News Release

Title

Rapid Detection of the *MYD88* L265P Mutation for Pre- and Intra-operative Diagnosis of Primary Central Nervous System Lymphoma

Key Points

- It is now possible to genotype the *MYD88* L265P mutation which is useful for the diagnosis of CNS malignant lymphoma in approximately 15 minutes.
- Since it can be analyzed with a simple procedure, it can be done in the operating room and is a very effective technique for intra-operative diagnosis.
- In the future, it is expected to be applied to liquid biopsy using cerebro spinal fluid.

Summary

The *myeloid differentiation primary response gene 88* (*MYD88)* L265P mutation is a disease-specific mutation of primary central nervous system lymphoma (PCNSL). Accordingly, this mutation is considered a reliable diagnostic molecular marker of PCNSL. As the intra-operative diagnosis of PCNSL is sometimes difficult to achieve using histological examinations alone, intra-operative detection of the *MYD88* L265P mutation could be effective for the accurate diagnosis of PCNSL. Herein, we aimed to develop a novel rapid genotyping system (GeneSoC) using real-time polymerase chain reaction (PCR) based on microfluidic thermal cycling technology. This real-time PCR system shortened the analysis time, which enabled the detection of the *MYD88* L265P mutation within 15 min. Rapid detection of the *MYD88* L265P mutation was performed intra-operatively using GeneSoC in 24 consecutive cases with suspected malignant brain tumors, including ten cases with suspected PCNSL before surgery. The *MYD88* L265P mutation was detected in eight cases in which tumors were pathologically diagnosed as PCNSL after the operation, while wild-type *MYD88* was detected in 16 cases. Although two of the 16 cases with wild-type *MYD88* were pathologically diagnosed as PCNSL after the operation, *MYD88 L265P* could be detected in all eight PCNSL cases harboring *MYD88 L265P*. The *MYD88* L265P mutation could also be detected using cell-free DNA derived from the cerebrospinal fluid of two PCNSL cases. Detecting the *MYD88* L265P mutation using GeneSoC might not only improve the accuracy of intra-operative diagnosis of PCNSL but also that of pre-operative diagnosis through liquid biopsy using cerebrospinal fluid.

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