PEOPLES'S DEMOCRATIC REPUBLIC OF ALGERIA
MINISTER OF HIGH EDUCATION AND SCIENTIFIC RESEARCH

UNIVERSITY OF FERHAT ABASS 1
FACULTY OF SCIENCES
Computer Sciences Department

MASTER THESIS

Presented with a view to obtain the Master's degree in Data Engineering and Web Technologies

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Machine Learning and Deep Learning for Autism Spectrum Disorder (ASD) detection

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2024 / 2025

Abstract

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder characterized by difficulties with social interaction, communication, and behavior. Early identification ensures timely intervention and improves quality of life. The present study proposes a two-modality binary classification approach for ASD detection based on facial image data and movement data. The image dataset consists of facial photos of children labeled as ASD or non-ASD. The movement data, captured using a Kinect v2 sensor, comprises 1,259 features per subject derived from 3D joint positions and gait metrics, with a total of 800 samples.

A DenseNet121 convolutional neural network (CNN) was employed for images, achieving 89% accuracy. Grad-CAM was used to provide visual explanations by highlighting important regions in the images. For the movement modality, a Multi-Layer Perceptron (MLP) trained on features learned via an autoencoder achieved 99.38% accuracy, with Shapley Additive exPlanations (SHAP) applied to identify key features influencing model decisions. Finally, a late fusion mechanism combining both models was evaluated, resulting in 88.12% accuracy. Results highlight the effectiveness of unimodal solutions, particularly the MLP with autoencoder, and indicate that multimodal fusion requires further optimization for enhanced overall performance.

Keywords: Autism Spectrum Disorder, Early detection ,Deep learning, Facial images,Movement..

Résumé

Le trouble du spectre de l'autisme (TSA) est un trouble neurodéveloppemental caractérisé par des difficultés dans les interactions sociales, la communication et le comportement. Une identification précoce permet une intervention rapide et améliore la qualité de vie. La présente étude propose une approche de classification binaire à deux modalités pour la détection du TSA, basée sur des données d'images faciales et des données de mouvement. Le jeu de données d'images comprend des photos de visages d'enfants diagnostiqués TSA ou non. Les données de mouvement, capturées à l'aide d'un capteur Kinect v2, comportent 1 259 caractéristiques par sujet, extraites des positions 3D des articulations et des métriques de la démarche, pour un total de 800 échantillons.

Un réseau de neurones convolutionnel DenseNet121 a été utilisé pour les images, atteignant une précision de 89 %. Grad-CAM a permis de fournir des explications visuelles en mettant en évidence les régions importantes des images. Pour la modalité mouvement, un perceptron multi-couches (MLP) entraîné sur des caractéristiques extraites via un autoencodeur a atteint une précision de 99.38%, avec l'application des explications additives de Shapley (SHAP) pour identifier les caractéristiques clés influençant les décisions du modèle. Enfin, un mécanisme de fusion tardive combinant les deux modèles a été évalué, aboutissant à une précision de 88.12 %. Ces résultats soulignent l'efficacité des solutions unimodales, en particulier le MLP avec autoencodeur, et indiquent que la fusion multimodale nécessite une optimisation supplémentaire pour améliorer la performance globale.

Mots Clée: Trouble du Spectre de l'Autisme,Détection précoce, Apprentissage profond,Images faciales,Mouvement.

الملخص

اضطراب طيف التوحد (ASD) هو اضطراب نمائي عصبي يتميز بصعوبات في التفاعل الاجتماعي، والتواصل، والسلوك. يضمن التشخيص المبكر التدخل المبكر ويحسن جودة الحياة. تقترح هذه الدراسة نهج تصنيف ثنائي يعتمد على نوعين من البيانات: صور الوجه وبيانات الحركة. تتكون مجموعة بيانات الصور من صور لوجوه أطفال تم تصنيفهم على أنهم مصابون بالتوحد أو غير مصابين. تحتوي بيانات الحركة، التي تم التقاطها باستخدام جهاز Kinect ،v2 على 1259 ميزة لكل فرد مشتقة من مواقع المفاصل ثلاثية الأبعاد ومقاييس المشي، بمجموع 800 عينة.

تم استخدام شبكة عصبية التفافية من نوع DenseNet121 لتحليل الصور، وحققت دقة بلغت 89%. تم استخدام تقنية Grad-CAM لتوفير تفسيرات بصرية من خلال تسليط الضوء على المناطق المهمة في الصور. بالنسبة لبيانات الحركة، تم تدريب شبكة Perceptron متعددة الطبقات (MLP) على ميزات مستخرجة عبر التشفير التلقائي ،(autoencoder) وحققت دقة بلغت 99.38 % ، مع تطبيق تفسيرات Additive Shapley (SHAP) لتحديد الميزات الرئيسية التي تؤثر على قرارات النموذج. أخيراً، تم تقييم آلية دمج متأخر تجمع بين النموذجين، وحققت دقة بلغت 88.12 % . تسلط النتائج الضوء على فعالية الحلول الأحادية النمط، خاصة MLP مع التشفير التلقائي، وتشير إلى أن دمج الأنماط المتعددة يحتاج إلى تحسين لتعزيز الأداء الكلي .

الكلمات المفتاحية: اضطراب طيف التوحد، الكشف المبكر، التعلم العميق، صور الوجه، الحركة.

Acknowledgements

In the name of Allah, the Most Merciful, the Most Compassionate, We express our gratitude and thanks to the Lord for His infinite blessings and mercy upon us. Without His guidance and help, We would not have been able to complete this research project.

We are also grateful to our supervisor, Dr.Semchedine , for his support and guidance throughout the research process.

We would like to extend our warm thanks to our families for their unwavering support and encouragement. May Allah subhanahu wa taala bless them all with happiness and success in this world and the Hereafter.

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LIST OF ABBREVIATIONS

1D CNN	One-Dimensional Convolutional Neural Network
ABIDE	Autism Brain Imaging Data Exchange
ADOS	Autism Diagnostic Observation Schedule
AI	Artificial Intelligence
AR	Augmented Reality
ASD	Autism Spectrum Disorder
ASQ	Autism Screening Questionnaire
CAD	Computer-Aided Diagnosis
CNN	Convolutional Neural Network
CSV	Comma-Separated Values
DL	Deep Learning
DNN	Deep Neural Network
DSM-5	Diagnostic and Statistical Manual of Mental Disorders, 5th Edition
ECARS	Early Childhood Autism Rating Scale
EEG	Electroencephalography
FC	Fully Connected
fMRI	Functional Magnetic Resonance Imaging
Grad-CAM	Gradient-weighted Class Activation Mapping
GRU	Gated Recurrent Unit
KNN	K-Nearest Neighbors
LR	Logistic Regression
LSTM	Long Short-Term Memory
MADE-X	Our modele name Multimodal ASD Detection with Explainability via Late Fusion
MEG	Magnetoencephalography
ML	Machine Learning
MLP	Multi-Layer Perceptron

mIoU	Mean Intersection over Union
NB	Naive Bayes
PCA	Principal Component Analysis
PET	Positron Emission Tomography
RBF	Radial Basis Function
ReLU	Rectified Linear Unit
RGB	Red, Green, Blue
RNN	Recurrent Neural Network
ROI	Region of Interest
SHAP	SHapley Additive exPlanations
SVC	Support Vector Classifier
SVM	Support Vector Machine
sMRI	Structural Magnetic Resonance Imaging
VR	Virtual Reality
WHO	World Health Organization
XAI	Explainable Artificial Intelligence
XGB	Extreme Gradient Boosting

GENERAL INTRODUCTION

Autism Spectrum Disorder (ASD) is a complex neurodevelopmental condition that affects millions of people worldwide. It is characterized by persistent difficulties in social interaction, communication (both verbal and non-verbal), and repetitive or restricted behaviors. ASD is not a single disorder but a spectrum, with symptoms and severity that vary greatly from one individual to another. This variability makes it extremely difficult to diagnose and understand. While some signs of autism may appear during early childhood, many cases are not identified until later in life, which limits the opportunities for timely and effective intervention.

Early detection of autism is critically important. Research has shown that early intervention, particularly before the age of five, can significantly improve cognitive, social, and emotional development in children with ASD. It can also reduce the long-term need for specialized services. Despite this, early diagnosis remains a major challenge. Current diagnostic methods rely heavily on clinical assessments, standardized behavioral observations, and interviews conducted by trained professionals. These methods, although scientifically validated, are often expensive, time-consuming, and not always accessible, especially in rural or under-resourced settings. The reliance on human expertise also makes the process subjective and potentially inconsistent across evaluators.

In response to these limitations, recent advancements in Artificial Intelligence (AI) have opened new possibilities. In particular, machine learning (ML) and deep learning (DL) technologies have demonstrated their ability to process and learn from large, complex datasets. These techniques can identify subtle patterns in data that are difficult or impossible for humans to detect. This is particularly relevant to ASD, where early signs might be reflected in minor facial expressions, body movements, or social behaviors captured in images or videos. AI can act as a powerful support tool in the diagnostic process, offering automated, fast, and scalable solutions that complement the work of clinicians.

One promising direction is the development of intelligent systems that combine different sources of data, such as static facial images, video recordings, motion sequences, and behavioral cues, to detect signs of autism. Convolutional Neural Networks (CNN) are well-suited for analyzing visual data like facial features and expressions,while Multi-Layer Perceptrons (MLP) and autoencoders are effective for processing structured data such as body movement features and behavioral metrics. When combined in a multimodal framework, these models can enhance diagnostic accuracy by capturing more dimensions of autistic behavior. Moreover, explainability tools such as GradCAM and SHAP can be integrated to interpret model outputs and increase trust among healthcare professionals.

The objective of this thesis is to explore and implement an AI-based approach using both ML and DL models to detect ASD in an automated, reliable, and early way. The system is designed to work with multimodal data (visual and behavioral), leveraging the strengths of each modality and ensuring high performance and interpretability. The study also includes a real-world immersion in a pediatric psychiatric center, providing qualitative insight and validation of the model from a clinical and ethical standpoint.

This thesis is organized into three chapters.

- Chapter 1: introduces Autism Spectrum Disorder, covering its clinical features, causes, and the importance of early diagnosis.
- Chapter 2: reviews the technological background, focusing on ML and DL techniques and their applications in ASD detection.
- Chapter 3: presents the original contribution of this research, detailing the development, experiments, and evaluation of a multimodal AI system for detecting ASD.

CHAPTER 1 : AUTISM SPECTRUM DISORDER (ASD)

Introduction

Over the past two decades, there has been a significant increase in the prevalence of autism spectrum disorder (ASD) diagnoses in our population. Autism is one of the most complex developmental disorders, profoundly affecting various aspects of a child's growth, including linguistic, social, and communicative abilities, as well as attention and perception processes. In addition to the individual, families are also impacted by autism, and they are compelled to cope with the situation and strive to provide the necessary support for their child [1].
This chapter offers an overview of autism spectrum disorder (ASD), covering its symptoms and signs, causes and risk factors, and early diagnosis. It also addresses ASD's comorbidities, the interventions and therapies available, and the role of technology in supporting individuals with autism.

1 Definition and overview

Autism spectrum disorder (ASD) is a neurological and developmental disorder that affects how individuals interact with others, communicate, learn, and behave. Although autism can be diagnosed at any age, it is referred to as a "developmental disorder" because symptoms typically emerge in the first two years of life. According to the Diagnostic and Statistical Manual of Mental Disorders (DSM-5), a guide created by the American Psychiatric Association that healthcare providers use to diagnose mental disorders and developmental disorders, individuals with ASD often exhibit:

- Challenges in social communication and interaction with others
- Limited interests and repetitive behaviors
- Symptoms that impact their ability to function in school, work, and other areas of life [2].

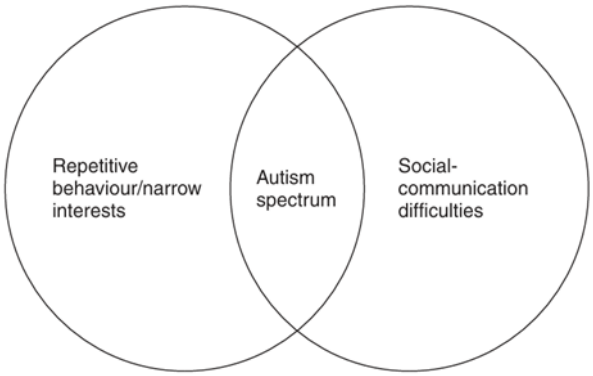

Figure 1.1: Identifying the autistic spectrum as the intersection of two features, i.e., those individuals who show both features.

[2]

Autism is known as a “spectrum” disorder because individuals with autism exhibit a range of characteristics, needs, strengths, and challenges. People of all ages, races, ethnicities, genders, and economic backgrounds can be diagnosed with ASD. Although ASD can be a lifelong disorder, treatments, services, and supports can enhance a person’s health, well-being, and daily functioning. The American Academy of Pediatrics recommends that all children undergo screening for autism. Caregivers should discuss ASD screening or evaluation with their child’s health care provider.

1.1 Historical perspective on autism

Classic autism is sometimes referred to as Kanner’s autism, named after Leo Kanner, the child psychiatrist who first described these children in 1943. In his clinic in Baltimore, Kanner observed 11 children exhibiting what he called ”autistic aloneness.” They showed such little interest in people that they might as well have been furniture in his office.
Kanner borrowed the term autism from Swiss psychiatrist Eugen Bleuler, who had originally used it to describe schizophrenia. Autism comes from the Greek word autos, meaning “self.” This term was aptly chosen because autism and Asperger syndrome involve profound difficulty in appreciating another person’s perspective, as if one’s view is the only correct one [2].
Simon Baron-Cohen, a professor of psychology and psychiatry and the Director of the Autism Research Centre at Cambridge University, and Patrick Bolton define autism spectrum disorder (ASD) in their book The Facts (Oxford Medical Publications). They state, “Autism is a condition that affects some children from either birth or infancy, and leaves them unable to form normal social relationships, or to develop normal communication. As a result, the child may become isolated from human contact and absorbed in a world of repetitive,

obsessional activities and interests" [3].

2 Prevalence and epidemiology of ASD

Epidemiological studies show that autism spectrum disorders (ASDs) are increasing in prevalence worldwide, largely due to better diagnosis, greater awareness, and improved study methods. A 2019 WHO study documented around 28.3 million cases globally, with more than 600,000 new cases, revealing marked disparities across sex, socioeconomic status, and geographical regions [4].
A large meta-analysis comprising over 74 studies reported a global prevalence rate of 0.6%, with divergence across regions: Asia 0.4%; Europe 0.5%; Americas 1%; Africa 1%; and Australia 1.7% [5].
In Africa, little research exists, especially in the sub-Saharan regions. High prevalence figures have been reported in Egypt and Tunisia for cases of developmental disorders, 33.6% and 11.5%, respec-tively, using M-CHAT [6, 7].
An increase in cases of autism spectrum disorder (ASD) in Algeria has been observed over the last decade. The previous rate was estimated to be around 1 in 10,000, while current estimates suggest that 1 out of 100 children is affected [8]. The Ministry of Health states that about 450,000, or nearly 1% of the total population, are living with ASD in Algeria, which aligns with the growing concern for public health worldwide [9].

3 Causes and risks

The causes of autism have not been clearly and comprehensively understood because there is no specific symptom; rather, there is a range of symptoms that vary in intensity and quality from one child to another.

3.1 Genetic factors

Autism spectrum disorder (ASD) is largely influenced by genetic factors; the likelihood of recurrence in siblings ranges from 2% to 8% [10, 11]. Monozygotic twins exhibit higher concordance rates than dizygotic twins[11], and studies estimate the heritability of ASD to be approximately 50% to 55% [12, 13]. Other genetic disorders, particularly Fragile X syndrome, occur in about 10% of children with ASD [14]. ASD features have been linked to oxytocin gene variations, specifically rs6084258 [15], and chromosomal abnormalities, such as deletions at 15q13.3, are implicated [16]. SHANK3 mutations, contactin-associated protein-like 2 antibodies, and sporadic mutations represent additional genetic contributors [17].

3.2 Environmental risk factors

Autism spectrum disorder (ASD) has been linked to several environmental factors. Pregnancy complications, maternal smoking, valproate exposure, pollution exposure, and nutritional factors are among the known risks [17].
Although results are not always in agreement, the CHARGE study found that individuals living close to highways had a higher prevalence of ASD, which may be related to air pollution from traffic [18]. ASD has been linked to mercury exposure [19], but heavy metal levels in hair samples did not significantly correlate with ASD [20]. Through neurotoxicity and changes in DNA methylation, environmental chemicals may contribute to ASD [17].
Valproate exposure during pregnancy is also well known to raise the risk of ASD [21]. Although the results are discordant, exposure to selective serotonin reuptake inhibitors (SSRIs) during pregnancy can similarly elevate the risk of ASD [22, 23]. Pregnancy-induced diabetes has been recognized as a risk factor that can impair fetal development either through epigenetic modifications or oxidative stress [24, 25]. While no significant relationship between direct maternal smoking and increased risk of ASD [26]is present, maternal secondhand exposure to smoke during pregnancy has been linked to a heightened risk [27]. In addition, ASD has been attributed to conditions of nutritional deficiency, such as decreased folic acid, zinc, and vitamin D concentrations [28, 29].

3.3 Parental age and premature birth

Many studies have demonstrated that older parents and preterm birth are significant risk factors for autism spectrum disorder (ASD), with extremely preterm birth (i.e., before 28 weeks of gestation) shown to have a fourfold increased risk (regardless of familial confounding factors) [30]. Being small for gestational age, which is often associated with preterm birth, has a higher risk of ASD; therefore, both gestational age and fetal growth are likely to be important factors [31].
Concurrently, advanced maternal and paternal age have been found independently to be associated with an increased risk of ASD. Paternal age greater than 50 years was found to have a 1.66-fold increased risk of ASD in offspring, while maternal age between 40 and 49 years exhibited a risk increase of about 15% [32]. A meta-analysis confirmed that for each 10-year increase in parental age, the risk of ASDs increased by 18% for mothers and 21% for fathers based on presumed de novo mutations and cumulative environmental exposures [33].

4 Signs and symptoms of autism spectrum disorder (ASD)

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder marked by distinct, persistent deficits in social communication and interactions, alongside associated limited, repetitive patterns of behavior, interests, or activities. Symptoms emerge early in development and show high variability in expression and severity. Social communication deficits are prominent aspects of ASD, identifiable through a range of characteristic behaviors. Individuals with ASD display unconventional uses of eye contact, such as a refusal to make eye contact or employing it in atypical modes of communication. Those with ASD often lack gestures to communicate, understand feelings, and express emotions. Furthermore, individuals with ASD struggle to form and maintain relationships with peers. The majority of individuals with ASD demonstrate a singular focus in social communication, seeking solitary activities while facing challenges in adapting their actions and behaviors to new environments[34].

One typical manifestation of the disorder is a state of restricted, repetitive behavior, which may include stereotypic motor actions such as hand-flapping, rocking, or spinning objects. Some individuals may also have a strong need for routines and can be very anxious about even slight changes in their environment or daily activities. Another characteristic generally refers to restricted, fixated interests that are abnormal in intensity or specificity; for instance, obsessing over certain subjects like train schedules, numbers, or maps. Furthermore, sensory processing abnormalities are often present. Patients may be hyperreactive or hyporeactive to sensory stimuli, exhibiting high sensitivity to sounds, textures, or lights, or conversely, showing indifference to pain, cold, or heat [34, 35] . Generally, language in children with ASD is abnormal compared to typical developmental patterns; a delay in speech acquisition is commonly observed early in a child's life. Some children may later start using their speech in unusual ways, in addition to being non-verbal or not using spoken language at all. Another characteristic is the repetition of words and phrases from other sources. Even when core language skills are mastered, individuals may struggle with pragmatic aspects of communication, such as converting speech reception into responsive speech, adjusting speech for various contexts, and understanding metaphors, jokes, or irony. These issues extend to broader challenges in establishing and maintaining social relationships[35].

The degree of variability in cognitive functioning associated with ASD is remarkable. Executive functioning is impaired in many individuals, resulting in difficulties with planning, flexibility, and impulse control. Attention is also atypical; individuals may focus intensely on minutiae while missing the broader contexts. Deficits in theory of mind—the ability to attribute mental states to

others—constitute another cognitive hallmark that complicates the interpretation and prediction of others' behavior and emotions. Emotional symptoms often co-occur with ASD. A high prevalence of anxiety disorders, depression, and mood instability complicates the clinical picture and often exacerbates the core autistic symptoms [35, 36].
Signs of ASD are often observable in infancy or toddlerhood. Infants later diagnosed with ASD exhibit diminished attention to social stimuli, including limited eye contact and a reduced response to human voices. A major early indicator is not responding consistently to one's name by 12 months of age. Another early sign includes a limited use of gestures, such as pointing and waving, to communicate with others. Additionally, children with ASD frequently display unusual play behavior, characterized by the absence of pretend or imaginative play skills that typically develop in toddlers with normal development [36].
The key characteristic of autism spectrum disorder is its heterogeneity, particularly regarding the severity of symptoms. Some individuals have profound disabilities in social and communication skills, as well as severely impaired cognitive abilities, and therefore require substantial support in daily life. In contrast, others, especially those with high-functioning autism or previously termed Asperger's syndrome, may achieve a significant degree of independence. Intellectual disability varies widely among individuals with ASD, with 30-40% co-occurring with it. However, some individuals do display "savant" skills, which are extraordinary abilities in very narrowly defined areas like memory, mathematics, art, or music [35, 36]. Such variability calls for diagnostic and intervention approaches that are individualized based on the strengths and weaknesses presented by each individual.

5 Diagnosis of autism spectrum disorder

Evaluating for ASD is primarily a multidimensional process that necessitates thorough evaluation, as symptoms can vary greatly among individuals. Although no medical test can confirm ASD, clinicians base their diagnostic judgment on behavioral assessments, developmental history, and the use of standardized screening tools.
Early indicators of autism spectrum disorder (ASD) may sometimes appear as early as 18 months; however, by age two, a diagnosis from a trained expert is widely accepted. Since ASD lacks a definitive test like a blood test to measure the disorder, clinical professionals base diagnoses on developmental histories, behavioral observations, and standardized screening tools[37].
Knowing developmental milestones is essential for diagnosing autism spectrum disorders (ASD). The CDC provides guidelines to track the social, communica-

tion, cognitive, and physical development of children from birth to five years. While individual children grow at their own rates, these milestones help identify possible delays. Screening does not diagnose ASD, but it may indicate the need for further evaluation by a specialist [38, 39].

Screening tools are an excellent way to help identify children who may be at risk for ASD. M-CHAT-R/F is widely used for children aged 16 to 30 months. It includes a 20-item questionnaire answered by parents or caregivers, which has very high sensitivity and specificity[40, 41].

Another tool is the SCQ, which is specifically designed for children aged 4 to 40 years and includes 40 yes-no questions answered by the caregiver about the child. The Parents' Evaluation of Development Status (PEDS) is a parent interview that assesses a child's development across motor, language, self-help, and many other domains to detect any potential delays [42].

The Childhood Autism Rating Scale (CARS) is a concise rating scale designed for use with children aged two and older. It is based on five well-known diagnostic models of ASD, each of which evaluates a different aspect, ability, or behaviour of the child[43].

More comprehensive techniques include the Autism Diagnostic Observation Schedule Second Edition (ADOS-2) and the Autism Diagnostic Interview-Revised (ADI-R), both of which provide structured evaluations of social behavior, communication, and stereotyped behaviors through direct observation and caregiver interviews [39, 44, 45].

In adults, ASD diagnosis remains complicated due to the lack of universally accepted screening tools. The National Institute for Health and Care Excellence (NICE) in the UK nonetheless recom-mends the use of the Autism Spectrum Quotient, a 10-item screening tool for adults deemed to exhib-it traits of ASD but not for moderate or severe intellectual disabilities, to help ascertain the need for further comprehensive assessment[46].

In addition to behavioral evaluations, genetic testing is often recommended, particularly when there is suspicion of ASD associated with other genetic syndromes. The first-tier genetic evaluations include chromosomal microarray (CMA) and DNA analysis for fragile X syndrome. Karyotyping may also be requested if the patient has a history of multiple miscarriages, as it could identify balanced translocations not visible on CMA[44, 47, 48, 49].

This means that an ASD diagnosis will be more accurate if it utilizes a combination of behavioral evaluations and genetic testing. Such a diagnosis can then be followed by early interventions and support strategies tailored to individuals or their families.

6 Importance of early diagnosis

Early diagnosis of autism spectrum disorder (ASD) helps enhance intervention strategies, thereby improving the development of children across various domains. Research suggests that early identification contributes to enhanced cognitive, language, and social-emotional development [50, 39]. Interventions including ABA and ESDM will be more effective when commenced early, in enhancing social skills, adaptive behaviour, and learning[51, 52].
Families also benefit from early diagnosis through reduced parental stress, improved access to support services, and increased parental participation in therapy[39, 53]. Programs that begin before the age of 4 years significantly enhance children's independence, communication, and social integration [54]. Research indicates greater adaptability and independent functioning later on with early intervention [55, 56].
With increased early identification, individuals can plan for individualized education and care, which may enhance the chances for school readiness and long-term achievement [53, 54]. Such interventions assist the healthcare system in better resource allocation, improving intervention outcome success at the population level. Standardized screening and surveillance systems ensure timely referrals while minimizing diagnostic delays, so children and families receive necessary support at the correct time [39, 50, 54].

7 Comorbidities of autism spectrum disorder (ASD)

Individuals with ASD also experience a variety of comorbidities that do not occur uniformly but instead come together to form distinct subgroups. These subgroups include seizures, multisystem disorders, and psychiatric disorders. Scientists categorize individuals with ASD into two groups: "essential or uncomplicated" and "complex" ASD, each associated with different outcomes and recurrence risks [57].

7.1 Common comorbidities

There are several medical conditions that often occur alongside ASD, especially in individuals with intellectual disabilities. These include epilepsy, psychiatric and behavioral challenges, and gastrointestinal (GI) symptoms.
Gastrointestinal (GI) comorbidities are commonly seen in individuals with ASD and include gastroesophageal reflux disease (GERD), constipation, diarrhea, food allergies, colitis, ulcers, and inflammatory bowel disease. Sleep disorders,

behavioral disorders, and connective tissue disorders, such as Ehlers-Danlos syndrome (EDS), are also frequently associated with GI distress in those with ASD. Another treatable comorbidity of ASD is obesity, which increases the risk of metabolic illnesses like diabetes, hypertension, and cardiovascular disease. Effective weight management and nutritional interventions can help reduce these risks[58].

7.2 Psychiatric conditions

These are also common in individuals with ASD. Some frequently occurring co-occurring disorders include anxiety, attention-deficit/hyperactivity disorder (ADHD), depression, and other mood disorders. These psychiatric comorbidities may significantly impact daily functioning and may require therapeutic interventions [59].

8 Interventions and treatments for ASD

Pharmacological and behavioral interventions are crucial in managing autism spectrum disorder (ASD), addressing both core and comorbid symptoms [60]. Although no known medications treat core autistic symptoms, drugs such as aripiprazole and risperidone are used to reduce behavioral and emotional dysregulation associated with aggression, irritability, and mood instability [61].
Psychotropic drugs have been largely employed among individuals with autism who have intellectual disabilities or other coexisting conditions, such as attention-deficit hyperactivity disorder (ADHD), anxiety, or other mental illnesses[62]. Most of the time, polypharmacy is necessary to treat coexisting symptoms because, for some conditions, neither improvement nor healing can be achieved with a single medication[63].
For instance, metformin, which downregulates the mTOR pathway, has been used alongside a stimulant for ADHD and with a Selective Serotonin Reuptake Inhibitor (SSRI) for anxiety[64].
Behavioral interventions, such as Applied Behavior Analysis (ABA), Discrete Trial Training (DTT), and Pivotal Response Training (PRT), focus on enhancing communication skills, social interaction, and adaptive behavior in the treatment of ASD. While the Early Start Denver Model (ESDM) emphasizes teaching children through naturalistic interactions, the Developmental, Individual Difference, Relationship-Based Model (DIR/Floortime) highlights the social and emotional aspects of development[65].
The Teaching and Educational Approaches for Children with Autism and Other Communication Handicaps (TEACCH) offers a structured teaching method

with visual supports that fosters academics, independence, and communication [66]. Cognitive Behavioural Therapy (CBT) is a therapeutic approach that has shown the most positive results in reducing anxiety and obsessive behaviour among autistic individuals [67, 68].
A comprehensive treatment model (CTM) integrates multidisciplinary approaches over the long term, providing individualized interventions in the home, school, or community. These models are designed to develop language, cognition, and functional skills in young children. Focused interventions target specific skills or behaviors, such as training in social behaviors, toilet training, or cognitive-behavioral strategies that involve prompting, reinforcement, and extinction. Social skills training for adolescents and young adults can assist with emotion regulation and social interactions [69].

9 The role of technology in autism spectrum disorder

Technology plays a key role in enhancing the quality of life for individuals with autism spectrum disorder (ASD). It includes features and elements that can assist those affected in communicating, learning, developing social skills, and fostering independent living.

- Assistive communication technologies:
  An augmented model of syntax and alternative communication (AAC) can be demonstrated through devices and types of assistive technology that enable nonverbal or communication-challenged individuals to express themselves using high-tech communication hubs, such as tablet applications, voice-generating devices, and specialized software. This positively impacts independence and social interactions [70].

- Mobile apps and wearable technologies:
  Mobile applications create personalized intervention schedules, enhancing time management and social skills among individuals with ASD. Wearable technologies, like smart devices, provide real-time monitoring and reminders to assist in improving emotional regulation [71].

- Virtual reality and augmented reality:
  VR and AR technologies have created immersive environments where individuals can practice social and behavioral skills under strict controls. These environments facilitate the safe replication of complicated social situations, helping users generalize skills beyond the immediate environment [72].

- Artificial intelligence (AI) and robotics:
  The application of AI in early diagnosis and the identification of appropriate intervention types is being explored. Social robots represent a new development that shows promise in teaching social skills through predictable and non-judgmental interactions, making them ideal for individuals with ASD [73].

- Educational technologies:
  Digital learning tools, such as serious games and online learning formats, are designed to meet the needs of autistic students. These tools provide customized and interactive learning experiences, enhancing engagement and motivation to learn [74].

Conclusion

In summary, this first chapter has provided a deeper understanding of autism, its symptoms, its impact on child development, and its influence on the family. We have also traced the evolution of diagnosis and scientific approaches that have allowed us to better understand autism spectrum disorder (ASD). The next chapter will provide an extensive background and literature review, covering the basic principles of machine learning and deep learning, highlighting their relevance to autism spectrum disorder (ASD) detection, and providing an overview of previous studies conducted in this domain.

CHAPTER 2 : BACKGROUND AND LITERATURE REVIEW

Introduction

Due to the complexities of their symptomatology and the limitations of conventional diagnostic methods, detecting autism spectrum disorder (ASD) remains a significant challenge. Most screening tools rely on varying degrees of subjectivity, while objective behavioural observation can be time-consuming and costly. In this context, Machine Learning (ML) and Deep Learning (DL) have emerged as viable alternatives, offering scalable, data-driven applications for early and accurate ASD detection. This chapter reviews the key concepts of ML and DL, analyzes their applications in ASD diagnosis, and describes the data sources, problems, and limitations associated with these approaches.

1 Machine learning (ML)

1.1 Definition and basic concepts of ML

Machine Learning (ML) is a subset of artificial intelligence that emphasizes creating programs and mathematical models designed to enable computers to learn from data and make decisions or predictions based on this data without requiring the explicit programming of every specific task.[75]
Various experts define ML as follows:
According to Ethem Alpaydin, programming the computers to optimize their performance based on past data or experience. As per Kevin P. Murphy, it is the creation of methods that detect data patterns and predict future or unknown values. As explained by Christopher M. Bishop, it is a subfield of pattern recognition enabling machines to recognize regularities related to the data and act accordingly[76].
Tom Mitchell said: "A computer program is said to learn from experience E with respect to some class of tasks T and performance measure P if its performance at tasks in T, as measured by P, improves with experience E." [77]

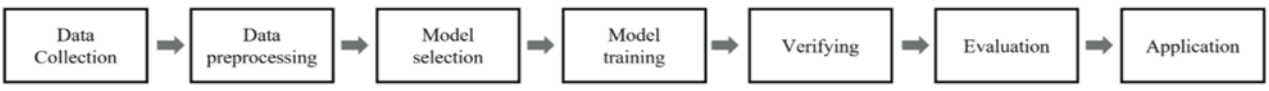

Figure 2.1: Machine learning procedure
[77]

ML relies on two main concepts: representing data in a meaningful way and generalizing learned patterns to new situations. Its applications range from image and speech recognition to medical diagnosis, financial forecasting, and

language processing. Overall, ML allows systems to adapt and evolve based on the information they receive[77].

1.2 Types of machine learning

Machine Learning can be divided into several categories based on the nature of the available data and the learning objective:

1.2.1 Supervised learning

Supervised learning is the training of a model on a labeled dataset, meaning that an output label is assigned to each training example. The purpose is for the model to learn a mapping from input x to output y, so that it can predict the output from the mapping when it is shown some new input it has never seen before. This is used in both classification (predicting categories) and regression (predicting continuous values) tasks [78].

Formally, supervised learning is the task of mapping an input $\mathrm{x} \in \mathbb{R}^d$ to an output label $y \in \mathcal{Y}$, through a function $f : \mathbb{R}^d \rightarrow \mathcal{Y}$ that depends on an independent variable, the training dataset $D = \{(\mathrm{x}_i, y_i)\}_{i=1}^N$ [79].

For example, suppose we are predicting the number of active users on an online platform. The input features may include the number of purchases or user reviews, and the output would be the number of active users in the next month. Supervised learning methods can include:

- Support Vector Machines (SVM): A powerful linear classification tool that separates data classes using a decision boundary [80].
- Naive Bayes (NB): A model based on Bayes' Theorem, assuming independence among features. It is highly efficient for high-dimensional datasets [81].
- Logistic Regression (LR): A statistical model used to predict binary outcomes.
- K-Nearest Neighbors (KNN): A simple instance-based learning method that classifies a data point based on the majority vote of its K nearest neighbors.
- Decision Trees, XGBoost (XGB): Tree-based methods useful for capturing non-linear relationships and addressing overfitting issues [82].

1.2.2 Unsupervised learning

Unsupervised Learning deals with unlabeled data, focusing on identifying hidden structures or patterns within the dataset without any pre-existing labels. This type of learning is best suited for data exploration, dimensionality reduction, or clustering similar data points together[77]. Unsupervised learning typically focuses on the following tasks:

- Clustering: Groups data points so that members of the same group are more similar to each other than to those in other groups. An example is customer segmentation in marketing.
- Dimensionality Reduction: Reduces the complexity of the data by decreasing the number of input variables, often using techniques such as Principal Component Analysis (PCA).

Unlike supervised learning, where correct outputs are specified, unsupervised learning allows the system to form an understanding of the internal structure of the data and to organize it into meaningful patterns [78]. Some common applications include market segmentation, anomaly detection, and recommendation systems.

1.2.3 Semi-supervised learnings

Semi-supervised Learning lies somewhere between supervised and unsupervised learning. It uses a small set of labeled data with a large volume of unlabeled data. This situation is particularly favorable where labeling observations is expensive or tedious, while there is abundant raw data.
In actual practice, a semi-supervised algorithm first trains on the labeled data and then uses this partial knowledge to infer labels for unlabeled data. This now enriched dataset is then used to improve the model's predictions. It is widely used in image recognition, where labeling every image by hand is simply impractical.
The semi-supervised approach, therefore, improves upon the accuracy of supervised learning while keeping intact the scalability and cost-friendliness of unsupervised learning[83].

1.2.4 Reinforcement learning (RL)

Reinforcement Learning (RL) is modeled on behavioral psychology and is based on understanding learning in relation to the environment. In RL, an

agent learns to define its actions within an environment in order to maximize a cumulative reward [79].

In contrast to supervised learning, where a model is trained on known labels, reinforcement learning relies on the consequences of actions for learning. The agent observes the state of the environment, takes an action, and receives a reward or penalty. Over time, the agent learns a policy that maps states to actions in a way that maximizes the expected reward [83].

A typical way to model RL problems is through a Markov Decision Process (MDP), which includes the following main components [79]:

- State: The current situation or configuration of the agent.
- Action: The set of all possible moves or decisions the agent can take.
- Reward: The feedback signal from the environment based on the agent's action.
- Policy: The strategy that the agent employs to decide its actions.

Applications of reinforcement learning are diverse, including playing games (e.g., AlphaGo), robotics, automated trading systems, and adaptive user interfaces.

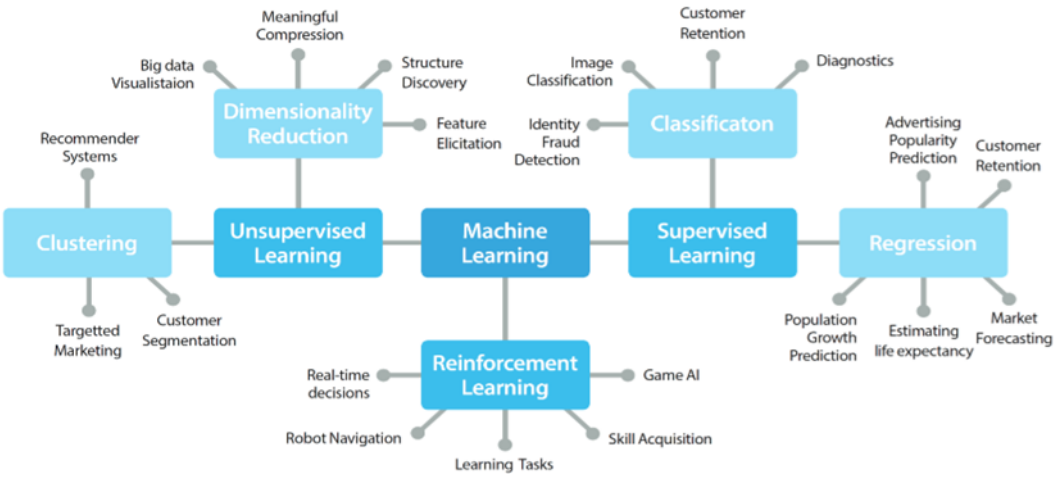

Figure 2.2: Classification of supervised and unsupervised learning [77]

2 Deep learning (DL)

2.1 Definition and basic concepts of DL

Deep learning is a part of machine learning (ML) that uses artificial neural networks with several hidden layers to train hierarchical representations of the

data. Networks learn to model data representation in a manner resembling the structure of a human brain, which consists of approximately 100 billion neurons and a maximum of 100 trillion synapses. The biological neurons send signals through electrical impulses; in a similar but artificial way, neurons in deep learning process inputs, activation functions, and output availability if certain thresholds are met [77]

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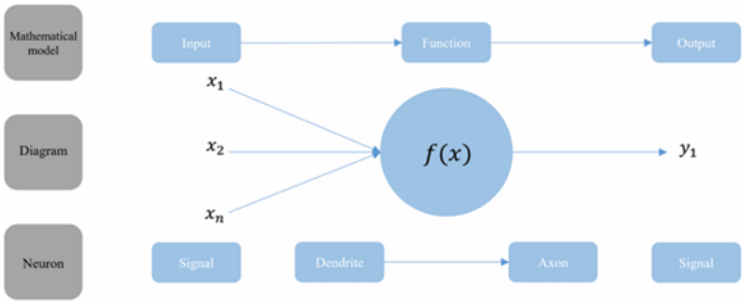

Figure 2.3: Example of neural network modeling [77]

Deep learning or DL processing of data traverses multiple layers, with the lower layers extracting simple features, such as edges or textures in an image, and the deeper layers combining such features into more abstract representations. This constructs a pathway for DL models to learn by themselves high-level abstractions directly from raw data without any upfront input within manual feature engineering [84].

2.2 Evolution of deep learning models

2.2.1 Perceptron

The first model of a neural net is a perceptron, which Frank Rosenblatt introduced in 1958. It comprises only a layer of input neurons that gives the output binary outcome, depending on weighted inputs. This paves the way for further advances in neural network research[77].

2.2.2 Multilayer perceptron

This technique overcomes the limitation of single-layer perceptrons by allowing the implementation of one or more hidden layers between the input and output layers. MLP can learn non-linear functions and handle more complex data than basic perceptrons. The classification of problems gets better with increasing hidden layers in the MLP, particularly for problems such as XOR that simple perceptrons fail to solve[77].

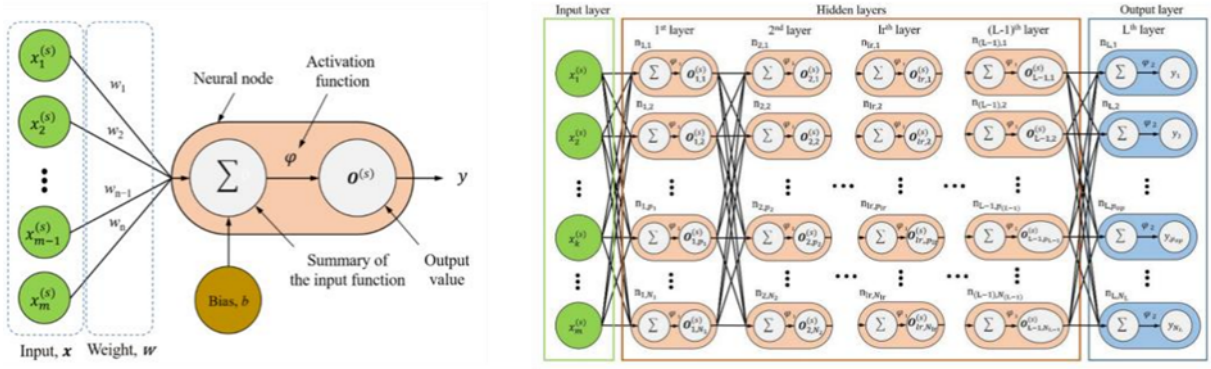

Figure 2.4: (a)Single-neuron perceptron model.(b) Structure of the MLP [85]

2.3 Types of artificial neural networks

Artificial neural networks can take many forms, often made up of multiple complex inputs, directional feedback loops (unidirectional or bidirectional), and various layers. They are generally classified into the following types:

2.3.1 DNN (Deep neural network)

A DNN consists of one input and one output layer, with multiple hidden layers in between. It is essentially a Restricted Boltzmann Machine (RBM), a simplified version of the Boltzmann machine with intralayer connections removed. The RBM calculates weights through a fine-tuning process after initial unsupervised learning. DNNs can be applied even when there is not enough labeled data.

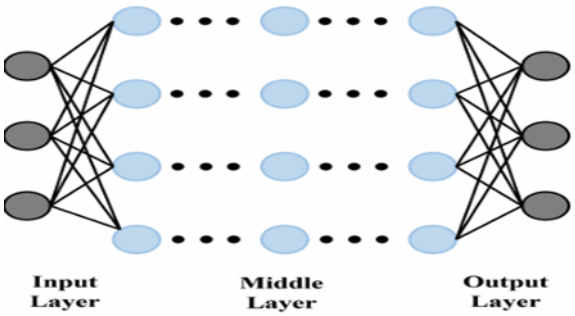

Figure 2.5: Common DNN Model [77]

DNNs adjust individual weights to output desired values for a given input using the backpropagation algorithm, providing slow but stable results [77].

2.3.2 CNN (Convolutional neural network)

CNNs are intended for processing spatial data like images or audio. These networks have convolutional layers that automatically learn spatial hierarchies of features. CNNs are most efficient in the areas of image classification, object detection, and speech recognition since they minimize manual feature extraction and can learn directly from raw data.

- Convolutional layers: Use filters (kernels) like 3x3 or 5x5 to extract features like edges and textures.
- Pooling layers: Downsample feature maps to reduce computational cost and overfitting, while also making features more stable to minor changes in input data.
- Fully connected (FC) layers: Serve as classifiers by connecting the features learned by convolutional layers to the final output neurons.

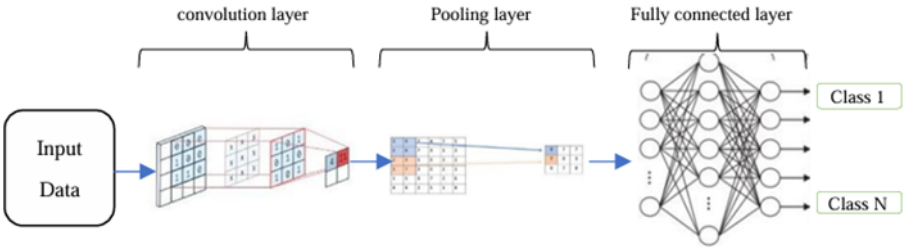

Figure 2.6: The pipeline of a Convolutional Neural Network [86]

Popular CNN architectures: Many CNN variants have been developed to improve learning and performance. Some of the most popular architectures include [86]:

- AlexNet: A breakthrough in deep learning that won the ImageNet competition in 2012. It introduced deep convolutional architectures and was highly successful in image classification tasks.
- VGG: Known for its simplicity and depth, VGG uses small 3x3 filters and deep networks to improve performance. It was a significant step in the evolution of CNNs.
- Inception: Also known as GoogLeNet, this model introduced Inception modules, which allow for more efficient computation by varying the filter sizes within a single layer.

- ResNet: Residual Networks introduced skip connections to allow deeper networks without the vanishing gradient problem. ResNet helped train networks with hundreds of layers effectively.
- MobileNet: A lightweight architecture designed for mobile and embedded devices, focusing on efficiency and reducing the number of parameters while maintaining accuracy.
- DenseNet: Connects each layer to every other layer in a dense manner. This results in a more efficient gradient flow, leading to better performance with fewer parameters.
- EfficientNet: A state-of-the-art CNN architecture that optimizes depth, width, and resolution in a scalable manner, resulting in higher accuracy with fewer resources.

2.3.3 RNN (Recurrent Neural Network)

RNNs are deep learning models for sequential data that maintain internal memory to capture time-based relationships. In processing sequences, results from previous steps are used as input for the next steps. Thus, this architecture becomes more convenient for tasks like language modeling, speech recognition, and video classification. RNNs keep some states, where an internal state is kept to model dynamic temporal characteristics. This means that RNNs are very good for processing sequential data.

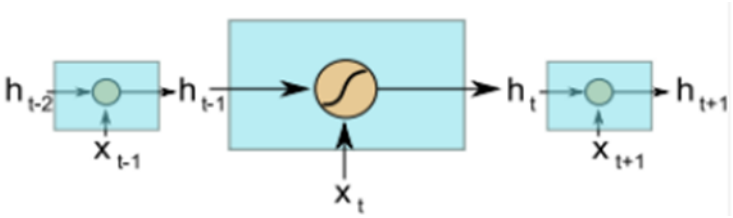

Figure 2.7: Simple RNN internal operation
[87]

Limitations: Basic RNNs commonly find it difficult to handle long-term dependencies due to short memory. To remedy this situation, advanced models for long-term memory have been developed: Long Short-Term Memory (LSTM), Bidirectional LSTM (Bi-LSTM), Gated Recurrent Unit (GRU), and Bidirectional GRU (Bi-GRU). These models try and help the network memorize some of these long-term dependencies, thereby increasing its performance in working with tasks associated with sequential data[86].

- LSTMs: This model was introduced in 1997 and contains three gates, that is input, forget, and output, that control the information entering and leaving each unit. A long-term dependency means these gates will hold information whenever relevant to do so. An S-LSTM is simply an LSTM with a tree structure; this variant can solve problems with more complex tasks, however, at a higher computational cost.
- Bi-LSTM: Extends the capabilities of LSTM by analyzing the sequence in both forward and backward directions. Through this bidirectional approach, it assists in numerous tasks such as natural language processing (NLP) and time-series analysis.
- The GRU is a light version of the LSTM that merges the functions of the input and the forget gates into a single update gate and does away with the separate cell state. Thus, it is more efficient while being able to catch long-term dependencies.
- Bi-GRU: Similarly to Bi-LSTMs, Bi-GRUs analyze data in both directions to facilitate understanding of the context in sequential tasks while remaining efficient.

2.3.4 Deep transfer learning

Deep Transfer Learning (DTL) is a machine learning technique that serves to transfer knowledge from a source domain to a target domain to address the issue of sparsely labeled training data. The primary purpose of DTL is to make use of pre-training so that less data needs to be labeled and less time is spent on training. It is inspired by the possible knowledge transfer humans affect from one skill to another: someone could learn the piano after having mastered the violin [86].
DTL is useful when a large labeled dataset is not available. It has found successful applications in image classification, speech, and video analysis. Some of the common pre-trained models that are used in DTL are Xception, MobileNet, DenseNet, EfficientNet, and NasNet systems, trained on large datasets like ImageNet.
The four major types of DTL are [86]:

- Instance-based: Where instances are selected from the source domain and suitably re-weighted to aid the learning in the target domain (e.g., TaskTrAdaBoost).
- Mapping-based: Where source and target data are mapped into a common feature space for training (MMD, MK-MMD).

- Network-based (Model-based): Where parts of a pre-trained model are reused and adapted to the target domain by freezing, fine-tuning, or adding different layers. Progressive Neural Networks (PNNs) do this while stacking new models on frozen previous ones, simulating human learning.
- Adversarial-based: Where various techniques, such as GAN, are used to discriminate features that are common to both source and target domains.

2.4 Evaluation metrics for deep learning and machine learning models

A handful of metrics are used for performance evaluation of deep learning models [86]:
Accuracy: This measures the prediction's correctness overall.

$$\text{Accuracy} = \frac{TP + TN}{TP + TN + FP + FN} \quad (2.1)$$

Precision: Proportion of true positives against all positive predictions made by the model.

$$\text{Precision} = \frac{TP}{TP + FP} \quad (2.2)$$

Recall: How the model performs well on the identification of true positives

$$\text{Recall} = \frac{TP}{FN + TP} \quad (2.3)$$

Balanced precision and recall measures can be obtained by calculating an F1-score.

$$\text{F1-score} = 2 \times \frac{\text{Recall} \times \text{Precision}}{\text{Recall} + \text{Precision}} \quad (2.4)$$

Specificity refers to "True Negatives." Specificity is calculated using formula :

$$\text{Specificity} = \frac{TN}{TN + FN} \times 100\% \quad (2.5)$$

Where: TP = True Positive; TN = True Negative; FP = False Positive; FN = False Negative.

Generally, higher values of Accuracy, Precision, Recall, F1-score, and Specificity are preferred, with values approaching 1 (or 100%)indicating excellent model performance.

2.5 Deep learning vs. traditional machine learning

Traditional machine learning algorithms often require manual feature engineering, where domain experts extract relevant features from raw data. In contrast, deep learning models learn hierarchical feature representations directly from raw data, reducing the need for manual intervention.
DL models perform better with large-scale datasets and complex tasks, whereas traditional ML models may struggle to generalize in such scenarios.

Category	Machine Learning	Deep Learning
Human Intervention	To achieve outcomes, machine learning requires more continuous human engagement.	Deep learning is more difficult to implement initially, but requires little intervention afterward.
Hardware	Machine learning programs are typically less complicated than deep learning algorithms and may frequently be executed on standard computers.	Deep learning systems necessitate significantly more robust hardware and resources. The increasing power demand has increased the utilization of graphics processing units. GPUs are advantageous due to their high bandwidth memory and thread parallelisms ability to conceal memory transfer latency (delays) (the ability of many operations to run efficiently at the same time).
Time	Machine learning systems can be installed and used quickly, but their results may not be as good as they could be.	Deep learning systems take more time to set up, but they can give results right away (though the quality is likely to get better as more data becomes available).
Approach	Typically, machine learning requires organized data and uses conventional techniques such as linear regression.	Deep learning utilizes neural networks and is designed to handle massive volumes of unstructured data.
Applications	Email, bank, and doctors office all currently utilize machine learning.	Deep learning technology enables more complicated and autonomous programs, such as self-driving automobiles and surgical robots.
Usage	There are numerous applications for machine learning, including regression analysis, classification, and clustering.	Deep learning is typically employed for complicated tasks such as picture and speech recognition, natural language processing, and autonomous systems.
Data	In general, machine learning algorithms use less data than deep learning algorithms, although data quality is more crucial.	Deep learning algorithms require enormous amounts of data to train neural networks, but may learn and improve autonomously as additional data becomes available.

Table 2.1: Comparison Between Machine Learning and Deep Learning [88]

3 ML and DL for ASD detection

Autism Spectrum Disorder (ASD) is a multifactorial neurodevelopmental disorder characterized by deficits in social communication and stereotyped behav-

ioral patterns [89, 90]. The heterogeneous nature of its presentations and lack of a conclusive medical test make it difficult to diagnose. Traditionally, behavioral assessment by trained professionals is still the most relied on method for diagnosis, but it is subjective, laborious, and prone to errors [91]. Thus, the need for improving objective, accurate, and efficient diagnostic tools has arisen, where machine learning (ML) and deep learning (DL) approaches have emerged as transformative solutions.

3.1 Advantages of ML/DL over traditional methods

Machine Learning and Deep Learning models improve the diagnosis of ASD through interpreting complex datasets like brain imaging, genetics, and behavioral patterns that are more consistent and accurate than traditional methods [92, 93]. These models will be enhanced with automated, large-scale screening efforts, particularly in regions with limited resources[94], allowing for continued updating with fresh information and research breakthroughs [95]. According to Nielsen et al. [96], ML/DL techniques uncover hidden correlations among several autism spectrum disorder-related factors, leading to much earlier and even more precise detection compared to fixed-criteria approaches.

3.2 Analyzing complex patterns in large datasets

ML and DL take advantage of high-dimensional data like functional MRI scans, EEG readings, and multimodal data that combines text, images, and videos [92]. These methods identify subtle neurobiological markers of ASD, thereby aiding in the early detection of and personalized interventions for ASD. Advanced models such as CNN and RNN show superior classification accuracy over many traditional statistical techniques [93]. Moreover, the integration of ML/DL into computer-aided diagnostic systems (CAD) presents a cost-effective alternative to traditional diagnostic techniques [97]. CAD methods improve the interpretability of complex brain imaging data and serve toward the automated screening process, hence lessening the dependence on expert evaluation [96]. The emergence of cloud computing and federated learning permits training the ML/DL-based diagnostic models on diverse datasets from different research centers, which enhances generalizability and robustness [95].

4 Machine learning techniques for ASD detection

The field of machine learning (ML) has emerged as an important aid to the detection and classification of autism spectrum disorder (ASD) and includes

neuroimaging as well as behavioural data to improve accuracy in diagnosis. Several ML techniques have been developed by researchers, including traditional, supervised classification models, hybrid approaches, and federated learning, to distinguish ASD individuals from typically developing controls. These advances have markedly improved early diagnosis of ASDs, but at the same time continue to hinder further adoption of the application in clinical settings due to issues associated with variability in datasets, computational demands, and interoperability.

4.1 Neuroimaging-based approaches

Data obtained through neuroimaging, in particular resting-state functional magnetic resonance imaging (rs-fMRI), has been widely utilized to classify individuals with autism spectrum disorder (ASD) based on functional connectivity patterns. The functional connectivity patterns have been classified using machine learning algorithms, allowing researchers to identify certain changes in neural correlates of the disorder between the ASD and control groups.
Binary classification dominates most research on ASD classification, with some studies describing gender differences. Gorriz et al.[98] investigated the classification of ASD from T1-weighted sMRI structural images using Support Vector Machines (SVMs) based on four gender-and-health groupings (male/female, healthy/autistic) to identify neuroanatomical differences. They made gross estimations of gray matter (GM) and white matter (WM) volumes, achieving 69.47% (GM) and 66.16% (WM) accuracy on a private dataset of 60 ASD and 60 HC subjects, aged 18–49 years, using SPM12 and CAT12 toolboxes.
To improve classification accuracy, Fu et al. [99] employed GentleBoost ensembles to classify surface anatomical features of the bilateral hippocampus using ABIDE I data. By applying boosting, subspace, and bagging classifiers, they worked with a dataset of 364 autistic and 381 healthy control participants, aged 6–34, achieving an accuracy of over 80%, although issues with feature interpretability and visualization persisted.
Lastly, the work of Kim et al.[100]aimed to detect early signs of ASD by employing multimodal imaging techniques, combining T1-weighted MRI and DTI data from the National Health and Nutrition Examination Survey (NHANES) and classifying 58 ASD preschoolers (ages 3–6) and 48 typically developing controls (TDC). Their machine-learning model achieved 88.8% accuracy, 93.0% sensitivity, and 83.8% specificity while identifying major biomarkers such as cortical thickness of the right inferior occipital gyrus, mean diffusivity of the middle cerebellar peduncle, and nodal efficiency of the left posterior cingulate gyrus.
To address multi-site classification challenges, Duan et al. [101]used SVM and Random Forest (RF) classifiers on ABIDE's multi-site sMRI framework and

detected gray matter changes in the middle temporal gyrus and angular gyrus, correlating with symptom severity, providing evidence for the promising utility of AI in the neuroimaging analysis of autism spectrum disorder.
Various ML-based classification models were explored by Chauhan et al. [102], who applied boosting, bagging, and neural networks on ABIDE sMRI data (N=740, 344 ASD). The authors identified Stochastic Gradient Boosting Machine (SGBM) as the best-performing algorithm, with 78.87% balanced accuracy, followed closely by RF and Averaged Neural Networks, proving the relevance of ensemble models and multi-feature integration.
Moreover, Bahathiq et al.[103]showed that they could construct an efficient machine learning model for diagnosing ASD using structural MRI data. The feature selection procedure required using either the Support Vector Machines (SVM), Random Forest, or k-Nearest Neighbors (k-NN). The model would produce a result of 94.8% accuracy, thus demonstrating the power of neuroimaging with machine learning in the diagnosis of the disorder and providing a faster and more accurate diagnosis than traditional means.
An explainable machine-learning approach for ASD classification through spatial EEG patterns was proposed by Saranya and Menaka[104]. The children included in this study were 10 with ASD and 10 Typically Developed (TD). The method includes spatial filtering, frequency band identification, and SVM with RBF kernel. The best frequency band is between the alpha and beta ranges (12-16 Hz), achieving an accuracy of 93.59%, a precision of 96.97%, and an F1-score of 95.52%.while Kabir et al. [105] harnessed contrastive machine learning to identify predictive EEG resting-state networks associated with the diagnosis of ASD. They carried out a contrastive variation autoencoder (CVAE) and support vector machine, random forest, deep neural networks (SVM-RF-DNN) approach to classifying age groups for ASD based on EEG functional connectivity. The research utilizes resting-state EEG records from the ABIDE datasets and other available public EEG repositories. Their model shows the greater potential for capturing subtle neural differences that are associated with ASD and classification accuracy greater than that of the traditional machine learning models.
In the study by Manjur et al. [106], they used electroretinograms as a modality for autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD) discrimination using multimodal time-frequency analysis. The sample comprised 286 individuals (146 controls, 94 ASD, 46 ADHD subjects). Data from ERG responses were fed into machine learning models, including support vector machines (SVM), random forest, k-nearest neighbors (KNN), and artificial neural networks (ANNs), to classify the groups based on their ERG responses. Thus, the SVM model achieved an overall accuracy of 70 percent,

obtaining more accurate results by working with higher flash strengths (80-300 Hz frequency). These findings point towards the efficacy of ERG-multimodal (machine learning) bi-omarkers in differentiating ASD and ADHD.

4.2 Behavioral data and feature-based models

The application of machine learning to behavioral data for the detection of autism spectrum disorder (ASD) constitutes an alternative approach to neuroimaging. Behavioral datasets are cheap and easily accessible when compared to neuroimaging and thus become ideal datasets for large-scale ASD screening. Vakadkar et al.[107]studied the Q-CHAT data (Polish Toddlers Q-CHAT, Q-CHAT-10 Responses) concerning ASD classification using different ML techniques such as Logistic Regression (LR), Naïve Bayes (NB), Support Vector Machines (SVMs), k-Nearest Neighbors (kNN), and Random Forests (RF). Their findings indicated strong performance for RF, but recent computations have stated that LR gave a maximum accuracy of 97.15%, followed by NB at 94.79% and SVM at 93.84%, which shows behavioral evaluations are a valid adjunct to the diagnosis of ASD without resorting to expensive imaging techniques.
This research was further taken up by Erkan and Thanh[108]to classify ASD on three datasets, which comprised AQ-10 for children, adolescents, and adults from the UCI repository using kNN, SVM, and RF. The most important finding is that RF produces very good results, achieving 100% accuracy, thus proving its potential as a sound and reliable tool for CD using ML as a diagnostic procedure for detecting ASD.
Akter et al. [109]developed works on ASD classification reliant on Kaggle and UCI repository behavioral datasets; feature transformation techniques such as logarithmic scaling, z-score normalization, and sine function transformation were applied to improve model performance. It has worked to test 250 classifiers, and 9 top models were selected from which Adaboost, FDA, C5.0, LDA, MDA, PDA, SVM, and CART are the few models. All of these have produced classification accuracies above 70 percent, indicating that behaviour datasets have great potential in making ML-based diagnostics of ASD.
The research work of Meng et al. [110]dealt with the early diagnosis of ASD on ML techniques grounded in eye-tracking data. In assessing the gaze behaviour of children with ASD versus typically developing children, the study used videos of cartoon characters as well as real human actors. The overall accuracy achieved by the model which used random forest for feature selection and classification 73%. Results reflected that eye-tracking patterns, such as paying more attention to human-related factors, were highly correlated with diagnosis for ASD and demonstrated the potential application of machine learning in ASD detection.

Simeoli et al. [111]performed systematic research on machine learning techniques that were applied for analyzing motion capture data for the early detection of ASD. The study achieved an accuracy of 90.5% for detecting ASD in children by implementing clinical motion data with Random Forest, SVM, and other algorithms. They indicate that, at the very least, there would be an increased diagnostic precision with motion-based features or provide more insight into the development of machine learning models for early intervention in ASD. While Ganai et al. [112] utilized a pose estimation method by Media Pipe, based on a single RGB camera motion capture system, to examine gait deviations in children with autism spectrum disorder. Step length was significantly reduced in the ASD group, and joint angles were altered in comparison to children with typical development. They applied machine learning algorithms on the ASD-Gait dataset, which consists of motion data obtained from clinical assessment, with binomial logistic regression yielding an 82% classification accuracy between ASD and typically developing children. Thus, their finding offers hope towards using gait analysis combined with machine-learning techniques to facilitate early detection of ASD.
Additionally, Haque et al.[113]investigated the various machine learning techniques in the direction of early detection of autism spectrum disorder (ASD). They worked on Logistic Regression, Support Vector Classifier (SVC), K-Nearest Neighbour (KNN), Decision Tree, and Random Forest, and gave outstanding results using open-source datasets for toddlers and children. The toddler data achieved the maximum mean Intersection over Union (mIoU) of 100% by SVC and 99.80% by Logistic Regression, while the children's dataset reached an mIoU of 100% by SVC and 99.96% by Logistic Regression. Finally, all algorithms reached 100% accuracy on real-world child-age datasets (4-11).

4.3 Hybrid and ensemble learning approaches

Behavioral data serves as an accessible screening tool; however, integrating it with other modalities through hybrid and ensemble learning methods significantly enhances classification accuracy and robustness. Omar and al. [114]created a hybrid ML framework combining Random Forest-CART (Classification and Regression Tree) and Random Forest-ID3 (Iterative Dichotomiser-3) models for classifying ASD. Using the AQ-10 dataset and a real dataset consisting of 250 survey responses collected from individuals with and without autistic traits. It recorded 92.26% accuracy for children, 93.78% for adolescents, and 97.10% for adults with their hybrid model, depicting that combining many ML models indeed boost classification effectiveness.
A study by Sharabash and Elghaish [115]researched the detection of potential signs of ASD and classified the subjects using machine learning tools, in-

cluding Support Vector Machines (SVM), k-nearest neighbors (KNN), decision trees, and Long Short-Term Memory (LSTM) networks on various datasets that consisted of ASD-related data such as eye-tracking data, EEG signals, brain imaging scans, exome sequencing results, and behavioral coding records. Their methodology combined both traditional diagnostic methods with AI predictive models to propose intervention plans that can potentially improve early detection. Their models achieved good classification performance, whereby SVM classified subjects with 95.2% accuracy, LSTM with 96.8% accuracy, and decision trees with 92.5% accuracy, showing the potential of a multimodal approach in ASD classification and early intervention.
Rownak Ara Rasul et al. [116]assessed the role of machine learning techniques in diagnosing ASD at an early stage using data from the Autism Screening Questionnaire (ASQ), Autism Diagnostic Observation Schedule (ADOS), and Early Childhood Autism Rating Scale (ECARS). Their proposed ANN model achieved 94.24% accuracy on a combined dataset, while SVM and Logistic Regression achieved the highest accuracies of 100% for children and 97.14% for adults. The tuning of ML models can enhance the detection of early-stage ASD. Naik et al. [117]have created an effective machine learning algorithm to diagnose autism spectrum disorder (ASD), utilizing datasets such as the Autism Screening Questionnaire (ASQ) and ADOS (Autism Diagnostic Observation Schedule). The hybrid model they utilized combines SVM and Decision Trees and achieved an accuracy rate of 91.3%. The results testified to the power of combining various machine learning techniques to improve the accuracy in an ASD diagnosis, hence stressing the importance of employing diverse datasets towards improving the classification of ASD. Farooq et al. [118] found an improvement in common ensemble learning techniques by integrating multiple Machine Learning models for ASD detection. By developing classifiers trained on both child and adult ASD datasets, they strove to improve the robustness of classification and reduce model bias.

5 Deep learning techniques for ASD detection

Due to its heterogeneity, the precise early detection of autism spectrum disorder (ASD) is not easy. Traditional diagnosis relies on behavioral assessments, which are time-consuming and subjective. New advances in artificial intelligence, particularly deep learning, are very promising in supplementing ASD classification and diagnosis. By utilizing various data modalities such as facial recognition, functional Magnetic Resonance Imaging (fMRI), and Electroencephalography (EEG), deep learning models can provide more precise and scalable diagnostic tools [119].

5.1 Convolutional neural networks for ASD classification

Convolutional Neural Networks (CNNs) have been most prevalently used in ASD detection, particularly with neuroimaging data. Compared to traditional methods such as Support Vector Machines (SVMs) and linear regression, CNNs have demonstrated higher accuracy for classification. From an fMRI data analysis accessed from the Neuroimaging Tools and Resources Collaboratory (NITRC), CNNs registered a maximum accuracy of 97.07% after 13 epochs, a significant improvement compared to the performance of SVMs, which only reached up to 66.36%. These findings emphasize the power of deep learning in ASD classification, and CNNs are a promising tool for automatic diagnosis [119].

In their study of the year 2024, Sharma and Tanwar from the Department of Computer Science, Manav Rachna International Institute of Research and Studies, proposed a deep-learning-based approach for the detection of autism spectrum disorder (ASD) using structural magnetic resonance imaging (sMRI) data. The authors took their data from the Autism Brain Imaging Data Exchange (ABIDE) initiative, which consists of a huge trove of neuroimaging data from diagnosed ASDs and neurotypical controls. The authors made a custom convolutional neural network (CNN) model instead of taking a ready-made pre-trained architecture like VGG or ResNet. The CNN was devised to take as input a set of 3D sMRI images resized to $300 \times 300 \times 3$, which have been generated by the researchers. The network architecture consists of a convolutional layer followed by a Parametric Rectified Linear Unit (PReLU) activation function. This function is known for allowing adaptive learning of activation parameters, followed by a 3D max-pooling layer. The design was intended to enhance the model's effectiveness in extracting discriminative spatial features from volumetric brain data. For the classification, the localization capability of the model was tested with regression metrics yielding a Mean Squared Error (MSE) of 0.30 and a Mean Absolute Error (MAE) of 0.29. This shows that there is little error in pointing out spatial patterns due to ASD in the brain regions Thus, this study shows how a lightweight custom CNN optimized using adaptive activation functions such as PReLU can be a robust tool for detecting ASD from structural neuroimaging. This further adds to the pile of literature documenting deep learning-based diagnostic systems in the field of mental health and neurodevelopmental disorders [120].

Ali and Shaker [121]have proposed a deep learning-based methodology for autism spectrum disorder (ASD) detection by facial features in images. The authors used Kaggle's Autism Face Dataset, which contained images that are coded based on whether or not a person has autism. Their method is based on a Convolutional Neural Network (CNN), which employs transfer learning on the

VGG16 architecture for the automatic extraction and classification of features. The model was trained to identify the faces of individuals who have autism and those without autism. Thus, no manual feature engineering was needed. The output of the model showed the promises of deep CNN in improving the detection of ASD by 94.7% accuracy on this dataset, but with non-trivial challenges of subtle feature variation and data scarcity observed.
Beary et al. [122] proposed a new deep learning model to analyze samples from children, achieving a remarkable accuracy of 94.6% in identifying healthy subjects and those potentially on the autism spectrum. They extracted features using MobileNet and connected them to two dense layers for classification. The study included a balanced dataset of 3,014 facial images from both autistic and non-autistic children. The data was divided with 90% allocated for training and 10% for testing. This research could open new avenues for using facial morphology as an accessible alternative biomarker in the preliminary screening of autism spectrum disorder (ASD).
Jahanara and Padmanabhan [123]investigated autism detection using facial images by employing a hybrid model that integrates traditional feature extraction with deep learning techniques. They utilized Histogram of Oriented Gradients (HOG) features in conjunction with Convolutional Neural Networks (CNNs) to improve performance. The research incorporated transfer learning with the VGG-19 architecture as the foundational model, achieving an accuracy of 96%. This combination of approaches demonstrates a promising advancement in the robustness and generalizability of the system designed for autism detection.

5.2 Transfer learning for ASD detection using facial features

Transfer learning techniques employing pre-trained deep models such as Xception, VGG16, Res-Net50, and DenseNet-121 have been applied to facial image datasets to detect ASD. Xception is a deep CNN with novel inception layers added, utilizing a depthwise convolution layer followed by a pointwise convolution layer. This achieves fewer losses in terms of accuracy. The Xception model employs feature extraction and fine-tuning, utilizing pre-trained weights to access the features of ASD datasets. It consists of two ReLU-activated dense layers (128 and 64 neurons, respectively), a global max pooling layer, batch normalization to prevent overfitting, and an early stopping function for termination during training in case of no improvement in validation loss [121].
ResNet50 is a residual neural network with four main steps, each containing multiple convolutional layers with residual connections that alleviate the vanishing gradient problem. It has 16 residual blocks, fully connected layers, pooling layers, and a Softmax output layer. Residual learning alleviates training complexity by adding the transformed input to its output, rendering deep learning

more efficient for the detection of ASD.
DenseNet-121 consists of 121 layers, which are grouped into dense blocks. Every layer is a connected layer, enhancing gradient flow and feature propagation. Transition layers manage down-sampling through batch normalization and ReLU activation after each convolutional layer. Global average pooling and a Softmax output layer are also used in the model, making it efficient in ASD image classification [124].
Rashid et al. utilized a dataset of 2,940 facial images obtained from Kaggle, achieving an accuracy of 91% via the Xception model, while VGG16 achieved 75%. Similarly, Khan et al. recognized that the DenseNet-121 model outperformed other models with a classification rate of 96%. Other models such as MobileNetV2 and NASNetMobile have also been utilized, with EfficientNetB0 and EfficientNetB7 exhibiting comparable accuracy [125].
The research work conducted by Zeyad A. T. Ahmed et al., published in 2022 in the book titled "Facial Features Detection System to Identify Children with Autism Spectrum Disorder: Deep Learning Models," indicates exploring the possibility of early ASD detection when using facial images through deep learning techniques. The research falls under an emerging field between healthcare and computer vision techniques, leveraging deep learning in challenging applications for neurodevelopmental disorders.
Aspects such as morphology would exhibit specific and dissimilar features that can potentially lead to identifying autism, hence the justification for image-based classification. Their study has references to previous work that relied on machine learning for the prediction of ASD, taking note of the rising trend concerning the use of transfer learning and Convolutional Neural Networks for ultra-high-dimensional image data.
In their approach, the researchers made extensive use of the following pretrained CNN architectures: MobileNet, Xception, InceptionV3.
All these have qualities making them favorite candidates for image classification tasks, as well as their generalization abilities over small data sets due to transfer learning. The input data thus consisted of a total of 3,014 facial images (1,507 from children with autism and 1,507 controls) downloaded from a public Kaggle dataset. Each of these models was fine-tuned on this dataset and evaluated in terms of accuracy concerning the classification of autistic from non-autistic faces.

Figure 2.8: Facial Differences Between ASD and Non-ASD Children [126]

Thus, the model that performed best, MobileNet, gained a validation accuracy of 95%, followed closely by Xception at 94% and InceptionV3 at 89%. Through this model, the authors further realized that deploying it into a web application using Flask makes it available for practical use in screening cases of an initial ASD screening [127].

5.3 Hybrid and multi-modal learning approaches

Several studies have explored hybrid models that combine CNNs with other deep learning models, such as Recurrent Neural Networks (RNNs) and Long Short-Term Memory (LSTM) networks. These methods aim to fuse different modalities, including eye-tracking data, EEG signals, and speech features, to enhance ASD classification. Multimodal fusion approaches that combine EEG, eye-tracking, and fMRI have been particularly effective in improving diagnostic accuracy [128].

6 Data sources for machine learning and deep learning in autism spectrum disorder

ABIDE Dataset:

The Autism Brain Imaging Data Exchange (ABIDE) dataset is the most widely used dataset in the analysis of ASD. It consists of two large datasets [129]: ABIDE I and ABIDE II.

- ABIDE I: Contains data from 17 global sites with 1,112 resting-state fMRI and anatomical datasets. The datasets span subjects aged 7 to 64 years, including 539 ASD patients and 573 normal controls. It demonstrated the possibility of aggregating neuroimaging data across different sources.

- ABIDE II: Expanding on ABIDE I, ABIDE II contains over 1,000 additional datasets with more comprehensive phenotypic characterization, including temporal MRI scans on 38 subjects over multiple years. This dataset explores ASD brain connectivity and heterogeneity in greater depth.

ABIDE Preprocessed: To ensure data consistency between studies, ABIDE provides preprocessed data through well-established pipelines, such as the Connectome Computation System, the Configurable Pipeline for the Analysis of Connectomes, and the Data Processing Assistant for Resting-State fMRI. Structural preprocessing is also available via ANTS, CIVET, and FreeSurfer.

Dataset	**Dataset Type**	**Creation Date**	**Creator**	**Availability**	**Link**
Autism Brain Imaging Data Exchange (ABIDE)	Functional-Magnetic-Resonance-Imaging(fMRI)	2012	Consortium ABIDE	Free	[130]
Autism Diagnostic Observation Schedule (ADOS)	Structured behavioral observation	1989(original-version),2012 (ADOS-2)	Catherine Lord et al.	Paid	[131]
Autism diagnostic Interview-Revised(ADI-R)	Structured interview with parents	1994	Michael Rutter et al.	Paid	[132]
MMASD:A Multimodal Dataset for Autism Intervention Analysis	Optical flow, 2D/3D skeletons, clinical assessment scores	2023	Jicheng Li et al.	Free	[133]
Autism Image Data	Facial images of autistic and non-autistic children	2020	Cihan Özturk	Free	[134]
AUTISM (Roboflow)	Facial images classified as autistic and non-autistic	2024	Gerald Piosenka	Free	[135]

Table 2.2: Publicly Available Datasets for Autism Diagnosis and Analysis

7 Challenges and limitations in ML/DL for ASD detection

Machine learning (ML) and deep learning (DL) are resourceful applications in the detection of autism spectrum disorder. But challenges such as limited neuroimaging datasets, complex algorithms, and hardware constraints hinder their full potential. Addressing these issues is key to improving ASD detection.

7.1 Unavailable MRI neuroimaging datasets of ASD patients

An essential issue concerning AI-enabled systems for ASD detection is the limited availability of neuroimaging datasets with enough variation. Most datasets in use today, such as ABIDE, provide mainly binary ASD-control classifications and primarily use fMRI or sMRI [136, 137, 138]. However, ASD is an extremely heterogeneous disorder, with several subtypes, all having different neurobiological and behavioural features. The lack of datasets that differentiate between these subtypes prevents the ML/DL models from better generalizing across the ASD spectrum, thereby impeding diagnostic accuracy. Additionally, the unavailability of multimodal datasets that intertwine MRI with some other neuroimaging techniques, such as MEG, PET, or EEG, remains a big concern. Such multimodal data could provide a better overall picture of brain function and the anomalies that might be associated with ASD [139, 140, 141].

7.2 Challenges in artificial intelligence algorithms in diagnosing autism spectrum disorder

Efficient and accurate algorithms are essential in making ASD diagnoses through AI. Nevertheless, developing ML-based CAD systems is time-consuming and complex. Algorithm selection is important enough since various methods can effectively extract and classify neuroimaging features differently [142, 143, 144]. In particular, DL models provide automated feature extraction, thereby minimizing pre-processing activities, but demand more data and computational resources. While ML techniques can support DL methods in increasing feature selection, optimizing these hybrid methods is a major challenge. Thus, the absence of standardized methodologies for AI-based ASD diagnosis further contributes to inconsistencies across research findings, hampering clinical implementation.

7.3 Challenges in hardware

Another serious obstacle regarding AI-based ASD detection is its use of high-performance hardware. For instance, public neuroimaging ASD datasets, such as ABIDE, may contain extensive data that renders storage and processing in standard computers [145]. Many researchers face constraints on the computational front, which affects the training and optimization of complex deep learning (DL) models. While cloud computing could resolve most of the hardware constraints, such developments have yet to be integrated into research involving ASD. Recently, deep compact convolutional neural networks (CNNs)

were developed to function using reduced hardware resources while keeping the performance value-adding [146, 147, 148].

Conclusion

This chapter provided a comprehensive background and literature review, establishing the theoretical underpinnings of Machine Learning (ML) and Deep Learning (DL) for the identification of autism spectrum disorder (ASD). It highlighted the potential of these techniques to go beyond the limitations of traditional methods in understanding complex patterns in large datasets. The following text in the subsequent chapter, "Contribution, Experiments and Results," will detail our suggested individual methodology, the experimental environment of hardware and software environment, the validation process with predetermined parameters and measures of evaluation, and detailed analysis of the at-tained results, thus demonstrating the applicability and effectiveness of our contribution provided to the field.

CHAPTER 3: CONTRIBUTION,EXPERIMENTS AND RESULTS

Introduction

This chapter discusses the main contribution of our research, which includes creating and assessing a multimodal deep learning model to detect autism spectrum disorder (ASD) in its early stages. Our strategy integrates facial image analysis through a Convolutional Neural Network (CNN) and behavioral movement information analyzed via a Multi-Layer Perceptron (MLP). We record the pre-processing routines, model structure, training process, and lastly, present and explain the experimental results. Finally, we will merge these two modalities into one for the sake of higher detection precision.

1 Development environment

1.1 Hardware

The experiments were conducted using two personal computers:

- Laptop 1
  - Mark: Hp
  - CPU: Intel Core i7
  - RAM: 8 GB
  - GPU: NVIDIA
- Laptop 2
  - Mark: Hp
  - CPU: Intel Core i5
  - RAM: 8 GB
  - GPU: NVIDIA

1.2 Software

The development and training of models were carried out using the following tools and platforms:

- Google Colab: for GPU acceleration and cloud storage via Google Drive.
- Jupyter Notebook: for local testing and debugging.
- Visual Studio Code (VSCode): as the primary code editor.

1.2.1 Libraries and Frameworks

The following libraries and frameworks were used throughout the project:

- Deep learning
  - TensorFlow / Keras: For building and training deep learning models (e.g., CNN, MLP).
  - Torch / PyTorch TabNet: For implementing the TabNet model on tabular data.
- Machine learning
  - Scikit-learn: For classical ML algorithms (e.g., SVM, Random Forest, Logistic Regression), data preprocessing, PCA, and model evaluation.
- Data handling
  - Pandas: Data loading and manipulation (e.g., Excel, CSV).
  - NumPy: Numerical computations and matrix operations.
- Visualization
  - Matplotlib & Seaborn: For generating plots, confusion matrices, and result visualizations.
- Image processing
  - ImageDataGenerator (Keras): For real-time image augmentation and loading.
  - OpenCV (cv2): For image handling and manipulation in tasks such as Grad-CAM heatmap overlay.
- Metrics and statistical analysis
  - Scikit-learn metrics: Accuracy, precision, recall, F1-score, and confusion matrices.
  - SciPy: Used for statistical testing (e.g., t-test).
- Model explainability
  - SHAP (SHapley Additive exPlanations): For explaining predictions made by tabular models.
  - TensorFlow GradientTape & Keras Model subclassing: Used to implement Grad-CAM for CNN model interpretability.

- Additional tools
  - Google Colab & Google Drive: Used for training with GPU acceleration and data storage.

2 Proposed methodology

2.1 Phase 1: Motion data analysis

In our MADE-X, we used a publicly available dataset titled Three Dimensional Dataset Combining Gait and Full Body Movement of Children with Autism Spectrum Disorders Collected by Kinect v2 Camera [149]. The dataset was designed to capture detailed motion patterns of children using a Microsoft Kinect v2 sensor.
Each subject is represented by 1,259 numerical features, extracted from 3D joint positions, joint angles, and gait-related metrics obtained during structured walking tasks. The dataset was later augmented through transformations to yield a total of 800 instances.
After removing missing values, the class labels were encoded, and the features normalized using StandardScaler. Dimensionality reduction was then performed using Principal Component Analysis (PCA), retaining 95% of the variance to reduce complexity.
We implemented and compared multiple machine learning (ML) and deep learning (DL) models. The traditional ML approaches were as follows:
PCA + MLP; PCA + SVM (RBF kernel); Random Forest; Logistic regression.
The latest DL approaches were:
Autoencoder + MLP: An unsupervised autoencoder was pre-trained to compress motion features into a 64-dimensional latent space, which was then modeled on an MLP.
1D CNN: This CNN model was designed with stacked convolutional layers and pooling layers for spatial extraction automatically across time-series motion features.
TabNet: A deep tabular learning model employing attentive feature selection and sparse regularization.
Each model was evaluated on accuracy and F1-score metrics, with an 80/20 training and testing split. Neural networks were further regularized with Dropout, EarlyStopping, and ReduceLROnPlateau for enhanced generalization.

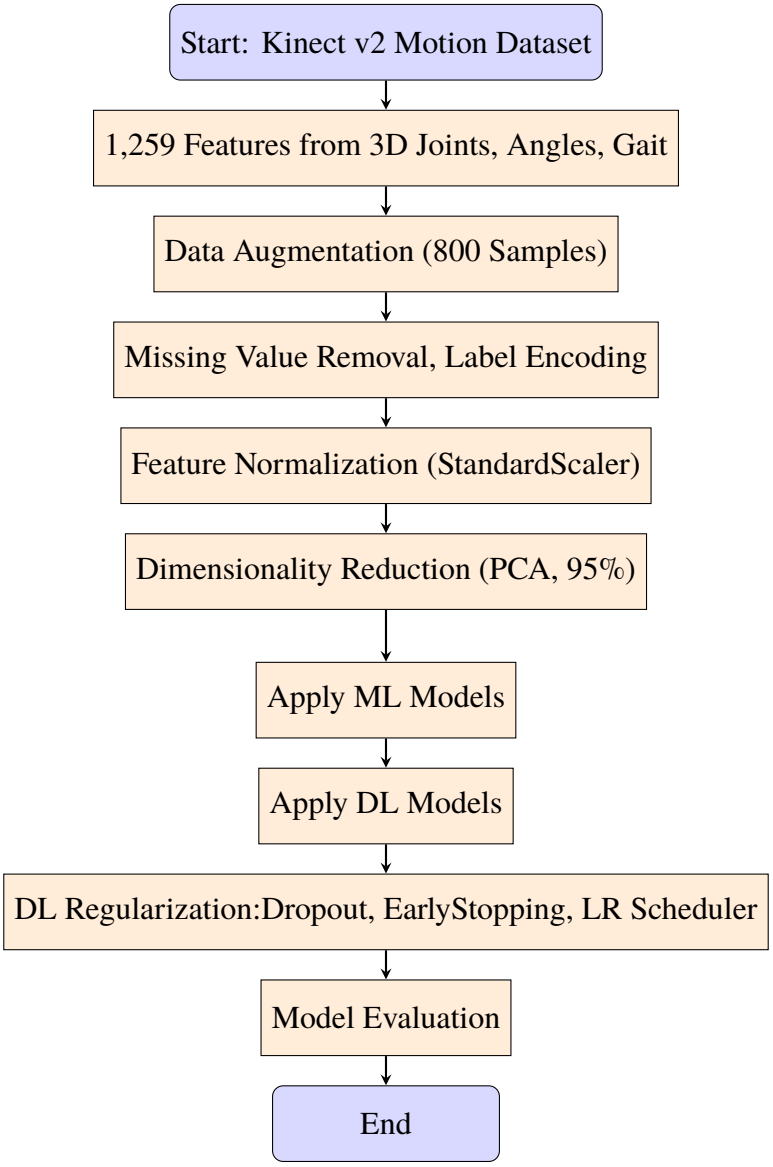

Figure 3.1: Workflow for ASD Detection from Motion Features

Model	**Accuracy (%)**	**F1-Score**
PCA + MLP	98.75	99
PCA + SVM (RBF)	98.75	99
Random Forest	98.75	98.73
Logistic Regression	98.75	99
MLP (without PCA)	98.12	98
Autoencoder + MLP	99.38	99
1D CNN	98.75	99
TabNet	91.88	92

Table 3.1: Model Performance Summary

2.1.1 Discussion and model comparison

These experimental results demonstrate the effectiveness of both traditional machine learning models and deep learning architectures in classifying Autism

Spectrum Disorder (ASD) patients based on gait characteristics. Although all models yielded high performance, certain differences emerged depending on the modeling approach.
As shown in Table 3.1, conventional supervised models including PCA + MLP, PCA + SVM (RBF), Random Forest, Logistic Regression, and MLP without PCA consistently achieved accuracies ranging from 98.12% to 98.75% and F1-scores between 98 and 99. This impressive performance can be largely attributed to a combination of effective preprocessing steps, such as standardization and dimensionality reduction via PCA. Given the very high dimensionality of the dataset (1260 features) relative to its size (800 samples), PCA played a critical role in reducing the curse of dimensionality by retaining the most informative variance while filtering out noisy or redundant features. This preprocessing facilitated a more manageable and accurate classification task.

The best overall performance was achieved using the Autoencoder + MLP combination, which attained an accuracy of 99.38% and an F1-score of 99. Unlike PCA, which captures only linear relationships, the autoencoder was able to learn nonlinear, compressed representations of the input data. This enabled the MLP to better generalize complex underlying patterns in the gait data, contributing to a measurable gain in performance.

The 1D CNN model also performed competitively, with an accuracy of 98.75% ,and an F1-score of 99. While CNNs are generally better suited for time-series or spatial data, their ability to extract local patterns even from standardized tabular gait data allowed for robust performance. Nonetheless, the limited size of the dataset constrained the CNN's potential. With a larger and more complex dataset, the CNN might have yielded even better results.

In contrast, the TabNet model lagged behind, with an accuracy of 91.88% and an F1-score of 92. Several factors may explain this lower performance:

- Dataset size: TabNet's attention-based learning generally requires large-scale datasets to unlock its full potential. With only 800 samples, its capacity could not be fully realized.
- Feature simplicity: The dataset's features are relatively direct and low in hierarchical complexity. TabNet's attention mechanism is most effective in more intricate feature spaces.
- Overfitting risk: The high number of features relative to the small number of samples increased the risk of overfitting to irrelevant patterns.
- Hyperparameter sensitivity: TabNet is particularly sensitive to hyperparameters such as n_steps, gamma, and sparsity regularization. Without optimal tuning, performance may significantly decline.

An important observation is that many models yielded nearly identical performance. This can be explained by the intrinsic nature of the dataset:

- Linearly separable or simple dataset: If the data is well-structured, clean, and the classes are easily distinguishable, many classifiers (even those based on different principles) can achieve similarly high results.
- Binary classification: With only two classes and minimal imbalance, models can easily distinguish between classes, leading to consistent performance across various algorithms.

Finally, the consistently high F1 scores across models reflect a strong balance between precision and recall. This is especially crucial in clinical applications where both false positives and false negatives can have serious consequences for the diagnosis and management of ASD.

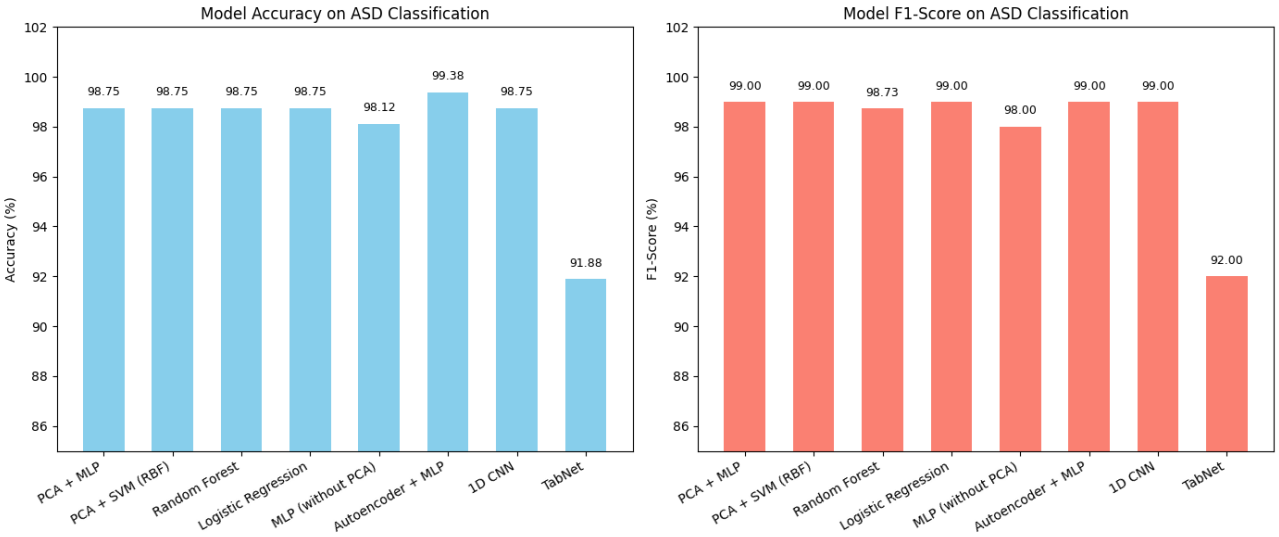

Figure 3.2: model comparison of Motion data

2.1.2 Comparative evaluation with prior studies

The Autoencoder + MLP model reached an accuracy of 99.38% in our experiment MADE-X , a number greater than that reported by Al-Jubouri et al.[150]. In their study called "Generating 3D Dataset of Gait and Full Body Movement of Children with Autism Spectrum Disorders Collected by Kinect v2 Camera," the authors achieved 95% accuracy on the classification between ASD and typically developing children using PCA and MLP. In a further study [151]with the same dataset, called "Gait and Full Body Movement Dataset of Autistic Children Classified by Rough Set Classifier," the authors obtained 92% accuracy with the Rough Set classifier. The superior performance of our model can be explained by the ability of the autoencoder to extract compact and meaningful

representations that preserve non-linear movement patterns much better than PCA .
Moreover, the MLP classifier can capture complex non-linear relationships in the data, unlike simpler models such as the Rough Set classifier. The integration of dropout layers in both the autoencoder and the classifier stages also contributed to regularization, helping to prevent overfitting and improving generalization. Lastly, the preprocessing and normalization techniques applied in our pipeline further improved the model's robustness and accuracy. These improvements together justify the superior performance achieved by our Autoencoder + MLP model.

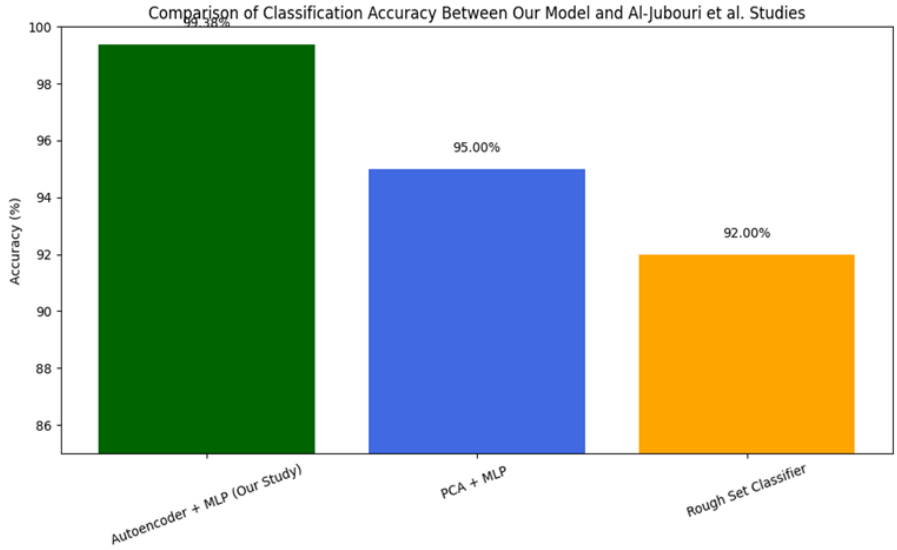

Figure 3.3: Comparison between our model and other studies

2.2 Phase 2: Analysis of image data using deep learning

This second step involves applying Convolutional Neural Networks (CNNs) on facial images collected from publicly available autism datasets [134]. Data preprocessing, comprising resizing, normalization, and augmentation for increasing dataset variability, has been done.
The following models have been used: DenseNet121, ResNet50, VGG16, and Custom CNN architectures.
Each of the models has been trained and validated on a dataset using stratified cross-validation techniques to ensure generalization. Additionally, the best model is selected based on its performance in terms of accuracy and F1-score.

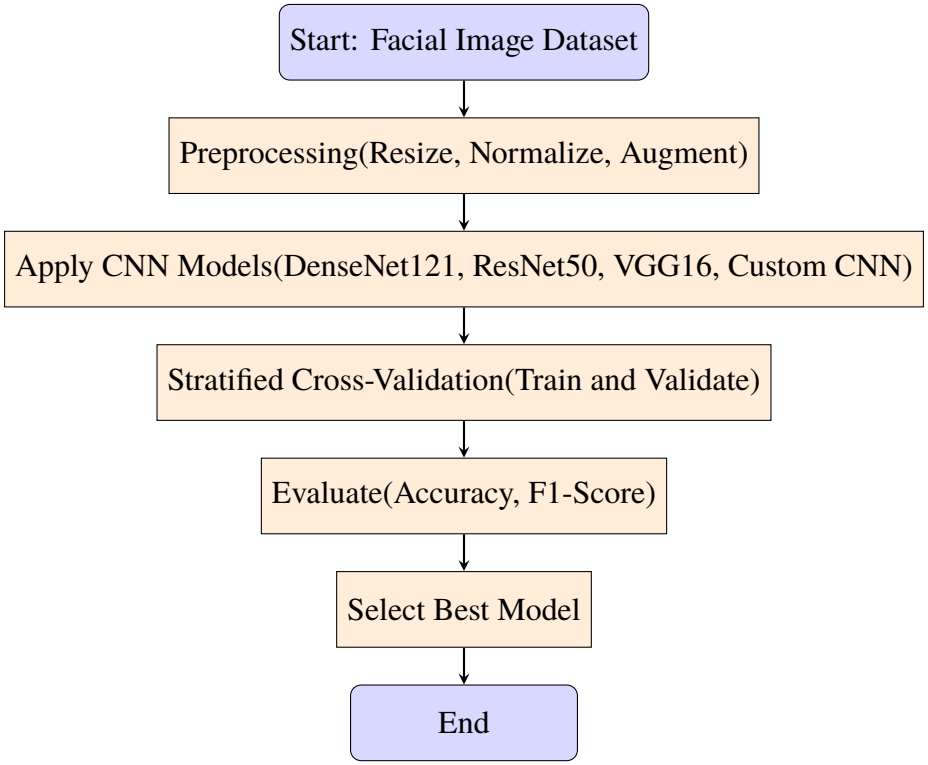

Figure 3.4: Workflow of CNN-based classification on facial images for ASD detection.

Model	**Validation Accuracy**	**F1-Score**
ResNet50	52	68
InceptionV3	85	82
DenseNet121	89	86
VGG19	79	76

Table 3.2: Comparison of Validation Accuracy and F1-Score for Different Models

2.2.1 Discussion of experimental results

Five pre-trained models ResNet50, InceptionV3, DenseNet121, VGG16, and VGG19 were tested for the binary classification problem of autism versus non-autism. Each model was trained and evaluated using the train and evaluate model function, which returned a dictionary of key performance metrics: accuracy, F1-score, recall, and precision. These metrics were displayed in a comparative bar chart, and the validation accuracy over epochs was plotted to observe the trend in learning.

The results show that DenseNet121 performs best overall, with a stable validation accuracy of 89. InceptionV3 follows closely, sitting at 85. Both models manage to learn well from the training data while remaining stable throughout the epochs. Their validation curves describe a linear ascent followed by a plateau, indicating good convergence.

ResNet-50, on the contrary, could only achieve a validation accuracy of 52. Early stopping of pre-trained layers during initial training, fewer training epochs,

or poorly tuned hyperparameters might have caused this relatively lower validation accuracy by limiting the model's ability to adjust adequately to the new dataset during the initial training phases.
VGG19 sometimes peaked at around 80 accuracy, but with significant instability. The validation accuracy curves fluctuate widely, perhaps pointing toward the overfitting nature of these networks or extreme sensitivity to data variations. Without further fine-tuning or some form of heavy regularization, the VGG architectures could be deemed unreliable in this case.
Hence, the experimental results verify that newer and deeper networks like DenseNet121 and InceptionV3 provide generalization and robustness to this binary classification task; thus, they are the best candidates for continued development in this research.

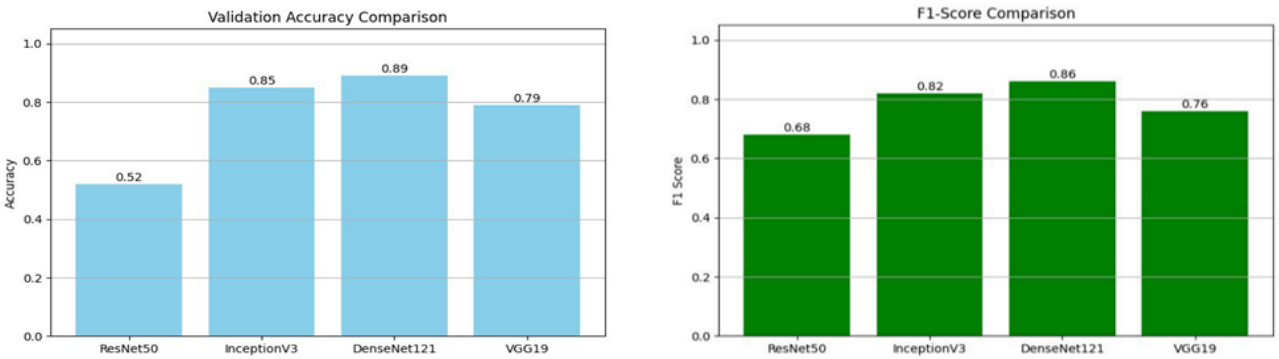

Figure 3.5: model comparison of Facial images data

2.2.2 Comparative evaluation with prior studies

Author	Year	Dataset	Method	Accuracy
Musser [152]	2020		VGG FACE	85%
Beary [122]	2020		MobileNet	94%
Jahanara [123]	2021	Detect autism from a facial image [134]	VGG19	84%
Zeyad [153]	2021		MobileNet	95%
Khan et al. [125]	2023		Dense-Net121	96%
MADE-X(Ours)	2025		Dense-Net121	89%

Table 3.3: Comparison between our model and other studies

Discussion
In our MADE-X , we used the DenseNet121 model to classify facial images for autism spectrum disorder (ASD) detection. This model achieved an accuracy of 89%, showing good performance in recognizing features linked to ASD. DenseNet121 was chosen because of its dense connections between layers, which help in improving feature learning and avoiding the vanishing gradient problem. Compared to other models like VGG-Face (85%) ,and VGG19 (84%), our Dense Net121 model performed better. However, it was slightly less accurate than MobileNet used by Zeyad (95%) ,and DenseNet used by Khan et al. (96%). Still, our model showed strong potential and can be improved further by using more data, better preprocessing, or fine-tuning the parameters.
Overall, DenseNet121 proved to be an effective and reliable model for ASD detection using facial images.

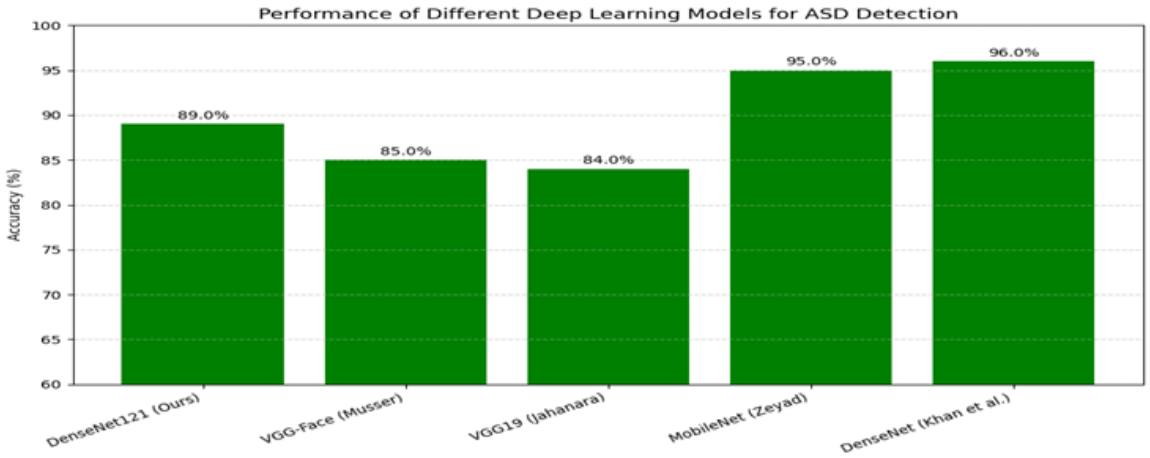

Figure 3.6: Comparison between our model and other studies

2.3 Phase 3: Multimodal learning via late fusion

We propose here a multimodal method that combines two very distinct kinds of data for the detection of autism: facial images and features related to body movements. These data come from two independent datasets and forward different people, thus disallowing fusion strategies of early or intermediate nature. Hence, our choice for late fusion meant that each modality is processed by a model dedicated to it, and these output decisions are then combined.
Since the two modalities are complementary, the rationale carries on. While subtle visual cues related to autism spectrum disorder (ASD) may be presented by facial expressions, body movements through Kinect-based motion analysis can provide kinematic cues often tied to ASD traits. Using both may, therefore, bring in the much-needed edge towards detection accuracy.
To interpret model decisions, we used explainability methods: Grad-CAM for the facial model (highlighting key facial areas like the eyes and mouth), and SHAP for the body movement model (showing the influence of each joint feature). These tools help validate predictions and offer insights aligned with clinical understanding.

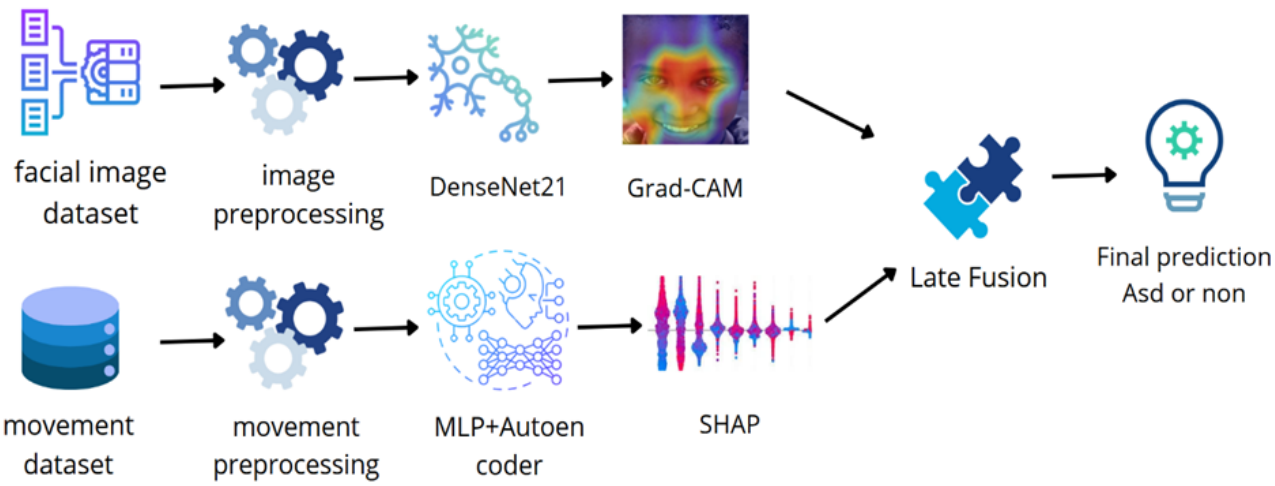

Figure 3.7: Multimodal ASD Detection via Late Fusion

2.3.1 Algorithm of the proposed system

The following pseudocode outlines the main steps of our multimodal deep learning pipeline:

Algorithm 1 Multimodal ASD Detection using Late Fusion + Explainability MADE-X

Require: *ImageDataset* ← Facial images (train/valid/test)
Require: *MotionDataset* ← Handcrafted features from Excel
Require: *fusion_weight* ∈ [0, 1]
Require: *threshold* ∈ [0, 1]
Ensure: *Accuracy, Classification Report, Visual Explanations*

// STEP 1: PREPROCESSING
PreprocessedImages ← PreprocessAndAugment(ImageDataset)
PreprocessedMotion ← PreprocessAndNormalize(MotionDataset)
// STEP 2: FEATURE EXTRACTION
ImageProbs ← DenseNet121(PreprocessedImages)
EncodedMotion ← Autoencoder(PreprocessedMotion)
MotionProbs ← MLP(EncodedMotion)
// STEP 2.1: EXPLAINABILITY
GradCAMHeatmaps ← ComputeGradCAM(PreprocessedImages, DenseNet121)
SHAPValues ← ComputeSHAP(PreprocessedMotion,MLP)
// STEP 3: PREDICTION FUSION
Align lengths of ImageProbs and MotionProbs
FusedProbs ← fusion_weight · ImageProbs + (1 - fusion_weight) · MotionProbs
for all fused_prob in FusedProbs **do**
    if fused_prob > threshold **then**
        Predict 1 (ASD)
    else
        Predict 0 (Non-ASD)
    end if
end for
// STEP 4: EVALUATION
Compute(Accuracy, F1-score, Classification Report using FusedLabels and TrueLabels)
// STEP 5: VISUALIZATION
Display(GradCAMHeatmaps, SHAPValues)
return Accuracy, Report

This pseudocode demonstrates how we integrate the two modalities, process them separately through CNN and MLP, and finally fuse their outputs for classification.

3 Validation Strategy

3.1 Dataset Preparation

3.1.1 Image Dataset

The image dataset [134] consists of faces of children who have been clinically diagnosed with ASD or labeled as non-ASD. These were resized to 224×224 resolution and normalized in the range [0, 1] by division of pixels by 255. Augmentation strategies such as random rotation, zooms, shifts, and horizontal flipping were performed during training to improve generalizability and prevent overfitting.

3.1.2 Motion Dataset

The motion dataset [149] includes behavioral characteristics from video captures of children's movements. The characteristics take into consideration motion properties such as limb positions, velocity, and movement patterns. The data was preprocessed, the missing values were replaced, and it was normalized before splitting into training and validation sets. Class labels of ASD and Non-ASD were labeled using LabelEncoder, and stratified sampling was applied such that class distribution was preserved in the training set as well as the validation set.

3.2 Model Architectures

The late fusion pipeline involves three main stages:

3.2.1 Independent Model Training

Image-Based model (CNN – DenseNet121 + Grad-CAM):

A pre-trained DenseNet121 convolutional neural network is fine-tuned to classify facial images as ASD or non-ASD. The network outputs class probabilities for each test image, which are saved for fusion.
To improve interpretability, we apply Grad-CAM (Gradient-weighted Class Activation Mapping) to visualize the model's attention on the face. Grad-CAM highlights the facial regions (eyes, mouth) that most influence the prediction, offering intuitive explanations for the classification outcome.

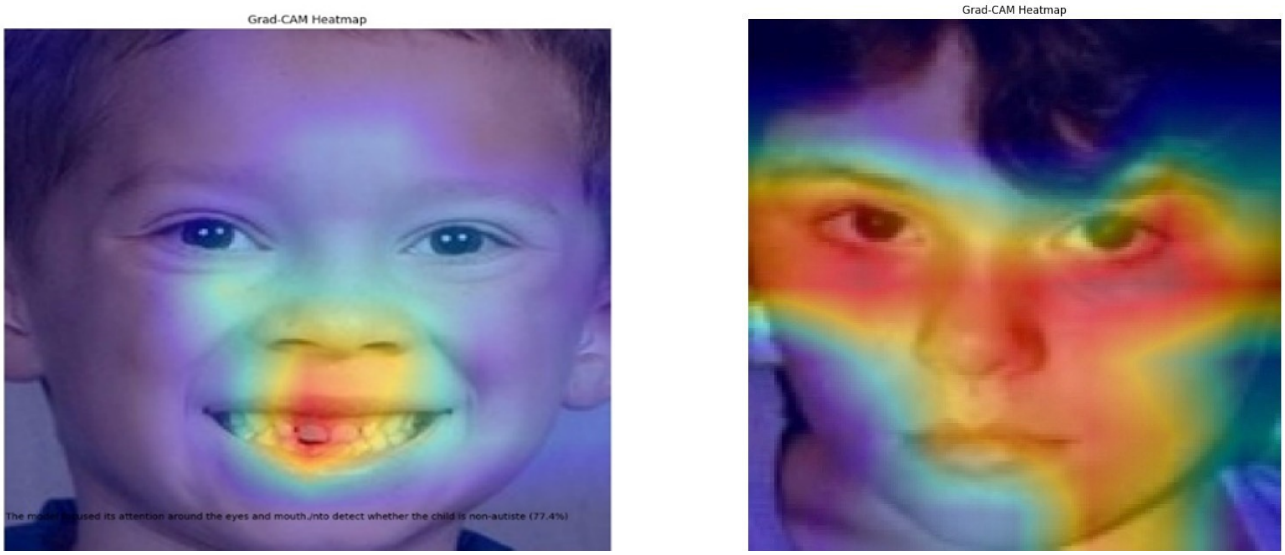

Figure 3.8: Explainable Autism Detection from Facial Features

Movement-Based model (Autoencoder + MLP + SHAP):

The motion dataset consists of 1260 handcrafted features extracted from Kinect sensors. We first apply standard scaling to normalize the features. An autoencoder compresses the high-dimensional data into a compact latent representation. A multilayer perceptron (MLP) classifier then predicts class probabilities from these representations.
To interpret the MLP model's predictions, we use SHAP (SHapley Additive exPlanations) values. SHAP identifies and ranks the most influential body movement features, helping to understand which joint movements (ankle, elbow, midspine) contribute to the classification of ASD or non-ASD.

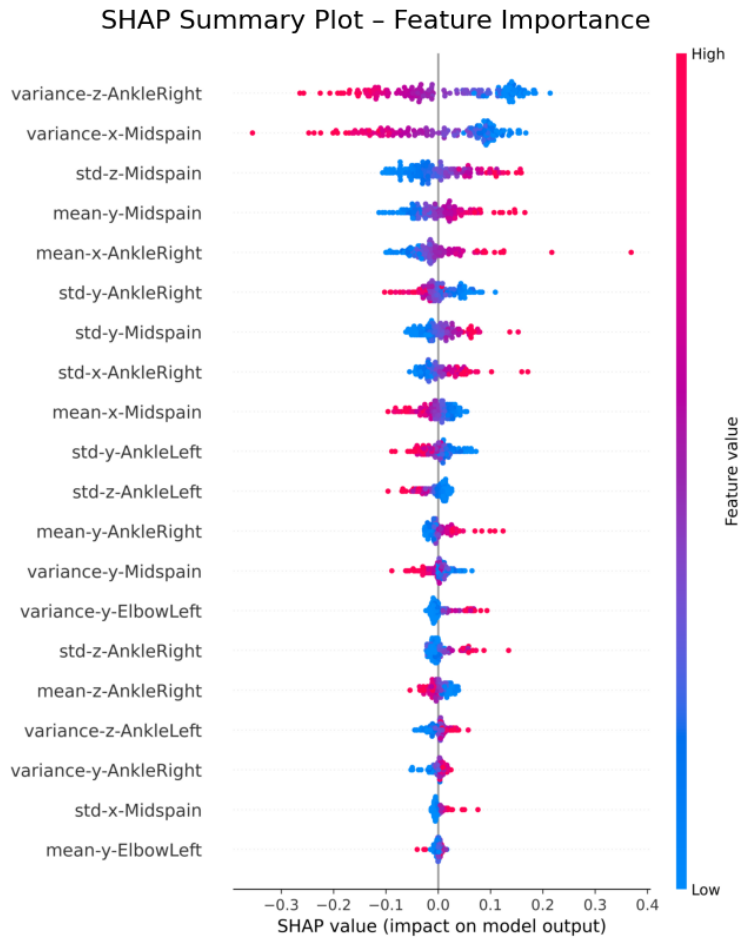

Figure 3.9: Explainable Autism Detection from Body Movements

3.2.2 Probability Fusion

Since the two models are trained separately, we use a weighted average of their output probabilities to perform fusion:

$$P_{\text{fused}} = w \cdot P_{\text{image}} + (1 - w) \cdot P_{\text{motion}} \quad (3.1)$$

where $w \in [0, 1]$ is a tunable fusion weight. We experimented with multiple values of w to identify the optimal balance.

3.2.3 Threshold Optimization:

We evaluated multiple classification thresholds ranging from 0.1 to 0.9 to convert probabilities into binary decisions. For each combination of w and threshold, we computed performance metrics (Accuracy, F1-score) to select the best configuration.

3.3 Experimental Settings

General Settings

Parameter	Value
Platform	Google Colab
Hardware	GPU-enabled (Google Colab)
Libraries	TensorFlow, Keras, Scikit-learn, NumPy, Pandas
Validation Split	20%
Metrics	Accuracy, F1-Score, Precision, Recall, AUC

Table 3.4: General Settings

Image Modality (DenseNet121)

Parameter	Value
Model	DenseNet121 (pre-trained on ImageNet)
Input Image Size	224 Œ 224
Epochs	50 (early stopping at 40)
Batch Size	64
Optimizer	Adam
Learning Rate	0.0001
Data Augmentation	Rotation, Flip, Zoom
Early Stopping	Patience = 5
Reduce LR on Plateau	Patience = 2
Explainability Method	Grad-CAM

Table 3.5: Image Modality Settings

Motion Modality (Autoencoder + MLP)

Parameter	Value
Input Features	Kinect-based motion features (Final dataset.xlsx)
Autoencoder	Input: size of motion features
Encoding Layers	128, 64, 32 neurons (ReLU)
Decoding Layers	64, 128 neurons (ReLU), linear output
MLP Hidden Layers	[128, 64], ReLU activation
Dropout	0.3
Epochs	50
Batch Size	64
Optimizer	Adam
Explainability Method	SHAP (SHapley Additive exPlanations)

Table 3.6: Motion Modality Settings

Fusion Strategy

Parameter	Value
Fusion Type	Late fusion (weighted average of probabilities)
Image Weight	0.7
Motion Weight	0.3
Decision Threshold	0.60

Table 3.7: Fusion Settings

3.4 Results and Analysis

3.4.1 Late Fusion Model

We evaluated several configurations of fusion weights and decision thresholds. The best result was achieved with an image weight of 0.7 and a classification threshold of 0.6.

Class	Precision	Recall	F1-score	Support
0	0.9586	0.9145	0.9360	152
1	0.1333	0.2500	0.1739	8
Accuracy		0.8812		**160**

Table 3.8: Classification Performance Metrics for Late Fusion

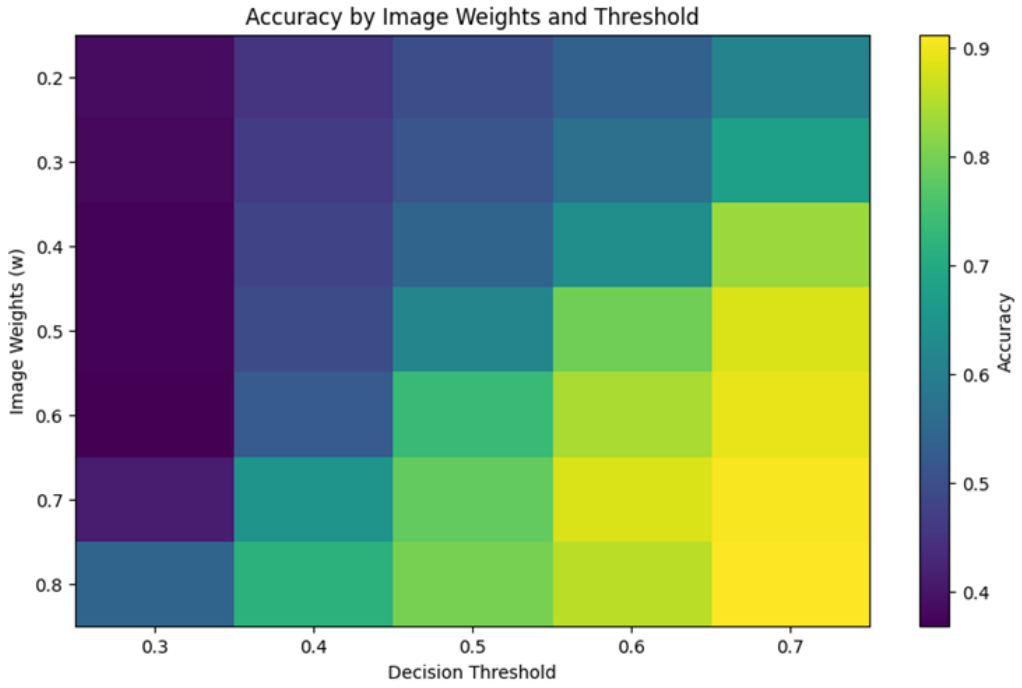

Figure 3.10: Accuracy by image weights and threshold

The point at weight 0.7 and threshold 0.6 achieves a high accuracy of 0.88, which is close to the maximum (0.91), while offering better stability and a

balanced trade-off between sensitivity and precision. Although the highest accuracy occurs at (0.8, 0.7), it leads to overfitting or becomes overly sensitive, making the intermediate choice more reliable and suitable for practical use.

Model	Accuracy
DenseNet121	89
Autoencoder + MLP	99.38
Fusion	88.12

Table 3.9: Model Accuracy Comparison

3.4.2 Discussion

The experiments brought forth the strengths and weaknesses of each individual modality and fusion method. The image-based model of the DenseNet121 portfolio achieved a medium-level accuracy of 89% but suffered from a low recall on ASD cases, reflecting a tendency toward making a non-ASD classification. This also suggests that facial features may not be robust enough as indicators for ASD, especially when image quality and various facial expressions are a little bit different.

Conversely, the motion-based model (Autoencoder + MLP), with an accuracy of 99.38% with fairly balanced precision and recall, shows that body-movement features derived from Kinect do well in capturing behavioral manifestations pertinent to autism. This also goes to show that motor behavior can indeed be an important biomarker in ASD detection.

The late fusion model achieved a slight improvement in overall accuracy to 88.12% by combining both modalities; however, recall for ASD cases dropped significantly to 25%. This was due to a heavy weighting (0.7) assigned to the poorly performing image model, causing many true ASD cases to be missed. This highlights the importance of carefully balancing the contributions of each modality in a multimodal system.

It is important to note that the fusion model's performance improves significantly when the image and motion data come from the same individual that is, when the data are homogeneous. This allows the model to better exploit the correlations between different feature types, resulting in higher detection accuracy and more reliable outcomes.

Overall, the results suggest that multimodal fusion techniques provide a powerful approach to leveraging the complementary strengths of different data types. With further refinement such as using trainable fusion networks that dynamically adjust modality contributions based on input features, or advanced training techniques and balanced datasets fusion models can achieve superior and

reliable performance in ASD detection. This opens exciting future research directions towards developing smart and clinically effective diagnostic systems.

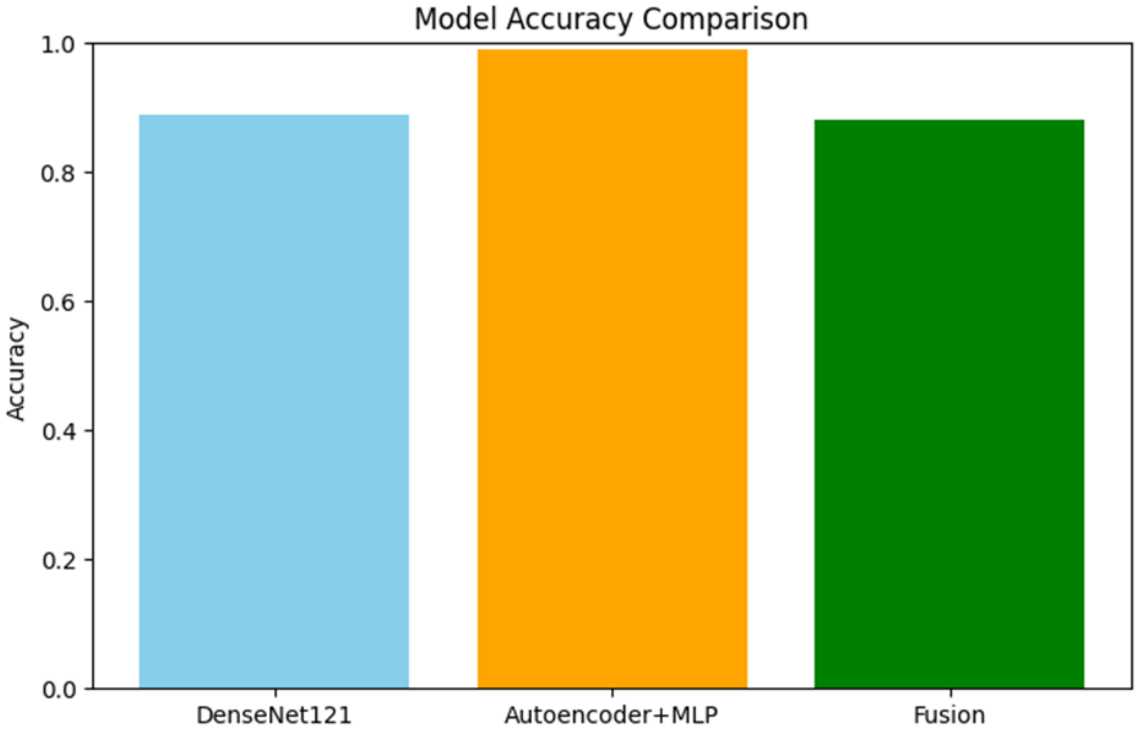

Figure 3.11: Model Accuracy comparison

Conclusion

This chapter outlines our method for identifying autism in facial image data using deep learning techniques. We detail the design of our proposed model, the validation process, and the development environment. The results of the trial demonstrated the effectiveness of our approach and highlighted areas for improvement. These findings provide a robust foundation for further research, especially with the inclusion of additional data sources to enhance classification accuracy.

CONCLUSION AND FUTURE WORK

In this thesis, we explored how machine learning (ML) and deep learning (DL) can improve the early detection of autism spectrum disorder (ASD), a complex condition that affects behavior, communication, and social interaction. Early diagnosis is critical but often challenging due to the wide range of symptoms and the requirement for specialist evaluation. Our goal was to demonstrate how AI techniques could assist in this process by making it faster, more accurate, and more accessible.

We proposed a method with name MADE-X that integrates two types of information: facial photographs and body movement data. To analyze facial features, we utilized a deep learning model called DenseNet121. For movement data, we used multilayer perceptrons (MLPs) and autoencoders. We then brought together the results from both data sources using a late fusion framework and added explainability methods like Grad-CAM and SHAP to make it clearer.

Our method improves diagnostic accuracy and provides visual explanations, with even better results when using facial and movement data from the same individual.

This is just the beginning. In the future, we plan to apply our method to larger, more diverse datasets to improve generalizability. We aim to develop a real-time tool usable in hospitals, schools, or homes for early screening. Adding data sources like voice, eye tracking, or brain scans could boost accuracy, while tracking individuals over time may help evaluate treatment outcomes.

Finally, we believe it is essential to collaborate with medical professionals and caregivers to ensure the tool is user-friendly, ethical, and fair for everyone. In conclusion, we assert that AI has significant potential to aid in the early detection of autism, and through ongoing research and collaboration, we can make meaningful contributions to this vital field.

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