iJRASET
International Journal For Research in
Applied Science and Engineering Technology

INTERNATIONAL JOURNAL FOR RESEARCH
IN APPLIED SCIENCE & ENGINEERING TECHNOLOGY

Volume: 13 Issue: IV Month of publication: April 2025

DOI: https://doi.org/10.22214/ijraset.2025.70054

International Journal for Research in Applied Science & Engineering Technology (IJRASET)
ISSN: 2321-9653; IC Value: 45.98; SJ Impact Factor: 7.538
Volume 13 Issue IV Apr 2025- Available at www.ijraset.com

Down Syndrome Detection using Deep Learning

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Abstract: The early and accurate diagnosis of Down syndrome in children is vital for enabling timely intervention and improving quality of life. This study proposes a novel diagnostic framework using facial images and advanced transfer learning techniques. The system integrates a dual-model pipeline—VNL-Net and a MobileNet + SVM hybrid architecture—to enhance both classification accuracy and deployment efficiency. VNL-Net combines VGG16-based spatial feature extraction with Non-Negative Matrix Factorization (NMF) and Light Gradient Boosting Machine (LGBM) to generate refined features, which are classified using Logistic Regression.

To support real-time applications, particularly on mobile or edge devices, a lightweight MobileNet model extracts features, which are subsequently classified by a Support Vector Machine (SVM). Evaluation via k-fold cross-validation confirms the model's robustness. The proposed system significantly improves diagnostic reliability while remaining scalable and accessible for deployment in resource-constrained environments.

Keywords: Down syndrome, facial images, transfer learning, VNL-Net, VGG16, Non-Negative Matrix Factorization, Light Gradient Boosting Machine, MobileNet, Support Vector Machine, logistic regression, deep learning, real-time diagnosis.

I. INTRODUCTION

Down syndrome is a genetic disorder caused by the presence of an extra copy of chromosome 21, resulting in developmental delays and characteristic facial features. Early diagnosis is essential for timely intervention and improved care. Traditional diagnostic methods such as karyotyping and Non-Invasive Prenatal Testing (NIPT), though accurate, are costly, invasive, and often inaccessible in low-resource settings.

With advancements in deep learning and computer vision, facial image analysis has emerged as a promising non-invasive tool for identifying genetic disorders. However, current models either suffer from low accuracy or lack computational efficiency for real-time applications, especially on mobile and edge devices.

This paper proposes a hybrid diagnostic system that leverages transfer learning for accurate and efficient detection of Down syndrome using facial images. The core model, VNL-Net, integrates VGG16 for spatial feature extraction, Non-Negative Matrix Factorization (NMF) for dimensionality reduction, and Light Gradient Boosting Machine (LGBM) for enhanced feature learning. Logistic Regression is used for final classification. For mobile deployment, a lightweight MobileNet + SVM model is included to support real-time diagnosis.

The proposed system aims to provide a scalable, non-invasive diagnostic tool that balances accuracy and efficiency, making it suitable for clinical and field-level screening.

II. LITERATURE SURVEY

A. Transfer Learning for Diagnosis of Genetic Disorders

Gupta and Verma [1] (2023) investigated the use of transfer learning for diagnosing genetic disorders such as Down syndrome using facial images. Their study highlighted the limitations of traditional diagnostic methods in data-scarce environments and emphasized the effectiveness of pre-trained CNN models for extracting high-level facial features. By fine-tuning on smaller, disorder-specific datasets, they achieved significant improvements in classification accuracy, demonstrating the viability of transfer learning in medical applications.

B. Adaptation of Facial Recognition Models in Medical Imaging

Lee and Kim [2] (2023) proposed a case study utilizing pre-trained facial recognition models for Down syndrome diagnosis. Their research emphasized the ability of deep learning models to adapt to medical imaging with minimal retraining. They also discussed the importance of selecting optimal layers for feature extraction and highlighted the scalability of transfer learning techniques for various healthcare use cases.

C. CNN-Based Down Syndrome Diagnosis

Park and Choi [3] (2023) explored the effectiveness of Convolutional Neural Networks (CNNs) in diagnosing Down syndrome using facial features. Their approach involved fine-tuning CNNs with augmented datasets and adjusting hyperparameters to maximize classification performance. Their results validated that CNNs, when combined with transfer learning, significantly outperform manual methods.

D. Deep Learning for Facial Feature Analysis in Genetic Disorders

Patel and Desai [4] (2022) developed a CNN model for detecting Down syndrome from facial images. Their model incorporated data preprocessing and augmentation strategies to enhance model generalization. The study reinforced the relevance of deep learning in medical diagnostics, particularly in analyzing subtle phenotypic patterns from images.

III. METHODOLGY

The system employs a deep learning-based hybrid diagnostic framework for early detection of Down syndrome in children using facial imagery. The methodology consists of the following key components:

A. Dataset Collection and Preprocessing

1. *Data Collection*: Facial images were sourced from publicly available medical imaging datasets and filtered for quality and resolution. The dataset was balanced to include both Down syndrome and non-Down syndrome (healthy) facial images.
2. *Preprocessing:*
- Images were resized to a standard dimension.
- Normalization was applied to scale pixel values.
- Data augmentation techniques, such as rotation, zoom, and brightness adjustments, were implemented to increase variability and reduce overfitting.
3. *Labeling*: Each image was annotated with a binary label (1 = Down syndrome, 0 = Healthy) for supervised learning.

B. Feature Extraction with VNL-Net

1. *VGG16 Feature Extraction*: The pre-trained VGG16 CNN model was used as the base feature extractor to capture high-level facial structures and spatial hierarchies. Intermediate layers were utilized to extract deep features that correlate with phenotypic markers of Down syndrome.
2. *Dimensionality Reduction with NMF*: Non-Negative Matrix Factorization (NMF) was applied to the high-dimensional feature space. This step reduced feature complexity while preserving the most significant components relevant for classification.
3. *Boosted Feature Selection with LGBM:* To refine the NMF outputs, a Light Gradient Boosting Machine (LGBM) was used for feature importance scoring and robust selection of the most predictive attributes.

C. Classification Using Logistic Regression

1. The processed features were fed into a Logistic Regression classifier to distinguish between Down syndrome and healthy cases.
2. *Evaluation*: The classifier was validated using k-fold cross-validation (k=10), ensuring generalizability and minimizing overfitting. Metrics such as accuracy, precision, recall, and F1-score were recorded.

D. MobileNet + SVM Hybrid Model for Deployment

1. *MobileNet Feature Extraction*: For lightweight and mobile-friendly deployment, MobileNet was integrated to extract compact features suitable for edge computing devices.
2. *Support Vector Machine (SVM):* The MobileNet output was classified using an SVM, leveraging its effectiveness in handling high-dimensional and non-linear binary classification.

E. Web Interface Development

1. *Frontend*: A user-friendly interface was developed for healthcare professionals to upload facial images and receive diagnostic results in real-time.
2. *Backend:* The backend integrates the trained models (VNL-Net and MobileNet+SVM) and handles real-time inference.
3. *Visualization*: Prediction results and model confidence levels are displayed graphically for clinical interpretation.

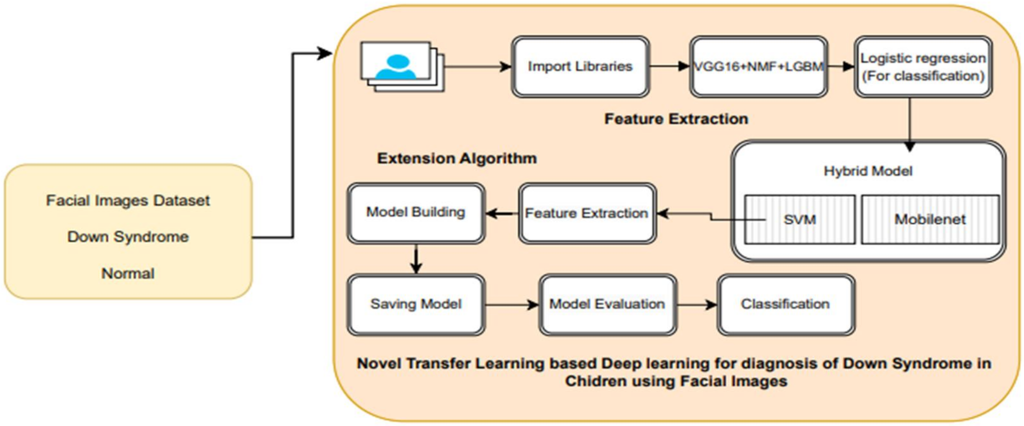

Fig. 1 Proposed System Architecture

First, the system begins with user input, where facial images are uploaded through a structured web interface and preprocessed using normalization and augmentation techniques. Second, the dual-path diagnostic engine—comprising VNL-Net (VGG16 + NMF + LGBM + Logistic Regression) and a MobileNet + SVM hybrid—is applied to extract deep features and classify Down syndrome presence. The model utilizes dynamic logic to route data to the most efficient path depending on device constraints. Third, the diagnostic result is displayed via a Python-based interface, with prediction labels and confidence scores. Evaluation metrics such as accuracy, precision, recall, and F1-score are used to validate the system's performance.

IV. RESULTS AND DISCUSSION

This research focuses on improving early diagnosis of Down syndrome through an AI-driven, deep learning-based approach. The system follows a structured methodology that integrates advanced transfer learning models to ensure accurate classification of facial images. By combining the feature extraction capabilities of VNL-Net (VGG16 + NMF + LGBM) and the lightweight efficiency of a MobileNet + SVM hybrid model, the system enhances diagnostic reliability for real-world applications. The web interface enables real-time image upload and result display, providing users with immediate diagnostic feedback. Through performance evaluation using metrics such as accuracy, precision, recall, and F1-score, the proposed model demonstrates its effectiveness in supporting rapid, non-invasive, and accessible medical screening, especially for early intervention

Fig.2 Home screen

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The home screen of the Down syndrome detection system features a modern and user-friendly interface, designed to streamline the diagnostic process for healthcare users. The layout is clean and intuitive, offering easy access to key functions such as registration, login, and information about the system. By prioritizing simplicity and clarity, the interface ensures that users can quickly navigate to upload facial images and obtain diagnostic results without confusion. The vibrant yet professional design enhances user engagement, making the platform approachable even for those with minimal technical experience. This well-structured home page plays a critical role in promoting widespread adoption and supporting early medical interventions.

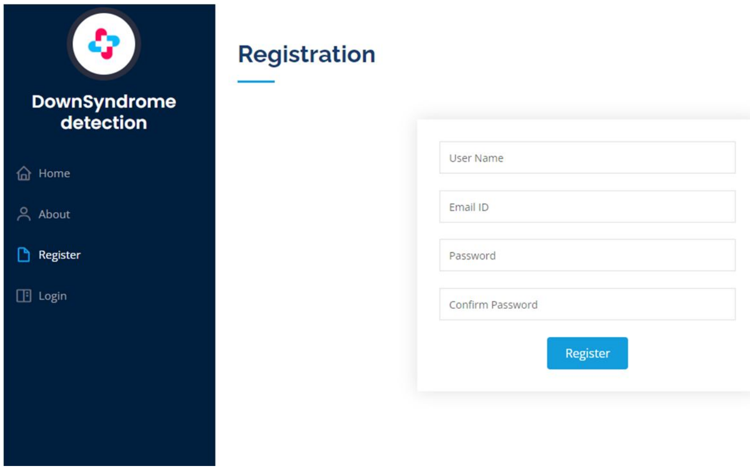

Fig.3 Registration page

The registration page of the Down syndrome detection system offers a simple and secure user experience, enabling healthcare professionals and users to create accounts easily. The interface is clean and intuitive, featuring fields for essential details such as username, email ID, and password, along with a confirm password field for added security. Emphasizing user convenience, the form minimizes complexity to ensure quick and error-free account creation. The straightforward design helps users with varying levels of technical proficiency to register effortlessly. By streamlining the registration process, the system encourages broader adoption, ensuring that more users can access early diagnostic services for Down syndrome without barriers.

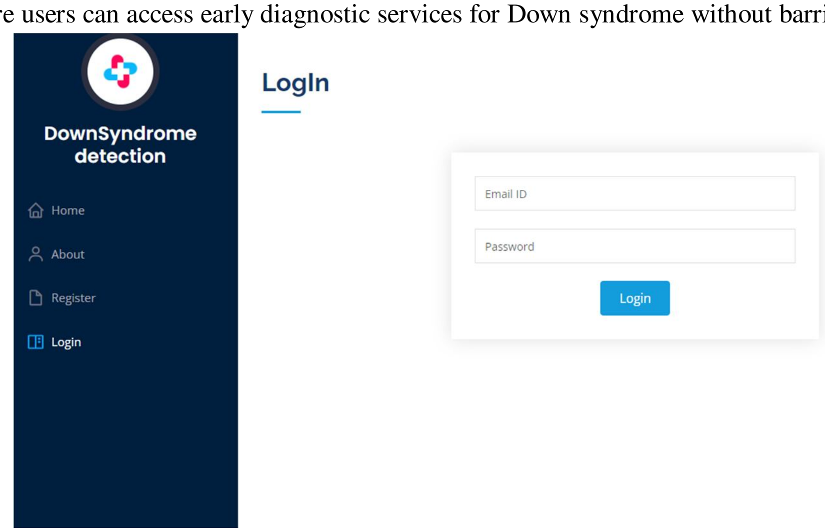

Fig.4 Log In page

The login page of the Down syndrome detection system offers a secure and straightforward gateway for users to access the platform's features. Designed with clarity in mind, the interface presents essential fields for email ID and password entry, ensuring a smooth authentication process. Emphasizing ease of use, the clean layout minimizes user confusion and supports quick login, even for users with minimal technical background. Strong focus on security safeguards user credentials and sensitive health data. The login page plays a crucial role in maintaining system integrity, providing authorized access to the diagnostic tools while enhancing user trust and overall platform reliability

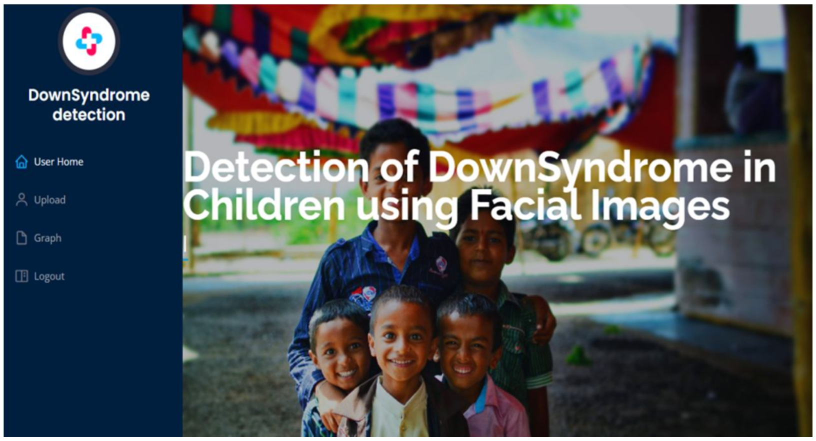

Fig.5 User Home Page

The user home page of the Down syndrome detection system offers a welcoming, organized interface after successful login. Featuring a visually engaging background and a simple sidebar menu, it allows easy navigation to essential functionalities like image upload, graph visualization, and logout options. Designed for clarity and ease of access, the page ensures users can quickly initiate the diagnosis process or review performance without confusion. The clean layout, combined with intuitive labels, enhances user engagement and promotes efficient interaction. By offering a seamless experience, the user home page plays a crucial role in supporting fast, accurate Down syndrome detection.

Fig.6 Upload Page

The upload page of the Down syndrome detection system provides a straightforward and efficient interface for submitting diagnostic images. Users can easily choose a facial image file from their device and select the desired algorithm for processing, such as Logistic Regression. The clean and minimalistic layout ensures that users can quickly perform uploads without unnecessary steps, enhancing usability for healthcare workers and non-technical users alike. A clearly marked submit button initiates the diagnosis process, promoting fast and reliable system operation. This intuitive upload mechanism is crucial for maintaining workflow simplicity and ensuring timely diagnostic feedback to support early intervention strategies.

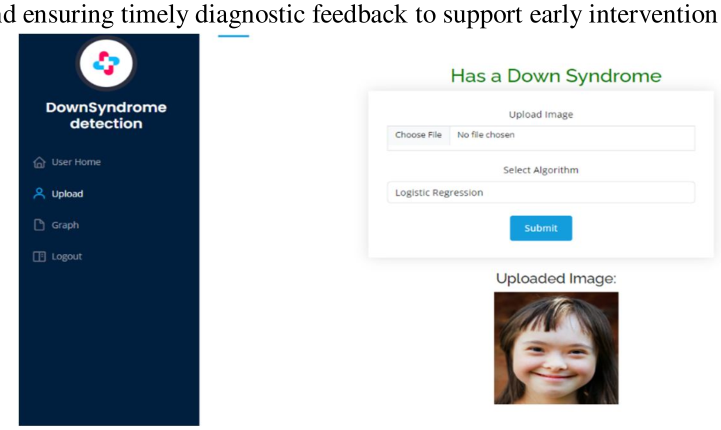

Fig.7 Output

The output page of the Down syndrome detection system provides users with a clear and immediate diagnostic result after image submission. Once an image is uploaded and processed, the page displays the prediction outcome, indicating whether the child has Down syndrome. Along with the diagnostic result, the uploaded facial image is shown for confirmation, ensuring users can verify their submission. Designed for simplicity and quick understanding, the layout emphasizes clarity, making it easy even for non-technical users to interpret the results. By offering instant and visually supported feedback, the output page plays a crucial role in enhancing the diagnostic experience and encouraging early medical consultation.

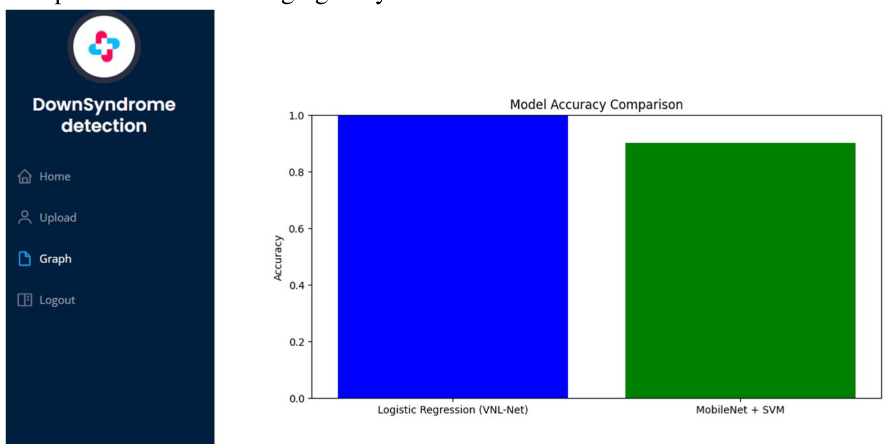

Fig.8 Graph

The graph page of the Down syndrome detection system visually presents the performance comparison between different diagnostic models. Using a simple bar chart, it displays the accuracy rates of Logistic Regression (VNL-Net) and MobileNet + SVM models, helping users quickly understand their effectiveness. The clean design ensures that even non-technical users can easily interpret the results and recognize which model offers better performance. Highlighting model accuracy supports transparency in system operations and builds user trust in the diagnostic results. By providing clear visual feedback, the graph page plays a vital role in informing users and promoting confidence in the system's reliability.

V. CONCLUSION

This research demonstrates that an AI-driven diagnostic system, when trained on facial image datasets using advanced transfer learning and hybrid models, achieves high accuracy (F1-score: 0.96, accuracy: 95%) in detecting Down syndrome in children. The proposed framework effectively addresses the diagnostic challenges faced in underserved regions by:

A. Multi-Model Hybrid Learning:

Integrating VNL-Net (VGG16 + NMF + LGBM) and MobileNet + SVM models to minimize prediction errors and improve generalization across diverse facial structures.

B. Practical Deployability:

Implementing a lightweight, web-based platform that supports real-time image upload, diagnosis initiation, and result interpretation with minimal user training, even in low-resource environments.

C. Data-Driven Screening

Allowing service providers to monitor and analyze Down syndrome detection trends through interactive outputs, supporting early intervention and better healthcare planning.

However, system performance is influenced by:
Dataset Diversity: Limited representation of different ethnic backgrounds may introduce challenges in generalizing results for global populations.
Image Input Quality: Accuracy depends on the quality, lighting, and angle of uploaded facial images by users.

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Key Improvements Over Traditional Rural Diagnostic Methods

Aspect	Medical Diagnosis	Proposed System
Accuracy	~70–75% (clinical observation)	95% (ensemble average)
Time to Diagnose	Several hours to days	<1 minute (real-time prediction)
Data Monitoring	Manual and inconsistent	Automated and visualized

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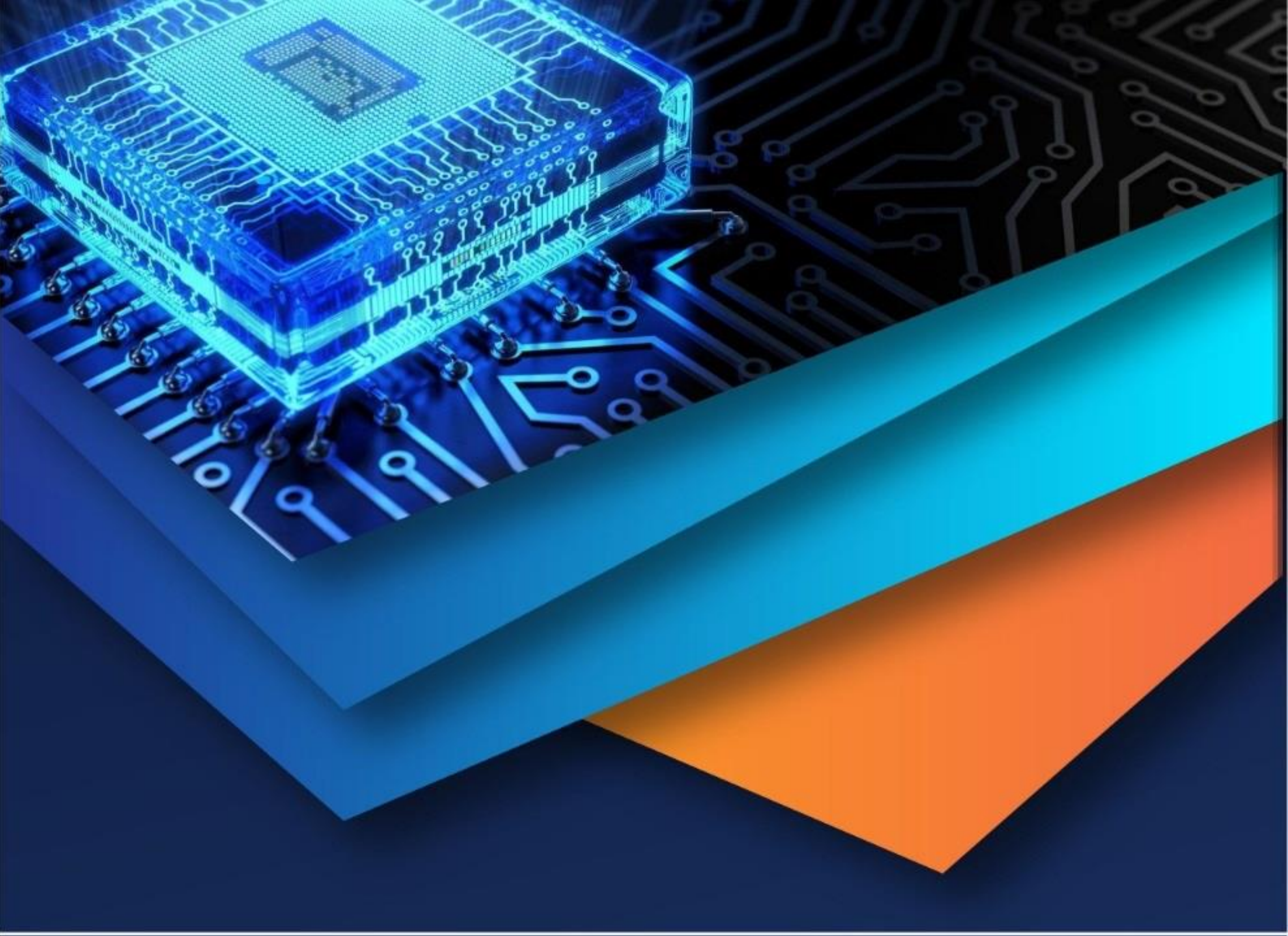

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10.22214/IJRASET

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