News Release

Ultra-Rapid Intraoperative Genetic Diagnosis of Brain Tumors Achieved in Just 25 Minutes
– A Breakthrough in Real-Time Brain Tumor Diagnosis During Surgery –

Key Points

• We established a novel intraoperative genetic testing method capable of detecting two key mutations critical for the diagnosis of gliomas—the most common type of brain tumor—within 25 minutes. This represents a significant reduction in turnaround time compared to conventional methods.

• The method demonstrated extremely high diagnostic accuracy, with 98.5% sensitivity and 98.2% specificity for detecting *IDH1* mutations, and 100% sensitivity and specificity for *TERT* promoter mutations.

• Intraoperative rapid genetic analysis at multiple tumor sites may contribute to the identification of tumor–normal boundaries and enable more precise determination of the extent of resection.

Summary

A research group led by Sachi Maeda, Fumiharu Ohka, and Ryuta Saito from the Department of Neurosurgery, Nagoya University Graduate School of Medicine, has developed a novel genetic testing system enabling rapid molecular diagnosis of brain tumors. This system is the world's first clinically applicable method capable of detecting tumor-specific genetic mutations within 25 minutes from tissue sampling. By combining the ultra-fast real-time PCR platform "GeneSoC®," which utilizes microfluidic technology, with an original protocol that extracts high-quality DNA through simple heat treatment, the team successfully detected two mutations critical for glioma diagnosis—*IDH1* mutations and *TERT* promoter mutations—with both speed and high accuracy. Intraoperative testing was conducted in 120 clinical cases. The system achieved a diagnostic sensitivity of 98.5% and specificity of 98.2% for *IDH1* mutation, and 100% sensitivity and specificity for *TERT* promoter mutations. Because these mutations are specific to tumor cells, real-time analysis of multiple intraoperative specimens allows for precise assessment of tumor–normal tissue boundaries based on the presence or absence of genetic alterations. This demonstrates the potential of the technology as a valuable tool for optimizing the extent of tumor resection during surgery. This innovative system is expected to enhance the accuracy of brain tumor diagnosis and improve the precision of surgical procedures.

The results of this research were published in the journal Neuro-Oncology on August 24, 2025.

Research Background

Among brain tumors, gliomas are particularly aggressive and have a high recurrence rate, making early and accurate diagnosis as well as appropriate treatment planning critically important. In recent years, molecular diagnosis based on genetic analysis has become central to brain tumor classification, in addition to traditional histopathological evaluation. In particular, *IDH1* mutation and *TERT* promoter mutations are not only important molecular markers of tumor cells, but also play a key role in determining tumor type and malignancy grade. If these molecular abnormalities can be identified during surgery, they may provide valuable information for intraoperative discrimination between tumor and normal tissue, and for determining the optimal extent of resection based on tumor biology. However, conventional genetic testing methods typically require 1–2 days to yield results, making it difficult to obtain molecular information in real time during surgery.

Research Results

In this study, we developed a novel genetic testing system for intraoperative molecular diagnosis of gliomas and evaluated its clinical utility in 120 brain tumor cases. Tumor tissue samples obtained during surgery were rapidly processed to extract DNA, and the presence of *IDH1* R132H and *TERT* promoter mutations (C228T/C250T) was assessed. When compared with postoperative Sanger sequencing results, the rapid genetic analysis showed high diagnostic performance—98.5% sensitivity and 98.2% specificity for *IDH1* R132H, and 100% sensitivity and specificity for *TERT* promoter mutations. The results also demonstrated excellent concordance with final pathological diagnoses, supporting the reliability of this system as an intraoperative diagnostic tool. The average time required per sample was 7.57 minutes for tissue collection and DNA extraction, followed by 14.29 minutes for *IDH1* analysis and 17.15 minutes for *TERT* promoter analysis. In all cases, results were obtained within 25 minutes, confirming the feasibility of using this system to inform surgical decision-making in real time. In addition, we conducted intraoperative assessments of tumor–normal boundaries by sampling multiple regions within the same tumor and evaluating the presence or absence of genetic mutations at each site. This approach was particularly effective in tumors harboring *IDH1* mutations, as the mutation serves as a reliable marker for distinguishing tumor cells from normal cells. A loss of the *IDH1* mutation signal indicated that the

sampling site likely extended beyond the tumor boundary, which was consistent with subsequent pathological findings. This demonstrates the potential of rapid intraoperative identification of molecular alterations to guide real-time surgical decisions and optimize the extent of tumor resection based on molecular markers. Furthermore, the DNA extracted using this rapid method was confirmed to be of sufficient quality for other molecular analyses, including whole-genome sequencing and DNA methylation profiling.

Research Summary and Future Perspective

We aim to expand the range of detectable genetic markers and further improve diagnostic accuracy. In addition, we are developing a multi-gene panel that enables the simultaneous detection of multiple mutations using this technology. Our goal is to establish a more detailed and user-friendly system for intraoperative molecular diagnosis.

Publication

Journal：Neuro-Oncology
Title：Rapid Intraoperative Genetic Analysis of Adult-type Diffuse Gliomas Using a Microfluidic Real-Time Polymerase Chain Reaction Device
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