

Machine-learning algorithm used to identify specific types of brain tumors

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An international team of researchers has used methylation fingerprinting data as input to a machine-learning algorithm to identify different types of brain tumors. In their paper published in the journal *Nature*, the team describes studying DNA methylation fingerprinting to create a system

that is able to identify central nervous system (CNS) tumors and report on its accuracy.

Accurately identifying a patient's specific tumor type is difficult for doctors because of the limited number of tests available and the large number of types—there are over 100 known, with some yet to be identified. In this new effort, the researchers studied methylation, in which methyl groups are added to DNA—the process induces changes in function, typically by repressing gene transcription. Methylation is a normal process, and one of its hallmarks is that it leaves a unique signature. Noting that the process also occurs in cancer cells, the researchers set out to use such signatures as fingerprints to identify the presence of [tumor cells](#) in a sample. They created a system based on a [machine-learning algorithm](#) that uses methylation fingerprinting data as its learning material.

To teach the system to recognize and then identify tumor types, the researchers entered data from 2,800 cancer patients and report that it is now capable of identifying 91 types of CNS tumors. They tested the system with tumor data from 1,104 patients with previously identified tumors. The system found that approximately 12 percent of the tumors had been misdiagnosed.

The researchers note that in addition to providing a new tool for tumor identification in new patients, the system is also able to discern whether a given tumor is one that has not been previously identified, circumventing the common occurrence of doctors feeling pressured into classifying tumors as belonging to known types. To make the system more approachable, the team has put their system [online free of charge](#). As of the publication of their paper, the system had been accessed over 4,500 times, suggesting people are finding it useful. Such widespread use, the [researchers](#) note, will only improve the accuracy of the system and hopefully aid in discovering new [tumor](#) types.

More information: David Capper et al. DNA methylation-based classification of central nervous system tumours, *Nature* (2018). [DOI: 10.1038/nature26000](https://doi.org/10.1038/nature26000)

Abstract

Accurate pathological diagnosis is crucial for optimal management of patients with cancer. For the approximately 100 known tumour types of the central nervous system, standardization of the diagnostic process has been shown to be particularly challenging—with substantial inter-observer variability in the histopathological diagnosis of many tumour types. Here we present a comprehensive approach for the DNA methylation-based classification of central nervous system tumours across all entities and age groups, and demonstrate its application in a routine diagnostic setting. We show that the availability of this method may have a substantial impact on diagnostic precision compared to standard methods, resulting in a change of diagnosis in up to 12% of prospective cases. For broader accessibility, we have designed a free online classifier tool, the use of which does not require any additional onsite data processing. Our results provide a blueprint for the generation of machine-learning-based tumour classifiers across other cancer entities, with the potential to fundamentally transform tumour pathology.

[Press release](#)

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