = 0.5 \wedge \mu_{\text{Second\_Opinion}}(i) \geq 0.5; \\ 1, & \text{if Target}(i) = 1 \wedge \mu_{\text{Referral}}(i) \geq 0.5; \\ 0, & \text{otherwise.} \end{cases}$$

Basic performance metrics of the system including the membership degree accuracy results are displayed in the Table 3. The overall accuracy of 92.91% demonstrates the system's general reliability in categorizing cases, with most of its outputs aligning with the expected decisions. The membership degree accuracy is very close to the overall accuracy at 92.47%, indicating that the correct results had high membership values in their respective fuzzy output sets. The system correctly identifies 15.74% of cases as referrals and 75.81% as non-referrals, indicating a conservative approach that minimizes false positives by preferring not to make unnecessary referrals. This aligns with the distribution of the AQ-10-ST, which is designed to produce approximately 70% non-referrals, reflecting real-world conditions. The proportion of cases correctly identified as needing second opinions is low at 1.36%, suggesting that the system seldom recommends second opinions and leans towards decisiveness. Incorrect classifications are relatively low, with false positives at 0.28%, false negatives at 0.79%, false positives for second opinions at 1.17% and false negatives for second opinions at 1.86%. A notable proportion of the dataset is incorrectly identified as needing second opinions in instances where no referral is necessary (1.97%), indicating a tendency of the system to over-recommend second opinions in these cases. Positive cases misclassified as needing second opinions (1.03%) also indicate that while errors in referral decisions are rare, there is room for improvement in handling ambiguous cases.

Table 3. Performance metrics: proportion of correctly and incorrectly identified classes in the fuzzy dataset.

Metric Description	Proportion (%)
Overall Accuracy	92.91
Membership Degree Accuracy	92.47
True Positives	15.74
True Negatives	75.81
True Second Opinions	1.36
False Positives	0.28
False Negatives	0.79
False Positive for Second Opinions	1.17
False Negative for Second Opinions	1.86
Positive Cases Misclassified as Second Opinions	1.03
Negative Cases Misclassified as Second Opinions	1.97

A basic statistical analysis of the fuzzy dataset, as shown in Table 4, reveals a mean of 27.77, with both the median and mode at 14. This predominantly categorizes cases as "No_Referral", reflecting the observation that most individuals in the dataset do not exhibit autism, consistent with findings in the AQ-10 dataset and the general population. This interpretation is reinforced by a significant mode count of 25,396, highlighting the system's frequent identification of cases without signs of autism spectrum disorder (ASD). Specifically, this indicates that there were 25,396 results with a defuzzified output of 14, categorically placing them in the "No_Referral" fuzzy set, meaning these cases did not present traits indicative of ASD. The substantial standard deviation of 28.02 reflects the wide variability in the data, indicative of the diverse symptoms and severities of ASD, and demonstrates the system's capacity to handle a broad spectrum of inputs.

Table 4. Basic statistical summary of fuzzy inference system output. The term "Count (Mode)" denotes the frequency with which the modal value of 14.0 occurs within the results dataset.

Statistic	Value
Mean	27.77
Median	14.0
Mode	14.0
Mode Count	25396
Standard Deviation	28.02

Referring to the confusion matrix shown in Table 5, the matrix reveals a high accuracy in identifying *no referral* cases, evidenced by 44,764 true negatives, which is likely due to their prolific representation in the dataset. However, the *referral* category, while showing a reasonable number of true positives (9295), also presents a concerning number of false negatives (164), indicating missed cases where *referrals* might be necessary, a significant issue in a medical context. The *second opinion* category demonstrates lower accuracy, with 1162 false negatives and only 804 true positives. This is possibly attributed to the category's complexity and its under-representation in the dataset, making it challenging for the system to correctly classify such nuanced cases.

Table 5. Confusion matrix.

	Predicted No Referral	Predicted Second Opinion	Predicted Referral
No Referral	44764	1097	465
Second Opinion	1162	804	608
Referral	164	690	9295

Figure 10 shows the distribution of the results and compares them with the original distribution in the fuzzy dataset. This figure demonstrates roughly the same distribution, albeit showing that the FIS cannot accurately place all data points into the correct output fuzzy set.

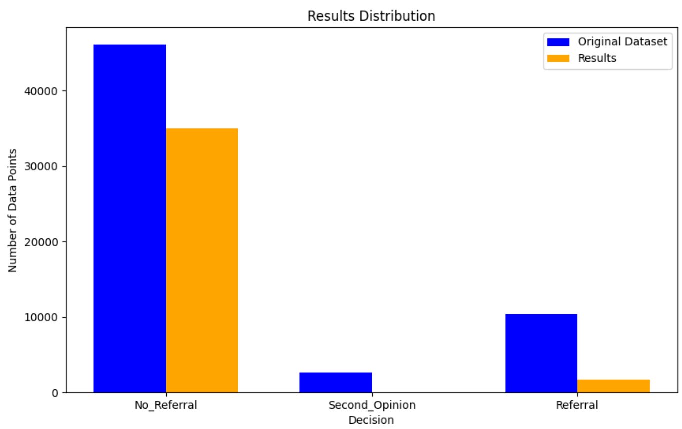
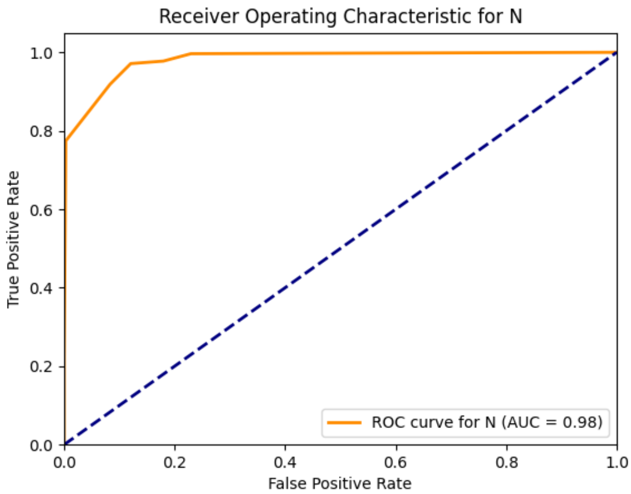
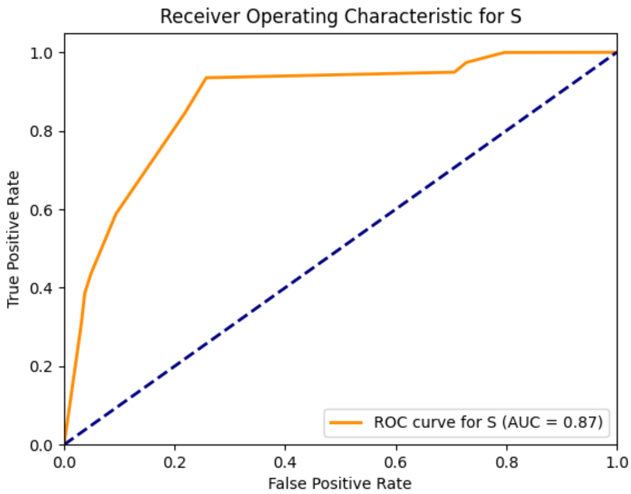


Figure 10. Results distribution compared to the original distribution.

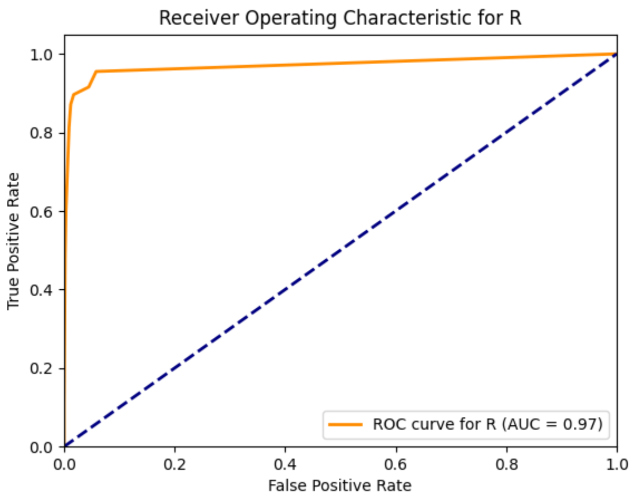
Besides the previous statistical insights, the medical domain often uses Receiver Operating Characteristics (ROC) and Precision Recall (PR) curves to determine the presence or absence of a disease [38]. PR is especially useful when dealing with highly skewed datasets, such as the datasets used in this study, given the majority of cases are negative for ASD according to AQ-10 criteria. Figure 11 shows the Areas Under Curve (AUC) for both ROC and PR.



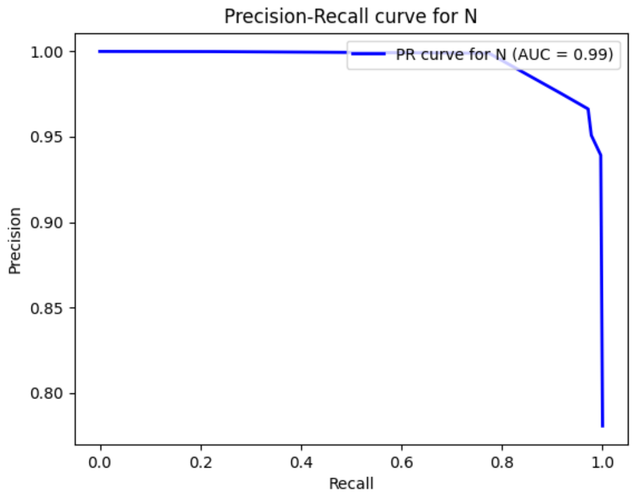
(**a**) AUC for N = No_Referral.



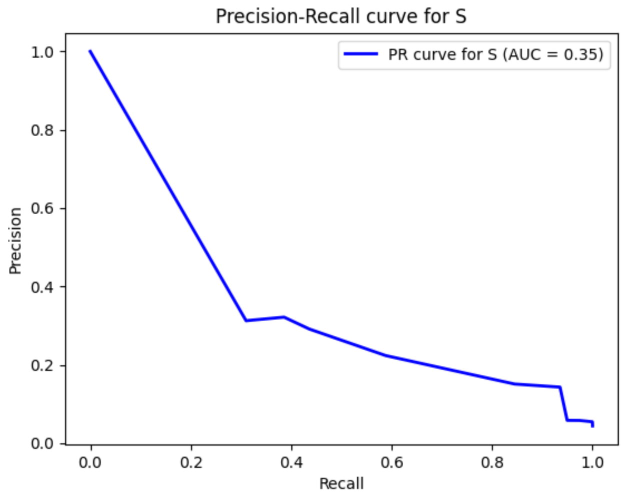
(**b**) AUC for S = Second_Opinion.



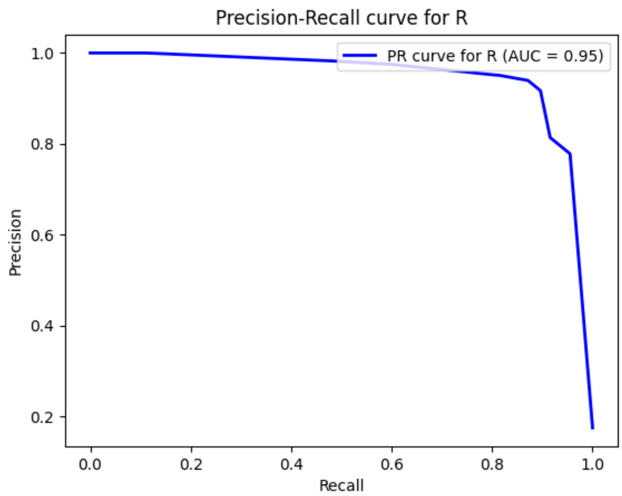
(**c**) AUC for R = Referral.



(**d**) AUC for PRN.



(**e**) AUC for PRS.



(**f**) AUC for PRR.

Figure 11. AUC for different categories.

Although the AUC for both the No_Referral and Referral decision categories are very high in both ROC and PR, this is likely due to the fact the dataset is very imbalanced and the FIS is struggling to correctly classify the minority class Second_Opinion decisions. The confusion matrix (Table 5) reinforces this.

8.4. Model Validation

To evaluate the robustness and statistical significance of the FIS, 100 datasets were randomly sampled from the original fuzzy dataset, each containing 1000 data points. Each value in these sampled datasets was perturbed slightly by ± 0.05 to ensure the FIS did not encounter the exact values it was fine tuned on during manual adjustments. The process for creating these datasets is as follows:

- **Random Sampling**: From the original dataset, 100 subsets, each with 1000 data points, were randomly selected.
- **Perturbation**: Each input value x corresponding to the five inputs to the five MFs in the sampled datasets were adjusted by adding a small random perturbation δ, where δ was uniformly distributed in the range $[-0.05, 0.05]$. Mathematically, for a value x, the perturbed value x' is given by:

$$x' = x + \delta, \text{where} \quad \delta \sim \mathcal{U}(-0.05, 0.05)$$

The summary statistics and standard deviation of the experiments are reported in Table 6.

Table 6. Summary statistics and standard deviation of FIS performance across 100 validation datasets.

Statistic	Value
Mean Overall Accuracy	0.9310
Standard Deviation of Overall Accuracy	0.0082
Maximum Overall Accuracy	0.9450
Minimum Overall Accuracy	0.9100

These results indicate that the FIS model maintains a high level of accuracy across multiple perturbed datasets. The mean overall accuracy of 0.9310 reflects consistent performance, while the low standard deviation of 0.0082 suggests minimal variation in accuracy across different datasets. The maximum and minimum accuracy values further highlight the system's stability, with the accuracy always remaining above 0.9100. These findings underscore the statistical significance of the FIS model, providing confidence in its reliability and applicability in real-world scenarios.

8.5. Non-Monotonic Defuzzified Output Values

Despite obtaining reasonable results, it was noted that the output values of the system do not increase exactly linearly with the dataset as expected. Given that each data point in the fuzzy dataset is summed and the membership degree to the associated output fuzzy set is expected to increase as the sum of the inputs increases, it would be expected that the defuzzified output value for an input [0.0 0.0 0.0 0.0 0.0] should be less than the defuzzified output value for the input [0.0 0.0 0.0 0.0 0.2], for example. Figure 12 shows all of the defuzzified output values for all 59,049 inputs in the dataset (ran through the FIS in sorted order according to the sum of their inputs). Clearly the output values of the FIS are not always increasing monotonically in proportion to the increase in the sum of the inputs. This indicates a potential misalignment between the expected growth in membership degree of the output fuzzy sets and the system's calculated output values.

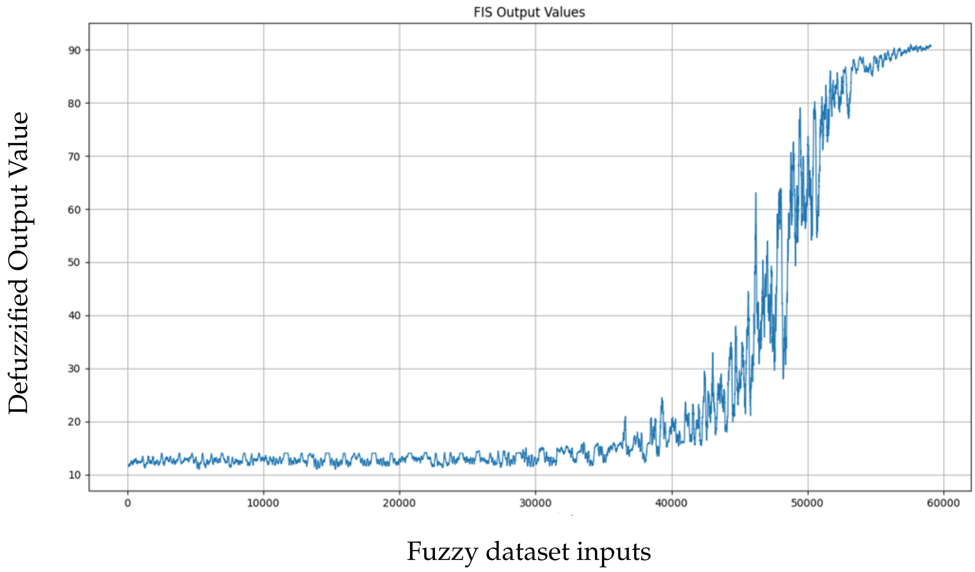


Figure 12. Output values for the whole dataset.

To be more explicit, referring to Table 7 which shows the first nine inputs of our fuzzy dataset with their corresponding defuzzified outputs and membership degress, although the output values are convincing majority membership of their correct No_Referral fuzzy set, they do not increase in a monotonic fashion.

Table 7. First 9 inputs of the fuzzy dataset, note the output values in the second column do not increase monotonically even though the sum of the inputs does increase.

Inputs	Defuzzified Output	Membership Degree
0.2 0.2 0.2 0.2 0.2	5.0	$\mu_{\text{No\_Referral}} = 0.97$
0.2 0.2 0.2 0.2 0.4	9.5	$\mu_{\text{No\_Referral}} = 0.91$
0.2 0.2 0.2 0.2 0.6	14.0	$\mu_{\text{No\_Referral}} = 0.80$
0.2 0.2 0.2 0.2 0.8	9.5	$\mu_{\text{No\_Referral}} = 0.91$
0.2 0.2 0.2 0.2 1.0	5.0	$\mu_{\text{No\_Referral}} = 0.97$
0.2 0.2 0.2 0.2 1.2	9.5	$\mu_{\text{No\_Referral}} = 0.91$
0.2 0.2 0.2 0.2 1.4	14.0	$\mu_{\text{No\_Referral}} = 0.80$
0.2 0.2 0.2 0.2 1.6	9.5	$\mu_{\text{No\_Referral}} = 0.91$
0.2 0.2 0.2 0.2 1.8	5.0	$\mu_{\text{No\_Referral}} = 0.97$

Further investigation reveals that this issue is present in the validation datasets. However, it is important to note that this has not compromised the overall accuracy. The predicted defuzzified output and the membership degrees to their respective output fuzzy sets still predominantly indicate correct results. Table 8 provides summary statistics for calculating the overall membership degree accuracy, which was determined as follows:

Table 8. Comparison of the accuracy results across 100 validation datasets (membership degree accuracy vs. classification accuracy).

Metric	Mean Accuracy	Standard Deviation
Membership Degree Accuracy	0.9263	0.0081
Classification Accuracy Metrics	0.9310	0.0082

Let T be the target label and μ represent the membership degree. A correct result is recorded if the following conditions are met:

1. $T = 0$ and $\mu_{\text{no referral}} > 0.5$.

2. $T = 0.5$ and $\mu_{\text{second opinion}} > 0.5$.
3. $T = 1$ and $\mu_{\text{referral}} > 0.5$.

In these conditions, $\mu_{\text{no referral}}$, $\mu_{\text{second opinion}}$ and μ_{referral} represent the membership degrees for the *no referral*, *second opinion* and *referral* fuzzy sets, respectively.

Table 8 shows the standard deviation low at 0.81. Furthermore, the table contrasts these membership degree results with the overall accuracy results obtained across all 100 validation datasets as previously reported in Table 6. This demonstrates that the membership degree results closely match the classification results, which suggests the FIS is not compromised by the non-monotonic anomaly.

9. Discussion

Following the presentation of the results, the discussion can now consider the implications of these findings. The extensive fuzzy dataset, which included 59,049 data points and a finer gradation of input values, presented a rigorous test for the system's ability to manage a vastly expanded input space. The system maintained high overall accuracy at 92.91%, with a membership degree accuracy of 92.47%, indicating robust performance under varied and less predictable conditions. The validation datasets support these findings. The ROC curves and Precision Recall results further highlight the system's strong capability in distinguishing *no referral* and *referral* cases, achieving AUC values of 0.98 and 0.97, respectively. However, the true positive rates for *referrals* and *second opinions* were lower, at 15.74% and 1.36%, respectively, suggesting a conservative bias in the system's decision-making process. The system effectively identified the majority of cases requiring *no referral*, with a true negative rate of 75.81%, crucial for minimizing unnecessary clinical evaluations. Yet, the classification of *second opinions*, as evidenced by a lower AUC of 0.87, remains a challenge, with the system correctly predicting *second opinions* in only 804 out of 2951 cases, illustrating a potential area for improvement in reducing false negatives and misclassifications. Notably, the FIS misclassified 1162 *second opinion* decisions as *no referrals*, and 608 as *referrals*, highlighting its struggle with these intermediate cases.

10. Conclusions

This study aimed to develop a FIS capable of making preliminary referrals for a full autism diagnosis, leveraging Fuzzy Logic to interpret responses in a manner aligned with the AQ-10 questionnaire. The system was designed with five membership functions representing key autism-related traits, each capable of categorizing responses into disagree, slightly agree and definitely agree. Its evaluation spanned three distinct datasets to validate the system's utility and robustness. The FIS demonstrated a promising alignment with the AQ-10 dataset, effectively categorizing the binary outcomes (no referral or referral) as intended. When expanded to the AQ-10+ dataset, which includes a "second opinion" category, the system continued to perform effectively, illustrating its adaptability to more nuanced decision-making frameworks. This capability suggests that the system can support existing diagnostic processes by providing an automated and consistent assessment tool.

However, several limitations were identified that could impact the findings and applicability of this study. The primary focus on high-functioning adults, while justified due to the difficulty in obtaining referrals for diagnosis and the adult-oriented AQ-10 model, may limit the generalizability of the results to other groups within the autism spectrum, such as children or individuals with more severe symptoms. The datasets used for validation, although extensive, might not fully capture the real-world complexities and variabilities of autism due to their synthetic nature. Real-world datasets focusing on behavior are fraught with difficulty to obtain due to privacy concerns, ethical considerations and the challenges of consistent data collection.

The findings from Phase 3 testing of the FIS indicate several areas for enhancement, particularly in improving its handling of nuanced scenarios. Enhancing the identification of "second opinions" could involve refining the Fuzzy Logic rules or increasing the granularity of the input categories to better differentiate these intermediate cases. Additionally, exploring machine learning techniques to dynamically adjust membership functions based on ongoing results could enhance the system's adaptiveness and accuracy. The manual adjustments to rule weights introduce subjectivity, potentially affecting reproducibility and limiting the system's adaptability. While the system excels in straightforward cases, its capability in handling complex scenarios presents a valuable area for ongoing research and development. Future work will focus on refining the model to improve sensitivity and precision across all categories of diagnosis referrals, integrating advanced computational techniques to ensure that the system can adapt to a wide range of clinical contexts and patient presentations.

11. Future Recommendations

- **Integrating Demographic Variants and Enhancing System Usability**: The next study could incorporate the children's and adolescent versions of the AQ-10, in addition to the adult AQ-10, to broaden its scope. Although expanding the system to include other demographics like children and adolescents might increase the number of membership functions and parameters, it is crucial not to complicate the system unnecessarily. Therefore, combining the features of different AQ-10 variants and utilizing data-mining techniques to determine the predictive power of AQ items across various demographics is recommended. By doing so, the study can employ other classification techniques, such as neural networks, to facilitate direct comparisons with Fuzzy Logic alone, Fuzzy Logic combined with neural networks (ANFIS) and deep learning independently. Currently, direct performance comparisons are challenging due to significant methodological and dataset differences among studies. Furthermore, developing a user-friendly graphical interface for the FIS will simplify data-input methods and provide clear result interpretations. Reinterpreting or renaming defuzzified output values and membership degrees to terms like "severity level" or "potential support required" will make the system more accessible to clinicians, caregivers and patients.
- **Incorporate Deep Learning Methods**: Fine-tune the FIS to enhance its accuracy, particularly for second opinion output, by utilizing automated techniques. Dynamically adjust the membership functions and rules based on ongoing data and outcomes. This adaptiveness can improve the system's accuracy in real-time scenarios. Ensure domain knowledge from the AQ-10 alignment is not compromised during this process. Although this may reduce domain knowledge and explainability, the performance improvements could justify these trade-offs.
- **Enhancing Realism in Synthetic Datasets for Autism Diagnosis:** Although the AQ-10 datasets used may be synthetic, they are designed to evaluate systems that are applied in real-world settings (AQ-10). Limitations still need to be addressed. To overcome the limitations of synthetic datasets in capturing real-world complexities for autism diagnosis, research should focus on enhancing the realism of synthetic data through advanced simulation techniques. By incorporating detailed behavioral patterns and variations observed in smaller, ethically sourced real-world samples, the synthetic datasets can be made more representative. Additionally, using techniques like generative adversarial networks (GANs) may improve the quality of synthetic data, especially for datasets like AQ-10, because GANs are designed to handle real-valued data [39]. GANs work by having two neural networks compete against each other: one generates synthetic data, while the other tries to distinguish between real and synthetic data. This process continues until the generated data are nearly indistinguishable from real-world data, making the synthetic datasets much more realistic and useful for validating computational models [39]. More robust validation frameworks that include stress-testing the models against diverse, hypothetical scenarios to ensure their reliability in varied real-world conditions will also be explored.

- **Thorough Non-Monotonic Anomaly Investigation:** No further enhancement to the system should go ahead without identifying the cause of the non-monotonic increasing defuzzified output issue, and finding a solution to it.

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Abbreviations

The following abbreviations are used in this manuscript:

ASD	Autism Spectrum Disorder
AQ	Autism Quotient
DSM	Diagnostic and Statistical Manual of Mental Disorders
FIS	Fuzzy Inference System
MRI	Magnetic Resonance Imaging
EEG	Electroencephalogram
CNN	Convolutional Neural Networks
DBN	Deep Belief Networks
ANFIS	Adaptive Neuro-Fuzzy Inference System
IFNN	Interpretable Fuzzy Neural Network
FHGO	Feedback-Henry Gas Optimization
DNFN	Deep Neuro-Fuzzy Inference System
ROI	Region Of Interest
ROC	Receiver Operating Characteristics
PR	Precision Recall
AUC	Area Under Curve
AI	Artificial Intelligence
UK	United Kingdom
TSK	Takagi–Sugeno–Kang
IQ	Intelligence Quotient

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