Atypical teratoid/rhabdoid tumor of the central nervous system in an 18-year-old patient.


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OBJECTIVE: Atypical teratoid/rhabdoid tumors are aggressive neoplasms of the central nervous system occurring mainly in the early childhood and rarely in adults. We described a case of this tumor in an 18-year-old male patient without previous medical history.

MATERIAL AND METHODS: The neoplasm was localized in the right frontotemporal area of the brain and was totally excised. The specimen was fixed in formalin and embedded in paraffin. The histological and immunohistochemical features of the neoplasm were assessed, while sequencing analysis as well as interphase fluorescence in situ hybridization (FISH) were performed. RESULTS: Histological and immunohistochemical analysis demonstrated atypical rhabdoid cells strongly and diffusely positive for EMA and Vimentin as well as focally immunoreactive for SMA and GFAP. Additionally, though no abnormalities detected in the coding sequence of the INI1 gene, interphase FISH studies were consistent with a homozygous deletion of the INI1 gene in the majority of examined nuclei. INI1 immunostaining demonstrated diffuse loss of nuclear INI1 expression in tumor cells. Taken together, the results were consistent with a diagnosis of atypical teratoid/rhabdoid tumor (ATRT). CONCLUSIONS: 26 previous cases of ATRT have been reported in adults, thus far. To our knowledge, this is the eighth case of an ATRT reported in an adult patient having genetic confirmation and the first one in which the tumor is, partly, localized in the right temporal area of the brain. This unusual presentation underlines the necessity of considering this devastating neoplasm in the differential diagnosis of malignant brain tumors of young adults.

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