

HD-RSNN: MRI-BASED HUNTINGTON DISEASE DETECTION USING SPIKING NEURAL NETWORK

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Abstract – Huntington's disease (HD) is a neurological condition caused by a trinucleotide replication extension in the Huntington gene. The degeneration of the corticobasal ganglia's white matter networks causes progressive impairment of motor, cognitive, and neuropsychiatric functioning. Magnetic resonance imaging (MRI) is increasingly being utilized to measure changes in the brain during the early stages of HD, as gene carriers for the disease demonstrate significant neuronal loss until the conclusion of the illness. The absence of tagged data is a significant challenge, particularly in the early phases of the illness. To overcome this, the study's proposed strategy is applied to MRI imaging of Huntington Disease. MRI pictures are pre-processed to reduce noise and improve image quality using normalization. ResNext is a deep learning system that uses MRI pictures to accurately restore hierarchical structures and extract features. After extracting the features, the SNN network is utilized to classify the detection. Finally, by classifying MRI scans as normal, far, mild, near, or HD, the model improves the accuracy of Huntington Disease detection. The proposed HD-RSNN detection accuracy of this optimized system measured at 98.83%. The HD-RSNN datasets were used to validate the suggested approach, yielding accuracy values of 1.81%, 2.40%, and 0.35%, respectively.

Keywords – Huntington's disease, deep learning, Magnetic resonance imaging, Labelled data.

1. INTRODUCTION

HD is a lethal autosomal dominant inherited disorder characterized by motor, behavioural, and cognitive symptoms. Structural changes in the striatum have been observed through imaging studies, with the volume of the putamen identified as a particularly sensitive marker for monitoring changes across the lifespan of individuals with HD [1]. Unlike many other degenerative conditions, the gene status is determined through a genetic test well in advance of the appearance of initial symptoms, rendering HD well-suited for the assessment of innovative imaging-based methods. Various advanced techniques have been developed to detect structural modifications earlier in the progression of neurodegenerative diseases using anatomical MRI [2, 3]. The

bulk of human illnesses are somewhat complicated, including a confluence of several genetic connections as well as epigenetic, environmental, and lifestyle factors. Still, there are sporadic instances of single-gene human disorders like SA and HD. These illnesses are referred to as monogenic diseases.

HD is a significant neurological illness because of its severe clinical symptoms, inherited origin, and detrimental effects on households [4]. The symptoms of HD, an autosomal mainly inherited illness, include uncontrollable motions, gradual impairments of motor and mental function, and cognitive impairment. The condition usually manifests in the following decade of life, although it can manifest at any age from infancy to beyond seventy years of age. The average time between the diagnosis of an illness to death is between 15 and 20 years [5]. All over the world, HD has been linked to numerous nations and localities. The most prevalent HD grades are seen in European countries and cultures possessing European origin, like the US and Australia. The severity of the disease in India is still difficult to determine, but researchers think the rate is similar to that of European populations with the same haplotype, which is estimated to be 3 to 5 scenarios per 100,000 people, or 40,000–70,000 cases of HD in India [6, 7]. Considering a statistic of 2.71 in 100,000 and a diagnosis of 0.38 in 100,000, HD is a progressive neurological condition. The illness impacts movement, cognitive abilities, behavior and inherits in a self-dominant fashion. The 50 areas of the HTT gene on DNA 4p16.3 are affected by an extension of the cytosine, adenine, and guanine (CAG) triplet repeat. Since there is now no cure for this illness, research is continuing, and as a result, there is a greater demand for metrics that could objectively demonstrate how the illness is progressing. [8, 9].

HD is a neurological condition that is inherited from parents and causes motor impairment, neuropsychiatric symptoms, and cognitive difficulties. It is also fatal. Those with a family history undergo gene testing to detect the HD-

causing mutation. Gene-positive individuals typically manifest clinically significant symptoms around their mid-40s, receiving an HD diagnosis [10]. Timely diagnosis, along with treatment, enables symptom management, as there is no cure for HD. At-home passive symptom monitoring aids in capturing real-world health data, facilitating a quicker HD diagnosis and improved treatment planning insight for clinicians [11]. The DNA analysis is one of the treatments for the identifying the HD patients, but still there is struggle in early identification of HD disease. HD patients benefit from a variety of therapies, which enhances their quality of life. The lack of a definitive biomarker for HD poses a challenge, as deep learning [12] models heavily rely on labelled data to learn patterns. In the absence of clear and consistent biological markers, training robust models becomes intricate, and the risk of misclassification increases. The primary goal of the research is to evaluate all current DL methods based on HD disease detection. By employing advanced deep learning models such as CNN [13], RegNet [14], YOLO [15] and so on contribute to detecting different types of diseases. The most recent deep learning research for detecting HD disease is discussed together with a thorough analysis of the advantages and disadvantages of the available approaches. The following lines provide a summary of the research's main findings.

- Researching all advanced deep learning networks on the identification of HD disease with the gathered images.
- To provide an in-depth review of many trustworthy data sources on HD disease.
- Image processing techniques for recognizing HD diseases discussed the shortcomings of existing review papers on disease detection.
- To present a future model for overcoming the current constraint and creating effective HD disease detection technology.

The rest of the paper is planned as follows; chapter II offers a systematic study of datasets and existing study related to HD disease. The preliminary results are presented in chapter III and the survey discussion is explained in chapter IV, and the conclusion is expressed in chapter V.

2. LITERATURE REVIEW

In 2020 Hett, K et al., [16] suggest two main additions: first, DL techniques based on patches. Second, create a new patch-based abnormalities measure and apply the patch-based grading technique to it. By using this technique, it is possible to identify specific structural anomalies in the test data from a range of normal standards. In comparison to conventional deep learning techniques, the study achieves 95.8% efficiency utilizing MRI intensities.

In 2022 Dang, K et al., [17] proposed a neural network method for classifying and segmenting neural tumors that makes use of MRI scan pre-processing techniques. For this task, three pre-processing methods are employed: information augmentation, WSO section, and gamma modification. Using Google Net and VGG techniques, the medical condition was detected with a maximum reliability

of 97.44%. A few photographs are eliminated during categorization and classifier, which is a drawback.

In 2022 Gopalsamy, A et al., [18] suggested predicting of neurodegenerative diseases using quadratic discriminant analysis using neuron imaging data and multilinear analysis of principal components. Wavelet transformations and histogram gradients are used to select qualities for further processing after the snapshot is handled using the Wie-ner filter and histogram equalization techniques, which also improve the image quality. With a 99.58% success rate, the test limits the quantity of visuals that can be produced utilizing the database.

In 2022 Nair, A et al., [19] proposed motor deficits and apathy in HD have been associated with a hypothesized insufficient connectivity between the basal ganglia. The first four EPI images were removed in order to maintain constant state stability while processing MRI data. The images were then realigned, unwrapped, and smoothed using a Gaussian filter. Co-registration of structural visuals and EPI photographs was done. The pre-processed pictures were used to create a fictitious GLM (General Linear Model) in order to remove unwanted data sets from the location and valves. The 85.2% success rate that the recommended strategy produced is still insufficient.

In 2022 Weiss, A.R et al., [20] suggested the rhesus macaque variant of HD reproduces important neuropathological changes along with motor and mental disorders. A gray matter mask, also known as a WM mask, is used for image pre-processing in all imaging modalities. The T2w SPACE MRI scanner was used to co-register the rest state images in a structural location. The study produced an 80% rate even though it did badly when compared to other states.

In 2020 Scannell, C.M et al., [21] suggested Realistic cardiac perfusion MRI pre-processing using deep neural networks. to determine which time frame in the image series corresponds to the maximum output increase of the LV. The LV canal and LV myocardium are identified using the illustration. MRI imaging uses fully convolutional neural networks (FCNs) to detect structural elements. yields a 93% success rate, has quantifiable diffusion data, and does not provide a ground truth that is testable.

In 2020 Li, H et al., [22] offered Livia NET variations, leveraging established methods for enhancing performance, to increase generalizability to individuals with significant neurodegeneration. In particular, stochastic elastic distortions for enhancing data and network input manipulation were investigated, together with Res-blocks in convolution neural systems. Utilizing pictures from the PREDICT-HD dataset, test the technique.

In 2022 Zeun, P et al., [23] suggested digging into the period of time before symptoms appear, when white matter loss in the corticobasal ganglia starts in preHD. Using diffusion tractography, the Fixel-based approach enables voxel-level precision of intersecting white matter fibres. The TrackON-HD, dataset is utilized for the detection of HD. Employing diffusion tractography-derived atlases of the thalamus and striatum, every network was divided into seven

sub-regions per quadrant according to the predominant portion of cortical connection in every sub-region.

In 2019 Zhang, S et al., [24] suggested a deep learning-based gigantic analysis in HD to classify each action's information-based disease level. To expand the dataset without creating a contemporary artifact, the embedded technique Image Data Generator was used. Image pre-processing is crucial for increasing model efficiency, and the results of 3D CNN are compared to ascertain the most dependable categorization. The method was unable to execute the actions with either the left or right foot.

In 2020 YİĞİT, A et al., [25] proposed the use of deep neural networks and MRI scans to forecast Alzheimer's stage. After reading volumes of MRI brain data, images were processed using Nifti and HDR's multidimensional data structures. Three different viewpoints of T1 graded voxel MRI data were separated into two different dimensions using pre-processing techniques. Using the OASIS data collection for diagnosis, the researchers achieved an 86% classification efficiency by employing CNN.

In 2019 Felix, J.P et al., [26] proposed the use of gait patterns as a predictive HD detection method. After being collected through a person's stride, time-based data is pre-processed and features are retrieved. By detecting outliers and eliminating them at the initial processing stage, the proposed method manages noisy data and enables the detection of recurring patterns that result in a precise diagnosis of sickness. The KNN method of the test achieves 97.2%, whereas several other methods perform appallingly.

In 2023 Ouwerkerk, J et al., [27] proposed Enroll-HD with machine learning methods to maximize participant and variable inclusion, and developed dataset-based ways to improve the age at start of AAO estimation. RNNs were used to compute roughly 42% of the missing data in Enroll-HD using simple pre-processing, demonstrating the value of ML approaches. The accuracy is still inadequate even if the proposed method outperformed the existing state-of-the-art equipment level of 85.2%.

In 2023 Maddury, S [28] Developing Huntington's Disease Reliable Prediction Using Medical Data and Simple Machine Learning. To be used for HD, the EEG and ECG data must first undergo pre-processing so that patterns and classification can be found. following data preparation and feature extraction from both the original and updated data. Even with the recommended method's 91.35% yield, it is still insufficient when compared to other methods.

3. PROPOSED METHODOLOGY

In this study the HD-RSNN proposed method is to detect the Huntington Disease detection using MRI images. The MRI image is taken as an input and preprocessed using Normalization to reduce the noise and increase image quality. The normalized images are fed into feature extraction exact hierarchical properties from MRI images using ResNext Network and the extracted features are classified as Normal, Far, Mild, Near, HD using Spiking Neural Network. Figure 1 shows the approach for detecting Huntington Disease.

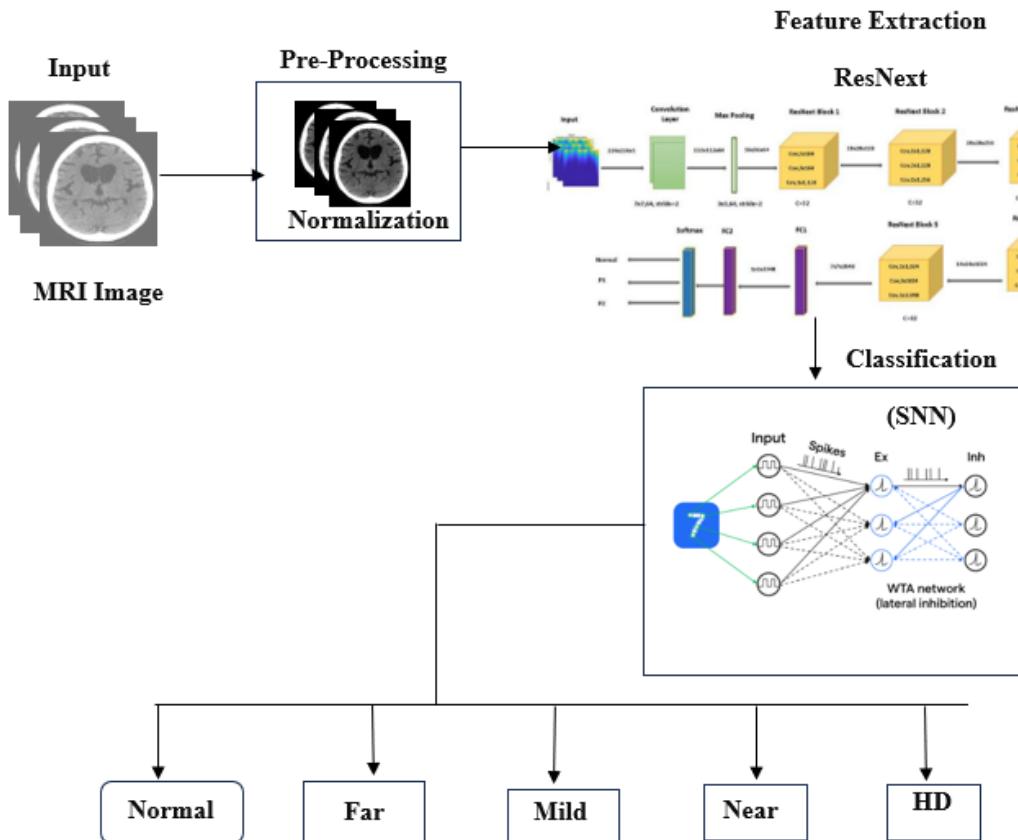


Figure 1. HD-RSNN Proposed Method

3.1. Dataset Description

The summary of studies conducted on HD, providing insights into the deep learning techniques for analysis. The datasets used in these studies vary, encompassing both publicly available datasets and self-generated datasets. PREDICT-HD, BraTS 2019, DS-66, DS-75, DS-160, and DS-255, TRACK-ON HD, OASIS and MIRIAD and self-generated datasets are utilized in the relevant study.

The PREDICT-HD contains 1091 images which involves individuals genetically predisposed to HD. The data are attaining from 96% Caucasian, 89% right-handed, 70% married, and 77% employed, with 64% females. Participants' average age was 42.1 years, and 90% had at least a high school education, averaging 14.5 years of education.

The 239 participants in the TRACK-ON HD dataset are divided into three groups: (1) 106 individuals without HD but with the mutant huntingtin gene, (2) 22 early HD patients,

and (3) 111 controls who are matched for age and sex. Newly enrolled preHD participants had to have a disease burden score greater than 250 and a CAG repeat length of at least 40. Manifest disease, age <18 or >65, significant neurological, mental, or medical disorders, and a history of serious head trauma were among the exclusion criteria. The tests use the datasets DS-66, DS-75, DS-160, and DS-255, which contain 66, 75, 160, and 255 images, respectively. T2-weighted MR brain scans with a 256×256 axial plane spatial resolution are represented by each image in these datasets. Because T2 model images have better contrast and clarity than T1 and PET, they are selected.

4. RESULTS AND DISCUSSION

The methodologies for classifying and identifying Huntington Disease Detection are briefly compared in the section below.

Table 1. Comparison of various techniques for finding of skin tumor.

Author & year	Dataset	Data description	Pre-processing methods	Segmentation	Classification	Validation results
2020 Hett, K et al., [16]	PREDICT-HD	750 MPRAGE images	non-local means filter and piece-wise linear histogram normalization	-	CNN	Accuracy- 95.8%
In 2022 Dang, K et al., [17]	BraTS 2019	335 images	gamma modification, WSO section, and information enhancement	U-Net	Google Net and VGG	Accuracy- 97.44%
2022 Gopalsamy, A et al., [18]	DS-66, DS-75, DS-160 and DS-255	66, 75, 160 and 255 images	Wiener filter, and histogram equalization	-	RDA	Accuracy- 99.58%
2022 Nair, A et al., [19]	TRACK-ON HD	102 Scans	Gaussian filter	-	Region of interest (ROI)	Accuracy- 83.8%
2022 Weiss, A.R et al., [20]	Self-Generated	5374 images	Gray matter mask, and WM mask	-	AAV-based approach	Accuracy- 80%
2020 Scannell, C.M et al., [21]	Self-Generated	1050 image	-	U-Net	CNN	Accuracy- 93%
2020 Li, H et al., [22]	PREDICT-HD dataset	750 MPRAGE images	Data augment,	Livia NET	ResNet	Accuracy- 93.7%
2022 Zeun, P et al., [23]	TrackON-HD, dataset	72 preHD disease image and 85 controls images	MRtrix3 and FSL	diffusion tractography	-	Accuracy- 92.4%
2019 Zhang, S et al., [24]	Self-Generated	180 images	Footprint Formation Process, Data augment	-	VGG	Accuracy- 89%
2019 Felix, J.P et al., [26]	Self-Generated	36 images	median filter	-	KNN	Accuracy- 97.2%,
2023 Ouwerkerk, J et al., [27]	Enroll-HD	21000 images	-	-	RNN	Accuracy- 85.2%

2023 Maddury, S [28]	Self-generated	69 images	discrete wavelet transforms	-	Random Forest (RF)	Accuracy- 91.35%
2024 Vethavikashini, A.M [29]	HD-RSNN	120 images	Spiking Neural Network	-	CNN	Accuracy- 98.83%

In this section, a deep learning model for diagnosing HD is mentioned and discussed. The pre-process models and various datasets that were utilized to identify HD are discussed. When contrasted with the models given below, RDA performs well and beats other models with 99.58% reliability. Table 1 below displays a comparison of various methods for pre-processing, segmenting and classifying HD.

The table summarizes various studies conducted in recent years that utilize deep learning approaches for the detection and classification of HD based on different datasets. It is evident from the table that various datasets such as PREDICT-HD, BraTS 2019, DS-66, DS-75, DS-160, DS-255, TRACK-ON HD, and Enroll-HD were used, each with its unique characteristics and sizes. Pre-processing methods included techniques like non-local means filtering, gamma modification, WSO section, and information enhancement.

The segmentation techniques varied, with U-Net being a popular choice. Classification models encompassed CNNs, RDA, AAV-based approaches, VGG, ResNet, KNN, and random forests. Notably, the reported accuracy rates ranged from 80% to 99.58%, highlighting the efficiency of deep learning in the context of HD detection. These studies collectively contribute to the growing body of research utilizing advanced computational methods for the early diagnosis and classification of HD.

Existing deep learning methods for categorizing HD have shown encouraging results, with models obtaining high accuracy in identifying healthy and abnormal patients. The variety of CNN was demonstrated to outperform more established machine learning techniques in a number of noteworthy comparative studies. MRI images of HD detection is shown in Figure2.

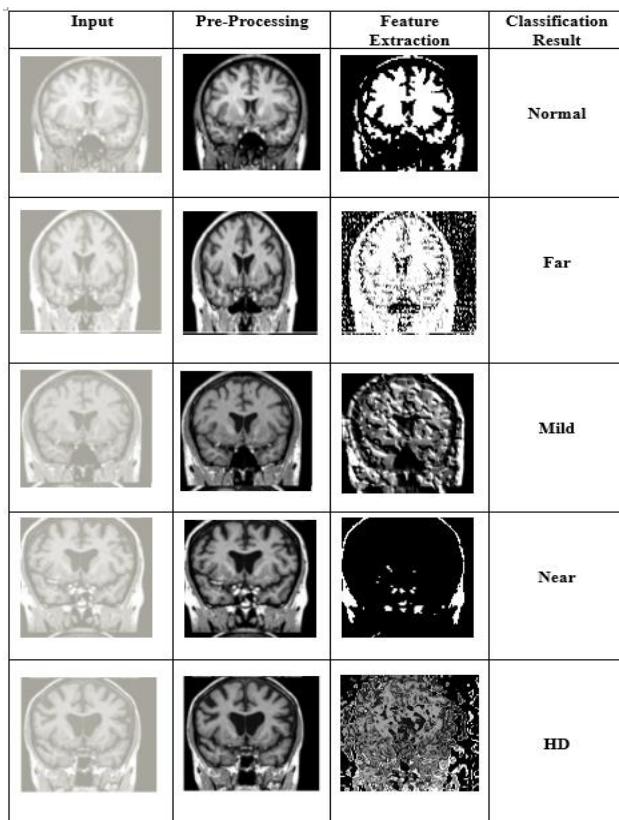


Figure 2. MRI Image for Huntington Disease diagnosis research

4.1. Performance Evaluation Measures

This section describes the metrics used to measure classification performance described in this section. Developed deep learning model is available in the dataset, which includes the number of samples. The metrics are accuracy, specificity, sensitivity (recall), F1-score. Macro_F

$$\text{Specificity} = \frac{\text{TN}}{\text{TN} + \text{FP}} \quad (1)$$

$$\text{Precision} = \frac{\text{TP}}{\text{TP} + \text{FN}} \quad (2)$$

$$\text{Accuracy} = \frac{\text{TP} + \text{TN}}{\text{TP} + \text{TN} + \text{FP} + \text{FN}} \quad (3)$$

$$\text{Recall} = \frac{\text{TP}}{\text{TP} + \text{FN}} \quad (4)$$

$$\text{F1-Score} = 2 \times \frac{(\text{Precision} \times \text{Recall})}{(\text{Precision} \times \text{Recall})} \quad (5)$$

This work used for basic evaluation measures, like accuracy, precision, and recall, F1 score, to evaluate the proposed SNN-based application. The classification is either normal, far, mild, near, HD of Huntington Disease Detection.

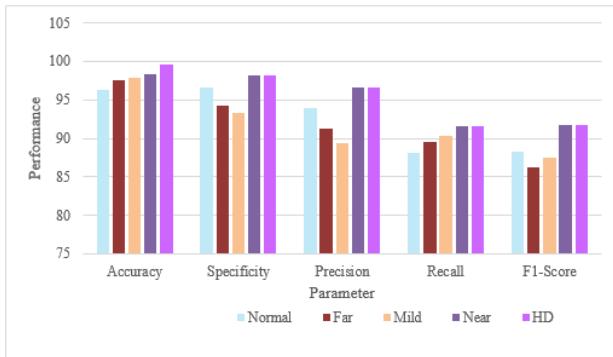


Figure 3. Performance Matrices

Figure 3 shows performance evaluation of Proposed Methodology. F1-Score, recall, accuracy, specificity, precision are to assess how effective the proposed methodology is. The proposed model's total accuracy is 98.83%.

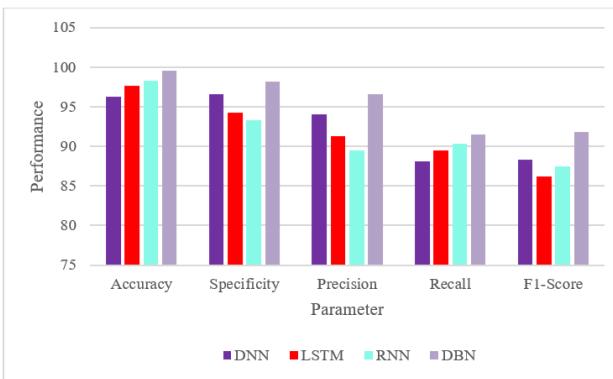


Figure 4. Performance metrics

Performance metrics of different segmentation were compared using a table shown in figure 4. F1-Score, accuracy, precision, specificity, and recall to evaluate segmentation performance. SNN outperforms other algorithms like ANN and CNN in terms of emotion identification accuracy, with a rate of 98.83%.

5. CONCLUSION

In this study, the proposed method is applied to MRI imaging to Huntington Disease. Input MRI images are pre-processed to decrease noise and increase image quality using Normalization. ResNext is a deep learning system that accurately recovers hierarchical properties from MRI images and is used for feature extraction. After extracting feature, the SNN network is used to classify the detection. Finally, by classifying MRI images as normal, far, mild, near, HD, the model aids in early detection and diagnosis of Huntington Disease detection accuracy is improved. The proposed HD-RSNN detection accuracy of this optimized system has been measured at 98.83%. The HD-RSNN datasets were utilized

to validate the proposed approach, with accuracy values of 1.81%, 2.40%, and 0.35%, respectively.

CONFLICTS OF INTEREST

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

FUNDING STATEMENT

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors

ACKNOWLEDGEMENTS

The author would like to express his heartfelt gratitude to the supervisor for his guidance and unwavering support during this research for his guidance and support.

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Arrived: 25.01.2024

Accepted: 27.02.2024