A patient with medulloblastoma in its early developmental stage.

Abstract
Medulloblastoma is the most frequent malignant brain tumor of the posterior fossa in children and is considered an embryonal tumor. It has been suggested that medulloblastomas be categorized into 4 distinct molecular subgroups—WNT (DKK1), SHH (SFRP1), Group 3 (NPR3), or Group 4 (KCNA1)—since each subgroup is distinct and there is no overlap. The authors report on a 13-year-old boy with medulloblastoma. He presented with sudden-onset nausea and vomiting due to intratumoral hemorrhage. The medulloblastoma was thought to be in an early developmental stage because the tumor volume was extremely small. Immunohistochemical analysis showed that the tumor was mainly composed of DKK1- and NPR3-positive areas. The individual areas of the tumor stained only for DKK1 or NPR3, with no overlap—that is, DKK1 and NPR3 expression were mutually exclusive. Samples obtained by laser microdissection of individual areas and subjected to mass spectrometry confirmed that the expression patterns of proteins were different. Fluorescence in situ hybridization for chromosome 6 showed there were 2 distinct types of cells that exhibited monosomy or disomy of chromosome 6. These results demonstrated that distinct subtypes of medulloblastoma may be present within a single tumor, an observation that has not been previously reported. Our findings in this case indicate that early-stage medulloblastoma may include more than 1 distinct subtype and hint at factors involved in the origin and development of medulloblastomas.

KEYWORDS: FFPE = formalin-fixed, paraffin-embedded; FISH = fluorescence in situ hybridization; LC = liquid chromatography; MS = mass spectrography; Rsc = ratio from spectral counting; genetic subgroup; heterogeneity; intratumoral hemorrhage; medulloblastoma; oncology; tumor development

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