A genotyping system is a tool used to determine an individual's genetic makeup, usually by analyzing their DNA. There are various genotyping methods, including PCR (polymerase chain reaction) and sequencing techniques, that can be used to identify genetic variations or mutations in a person's DNA.

Genotyping systems are widely used in research, clinical diagnostics, and forensic investigations. They can be used to study genetic diseases, identify individuals at risk for certain conditions, and track the spread of diseases in populations.

There are several commercial genotyping systems available, such as Illumina's BeadChip and Thermo Fisher Scientific's TaqMan assays, that can genotype thousands of genetic markers at once. These systems are often used in large-scale genetic studies to identify genetic associations with disease or other traits.

Yamaguchi et al. 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 10 cases with suspected Primary central nervous system lymphoma 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. Detection of the MYD88 L265P mutation using GeneSoC might not only improve the accuracy of intra-operative diagnosis of PCNSL but also help the future pre-operative diagnosis through liquid biopsy of cerebrospinal fluid [1)]

1)
Yamaguchi J, Ohka F, Kitano Y, Maeda S, Motomura K, Aoki K, Takeuchi K, Nagata Y, Hattori H, Tsujiuchi T, Motomura A, Nishikawa T, Kibe Y, Shinjo K, Kondo Y, Saito R. Rapid detection of the MYD88 L265P mutation for pre- and intra-operative diagnosis of primary central nervous system lymphoma. Cancer Sci. 2023 Mar 1. doi: 10.1111/cas.15762. Epub ahead of print. PMID: 36859777.