Medulloblastoma with multi-lineage differentiation including myogenic and melanotic elements: a case report with molecular data.

Stefanits H, Ebetsberger-Dachs G, Weis S, Haberler C.

Abstract

We present an unusual medulloblastoma in a 3.9-year-old boy who had a 2-week history of nausea and vertigo. MRI revealed a 5×5.5×5 cm sized tumor located in the fourth ventricle and spinal leptomeningeal dissemination. The patient was treated according to the MET-HIT 2000-BIS4 protocol but showed tumor progression after 6 months and died 9 months postoperatively. Histopathologically and immunohistochemically, the tumor showed PNET-like areas with focal anaplasia, admixed rhabdomyoblastic and pigmented elements, cartilage and bone formation, as well as areas with neurocytic and glial differentiation. Neither CTNNB1 mutation nor MYCC/MYCN amplification was detected. The combination of rhabdomyoblastic and melanotic elements in medulloblastoma is exceptionally rare. Although the histopathological features suggested a teratoid tumor, the endodermal cell lineage required for this diagnosis was not present. An atypical teratoid-rhabdoid tumor was ruled out due to the presence of the INI1-protein. Regarding the molecular profile with 1q and 17q chromosomal gains and loss of chromosome 8, this tumor could be compatible with a molecular medulloblastoma Group 3 or 4. Yet, it cannot be definitively ruled out that medulloblastomas with multi-lineage differentiation represent a distinct subgroup of medulloblastoma, and it remains to be clarified whether these tumors are associated with a distinct clinical behavior.

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