

An Overview of Huntington's Disease using Machine Learning

Mr. John Felix V¹ and Dr. K. Sharmila²

Research scholar, Department of Computer Science¹

Associate Professor, Department of Computer Science²

Vels Institute of Science, Technology & Advanced Studies (VISTAS), Chennai, Tamil Nadu, India

felixsvj@yahoo.co.in and sharmila.scs@velsuniv.ac.in

Abstract: *Huntington's disease (HD) is one of the rare neurodegenerative diseases caused because of genetic mutation of the Huntington gene and also characterized by progressive motor dysfunction, cognitive impairment, and psychiatric symptoms. Since no cure is possible and only symptomatic therapy is available, early detection and individualized therapy are guaranteed for effective disease treatment. Artificial intelligence has emerged as a transformative tool in healthcare, revolutionizing many aspects of medical practice and research, enabling the detection, monitoring and treatment of Huntington's disease. Additionally, this paper serves as a valuable resource for researchers in the field of machine learning and neurodegenerative disease detection, as well as healthcare professionals.*

Keywords: Huntington's disease (HD), neurodegenerative diseases, Artificial intelligence

I. INTRODUCTION

In 1872 George Huntington wrote an account of hereditary chorea, which is now known as Huntington's disease (HD). Huntington's disease is a profoundly impactful neurodegenerative disorder [1] that not only affects individuals but also casts a long shadow over their families [2]. The gravity of HD has sparked growing concern among the medical and research communities worldwide, triggering multifaceted efforts to not only unravel its etiological and pathophysiological intricacies but also to pioneer advancements in its early detection and management [3]. Research in other neurodegenerative diseases such as Alzheimer's [4,5,6] and Parkinson's [7] has similarly aimed to decode their intricate mechanisms, leading to strides in understanding their pathology and paving the way for potential treatment breakthroughs.

HD is one of the most common diseases with severe symptoms that affect people of all ages and gradually lead to severe disability. The genetic basis of this disorder has been elucidated and known for several decades, and recent studies have shown promising mechanisms to reverse symptoms of HD (8). In addition, HD can be considered as a neurodegenerative disease model to study other cases with common symptoms, and knowledge about other diseases can be useful in the diagnosis and treatment of HD. This paper represents the pioneering effort to comprehensively compare the effectiveness of AI powered approaches for HD diagnosis.

Artificial Intelligence:

AI is classified into two main branches: virtual and physical. The physical branch of AI comprises medical devices and robots that assist in complex surgeries and healthcare delivery. The virtual branch majorly consists of machine learning (ML) and deep learning (DL). ML is further classified into 3 types: (1) Unsupervised learning (2) Supervised learning and (3) Reinforcement learning. [9,10]. ML is a learning and predictive technique that finds a meaningful pattern in each set of data with little human interference. However, a part of ML is dependent on human knowledge for the selection of features in data, multitasking, and transferring knowledge [11]. The major difference between the three subclasses of ML is the data interpretation method.

AI Methodologies in Huntington's disease studies

HD includes characteristic symptoms such as psychiatric decline, cognitive impairment, and movement disorders like chorea (involuntary and jerky movements), motor in coordination, and bradykinesia [12]. Detection or diagnosis of HD at an early stage is difficult and mostly the symptoms are considered the normal symptoms of aging and dismissed. Since artificial intelligence consists of ML as virtual branch, it has merged as valuable tools in the diagnosis and assessment of Huntington's disease.

These techniques utilize various algorithms and computational approaches to analyze complex data sets, offering clinicians and researchers new insights into the disease [13]. One of the primary applications of machine learning in HD diagnosis is the identification of biomarkers and patterns within medical images, such as magnetic resonance imaging (MRI) and functional MRI (fMRI) [14].

Machine learning algorithms can use genetic information and other important factors to predict a person's likelihood of developing HD, aiding in early detection and counseling. Early diagnosis and counseling can be achieved by machine learning algorithms that consider genetic information and other important factors, such as the likelihood of a person developing HD.

Machine learning techniques are advancing the field of HD diagnosis by facilitating the extraction of valuable insights from clinical and genetic data, ultimately leading to earlier detection and improved management of this devastating neurodegenerative disorder. The various machine learning techniques that involved in predicting the HD risk assessment, namely Naive Bayes, Decision Tree, etc.

The role of AI and numerical simulations in neurodegenerative diseases is multifaceted and holds significant promise for improving our understanding, diagnosis, treatment, and management of these complex conditions. AI algorithms can analyze diverse data sources, such as genetic information, neuro imaging scans, and clinical assessments, to identify early biomarkers and patterns indicative of neurodegenerative diseases.

Machine learning, which aims at developing algorithms to discover repetitions or trends in present data and form new prediction data, the repetitive motor, historical patterns, and other features of HD could be used to predict the disease. It can also classify HD from other neurodegenerative disorders and provide information on the progression of the disease, as well as measure the effectiveness of the drug thereby confirming medication adherence. This could only be done by computational statistics and mathematical optimization [15].

II. CONCLUSION

The incorporation of AI in the field of HD is an enticing and transformational strategy to address the difficulties of this neurodegenerative disorder. Significant progress has been made in understanding HD, enabling early identification, and data analysis. AI provide the doctor's judgements more precision and legitimacy, so both of us can cooperate to produce the intended results. For HD, switching from traditional to computerized methods is encouraging and will improve patient treatment.

REFERENCES

- [1]. Bhachawat S., Shriram E., Srinivasan K., Hu Y.C. Leveraging Computational Intelligence Techniques for Diagnosing Degenerative Nerve Diseases: A Comprehensive Review, Open Challenges, and Future Research Directions. *Diagnostics*. 2023;13:288. doi: 10.3390/diagnostics13020288. [PMC free article] [PubMed] [CrossRef] [Google Scholar]
- [2]. Walker F.O. Huntington's disease. *Lancet*. 2007;369:218–228. doi: 10.1016/S0140-6736(07)60111-1. [PubMed] [CrossRef] [Google Scholar].
- [3]. Ghosh R., Tabrizi S.J. Polyglutamine Disorders. Springer; Cham, Switzerland: 2018. Clinical features of Huntington's disease; pp. 1–28. [PubMed] [Google Scholar]
- [4]. Mahendran N., PM D.R.V. Deep belief network-based approach for detecting Alzheimer's disease using the multi-omics data. *Comput. Struct. Biotechnol. J.* 2023;21:1651–1660. doi: 10.1016/j.csbj.2023.02.021. [PMC free article] [PubMed] [CrossRef] [Google Scholar]

- [5]. Mahendran N., PM D.R.V. A deep learning framework with an embedded-based feature selection approach for the early detection of the Alzheimer's disease. *Comput. Biol. Med.* 2022;141:105056. doi: 10.1016/j.compbiomed.2021.105056. [PubMed] [CrossRef] [Google Scholar]
- [6]. Mahendran N., Vincent P.D.R., Srinivasan K., Chang C.Y. Improving the classification of alzheimer's disease using hybrid gene selection pipeline and deep learning. *Front. Genet.* 2021;12:784814. doi: 10.3389/fgene.2021.784814. [PMC free article] [PubMed] [CrossRef] [Google Scholar]
- [7]. Dixit S., Bohre K., Singh Y., Himeur Y., Mansoor W., Atalla S., Srinivasan K. A Comprehensive review on AI-enabled models for Parkinson's disease diagnosis. *Electronics.* 2023;12:783. doi: 10.3390/electronics12040783. [CrossRef] [Google Scholar]
- [8]. Kumar A, Kumar V, Singh K, Kumar S, Kim YS, Lee YM and Kim JJ: Therapeutic advances for Huntington's disease. *Brain Sci.* 10(43)2020.PubMed/NCBI View Article : Google Scholar.
- [9]. Londhe VY, Bhasin B (2019) Artificial intelligence and its potential in oncology. *Drug Discov Today* 24:228–232. <https://doi.org/10.1016/J.DRUDIS.2018.10.005>.
- [10]. Wang F, Preininger A (2019) AI in health: state of the art, challenges, and future directions. *Yearb Med Inform* 28:016–026. <https://doi.org/10.1055/s-0039-1677908>.
- [11]. Fu G, Levin-schwartz Y, Lin Q et al (2019) Machine learning for medical. *Imaging* 2019:10–12.
- [12]. Gordon MF, Grachev ID, Mazeh I et al (2019) Quantification of motor function in Huntington disease patients using wearable sensor devices. *Digit Biomark* 19355:103–115. <https://doi.org/10.1159/000502136>.
- [13]. Mohan A., Sun Z., Ghosh S., Li Y., Sathe S., Hu J., Sampaio C. A machine-learning derived Huntington's disease progression model: Insights for clinical trial design. *Mov. Disord.* 2022;37:553–562. doi: 10.1002/mds.28866. [PubMed] [CrossRef] [Google Scholar]
- [14]. 71. Lois C., González I., Izquierdo-García D., Zürcher N.R., Wilkens P., Loggia M.L., Hooker J.M., Rosas H.D. Neuroinflammation in Huntington's disease: New insights with 11C-PBR28 PET/MRI. *ACS Chem. Neurosci.* 2018;9:2563–2571. doi: 10.1021/acchemneuro.8b00072. [PubMed] [CrossRef] [Google Scholar]
- [15]. Cohen S, Waks Z, Elm JJ et al (2018) Characterizing patient compliance over 6 months in remote digital trials of Parkinson's and Huntington disease. *BMC Med Inform Decis Mak* 18:1–10. <https://doi.org/10.1186/s12911-018-0714-7>