Medullomyoblastoma: a case report and literature review of a rare tumor entity.

Smolle E, Al-Qubati S, Stefanits H, Haberler C, Kleinert R, Haybaeck J.
Department of Neuropathology, Institute of Pathology, Medical University Graz, Auenbrugger-Platz 25, A-8036 Graz, Austria.

Abstract

BACKGROUND: Medullomyoblastoma (MMB) is a very rare medulloblastoma (MB) variant consisting of primitive neuroectodermal cells intermixed with cells featuring myogenic differentiation. MMBs are a subtype of primitive neuroectodermal neoplasm (PNET) predominantly occurring in children.

CASE REPORT: We describe a case of a one-year-old girl who presented with headache, emesis and ataxia. The symptoms had started seven weeks before hospital admission. Magnet resonance imaging of the brain was performed, and revealed a lesion with a maximal diameter of 5 cm, located in the cerebellum close to the vermis. Histologically, the poorly-differentiated lesion was diagnosed as a type of PNET, but it was the immunohistochemical staining that assured the diagnosis of MMB.

RESULTS: Immunohistochemistry and interphase fluorescence in situ hybridization (I-FISH) were performed on formalin-fixed paraffin-embedded tissue. FISH did not reveal any amplification of CMYC or NMYC. No nuclear expression of β-catenin was detectable.

DISCUSSION: Since MMB is a very rare tumor entity, standard treatment today is the same as that for conventional MB due to the lack of larger study series. Some authors assume that MMBs behave especially aggressive in comparison to conventional MBs. Therefore, new treatment regimes should be tested to optimize the prognosis of MMB. Further data is needed to determine the differences between MB and MMB.

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