ARIA
APPLIED RESEARCH IN ACTION

Prediction of Genetic Mutations in Pediatric Brain Tumors Using Pathology Images

Multimodal Genetic Mutation Prediction: Leveraging Convolutional Neural Networks for Advanced Histology Analysis and Evaluating the Impact of Clinical Data Integration

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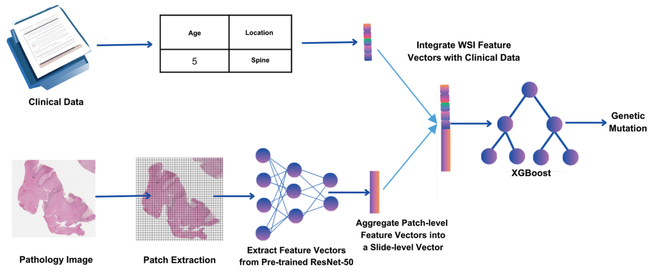

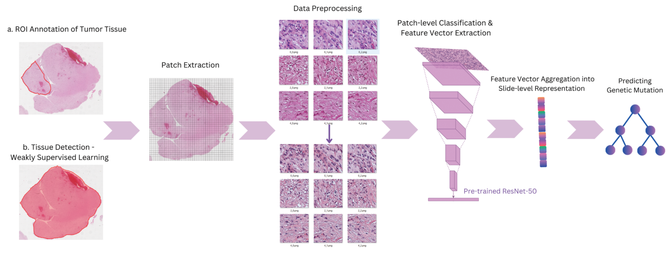

PROJECT SUMMARY

Brain tumors are the leading cause of cancer-related childhood mortality. Identifying genetic driver mutations in these tumors is increasingly important to guide treatments. We hypothesized that mutations could be predicted from routine pathology images in pediatric low-grade gliomas (LGGs), providing a potentially faster and cheaper alternative to genetic sequencing. We used 129 whole slide images (WSI) from LGG cases across the three most common genetic drivers: BRAF fusion (73), BRAF V600E (37), and FGFR alterations (19). 90 slides were allocated to the training set, with 19 for validation and 20 for the hold-out test set. Specifically, gigapixel whole slide images (WSIs) are tessellated into smaller patches, which are analyzed by a pre-trained ResNet50 convolutional neural network to extract feature vectors from the penultimate layer. Patch-level feature vectors from each slide are aggregated to form a single, comprehensive slide-level feature vector. Using these vectors, we train XGBoost for robust genetic mutation classification. Our preliminary results indicate an accuracy of 85%, an ROC AUC score of 0.88, and an F1-score of 0.84 in predicting the driver mutation in WSIs in the test set. These results offer a proof of principle of the ability to predict genetic mutations in LGGs using WSIs, which could be leveraged for rapid diagnosis, particularly in lower income countries.

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