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Utility of genome-wide DNA methylation profiling for pediatric-type diffuse gliomas

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Abstract

Despite the current progress of treatment, pediatric-type diffuse glioma is one of the most lethal primary malignant tumors in the central nervous system (CNS). Since pediatric-type CNS tumors are rare disease entities and highly heterogeneous, the diagnosis is challenging. An accurate diagnosis is essential for the choice of optimal treatment, which leads to precision oncology and improvement of the patient's outcome. Genome-wide DNA methylation profiling recently emerged as one of the most important tools for the diagnosis of CNS tumors, and the utility of this novel assay has been reported in both pediatric and adult patients. In the current World Health Organization classification published in 2021, several new entities are recognized in pediatric-type diffuse gliomas, some of which require methylation profiling. In this review, we investigated the utility of genome-wide DNA methylation profiling in pediatric-type diffuse glioma, as well as issues in the clinical application of this assay. Furthermore, the combination of genome-wide DNA methylation profiling and other comprehensive genomic assays, which may improve diagnostic accuracy and detection of the actionable target, will be discussed.

Keywords: Genome-wide DNA methylation profiling; Pediatric brain tumor; Pediatric-type diffuse glioma.