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Extraneural Metastases of Diffuse Midline Glioma, H3 K27M-Mutant at Diagnosis: Case Report, Review of the Literature, and Identifying Targetable Alterations

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Abstract

Extraneural metastases are rare in pediatric high-grade gliomas and little is known about the genomic profiles of tumors that disseminate beyond the central nervous system. We describe a pediatric patient with H3 K27M-mutant diffuse midline glioma of the brain and spine with biopsy-confirmed osseous metastases present at diagnosis and suspected metastatic parenchymal pulmonary disease. Several potentially clinically and/or therapeutically relevant genomic alterations were identified, including H3F3A and TP53 mutations as well as MET, CDK6, EMSY, and PIK3CG amplifications. Sequencing is critical to improve our understanding of the molecular drivers of distant metastases and discover therapeutic targets that penetrate all disease sites.

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1 di 1 03/06/2021, 16:02