AUTISM SPECTRUM DISORDER SCREENING USING DSM-5 FULFILLMENT AND MACHINE LEARNING ADAPTATION

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DECLARATION

I declare that this thesis entitled "Autism Spectrum Disorder Screening using DSM-5 Fulfillment and Machine Learning Adaptation" is the result of my own research except as cited in the references. The thesis has not been accepted for any degree and is not concurrently submitted in candidature of any other degree.

Signature : ....

Name : Wan Azamudin Bin Wan Zaini

Date : 7/5/2025

APPROVAL

I hereby declare that I have read this thesis and in my opinion this thesis is sufficient in terms of scope and quality for the award of Master of Science in Electronics Engineering.

Signature : ....

Supervisor Name : Profesor Madya Dr. Ir. Syafeeza Binti Ahmad Radzi

Date : 7/5/2025

DEDICATION

I dedicate this thesis to my Dad, Mom, my siblings and also my beloved wife.

ABSTRACT

Autism Spectrum Disorder (ASD) is a complex neurodevelopmental condition characterized by persistent challenges in social communication, restricted interests, and repetitive behaviours. The prevalence of ASD has increased globally, prompting the need for more reliable, objective, and scalable screening and diagnostic methods. Traditional diagnostic tools, such as the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5), remain widely used in clinical settings. However, these tools are inherently dependent on subjective human judgment and clinician expertise, which can lead to inconsistencies in diagnosis and delayed interventions, particularly in early developmental stages. To address these limitations, this study explores a data-driven approach by integrating DSM-5 diagnostic criteria with advanced machine learning (ML) and deep learning (DL) models to enhance ASD detection and severity classification. Two datasets were employed in this research: the Q-Chat-10 online screening dataset, consisting of 1054 toddler data, 104 adolescence data, and 704 adult data samples, used for binary classification between ASD and non-ASD individuals; and the DSM-5 Diagnostic Dataset from Hospital Universiti Kebangsaan Malaysia (HUKM), comprising 177 clinical samples after oversampling, used for multi-class classification of ASD severity (mild, moderate, and severe). Given the imbalance in class distribution, particularly in the severity-level dataset, oversampling techniques were implemented to improve model fairness and performance across all severity categories. The machine learning models evaluated in this study include Support Vector Machine (SVM), Decision Tree, and *k*-Nearest Neighbour (*k*NN). A Deep Neural Network (DNN) architecture was also designed and trained for comparative analysis. Model performance was assessed using standard classification metrics such as accuracy, precision, recall, and F1-score. The results demonstrate that the DNN model outperformed traditional ML models in both binary and severity-level classification tasks. Notably, the DNN achieved 100% accuracy in detecting ASD among younger children, reinforcing its potential as a tool for early screening. Furthermore, the severity classification results showed improved granularity and consistency compared to outcomes generated by manual assessments alone. This research highlights the value of integrating clinical standards with artificial intelligence to improve the speed, accuracy, and objectivity of ASD screening processes. The findings suggest that such hybrid approaches could support clinicians in making more informed decisions, reduce diagnostic delays, and enable timely interventions. Future research should explore larger and more diverse populations, refine model generalizability, address ethical considerations such as data privacy and bias, and assess real-world clinical deployment feasibility.

PENAPISAN GANGGUAN SPEKTRUM AUTISME MENGGUNAKAN PEMENUHAN DSM-5 DAN PENYESUAIAN PEMBELAJARAN MESIN

ABSTRAK

Gangguan Spektrum Autisme (ASD) ialah satu keadaan neuroperkembangan yang kompleks yang dicirikan oleh cabaran berterusan dalam komunikasi sosial, minat yang terhad, dan tingkah laku berulang. Kadar prevalens ASD telah meningkat di seluruh dunia, sekali gus mendorong keperluan terhadap kaedah penapisan dan diagnosis yang lebih boleh dipercayai, objektif, dan boleh diskalakan. Alat diagnostik tradisional seperti Diagnostic and Statistical Manual of Mental Disorders, Edisi Kelima (DSM-5) masih digunakan secara meluas dalam persekitaran klinikal. Namun begitu, alat-alat ini bergantung kepada penilaian subjektif manusia dan kepakaran klinikal, yang boleh menyebabkan ketidakkonsistenan dalam diagnosis serta kelewatan intervensi terutamanya dalam peringkat awal perkembangan kanak-kanak. Bagi menangani kelemahan ini, kajian ini meneroka pendekatan berasaskan data dengan mengintegrasikan kriteria diagnostik DSM-5 bersama model pembelajaran mesin dan pembelajaran mendalam untuk meningkatkan pengesanan ASD dan klasifikasi tahap keterukan. Dua set data telah digunakan dalam kajian ini: set data penapisan dalam talian Q-Chat-10, yang terdiri daripada 1054 data kanak-kanak kecil, 104 data remaja, dan 704 data dewasa, digunakan untuk klasifikasi binari antara individu ASD dan bukan ASD; serta set data Diagnostik DSM-5 dari Hospital Universiti Kebangsaan Malaysia (HUKM), yang terdiri daripada 177 sampel klinikal selepas proses oversampling, digunakan untuk klasifikasi berbilang kelas bagi tahap keterukan ASD (ringan, sederhana, dan teruk). Memandangkan ketidakseimbangan dalam taburan kelas, khususnya dalam set data tahap keterukan, teknik oversampling telah dilaksanakan bagi meningkatkan keadilan dan prestasi model merentas semua kategori keterukan. Model pembelajaran mesin yang dinilai dalam kajian ini termasuk Support Vector Machine (SVM), Decision Tree, dan k-Nearest Neighbour (k-NN). Satu rangka kerja Deep Neural Network (DNN) turut direka bentuk dan dilatih bagi tujuan perbandingan. Prestasi model dinilai menggunakan metrik klasifikasi standard seperti ketepatan, precision, recall, dan skor F1. Keputusan menunjukkan bahawa model DNN mengatasi model ML tradisional dalam kedua-dua tugas klasifikasi binari dan klasifikasi tahap keterukan. Menariknya, DNN mencapai 100% ketepatan dalam mengesan ASD dalam kalangan kanak-kanak kecil, mengukuhkan potensinya sebagai alat untuk penapisan awal. Selain itu, keputusan klasifikasi tahap keterukan menunjukkan perincian dan konsistensi yang lebih baik berbanding penilaian manual semata-mata.Kajian ini menekankan nilai penggabungan piawaian klinikal dengan kecerdasan buatan bagi meningkatkan kelajuan, ketepatan, dan objektiviti proses penapisan ASD. Dapatan kajian mencadangkan bahawa pendekatan hibrid seperti ini boleh menyokong pengamal klinikal dalam membuat keputusan yang lebih tepat, mengurangkan kelewatan diagnosis, dan membolehkan intervensi dilaksanakan dengan lebih awal. Penyelidikan masa depan disarankan untuk meneroka populasi yang lebih besar dan pelbagai, memperhalusi keumuman model, menangani pertimbangan etika seperti privasi data dan bias, serta menilai kebolehlaksanaan pelaksanaan di persekitaran klinikal sebenar.

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LIST OF ABBREVIATIONS

HUKM	-	Hospital Universiti Kebangsaan Malaysia
ASD	-	Autism Spectrum Disorder
ADOS	-	Autism Diagnostic Observation Schedule
ADI-R	-	Autism Diagnostic Interview-Revised
CARS	-	Childhood Autism Rating Scale
GARS	-	Gillian Autism Rating Scale
DISCO	-	Diagnostic Interview for Social and Communication Disorder
3DI	-	Developmental, Dimensional & Diagnostic Interview
DSM	-	Diagnostic and Statistical Manual of Mental Disorder
DNN	-	Deep Neural Network
SVM	-	Support Vector Machine
*k*NN	-	*k*-Nearest Neighbour
TP	-	True Positive
TN	-	True Negative
FP	-	False Positive
FN	-	False Negative
PCA	-	Principal Component Analysis (dimensionality reduction)
AUC	-	Area Under the ROC Curve
ACC	-	Accuracy

LIST OF SYMBOLS

TP_i	-	True Positive for class i (e.g., TP_1 for Mild)
TN_i	-	True Negative for class i (e.g., TN_1 for Mild)
FP_i	-	False Positive for class i (e.g., FP_1 for Mild)
FN_i	-	False Negative for class i (e.g., FN_1 for Mild)
M_{ij}	-	Confusion matrix entry: actual class i, predicted class j
X	-	Input features or data (e.g., QChat-10 responses)
Y	-	Target label or class (ASD, severity class)
$\hat{Y}$	-	Predicted label or output from model
L	-	Loss function (e.g., categorical cross-entropy)
σ	-	Activation function (e.g., sigmoid, ReLU, softmax)
k	-	Number of neighbors in k-Nearest Neighbour (kNN)
C	-	Regularization parameter in SVM

LIST OF APPENDICES

LIST OF PUBLICATIONS

The followings are the list of publications related to the work on this thesis:

1. Azamudin, W.W., Syafeeza, A.R. and Ali, N.A., 2022. A Review of Autism Spectrum Disorder Diagnostic Tools. *International Journal of Integrated Engineering*, *14*(6), pp.329-345.

CHAPTER 1

INTRODUCTION

1.1 Background

1.1.1 Brief introduction of Autism Spectrum Disorder (ASD)

Autism Spectrum Disorder (ASD) is a lifelong neurodevelopmental condition that typically manifests in early childhood and continues across a person's lifespan. It is primarily defined by persistent deficits in social communication and interaction, along with restricted, repetitive patterns of behavior, interests, or activities. These core characteristics significantly affect an individual's ability to function in everyday life, including their interactions within family units, academic environments, and broader social contexts (American Psychiatric Association, 2013; Mayes & Calhoun, 1999).

The term "spectrum" in ASD reflects the wide heterogeneity in the condition's presentation, severity, and functional impact. Some individuals may exhibit profound impairments in verbal and non-verbal communication and require intensive lifelong support, while others may possess average or even above-average intelligence with subtle social and behavioral difficulties. Historically, such variation was represented through sub-diagnostic categories such as Autistic Disorder, Asperger's Syndrome, and Pervasive Developmental Disorder – Not Otherwise Specified (PDD-NOS), as proposed in earlier versions of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV). However, the DSM-5, published in 2013, consolidated these categories under a single diagnosis, Autism Spectrum Disorder to provide a more unified and flexible diagnostic framework (APA, 2013).

The origins of the condition trace back to early 20th-century clinical observations. Leo Kanner first described autism in 1943, characterizing children with severe language delays, social withdrawal, and repeatable behaviours. Around the same time, Hans Asperger documented children who displayed more fluent speech and higher cognitive functioning, but who still struggled with social reciprocity and exhibited intense interests and repetitive behaviuors (Kanner et al., 1943; Asperger, 1944). These foundational studies shaped early understanding and classification of ASD, which has since evolved with growing research and clinical insight.

Beyond the primary diagnostic criteria, ASD is often accompanied by a range of comorbidities that further complicate diagnosis and management. Common co-occurring conditions include intellectual disabilities, attention-deficit/hyperactivity disorder (ADHD), anxiety disorders, sensory processing difficulties, epilepsy, gastrointestinal issues, and sleep disturbances (Zachariah et al., 2017). These associated conditions can intensify the challenges faced by individuals with ASD and often require multidisciplinary treatment approaches involving neurologists, psychiatrists, occupational therapists, and special educators.

The prognosis for individuals with ASD is highly variable and influenced by several factors, including the severity of symptoms, presence of comorbidities, cognitive ability, access to interventions, and age at diagnosis. Early identification plays a critical role in improving outcomes. Studies have consistently shown that intervention during the sensitive developmental window, typically between ages 2 to 3 years that can lead to significant improvements in communication skills, adaptive functioning, and overall quality of life (Dawson et al., 2008). For example, Applied Behavior Analysis (ABA), one of the most widely validated intervention models, involves highly structured and individualized therapy sessions that can range from 20 to 40 hours per week. These programs are designed to reinforce desirable behaviors and reduce maladaptive ones, while also equipping children with functional skills necessary for daily living (Lovaas, 1987).

Despite the documented benefits of early and intensive intervention, the diagnostic and therapeutic process remains challenging. Diagnosis often depends on the subjective assessment of behavioural symptoms, which can vary widely in presentation and may not fully emerge until later in development. Moreover, waiting times for formal diagnosis and access to intervention services can be long, placing emotional and financial strain on families (McKinney & Peterson, 1987).

To compound this, the underlying biological mechanisms of ASD remain incompletely understood. Although genetic and neurobiological factors are believed to play major roles, no single cause has been identified, and the condition is widely considered to be the result of complex interactions between genetic susceptibility and environmental influences (Vorstman et al., 2017). This ongoing uncertainty makes diagnosis and treatment planning even more complex, leading to variability in care quality across different regions and populations.

In summary, Autism Spectrum Disorder represents a diverse group of developmental conditions that affect multiple domains of functioning. The disorder’s complexity demands a nuanced approach to diagnosis, early screening, intervention, and long-term support. Understanding the broad spectrum of presentations and needs associated with ASD is essential for developing effective educational plans, therapeutic strategies, and support systems that can empower individuals with autism to lead fulfilling lives.

1.1.2 Standard ASD screening and diagnostic tool

The current methods of ASD screening and diagnosis have several limitations that warrant scholarly attention. These conventional methods often rely on human judgment, leading to inconsistent diagnoses and reduced reliability. Moreover, they tend to focus on easily observable behaviors, which might miss the important underlying aspects of the disorder and its various forms. The reliance on behavioral indicators alone may not fully encompass the multidimensional nature of the disorder, potentially overlooking the underlying cognitive, sensory, and emotional aspects (Sahafi et al., 2023). These methods may also lack the sensitivity required to identify early signs of ASD in young children, delaying critical interventions for developmental outcomes (Whitehouse et al., 2021). Additionally, these methods might not adequately address the wide spectrum of ASD manifestations, leading to underdiagnoses or misclassifications in cases that deviate from standardized criteria (Johnson et al., 2022). Furthermore, dependency on clinical expertise and longer assessment times could limit the scalability of diagnostic services (Corona et al., 2021). These shortcomings underscore the need to explore alternative methodologies that offer a more comprehensive and objective assessment of ASD, transcending the limitations of traditional screening and diagnostic paradigms.

1.1.3 AI-based approach

The current AI approaches in ASD detection often involve the application of machine learning algorithms to analyze large datasets, which can include clinical, behavioral, genetic, and environmental data. The use of deep learning, natural language processing, and