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Update on cancer predisposition syndromes and surveillance guidelines for childhood brain tumors

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Abstract

Tumors of the central nervous system (CNS) comprise the second most common group of neoplasms in childhood. The incidence of germline predisposition among children with brain tumors continues to grow as our knowledge on disease aetiology increases. Some children with brain tumors may present with non-malignant phenotypic features of specific syndromes (e.g. nevoid basal cell carcinoma syndrome, neurofibromatosis type 1 and type 2, DICER1 syndrome, and constitutional mismatch repair deficiency), while others may present with a strong family history of cancer (e.g. Li-Fraumeni syndrome), or with a rare tumor commonly found in the context of germline predisposition (e.g. rhabdoid tumor predisposition syndrome). Approximately 50% of patients with a brain tumor may be the first in a family identified to have a predisposition. The past decade has witnessed a rapid expansion in our molecular understanding of CNS tumors. A significant proportion of CNS tumors are now well characterized and known to harbor specific genetic changes that can be found in the germline. Additional novel predisposition syndromes are also being described. Identification of these germline syndromes in individual patients has not only enabled cascade testing of family members and early tumor surveillance but increasingly has also impacted cancer management in those patients. Therefore, the AACR Cancer Predisposition Working Group chose to highlight these advances in CNS tumor predisposition and summarize and/or generate surveillance recommendations for established and more recently emerging pediatric brain tumor predisposition syndromes.