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Current status of DNA methylation profiling in neuro-oncology as a diagnostic support tool: A review

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Abstract

Over the last 2 decades, high throughput genome-wide molecular profiling has revealed characteristic genetic and epigenetic alterations associated with different types of central nervous system (CNS) tumors. DNA methylation profiling has emerged as an important molecular platform for CNS tumor classification with improved diagnostic accuracy and patient risk stratification in comparison to the standard of care histopathological analysis and any single molecular tests. The emergence of DNA methylation arrays have also played a crucial role in refining existing types and the discovery of new tumor types or subtypes. The adoption of methylation data into neuro-oncology has been greatly aided by the development of a freely accessible machine learning-based classifier. In this review, we discuss methylation workflow, address the utility of DNA methylation profiling in CNS tumors in a routine diagnostic setting, and provide an overview of the methylation-based tumor types and new types or subtypes identified with this platform.

Keywords: CNS tumors; DKFZ classifier; DNA methylation; WHO CNS 5 classification; new type/subtype.

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