Atypical teratoid/rhabdoid tumor arising in pleomorphic xanthoastrocytoma: a case report.

Jeong JY, Suh YL, Hong SW.

Abstract
Atypical teratoid/rhabdoid tumor (AT/RT) is a rare, highly malignant, true rhabdoid tumor in the central nervous system predominantly presenting in young children. AT/RT typically shows rhabdoid cells which can also be seen in other tumors, but it is differentiated from other tumors by the specific genetic alteration involving the SMARCB1 gene. Only a few cases of AT/RT arising in low-grade glioma have been reported. A 13-year-old girl presented with headache, dizziness, nausea and vomiting. A 4.7 cm cerebellar mass was found on MRI. The mass was totally removed. Histologically, the tumor revealed two distinct morphologic appearances: central areas of AT/RT containing rhabdoid cells and sarcomatous component in the background of pleomorphic xanthoastrocytoma (PXA). Immunohistochemically, PXA areas retained nuclear expression of INI-1 and low Ki-67 proliferation index, whereas AT/RT component showed loss of INI-1 nuclear expression and markedly elevated Ki-67 proliferation index. Epithelial membrane antigen (EMA), smooth muscle actin (SMA), and p53 protein were positive only in AT/RT. BRAF V600E mutation was identified in PXA by real-time polymerase chain reaction. We report a rare case of AT/RT arising in PXA which is supposed to progress by inactivation of INI-1 in a pre-existing PXA.

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