



Diagnosis of Autism Spectrum Disorder Based on Symptoms and Face Recognition

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Abstract: Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder that has various effects on language, speech, and communication of individuals. When ASD is detected in the earlier stages of life, especially in childhood, there would be many identifiers that would aid in the strategizing of the right therapeutic plan at the right time period.

Human faces have important markers that would aid in the identification of ASD through the analyzation of facial features and eye contact. There are other Artificial Intelligence-aided means of detection of ASD through the studying of various symptoms and finding patterns. In this research, we developed two systems that would aid in the diagnosis of Autism Spectrum Disorder in children, one of which uses a transfer-learning-based face detection framework. And the other system uses a decision-tree-based system to identify Autism in children based on symptoms. Various machine learning and deep learning techniques were applied.

Chapter 1: Introduction

1.1 Introduction

Autism Spectrum Disorder (ASD) is a neurodevelopmental disability that causes significant communication and behavioral challenges to patients. It consists of five main disorders: Autistic Disorder, Rett's Disorder, Asperger's Disorder (which is typically characterized by less severe symptoms and the absence of language delays), Childhood Disintegrative Disorder and Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS). According to the Centers for Disease Control and Prevention, some of the main signs of ASD in children include the avoidance of eye contact, delayed communication, delayed movement, impaired cognitive and learning skills, the inability to implement facial expressions in their communication, a lack of interest or total indifference to peers, not responding to their name being called by the age of nine months, and not being able to play common children games by certain ages during their development in the early stages of childhood. ASD patients also show repetitive behaviors like repeating words, flapping their hands, spinning themselves in circles, being obsessive over small details and routines, and getting upset over minor changes, as well as myriad other symptoms.

Today, there is extensive medical research dedicated for the disease all around the world.

1.2 Project Aims

This goal of this project is to provide a quick and reliable system for medical personnel to predict the chances of Autistic Disorder in children, which would allow for higher chances of success in the

treatment process, which means patients that are diagnosed with ASD at earlier stages in their lives might have significantly better communication abilities and a more-integrated lifestyle as they grow up. This project aims to use cutting-edge Machine Learning and Deep Learning methods to sufficiently meet the needs of the public health systems in many areas of the world, especially in developing countries, where access to a good healthcare system is significantly harder.

Furthermore, the diagnosis of ASD in children is a hard process even when there are lots of evidence for it, thus, software like these would greatly aid medical professionals in their choices.

1.3 Project Motivation

In 2021, The Center for Disease Control and Prevention in the United States stated that 1 in every 44 children in the U.S. is diagnosed with Autism-Spectrum Disorder. Around a third (31%) of the total number of children with ASD have intellectual disabilities characterized as having an IQ level of below 70, and an estimated 40% of ASD patients are nonverbal. The lifestyle of patients with ASD that is significantly different from the lifestyle of neurotypicals is still leading to major difficulties in the lives of patients that last even to their adulthood like being constantly harassed and bullied, the elevated chances of practicing self-injurious behavior, which ranged from skin-scratching, banging head against a wall, to running away from safety, which is the cause of 90% of deaths in ASD-diagnosed children.

1.4 Project Outline

Chapter One: Introduction

- This chapter introduces the objective and the motivation of the Autism disease.

Chapter Two: Autism Disorder Disease

- This chapter presents the theoretical background and the literature review for this project.

Chapter Three: Identification of Autism Spectrum Disorder

- This chapter demonstrates the techniques used for ASD identification. In its first part, it discusses the Facial imaging system that was used. And in the second part, it goes in detail about the symptoms recognition Decision Tree method we had used.

Chapter Four: Conclusions and Future Works

- This chapter concludes the research and asks further questions in the developing field of AI.

Chapter 2: Background and Literature Review

2.1 Introduction

The roots to the identification and classification of autism spectrum disorder go far back to the first two decades of the twentieth century, and since then, numerous changes to the definition, classification, and symptom identification have been made, and new ways of identification are being discovered lately that range from direct professional physiological and mental evaluation to fully-automated Machine-Learning-aided methods that require facial feature extraction, neuroimaging, EEG reading, or various other techniques.

2.2 Autism Disorder Disease

2.2.1 Symptoms

Children with Autism Spectrum Disorder (ASD) typically show a wide range of symptoms, varying from being mild communication difficulties to extreme impairment in cognitive functions and sensory abilities as well as showing repetitive behavior, language problems, and epilepsy in some cases.

Patients with ASD face challenges in their behavior from the early stages of their lives, and their symptoms persist as they grow. If the symptoms are overlooked, they might have the tendency to cause more discomfort to the patients with time, thus, some patients need consistent care. Most ASD symptoms in children are commonly subdivided into communication-related issues, repetitive

behavior, and physical challenges. The communication problems are typically characterized as having a below-average capacity to communicate with peers and parents since a third of ASD-diagnosed patients are nonverbal. They also show great difficulty in communication tasks that are nonverbal, like hand gestures, developing and reading facial expressions, and eye contact. They also typically cannot properly control their tone of voice. Patients with Autism Spectrum Disorder also show multiple signs that pose serious social challenges to them, some of which including the inability to accurately recognize and express their own emotions, the inability to seek emotional comfort from others, as well as recognizing the emotions of others. It is common for children with ASD to be diagnosed with Social Anxiety. Secondly, the repetitive behavior symptoms include repetitive body movements (e.g., rocking, running back and forth), staring at lights, spinning objects (e.g., spinning wheels, shaking sticks, flipping levers), ritualistic behavior (e.g., obsessively lining up objects and touching objects in specific orders), sticking to specific routines obsessively, and extreme interest in specific topics. And thirdly, some of the physical challenges patients with ASD might face are digestive issues, epilepsy, and poor muscle control.

2.2.2 Human Brain of Autism Spectrum Disorder (ASD)

The human brain is divided into three main regions: the cerebrum, the cerebellum, and the brain stem. Each region has its specific location and roles. The cerebrum is divided into two hemispheres that are subdivided into the cerebral lobes, which consist of the frontal lobe, the parietal lobe, the occipital lobe, and the temporal lobe, each with its distinct characteristics and functions. Autism Spectrum Disorder originates in the brain. Even though the exact cause for autism has not been found yet, but multiple distinctions have been found in the structure of certain regions in the brains of children with autism. For example, the hippocampus (which is in the medial region of the temporal lobe) of patients with autism has been found to have an increased size. Patients with autism also have a decrease in the amount of brain tissue in parts of the cerebellum. They also might have a decrease in the size of their amygdalae, which are located adjacently to the hippocampus, even though some studies suggest that autism is connected to an increase in the size of the amygdala. Furthermore, studies show that in the early stages of development, patients with autism show an enlargement in their head and brain sizes, as well as an unusual growth rate of the brain in specific regions.

2.3 Classification Techniques & Image Processing

There are multiple techniques for the initial diagnosis and classification of ASD in children, ranging from image processing-related techniques that use facial features or MRI images, which can aid in the final diagnosis through the search for specific measurements that might be increased/decreased activity levels in certain brain regions, or certain facial features that characterize autism patients, which include an increased size in the area between the mouth and the nose or a large forehead. These imaging techniques have been on the rise in the last ten years, and they are only increasing in research. They provide a valuable, ML-based methodology for the detection of autism in patients.

2.4 Literature Review

There is a significant rise in research for autism spectrum disorder diagnostic techniques, lots of which heavily rely on machine learning methods through the usage of neuroimaging [1], facial feature extraction [2], or questionnaires [3] that assess the symptoms based on the experience of the children's parents or guardians. Some of these questionnaires, like the Modified Checklist for Autism in Toddlers, Revised (M-CHAT-R) which is used for toddlers between the age range of 16-30 months, Childhood Autism Rating Scale (CARS), and the Support Intensity Scale (SIS), Eating and Drinking Ability Classification System (EDACS), and Communication Function Classification System (CFCS), which are used for the determination and examination of cerebral palsy in older children, have been used by Bertonecelli, Altamura, Vieira, Bertonecelli, and Solla, 2018 [4] in a machine-learning-based system to assess the symptoms of patients diagnosed with cerebral palsy and predict their chances of having autism. The system they developed used systems for the classification of cerebral palsy severity like SIS, EDACS, CFCS, as well as MACS and GMFCS, and had a precision rate of 75%, and provided specific scores in those systems that mostly associated with a higher presence of autism in patients. Achenie et al. [5] used FNN (Feedforward Neural Network) on

data of M-CHAT-R tests on 14,995 toddlers. Their best results showed high accuracy levels and they concluded that Machine Learning provides an advantage over previous screening methods and can ease the process of assessment and diagnosis. Shahriri and Thabtah, 2019, developed a questionnaire-based mobile app called *Autism AI* [6] that used Convolutional Neural Networks and determined autism presence with good accuracy. Moreover, image processing methods are also gaining popularity among researchers, a good example is the eye-tracking tool by Vargas Cuentas et al., 2017, [7] which followed the gaze preference of children diagnosed with autism spectrum disorder and differentiated between them and typically-developed children with sufficient accuracy. This system required no restriction of the children's head movement, making it extremely easy to work with alongside children diagnosed with ASD. Liu, Li, & Yi, 2016 [2], also developed an eye-tracking feature extraction-based machine learning system. Various other studies have been made that use similar methodology, which follows Artificial Intelligence techniques for the identification of autism in children (Rahman & Subashini [8], 2022) and (Lu & Perkowski [9], 2021) which takes ethno-racial factors into consideration. It is also worth mentioning that Artificial Intelligence use is being researched heavily for a myriad of neurodevelopmental diseases as well as autism (Uddin, Wang, Woodbury-Smith [10], 2017).

Chapter 3: Identification of Autism Spectrum Disorder

3.1 Introduction

The goal of the majority of this research is to assist medical professionals by automating parts of the diagnosis process. Among these methods is using Convolutional Neural Network (CNN) systems, which heavily integrate part-based and holistic information.

This study is about a transfer-learning-based framework that uses facial recognition for the detection of ASD in children with high precision. It classifies input images of children into two fields, autistic, and non-autistic. This software can be used in hospitals with heavily-available imaging equipment.

In this project, there is an improved transfer-learning-based facial recognition system that has high accuracy, while using an improved MobileNet model that has relatively-high levels of accuracy when compared to other machine learning models, deep-learning models, and various other AI systems. Furthermore, a range of different machine learning and deep learning models was used, with the aid of datasets of facial images of ASD-diagnosed children and neurotypical children. [11]

3.1.1 Methods

In this study, a transfer-learning-based classification system is implemented, it used machine learning and deep learning which automatically detected parameters with extremely-high accuracy.

A clinical data set which had 1468 images of children with autism and 1468 images of neurotypical children was acquired from the Kaggle data repository, and had a total of 2936 images. These images were curated by Piosenka [12] and were downloaded from various websites as well as Facebook pages that were associated with autism. There were minor disadvantages in the dataset, since a significant number of the images had low quality or consistency with facial alignment, location, or image size.

A Python script is developed in this study, and it automatically crops images so that the input dimensions of facial images was 224 x 224 x 3. The resulting dataset was categorized into three groups; training, validation, and test groups. A training set for model training was used, and it contained a total number of 2536 images that comprised (86.38%) of the total images, 1268 of the images were images of ASD-diagnosed children, and 1268 of the images were images of neurotypical children. The validation set comprised 3.41% of the total images, and it was contained 50 images of ASD-diagnosed children and 50 images of neurotypical children. And the test set comprised 10.22% of the dataset, and the number of autistic and neurotypical children was 150 each.

3.1.2 Improved Deep-Learning-Based Framework for Autism Detection

This framework was used to aid the system in analyzing facial patterns of children using various machine learning methods through the following steps:

- **Data Acquisition:** The primary dataset was gathered and cleaned.
- **Training:** the training dataset was used to train a Convolutional Neural Network (CNN), pretrained CNN models, and other machine learning classifiers.
- **Performance of Validation and Test Set:** Here, various classifiers in the validation set were used to assess the performance of training activities. Baseline classifiers were also implemented into the test set and to evaluate the performance of classifiers.
- **Evaluation Metrics:** various metrics were used in the evaluation process, such as accuracy, area under the curve (AUC), f-measure, g-mean, sensitivity, specificity, fall-out and miss-rate.
- **Comparison and Evaluation:** After evaluating the performance of individual classifiers in the validation and test set, an improved MobileNet-VGG16 showed the best result among all other classifiers.
- **Clustering-based Autism Sub-typing:** This reduces data dimensionality and wraps strong and significant features that can appear in multiple clusters [13]. This decreases the number of false-positive and false-negative results of autism for further supervised learning tasks. K-means algorithm was implemented into only the images of children with ASD, because k-means algorithm is used in different areas in machine learning to generate the number of clusters from the working dataset [14]. Images of children with autism were gathered from training, validation, and testing sets, and generated numerous autism sub-types in each iteration by changing the values of k from 2 to 10.

3.1.3 Baseline Classifiers

Multiple classification algorithms in the primary facial datasets were implemented. The proposed framework uses 17 classifiers, 10 of which are machine learning models [15], and the remaining are pre-trained transfer learning models such as Adaboost [16], Decision Tree (DT) [17], Gradient Boosting (GB) [18], K-Nearest Neighbor (KNN) [19], Logistic Regression (LR) [20], Multi-layer Perceptron (MLP) [21] [22], Naïve Bayes (NB) [23], Random Forest (RF) [24] [25], Support Vector Machine (SVM) [26], Gradient Boosting (XGB) [27], Convolutional Neural Network (CNN) [28], and pre-trained Convolutional Neural Networks: DenseNet121 [29], ResNet50 [30], VGG16 [31], and MobileNet-V1 [32]. The results of default pre-trained models were not promising initially, then several additional layers were appended in each of the models. In every transfer learning model, three batch normalization (BN) and two fully connected (FC) layers are appended one after another before the output layer to classify facial images into the autistic and non-autistic groups, the BN layers therefore kept their default setting. Additionally, the first FC layer is applied with 128 neurons and a second FC layer was employed with 16 neurons. These classifiers are called baseline classifiers.

3.1.4 Improved Convolutional Neural Network VGG16

An improved version of the MobileNet-VGG16 [33] was used in our study. This model has been used to have additional layers to increase the performance of the traditional MobileNet-VGG16 model. The batch normalization layer normalizes the output of global average pooling layer by re-centering and re-scaling input values. It also appends three batch normalization (BN), two fully connected (FC) layers one after another before the output layer. Regarding the dimensions, the primary facial images had their input dimensions as 224 x 224 x 3. With the input filter dimensions as 3 x 3 x 32 when the improved MobileNet-VGG16, the dimensions of input images are reduced according to the regular conversion of MobileNet-VGG16 depending on the depthwise separable convolution operation. It also generates one-dimensional output for the given input image, depending on the number of classes to be predicted. (See Figure 1)

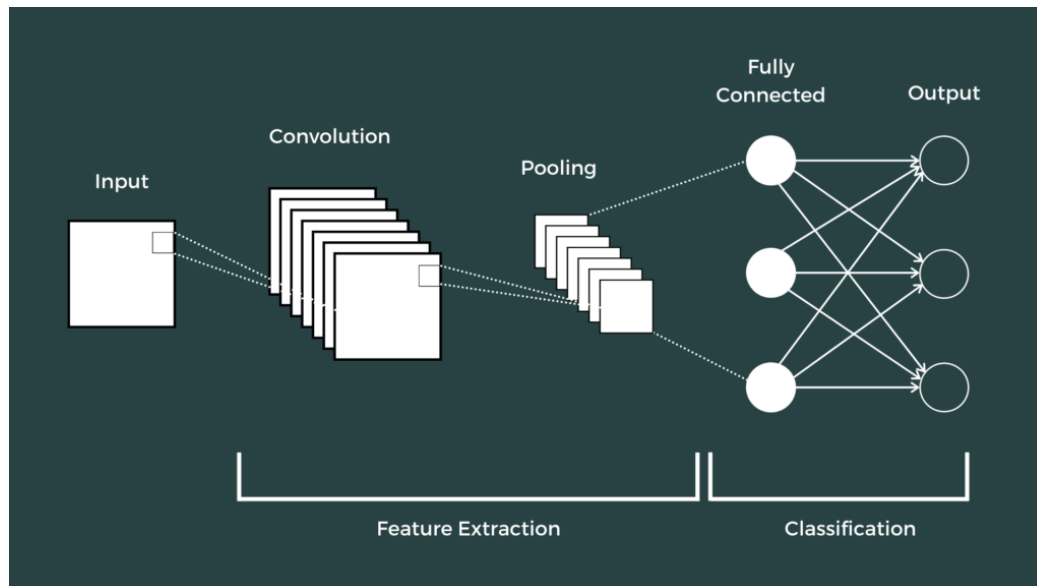


Figure 1: Architecture of Convolutional Neural Network CNN [Source: Google Colab/ CNN explanation]

3.2 Identifying ASD in patients through symptoms

In this section, a dataset [34] related to autism screening of adults was used and contained 20 features to be utilized for further analysis especially in determining influential autistic traits and improving the classification of ASD cases. In this dataset, ten behavioral features (AQ-10-Adult) were recorded, plus ten individual characteristics that have proved to be effective in detecting the ASD cases from controls in behavior science.

The 10th scores included in the dataset are officially used by National institute for health research and published on a website as a guaranteed test for ASD and ASD levels diagnosis including:

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

3.2.1 Code

While the answers for the questions are (yes, no, agree, disagree), each of the converted to 0,1 binary language so it can be workable by the computer.

Then, the data were separated to features (scores and other patient data) and target (autism). All the data preprocessed by Standard Scaler method is built-in in python.

Furthermore, the Decision tree algorithm was called from model selection library; all the processed data automatically distributed into DT field then after 12 minutes of progressing, the model finished the prediction with excellent results according to the metrics calculations.

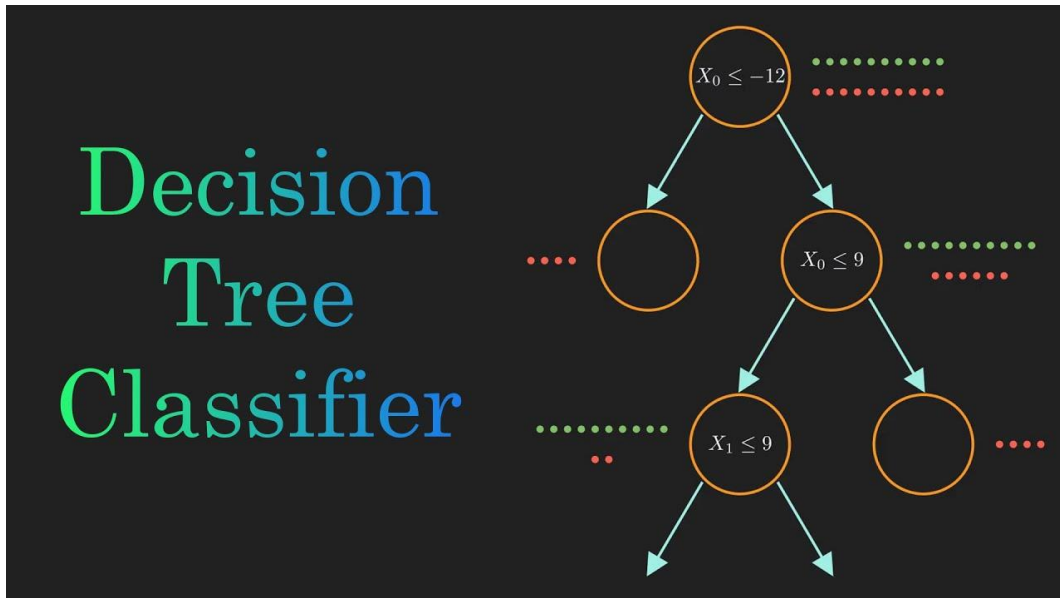


Figure 2: Architecture of Decision Tree. [Source: Scikit-learn.org]

3.3 Results

Our system showed great results in telling between children with ASD neurotypical children. This system can be used in developing-countries to aid short-staffed medical personnel, or for various other reasons.

The model showed results of: overall accuracy, which shows the rate at which the model was accurate. Precision, which is the accuracy of the model in predicting a specific topic. Furthermore, recall is the ability of the model to detect a specific topic. And F1-score, combines precision and recall using the following formula:

$$F1\text{-score} = (\text{precision} * \text{recall}) / (\text{precision} + \text{recall})$$

3.3.1 Results for The Facial Recognition Model

Individual classification models were trained by the training set, the validation set, and the test set, to evaluate the performance of the classifiers. The performance of these classifiers was measured by measuring evaluation metrics like sensitivity, specificity, g-mean, f-measure, accuracy, AUC, fall-out, and miss rate.

The metrics were:

- Overall accuracy: 0.854167
- Accuracy per epoch: 0.8854
- Precision: 0.8697431
- Recall: 0.80007
- F1-score: 0.83236

3.3.2 Results for The Symptoms-Based Model

The results that were acquired were: accuracy, precision, recall, and f1_score. They are shown below:

- Accuracy: 0.900709219858156
- Precision: 0.8235055724417426
- Recall: 0.900709219858156
- F1-score: 0.8603789562824178

3.3.3 Discussion

Both of the research techniques that were proposed in this research show promising results that would have a significantly-important uses in the real world. The accuracy levels were high (85% for the facial imaging technique, and 90% for the symptom-analysis technique) although the accuracy level of the symptom-based system was higher. Other metrics also prove that both models are good.

Moreover, although both techniques showed good results, more complex and larger databases with the aid of different Deep Learning models might aid in providing better results [25]. Furthermore, using 3D imaging techniques for facial structures might provide better data for the AI systems used [18], and different symptom-acquisition techniques can also have better results.

Chapter 4: Conclusions and future works

In this work, we proposed a well-assembled transfer learning-based autism face-recognition framework, where the improved VGG-16 model shows result of Metrics Performance for The Image Facial Recognition Model which is (85.4% Overall accuracy) in a range of machine learning and deep learning models to identify autism in children more accurately. Later, by k-means clustering method has been applied to autism faces to fabricate various sub-types (depending of the values of k in k-means algorithm) and used improved VGG-16 to predict high accuracy for binary sub-types.

The proposed framework can play a significant role in early autism detection and can be as a useful tool for physicians and health-workers. It can also be used to detect autism without or with less training in the domestic environment. Some limitations are noted in the proposed framework; for instance, a few facial images has been used and their quality is not promising. However, we cannot associate this work with activity recognition/video sequencing/3D images analysis to formulate more auspicious results. In addition, improved MobileNet-VGG16 has not provided more stable predictive performance for greater number of autistic sub-types.

In the future, there is a possibility to diminish these shortcomings by ameliorating this model with more standard facial images and mingled dynamic recognition techniques (e.g., activity/motion picture/video sequence identification) to perceive autism more precisely. Moreover, we will further develop our “improved VGG-16” to have more stable predictive performances for a greater number of autistic sub-types, by boosting multi-class classification tasks to obtain more accurate predictive performance to define autism sub-types.

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