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Exploring the use of Artificial Intelligence in genetic prognosis within Medical Field: A case study on Huntington's Disease (HD)

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Introduction

The use of Artificial Intelligence in the medical field: Artificial Intelligence has been growing in popularity in society, especially the medical field, with its ability to perform rapid analysis, mass data readings as well as determining the most viable outcomes for suffering patients with all its ability to outperform humans within the medical field. Currently, AI is most prominently used in analyzing medical images to detect diseases (ie: brain scans, MRIs as well as X-rays), being able to account for various factors in order to diagnose conditions as accurately if not more accurately than human experts in most cases **[1]**. Most crucially, AI is able to predict a patient's future health and life outcomes such as, diseases progression, condition and any other aspects within the human body that can alter one's condition, this ability as a result, sets the ability of AI apart from humans as Artistic Intelligence continues to take the lead within key medical advances as well as diagnostic elements.

***An overview of Huntington's Disease (HD)*:** The Huntington's Disease is a hereditary neurodegenerative disorder that primarily affects movement, cognitive and behavioral functions. It is primarily caused by a faulty gene that leads to the degeneration of nerve cells within the brain. **[2]** In detail, a mutation occurs in the HTT gene which causes the huntingtin protein to misfold and aggregate as a result of an abnormally long polyglutamine tract within the protein. The misfolding of the HTT gene hinders the efficiency of the ubiquitin-proteasome system and the autophagy-lysosomal pathway, which are core pathways that continue to generate misfolded proteins, leading to the conditions we see in HD. Importantly, the misfolded HTT protein disrupts the normal movements of vesicles and organelles, as well as axons and dendrites leading to a lack of cognitive movements in patients who suffer from HD. **[3]** However, symptoms usually only appear when one reaches adulthood (between ages 30-50), yet can occur earlier in life. Currently, there are no immediate nor direct cures for HD but treatment can help manage symptoms.

Importance of the Research: Understanding the use of Artificial Intelligence not just in societal duties but in the medical field as well will allow more of the population to identify the benefits of using AI within the process of determining treatments and fully relying on AI for future medical advancements. This research's focus on Artificial Intelligence's ability in prognosis will allow many to differentiate the abilities between humans and AI, hence allowing more to acknowledge the benefits of using AI.

Goals of the Research: This research aims to provide a variety of literature analysis as well as in depth case study of HD in order to raise awareness of the benefits of using Artificial Intelligence within the healthcare system.

Research Question

How does the usage of Artificial Intelligence directly the process of prognosis within the medical field and more specifically, through the Hunginton's Disease?

Literature Review:

Research on the rise of Artificial Intelligence within the medical field: Research has noted that the usage of Artificial Intelligence not just in daily lives, but the healthcare system as well as the medical field has been more prominent than ever. While it is said that AI can not guarantee 100% accuracy, neither can humans within the medical care, especially diagnostic responsibilities, hence automatically inducing more people to accept the use of AI as a means for more rapid and efficient medical advances within procedures. Recently, extensive research was done on the use of AI within prognosis by evaluating the cardiovascular risk predictions within the patient. Through the evaluation of electronic machine that involves AI when assessing the possibilities of cardiovascular events such as heart attacks or strokes), it is proven that AI and the machine learning models demonstrated the ability to outperform traditional models such as humans or even risk scores evaluated by professionals by uncovering complex interactions within the patients' data that can often be missed by simpler statistical methods in which the medical field often resort to today **[4]**. Furthermore, it is clear that AI has the ability to outperform human capabilities when an objective and rapid standardization is required, especially in urgent patient cases that require immediate and the best actions in order to guarantee survival, in which AI supports as seen through various research done. The research paper also suggests that by using machines and AI to accurately identify high-risks individuals before we as humans, begin to see the symptoms, resource for intensive interventions can be better targeted to those who need them the most, differing from the current healthcare system we see today, where in most cases, many die before they receive treatment due to the delay in diagnosis of serious conditions **[5].** Lastly, research also identified the possibility to integrate the algorithm and data organization of AI into existing clinical workflows or Electronic Health Record systems so the medical field does not require an immense change in order to achieve benefits in the best interest of the patients **[6].**
Research on Huntington's Disease (HD) and the use of Artificial Intelligence in prognosing HD: Research has shown that the first step for Artificial Intelligence to prognose HD is through Genomic data analysis, indicating that AI algorithms can be trained through multiple datasets of human genomes (in order to have a dataset of normal sequencing to compare and allow AI to find the mutations before symptoms are expressed), to identify mutations such as single nucleotide variations, insertions/deletions or copy number variants. Starting from 2023, the method of Genomic data analysis is already being put in use for prognosing HD since HD is caused by a specific gene mutation – an elongated CAG trinucleotide that repeats in the HTT gene, leading to misfolding and hence resulting in the symptoms we see in HD **[7].** In summary,

because HD symptoms such as lack of cognitive mobility only expressed through late adulthood, it is often that many only realise they have HD when these symptoms are expressed, in which is often too late for adequate treatment and adjustment **[8]**, yet, by integrating advanced artificial intelligence within the prognosis of HD, it allows for patients to easily and quickly acknowledge that they have the possibility of having HD future in life and having enough time and resources to receive treatments and adjust accordingly, allowing a less painful adjustment.

Proposed Methodology

This study will employ mixed-method approaches to ensure a practical analysis. A comparative experiment will be held, where 200 adults who have tested positive for the HD mutation but are not yet showing motor symptoms (n=200), then two groups will be established with two distinct treatments. In Group A (Standard Group, without AI): the 100 samples randomly assigned will receive the standard care where their prognosis is solely based on the mutation (CAG expansion) and the doctor's clinical experiences, where the doctor can only provide general and broad possibility assessments (ie: "Based on the amount of CAG repetition, you will most likely express symptoms by age X). In Group B (AI-Assisted Group): the other 100 samples randomly assigned will receive a personalized prognosis report generated by machines integrated through AI models that not only analyzes their CAG count but various data through body reports, giving the patients a more realistic and practical timeline of the symptoms expressions. Before running the experiment, both groups (n=200) will be given a starting test with questionnaires that require self evaluations of various aspects, such as uncertainty in illness, quality of life, current cognitive mobilities as well as anxiety and depression level, all ranked on a scale of 1-10. After Groups A and B have been established with 100 samples in each, follow-up assessments will be scheduled every 6 months after their prognosis session with the same questionnaire, evaluating the difference in certainty and anxiety level caused by the type of treatment and prognostic reports received by distinct patients.

Project Practicalities

Data collection will take place over a period of 1 year, with each follow-up assessment occurring 6 months after the prognosis sessions. At the end of the experiment, there should be 3 sets of data from each patient to analyse. This experiment will adhere strictly to the privacy of patients as well and the ethical considerations when inducing treatments and the survey.

Roadblocks and potential limitations

This study/experiment may face challenges in having enough samples to conduct the experiment, since the experimental sample is n=200, it may be difficult to find 200 HD diagnosed patients given that HD is a rare condition and require those who have not experienced symptoms yet who are also willing to participate in an experiment that occupies much of their time, hence it may be difficult to induce the experiment at the very beginning when looking for samples, however, this can be resolved through connecting to HD specialised clinics who may have a better grasp of connections to patients. In additional, the experiment may also face challenges in finding representative samples to ensure that the results and concussions made are not too narrow, especially because HD is such a rare condition, is is likely that many of the samples can have similar traits that caused them to have HD, as a result, data conducted may be nonrepresentative of the true population of patients that experiences HD as well as misinterpreting the use of AI within prognosis when there can be potential bias within the experiment, this can result in misinterpretation and nonrepresentative conclusions.

Conclusion

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This research aims to enhance the understanding of society on the benefits Artificial Intelligence integrates within the medical field as well as the healthcare systems. As AI continues to advance and be utilized in daily chores, it is crucial that we understand both the implications and abilities of AI that can often outperform human capabilities, especially within the medical field, regarding diagnosis, determining risks and deciding the best outcome and treatment given the patients' conditions. Through the case study of the usage of AI in the prognosis of the rare condition, Huntington's Disease, it is clear that AI not only provides more accurate and rapid results within risk analysis but provides patients with the advantage of more time and adequate resources to adjust accordingly, even when HD currently still has no direct cure. Ultimately, this research aims to allow society to gain insights of the many hidden benefits of AI integrated machines within the medical field and reminds many of how we can rely on AI in future medical care advancements.

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