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A Rule Based Expert System for Autism Diagnosis/Screening: Prototype Development

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ABSTRACT

Autism is one of the challenging neurodevelopment disorders affecting individuals from childhood up to their adulthood. Most individuals with ASD have issues with social communication, and social interaction, exhibit some repetitive behavior, and have sensory issues. According to the DSM5 criteria, there are 3 severity levels of autism: level 1, level 2, and level 3 depending on the individual's behavioral features. Diagnosing and screening autism is very challenging in terms of cost, the time it takes and the lack of enough experts to do it, especially in the low medium-income countries (LMIC) such as Nigeria. In this study, we have developed a prototype of a mobile-based expert system named Autism Tracker which can be used for parents and caretakers for screening autism. Primary healthcare workers and special needs educators can also use it for Diagnosis. The system can also be used for knowing the severity level of autism in the individual after diagnosis. It is a rule-based system where JRip classification algorithm was used for pruning rules used in developing the expert system for both diagnosis and severity. This expert system is in English language and has also been translated into three of the major languages spoken in Nigeria and some West African countries.

Key words: Autism, Diagnosis, Severity, Expert system, Rule based

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INTRODUCTION

Autism Spectrum disorder (ASD) is one of the neurodevelopment disorders among children that have neurologic conditions along with other co morbidities associated particularly hyperactivity and epilepsy [1] but many others may exist too [2]. According to the World Health Organization (WHO) 1 out 0f 160 children has ASD [3] and its prevalence have said to be increasing according to the autism society between 6% and 15% each year from 2002 to 2010 and will likely continue to rise at this rate. And over 3.5 million people in the U.S. have an autism diagnosis [3]. In Nigeria, about 600,000 children are said to be diagnosed with autism which is one out of every 125-150 children is living with the condition as at 2014 according to Lesi, et al. [4].

Some of the early signs of ASD include lack of eye contact, lack of imitative play, lack of interest in other children, lining up of toys and repetitive behaviors. Early diagnosis helps in reducing these behaviors with appropriate therapy but there is no medical test for autism diagnosis [5]. So many tools have been developed to screen and diagnose autism but most of them are manual and required expertise before it can be used [6]. An most of these tools are not freely available, therefore researchers have developed tools that can help with the diagnosis for their own countries which are based on the languages used in the country [7].

According to the Diagnostic and Statistical Manual of Mental Disorders, 5th edition (DSM5) [2], ASD can be classified into 3 levels of severity. The first level is Level 1 (Mild), this is the mildest form of autism. Children with level 1 ASD have a hard time communicating appropriately with others, may also have trouble moving from one activity to another or trying new things. And also they may have problems with organization and planning, which may prevent them from being as independent as other people their age. Second is Level 2 (Moderate), individuals with ASD level 2 have problems with verbal and social communication, they find it harder to change focus or move from one activity to the next, they engage in repetitive behaviors that can make it difficult for them to function in certain situations.

And the Third is Level 3 (Severe), individuals in this category will have many of the same behaviors as those with levels 1 and 2, but to a more extreme degree. They have problems expressing themselves both verbally and nonverbally can make it very hard to function, interact socially, and deal with a change in focus or location. They also engage in repetitive behaviors.

This research study is aimed at addressing the main issue with ASD diagnoses in Nigeria which is lack of qualified professionals and tools in our healthcare centers to diagnose it properly [8]. And many of the medical practitioners in Nigeria lack the knowledge about the condition [1,8,9] which is another reason that may cause late diagnosis of the condition. And also as a tool for data collection, this will help to know the prevalence of autism in Nigeria because most of the research conducted in Nigeria did not provide any statistics or means of getting the number of people who are on the spectrum [1,10,11] rather provided statics for a particular state in the country.

The mobile based expert system is an important tool that can help parents to easily identify their children condition and also for teachers to carry out some assessment which they can notify the parents for early Autism diagnosis. The application also classify the patient's severity level based on the DSM5 criteria, which will help in giving the appropriate therapy on time to the individual. Autism Tracker is also intended for use in remote areas where health care facilities are very limited, because of this reason this expert system is in the 3 major languages (Hausa, Yoruba and Igbo) and some of the West African countries like Ghana, Niger and Cameron.

This application will save the time of people waiting on the queue for Autism screening and also cost that is said to be very high in most developing countries [12].

LITERATURE REVIEW

In this section we did a literature review of studies that have suggested the development of tools for Autism diagnosis, how machine learning can be used to improve the accuracy of autism diagnosis, the need for data in this field of study and some of the studies that have classified autism into the severity levels according to the DSM5 criteria. There are many tools used for Autism diagnosis and screening, but very few are for use in low Medium Income Countries (LMIC) according Marlow et al [13], they identified only 3 tools which are being used for Autism Screening in LMIC which are M-CHAT –R/F, PAAS and TIDOS.

In two of our studies [7,14], we reviewed some Mobile Application and Expert system which are developed for Autism diagnosis and screening and we found out that some low medium countries have developed screening/ diagnosis tools that can be used in their countries in their own languages. PCADEX [15] is one of the Expert system developed to be used in Pakistan, in Sri Lanka [16] and ASES [17] in Iran. Then BRBES [18], Autism [19,20] all in Bengali for use in Bangladesh. There was none developed for use in Nigeria as seen from the literature or Africa and [16] suggested that a fully transparent algorithm (Rule based model) may prove to be advantageous because of its ease of interpretation which is what we have implemented in this work.

Some of the Studies that recommended the need for more research to be done on ASD in Nigeria and the entire Sub-region so as to know the exact figures are [1,21-23], the authors also recommended more awareness on ASD conditions so that diagnosis can be done on time. Barbaro, et al. [24] suggested that the Knowledge of Autism and Its diagnosis tools should adopt the open source collaboration where each person should have access to it when needed. Lack of affordable tools for use, insufficient diagnosticians and resources in Africa were some of the findings of Wannenburg, et al. in their study [23].

For decision support tools, Bakeron, et al. [25] reported that very few studies have been done on using machine learning to provide decision support tools that will help health care practitioners to diagnose ASD. He used machine learning method on a certain data set to determine the main conditions that will help in classifying a person as having ASD or Not but finds it difficult to determine the severity of ASD using the conditions. Kanimozhiselvi, et al. [26] in his work also highlighted the need for applications that can be used as a support for ASD diagnosis due to scarcity of professionals in the field. He developed models using few datasets collected manually that can help in classifying Autism at an earlier stage. And suggested mobile applications to be developed which can help parents and health workers to perform preliminary evaluations at a very early stage.

Romero-Garcir, et al. [27] developed a robot human interactive application called Q-Chat NAO for screening ASD using machine learning algorithm to select the questions from the Q-CHAT 10 questionnaire that best classify a person as having Autism or not. The Authors suggested for further validation of the system on a larger dataset of already diagnosed individuals before it can be used for real life diagnosis. This is one of the studies that highlighted the need for more data to be collected in this field of study.

We also did a review of studies that have tried to classify autism into its severity levels and very few in the literature have done that. Aldharbi et al [18] developed an expert system for autism diagnosis and severity level classification. Real patients' data was collected for the study and Belief Rule-Based Inference methodology using the Evidential Reasoning (RIMER) approach was adopted for the study. The autism factors measured in the study for the construction of the rules were social interaction, communication, behavior, sensory, motor and measured intelligence. The authors reported good result generated by the system compared to expert opinion and other fuzzy systems.

Isa, et al. [28] developed an expert system for classifying autism based on the severity level, it is a fuzzy based expert system named Autysis. Rules were generated based on only two main criteria which are social communication impairment and restricted behaviors. The authors reported that the result obtained by the system is 60% similar results given by the clinical psychologist. And lastly, very few machine learning studies were seen for autism severity classification in literature compared to the autism diagnosis studies.

Knowing the severity level of ASD is very important so that a good therapy plan can be recommended for the individual. It will also help parents to understand that not all the individuals on the Spectrum are the same, most of the individuals with mild ASD (Level 1) have very high IQ and thus make the parents to put more effort in supporting their children to reach their potentials. This will also help some parents from denying their children's diagnosis.

METHODOLOGY

This expert system is designed based on the questions of the commonly used questionnaire AQ-10 screening tool whose dataset is mostly used by researchers for machine learning and deep learning research work. The AQ-10 questionnaire is also one of the questionnaire that considers Children, Teenagers and Adults while the Q-CHAT was used for toddlers at 18months to 24 months [29]. The AQ-10 questionnaire was actually designed to help front line health workers easily asses ASD in individuals as early as possible before a final diagnosis can be made by a specialist after that referral [29]. Research has also shown that AQ-10 performed well as an instrument for screening ASD [30] and so many machine learning studies that used data set collected using the questionnaire have reported good results using different machine learning algorithms [31-34].

Our mobile based expert system is developed to screen individual based on their age range with different questions for each age range. The age ranges are Toddler (0-3 years), Child (4-11 years), Adolescent (12-16 years) and Adult (17 to 50years). It will also classify the individuals based on the DSM5 severity level criteria: Level 1 (Mild ASD), Level 2 (Moderate) and Level 3 (Severe).In this section, we will discuss on the dataset used for the classification and the rules generated using JRip algorithm (RIPPER).

Repeated Incremental Pruning to Produce Error Reduction (RIPPER) Algorithm is a more efficient algorithm used for generating rules on large noisy datasets. This algorithm was designed by Cohen in 1995 namely. There are two types of loops in the algorithm, the outer loop and the inner loop. The outer loop is used for generating a single rule at a time while and Inner loop increase one condition at a time to the current rule. The information gain measure is maximized by adding the conditions to the rule. This process is continued until it covers no negative example [35].

WEKA (Waikato Environment for Knowledge Analysis) tool was used for generating the rules in this study. WEKA is a collection of machine learning algorithms and data processing tools that can be used for different data mining problems such as classification, regression, clustering and association rule mining. WEKA is written in java and was developed at the University of Waikato in New Zealand. It is a user friendly platform that provides users with extensive support for the whole process of experimental data mining, from preparing the data, evaluating learning schemes statistically, data visualization, it also has a wide variety of learning algorithms and wide range of preprocessing tools [36].

Autism diagnosis classification

In this section, we will discuss how the rules for the diagnosis were obtained. Our expert system is designed to make diagnosis based on the age group of the individual, therefore each age group has different

Table 1: Autism diagnosis toddler dataset description.

Variable	Type	Description
A1	Binary (0, 1)	Does your child look at you when you call his/her name?
A2	Binary (0, 1)	How easy is it for you to get eye contact with your child?
A3	Binary (0, 1)	Does your child point to indicate that s/he wants something?
A4	Binary (0, 1)	Does your child point to share interest with you?
A5	Binary (0, 1)	Does your child pretend?
A6	Binary (0, 1)	Does your child follow where you are looking?
A7	Binary (0, 1)	Does your child show signs of wanting to comfort someone upset?
A8	Binary (0, 1)	Description of child first words
A9	Binary (0, 1)	Does your child use simple gestures?
A10	Binary (0, 1)	Does your child stare at nothing with no apparent purpose?
Age	Number	Toddlers (months)
Score by Q-Chat-10	Number	1-10 (Less than or equal 3 no ASD traits; > 3 ASD traits
Sex	Character	Male or Female
Ethnicity	String	List of common ethnicities in text format
Born with Jaundice	Boolean (Y/N)	Whether the case was born with jaundice
Family Number with ASD history	Boolean (Y/N)	Whether any immediate family member has a PDD
Who is completing the Test	String	Parent, self, caregiver, medical staff, clinician, etc.
Class Variable ASD	String	ASD traits or No ASD traits (Yes / No)

question and different rules are applied. The rules for the Child, Adolescence and Adult age group were adopted from a research by Fadi, et al. [37]. In this work we pruned the rules for the Toddler age group using a secondary dataset.

Dataset for diagnosis classification

The dataset used for generating rules for the toddler age group was collected by the same Fadi, et al. [37] and was obtained from kaggle (https://www.kaggle.com/fabdelja/autism-screening-for-toddlers). JRIP algorithm on WEKA platform was used to generate the toddler rules used for our expert system. The toddler dataset contains 1054 instances (319 females and 735 are males), 728 have ASD result as Yes and age between 12-36 months. The table 1 below gives the description of the features of the toddler data set.

Rule pruning for diagnosis

WEKA tool was used for the rule pruning using the JRip algorithm and K-fold cross validation (k=10) method was adopted for training and testing. This method runs the test 10 times and the first 9 times is used for training while the final fold is for testing. The algorithm generated 6 rules and was evaluated in terms of recall, precision, F-measure and ROC Area as shown in Table 2 below.

The rules for the Toddler are:

Rule 1: (A6=0) and (A7=0) and (A2=0) and (A3=0) => Class/ASD Traits =No

Rule 2: (A5=0) and (A1=0) and (A9=0) and (A8=0) => Class/ASD Traits =No

Rule 3: (A6=0) and (A2=0) and (A5=0) and (A10=0) and (A3=0) => Class/ASD Traits =No

Rule 4: (A9=0) and (A7=0) and (A10=0) and (A8=0) => Class/ASD Traits =No

Rule 5: (A4=0) and (A5=0) and (A2=0) and (A9=0) => Class/ASD Traits =No

Rule 6: Else => Class/ASD Traits

Autism severity classification

Our expert system gives the severity of ASD based the DSM 5 criteria. For this study, we collected our own primary data using google forms questionnaire, the link to the forms was shared to autism parents support groups on WhatsApp, Facebook and personal chat to some of them. The questionnaire was designed based on the DSM 5 criteria for diagnosing the severity of ASD.

Dataset for severity

The datasets we collected has a total of 72 instances, but 3 instances have missing values (Parents did not specify the DSM 5 Diagnosis) which were removed from these experiments and then we are left with 69 instances (50 males & 19 Females) and of different age groups mostly 3-8years of age. In our datasets, 6 behavioral features were recorded in addition to some features that will help in understanding the data and its validity. Table 3 below is the description of our primary dataset.

Rule pruning for severity

The first step of any machine learning experiment is usually preprocessing, in this study we first removed all missing value instances before uploading to WEKA and other preprocessing were done on WEKA platform. Secondly, our dataset have the problem of

Table 2: Result for toddler rules pruning.

Diagnosis	Precision	Recall	F- Measure	ROC Area
No	0.874	0.893	0.883	0.925
Yes	0.951	0.942	0.947	0.925

Table 3: Autism severity dataset description.

Features	Description	Type
Age_years	Numbers indicating the age	Number
Sex_of_Child	Gender of the Patient	String (Male or Female)
Age_Of_ Mother	Age of Mother at Birth of Age	Number
Formerly_ diagnosed	If Child has undergone a formal diagnosis	Boolean (Yes or No)
DSM_4_Diagnosis	Child DSM 4 Diagnosis	String
Age_Child_ diagnosed	The age the child was Diagnosed	Number
Country_of_ diagnosis	The Country the Child was Diagnosed	String (Nigeria or Abroad)
A	Answer to if the child is verbal or not	String (Verbal, nonverbal, Few words)
B	Answer to: How can you rate your child's deficits in social communication and social interaction	String (Mild, Moderate, Severe)
C	Answer to : How can you rate your Childs nonverbal communicative behaviours capabilities	String (Poor, Good, Very Good)
D	Answer to: If the child has difficulties in sharing imaginative play or in making friends and absence of interest in peers.	String (Yes, No, Sometimes)
E	Answer to: Do your Child has Stereotyped or repetitive motor movements and echolalia	String (Yes, No, Sometimes)
F	Answer to: How is his/her Hyporeactivity to sensory aspects of the environment	String (Mild, Moderate, Severe)
Comorbid Diagnosis	Answer to: Please what other challenges is your Child diagnosed apart from Autism	String
DSM5_Diagnosis	DSM 5 Diagnosis of the child (Level 1, Level 2, Level 3)	String

data imbalance, our level 3 class has a total number of 7 instances while the other classes have 30 and 32. So we used Synthetic Minority Over-sampling Technique (SMOTE) to illuminate the problem of class imbalance which we have in our dataset.

SMOTE is a method that was proposed by Chawla, et al. [38] to take care of class imbalance in a dataset. It is an oversampling approach in which the minority class is over-sampled by creating "Synthetic" examples, along the line segments joining any/all of the minority class nearest neighbors. Depending on the amount of oversampling required, neighbors from the k nearest neighbors are randomly chosen. Percentage split was the method adopted for this experiment, 90% of the dataset was used for training and 10% of the dataset was used for testing. The Performance of the algorithms were measured by their precision, recall, F-measure and Receiver Operating Characteristic (ROC) area which are the most commonly performance evaluation measures used for text classification as shown in Table 4 below.

The algorithm generated 4 rules which are shown below:

Rule 1: (D=No) and (A=Non- Verbal) and (C=Good) => DSM5=Level 3 (Severe Autism)

0Rule 2: (B=Mild) => DSM5=Level 1 (Mild Autism)

Rule 3: (C=Poor) and (B=Moderate) => DSM5=Level 1 (Mild Autism)

Rule 4: Else, DSM5=Level 2 (Moderate Autism)

EXPERT SYSTEM COMPONENT AND DISCUSSION

Our expert system comprises of 2 main components, the Knowledge engineer interface and the user interface which is a mobile based interface. This sections below will briefly discus the functions of the interface and how we develop them.

The knowledge engineer interface

The Knowledge engineer interface is a web-based interface developed using PHP programming language. The Knowledge base of the system which is the questions and rules are all embedded on the interface. The inference of the system is also done on the interface. The interface is accessible via the web pages using a username and password. Appendix figures are screen capture of the interface.

The user interface

The User interface is a mobile based interface which is an android based interface developed using Kotlin programming language. The user uses the interface to access the system in 4 different languages which are English, Hausa, Igbo and Yoruba. Figures 1 to Figure 3 below are screen shot of the user interface.

The user chooses the preferred language of usage on the landing page of the mobile application immediately after installation which is shown in Figure 1 below. There are 3 main menus for users to choose from which are shown in Figure 2 below, the first menu is for new users who have someone they want to Diagnose, the second is for self- diagnosis and the last is for people who have once used the system.

DISCUSSION

This application is developed to help individuals screen Autism for different age groups and also gives the severity of ASD. The users answer 10 questions for each individual and results are produced based on the rules. Responses and result are stored on SQL database and the application also captures responses and level of Autism of individuals who have conducted a formal diagnosis.

The locations of the individual being assessed is also captured which can be used for easy referral. We have also translated the application in the 3 commonly languages used in Nigeria apart from English which are the Hausa, Yoruba and Igbo languages. These translations were done by fluent speakers /writers of the various languages. Users switch from one language to another while using the application.

Figure 1: Landing page of the mobile application.

Table 4: Result for severity rules pruning.

Severity	Precision	Recall	F- Measure	ROC Area
Level 1	1	0.75	0.857	0.9
Level 2	0.6	1	0.75	0.833
Level 3	1	0.5	0.667	0.857

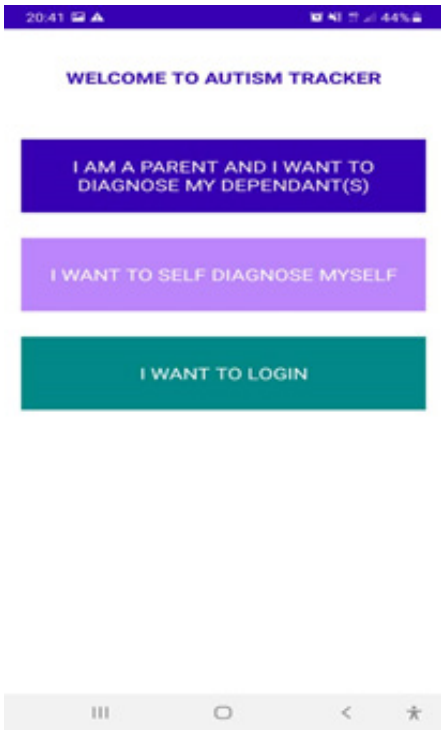

Figure 2: Main menu of the mobile application.

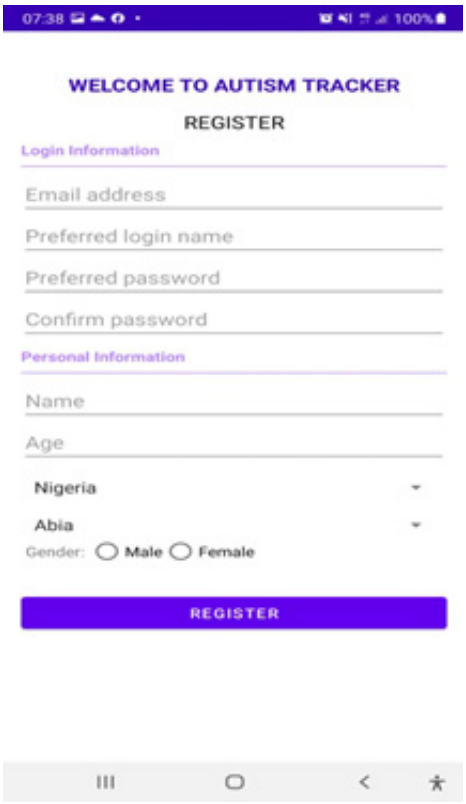

Figure 3: Registration page for self-diagnosis.

CONCLUSION AND RECOMMENDATIONS

Autism is a neurological development that affects children from childhood. Early intervention has proven to help both the individuals on the spectrum and their parents. In this study, we have developed a prototype mobile based expert system that can be used for screening Autism based on one of the existing Autism screening tool name AQ10 which is based on certain age groups. We have used machine learning to generate rules which were implemented. The Machine learning algorithm was measure based on recall, precision and F-measure which have shown good performance on both dataset.

In Nigeria Autism screening takes longer time, it is expensive and we also lack enough expert to handle the assessment/Screening efficiently therefore this will help in addressing such issue. The expert system can be used in 4 different languages including English and available on play store for testing by stakeholders.

In the future, integration of more modules like therapy suggestions and features will be considered so as to make it more beneficial to its users. A clinical validation of the system will also be conducted. Further research can be done by comparing the rule based expert system using decision tree or other machine learning models.

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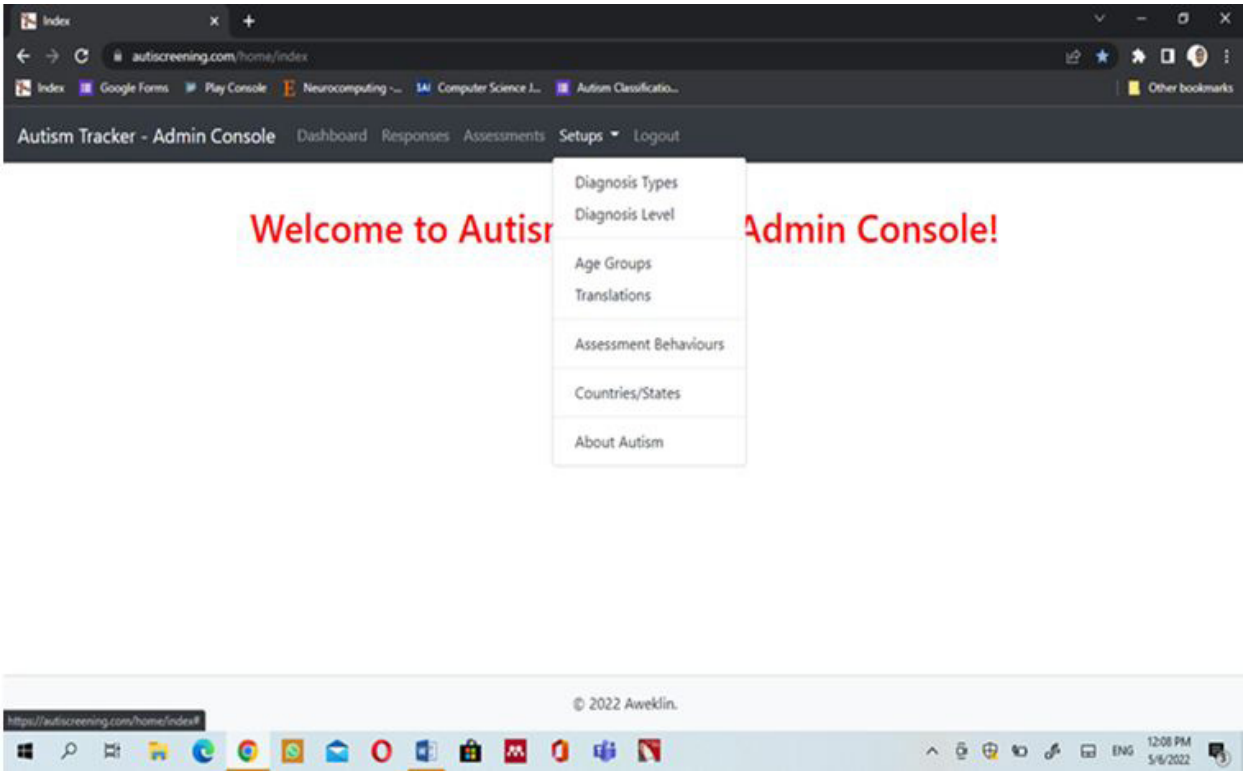
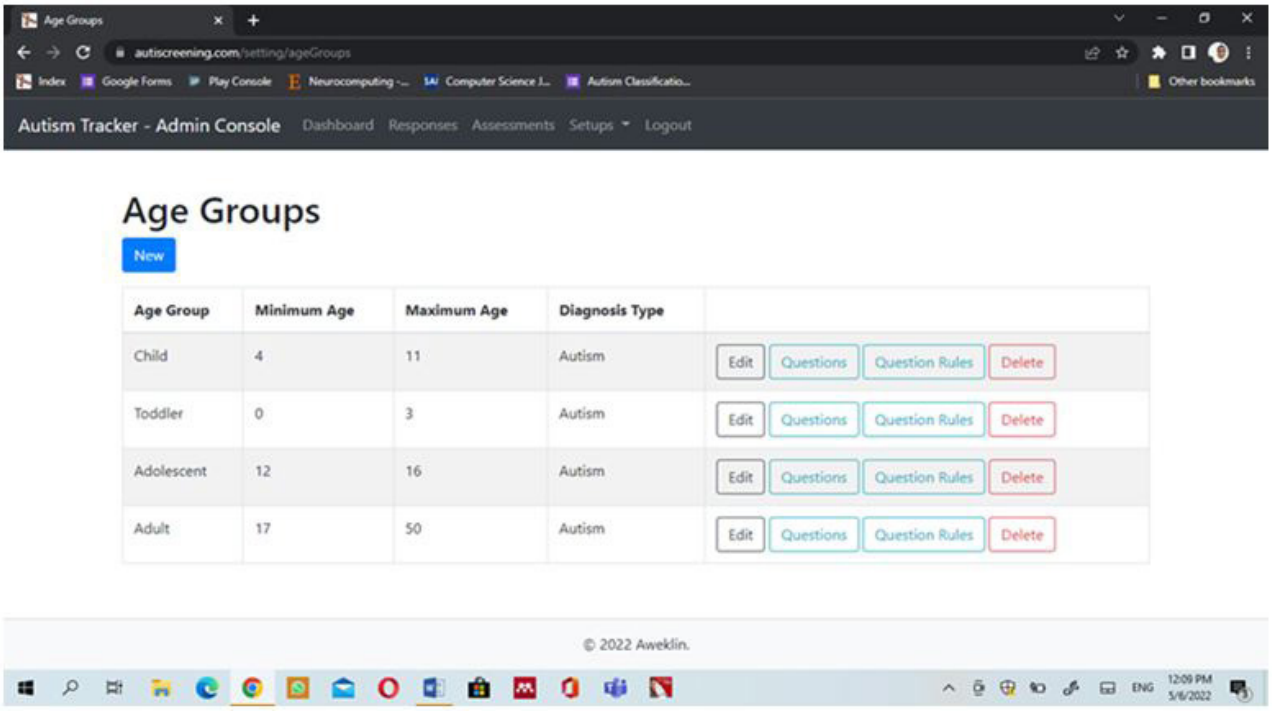
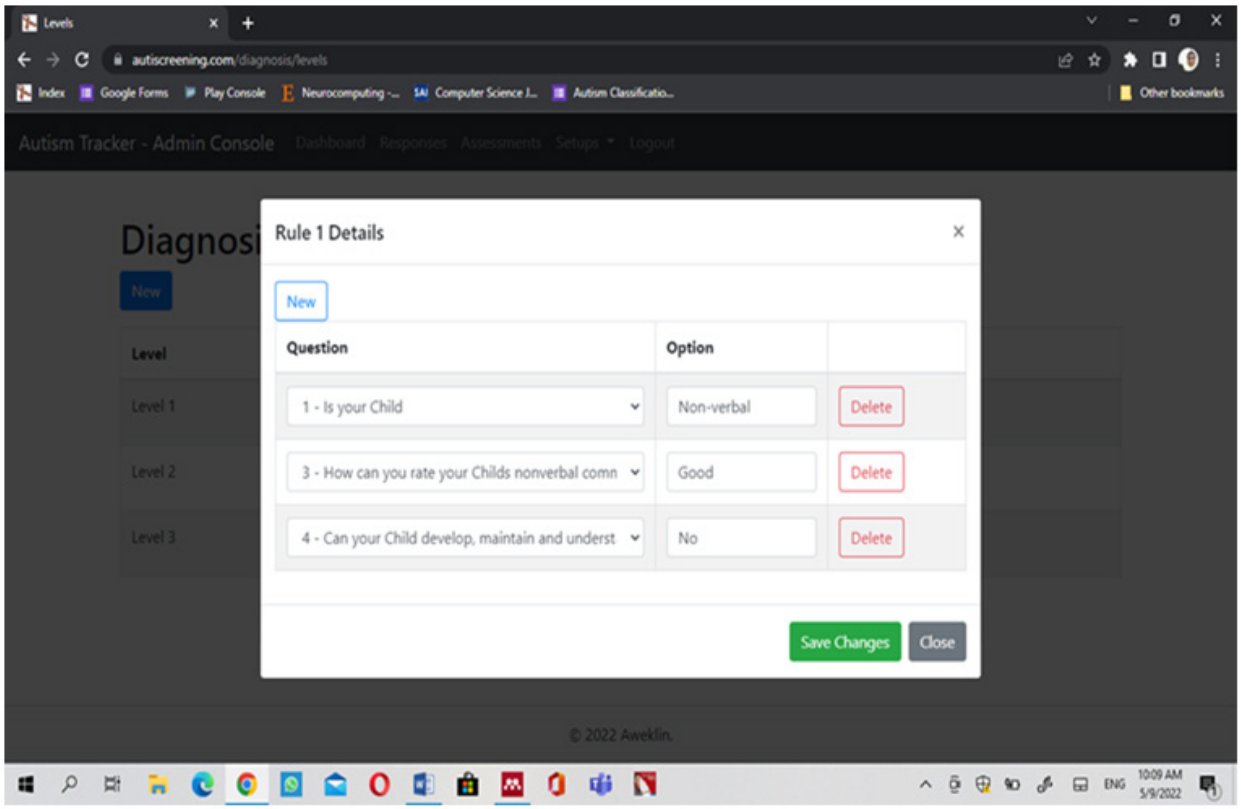
APPENDIX



Appendix 1: Screen shot of the knowledge engineer interface showing main menus.



Appendix 2: Screen shot of the knowledge engineer interface showing the knowledge base of the system.



Appendix 3: Screen shot of the knowledge engineer interface showing the implementation of the rules for severity level.