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Neuroepithelial tumor with EWSR1::PATZ1 fusion: A literature review

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Abstract

We present the clinicopathological and molecular genetic characteristics of a neuroepithelial tumor (NET), EWSR1::PATZ1 fusion-positive with a literature review. This fusion has recently been discovered in rare central nervous system tumors and soft tissue sarcomas and was not included in the fifth edition of the WHO classifications. We identified this fusion in 2 NETs. The first case involved a 7-year-old girl and the second case occurred in a 53-year-old man; both presented with headaches and vomiting. The pediatric case initially showed an intermediate grade of the tumor, but upon recurrences, it transformed into a high-grade tumor with 2 relapses in 8.3 years. This case exhibited high mitotic activity (20/10 high-power fields), and a high Ki-67 index (21%). The TERT promoter (TERTp) mutation was present in both initial and recurrent tumors. In contrast, the adult case was a low-grade tumor with no mitotic activity or recurrence over 13.5 months after subtotal resection and gamma knife surgery. Interestingly, the pediatric case demonstrated a longer survival time compared to conventional glioblastoma. The TERTp mutation, similar to being a molecular signature in adult-type glioblastoma, could also be an indicator of high-grade behavior in PATZ1 fusion NET.

Keywords: EWSR1::PATZ1 fusion; Brain tumor; Neuroepithelial tumor; Next-generation study.