From Pixels to Prognosis: Artificial Intelligence and Machine Learning Models in Brain Tumour Mutation Prediction

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Abstract

Brain tumours are a leading cause of death and disability, impacting individuals across all ages, genders, and ethnicities. They are primarily diagnosed using MRI but a precise diagnosis is dependent on the molecular biology of the tumour studied on the pathological specimen. Artificial intelligence and machine learning are forging new paths through diagnostic obstacles, offering the intriguing benefits of non-invasive diagnosis, pattern recognition, and outcome prediction from imaging data. Here, we present a literature review on the role of machine learning in tumour mutations using imaging alone.

Keywords: brain tumor, artificial intelligence, machine learning, tumor mutation

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Introduction

Central nervous system neoplasms rank as the second leading cause of cancer deaths. Brain tumours represent a diverse array of over a hundred molecular subtypes, each associated with distinct clinical and biological behaviours. Traditionally, invasive methods such as biopsy, IHC studies, direct sequencing, FISH, NGS, and methylation-specific PCR have been fundamental for precise diagnosis. However, these methods are not only expensive and time-consuming, prone to human error. Artificial intelligence presents the potential to overcome aforementioned limitations and enhance diagnostic precision by forecasting molecular alterations in tumours through radiological imaging, thereby improving the safety, efficiency, and effectiveness of treatment. This can be accomplished through the application of deep learning (DL) or machine learning (ML).[1]

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The conversion and comparison of imaging data to conclude a comprehensible result has been achieved through machine learning (ML). ML develops algorithms to enable computers to learn from existing data and apply this function to analyze new data. Artificial Neural Networks (ANNs) and Convolutional Neural Networks (CNNs), also known as deep learning methods, are the most cited supervised learning techniques in neuro-oncology. Radiomics is revolutionizing diagnosis by uncovering non-invasive links between imaging and biological markers. Its vast use and results have not been critically analyzed in the prediction of tumour mutations in neuro-oncology.

Literature Search

Glioblastoma, the most common malignant brain tumour, is characterised by the mutational status of IDH, TP53, and ATRX. Typically, results take one to two weeks to obtain. Moreover, conventional diagnostic methods often require surgical procedures that have risks involved. To mitigate these risks the adoption of an artificial CNN system can be incorporated as it requires radiological imaging alone. Choi YS et al. assessed[1],166 preoperative MRI images (including T1 with contrast and FLAIR sequences) of gliomas, and the implementation of an artificial CNN system achieved over 90% accuracy in predicting IDH status based solely on imaging data.[2]

Yoganda et al. utilized a deep learning method for the classification of IDH mutation status in gliomas utilising data from 214 patients and two separate networks involving T2W image network-only and T2W, T1 post-contrast, fluid-attenuated inversion recovery. Interestingly, high IDH classification accuracy was achieved using only T2-weighted MR images with a mean cross-validation accuracy of 97.14% ± 0.04.[3] Another meta-analysis including 9 studies analysed over 996 patients concluded that ML in predicting IDH status for gliomas stated sensitivity and specificity ranging from 87% to 93%.[4]

Another retrospectively cohort of 199 adult patients with glioblastoma was analysed using pre-operative MRI and 9 different molecular biomarkers. The imaging protocol included 3D T2, T2/FLAIR, SWI, DWI, pre- and post-

contrast T1, 3D ASL, and 2D 55-direction HARDI. Three key components of glioblastoma, enhancing, non-enhancing tumour, and surrounding oedema were used to create a 3D augmented segmentation. Out of all, genetic biomarker predictions were most precise for ATRX mutations with a sensitivity of 0.94 ± 0.07 and AUC of 0.97 ± 0.02.[5]

Oligodendrogliomas can have 1p/19q co-deletion and mutations in IDH, CIC, and FUBP1. 1p/19q has a prominent diagnostic and prognostic value plus a pertinent biomarker for surgical planning to avoid non-maximal resection. Matusi et al., predicted 3-molecular subtypes using MRI, PET, and CT to develop a deep learning model which could assess the subtype with 96.6% for the training dataset and 68.7% for the test dataset with an overall accuracy of 68.7%.[6] Brown et al. reported that the prediction of 1p/19q co-deletion through the analysis of MRI image texture achieved a sensitivity of 93% and a specificity of 96%. While radiological imaging alone yields impressive results, integrating machine learning could unlock new and powerful advancements in diagnostics and treatment.[7]

It is pertinent to select the most accurate machine learning model, as this minimizes errors and maximizes data utilization. Urooj et al. reviewed 47 articles on the role of AI in radiomics and assessed four machine learning models, reporting that the Support Vector Machine (SVM) combined with DL techniques exhibited superior accuracy and precision compared to Cox regression and Random Forest algorithms.[8]

The emphasis on the integration of genetic information for gliomas has been reinforced by the WHO keenly. Chang et al., trained CNN network to independently predict the underlying molecular abrasion using data from 259 patients and achieved a high accuracy of 94% for IDH 1 mutations, 92% for 1p/19q co-deletion, and 83% for MGMT promotor methylation status.[9] Lu et al. conducted a retrospective study including 181 MRI studies. A total of 333 radiomics features and 16 Visually Accessible Rembrandt Images (VASARI) features, when combined with clinical data, demonstrate significant potential for predicting overall survival. This approach achieves high predictive accuracy, with an average AUC of 96.2 ± 1.7 and a C-index of 90.0 ± 0.3.[10]

Conclusion

The compelling role of AI and ML in predicting and diagnosing molecular pathology with strong accuracy seems to be a preferable method due to its non-invasive and cost-effective diagnostic technology compared to surgical biopsy and histopathological evaluation. Diagnosis cannot rely solely on machine learning due to insufficient data and the absence of standardized, specialized software. Both of these avenues are pioneering and inspire researchers to develop novel modalities that could revolutionize the diagnosis and treatment of brain tumours.

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