

Central nervous system tumor diagnosis

see [Intracranial tumor diagnosis](#)

Accurate diagnosis is essential for optimal clinical management. The increasing use of molecular diagnostics has opened up novel possibilities for more precise classification of CNS tumors.

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.

Bächli et al., report a single-institutional collection of [pediatric brain tumor](#) cases that underwent a refinement or a change of diagnosis after completion of molecular analysis that affected clinical decision-making including the application of molecularly informed targeted therapies. 13 pediatric CNS tumors were analyzed by conventional histology, immunohistochemistry, and molecular diagnostics including DNA methylation profiling in 12 cases, DNA sequencing in 8 cases and RNA sequencing in 3 cases. 3 tumors had a refinement of diagnosis upon molecular testing, and 6 tumors underwent a change of diagnosis. Targeted therapy was initiated in 5 cases. An underlying cancer predisposition syndrome was detected in 5 cases. Although this case series, retrospective and not population based, has its limitations, insight can be gained regarding precision of diagnosis and clinical management of the patients in selected cases. Accuracy of diagnosis was improved in the cases presented here by the addition of molecular diagnostics, impacting clinical management of affected patients, both in the first-line as well as in the follow-up setting. This additional information may support the clinical decision making in the treatment of challenging pediatric CNS tumors. Prospective testing of the clinical value of molecular diagnostics is currently underway ¹⁾.

¹⁾

Bächli H, Ecker J, van Tilburg C, Sturm D, Selt F, Sahm F, Koelsche C, Grund K, Sutter C, Pietsch T, Witt H, Herold-Mende C, von Deimling A, Jones D, Pfister S, Witt O, Milde T. Molecular Diagnostics in Pediatric Brain Tumors: Impact on Diagnosis and Clinical Decision-Making - A Selected Case Series. *Klin Padiatr.* 2018 Jul 11. doi: 10.1055/a-0637-9653. [Epub ahead of print] PubMed PMID: 29996150.

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