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A pathologist's guide for the diagnostic workup of paediatric central nervous system tumours

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Abstract

Advances in precision medicine and our understanding of the molecular drivers of central nervous system (CNS) tumorigenesis in children have broadened the scope of diagnostic testing that is required on paediatric CNS tumour samples. The pathologist plays a central role in ensuring that the correct test is ordered, in the integration of test results into the diagnosis and in recognising therapeutic targets to guide targeted treatment planning. The diagnostic and molecular workup of many of the prototypical paediatric CNS tumours differs from that required for adult CNS tumours and can be particularly challenging when tissue is limited. Many paediatric CNS tumours are driven by Rat sarcoma virus (RAS)-mitogen-activated protein kinase (MAPK) pathway or histone alterations, a subset are fusion or single-nucleotide variant (SNV) driven, whereas others require specific molecular subgrouping for treatment planning. This review summarises the clinicopathological and molecular features of some of the more prototypical paediatric CNS tumours and provides a practical guide for the pathologist regarding the molecular workup of paediatric CNS tumours. Common diagnostic dilemmas relevant to the diagnosis of paediatric CNS tumours encountered by the paediatric neuropathologist will be explored, together with some suggested approaches to overcoming these. It is hoped this will aid the pathologist to reach a more accurate and clinically informative diagnosis for paediatric CNS tumours.

Keywords: DNA methylation profiling; NanoString; diffuse midline glioma; embryonal tumour; molecular testing; next-generation sequencing; paediatric CNS tumour; paediatric high-grade glioma; paediatric low-grade glioma; targeted therapy.

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