Extra-axial Desmoplastic/Nodular Medulloblastoma, SHH-activated in an adult: a case report

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Introduction

Medulloblastoma (MB) is an embryonal central nervous system (CNS) tumor located in the posterior cranial fossa. Medulloblastoma is one of the most common malignant pediatric brain tumors, accounts for 25% of all pediatric intracranial tumors, but rare in adults; and constitute 0.4%-1% of adult primary brain tumors. The incidence of medulloblastomas in adults is approximately 0.5 per million per year and decreases with age with male predominance. Medulloblastomas classically appear as well-defined masses in the cerebellum that enhance both computed tomography (CT) of the head and brain magnetic resonance imaging (MRI) with gadolinium contrast. The clinical symptoms and signs of MB in adults are associated with the location of the tumor, increased intracranial pressure, and/or obstruction of the cerebrospinal fluid pathway leading to headache, dizziness, nausea, ipsilateral cerebellar signs, and ataxia.

Case Presentation

A 31-year-old female presented with 3 weeks of intermittent headaches with dizziness, followed by vomiting. The neurological exam was unremarkable. The patient was started on dexamethasone after a non-contrast brain CT revealed a 5 cm ill-defined cerebellar mass and was suggestive of early herniation. Follow-up magnetic resonance imaging with contrast showed a 4.7x4.1x4.3 cm enhancing extra-axial mass (Fig. 1)

Results

Histopathologic evaluation of the tumor biopsy showed a highly cellular embryonal neoplasm with prominent nuclear molding and brisk mitotic activity, and also scattered karyorrhexis; and occasional areas of the tumor show apparent nodular growth(Fig. 2 and 3). Some tumor cells were immunoreactive for GFAP while synaptophysin shows weak reactivity in much of the tumor with slightly stronger staining of tumor nodules. A reticulin-rich stroma is apparent in many areas (Fig. 3). Molecular studies showed p53 wild-type pattern with IHC, no amplification of MYC by FISH, no amplification of MYCN by FISH, SHH molecular group. This case is diagnosed as Medulloblastoma, desmoplastic/nodular, SHH-activated, CNS WHO grade 4.

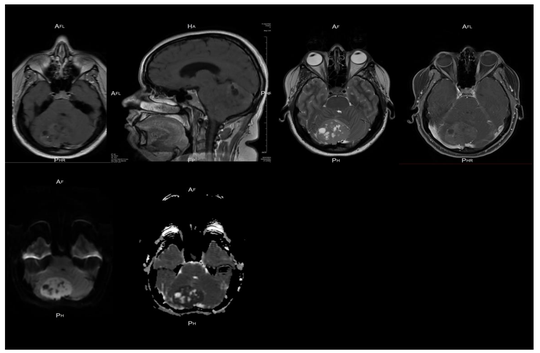

Figure 1: Top row: T1 post contrast without fat saturation axial - T1 post contrast without fat saturation sagittal - T2 axial - T1 post contrast with fat saturation axial Bottom row: Diffusion weighted imaging (DWI; abnormal is bright) - Apparent Diffusion Coefficient Map (ADC 10^-6 mm2/s; abnormal is dark)

Results (Continued)

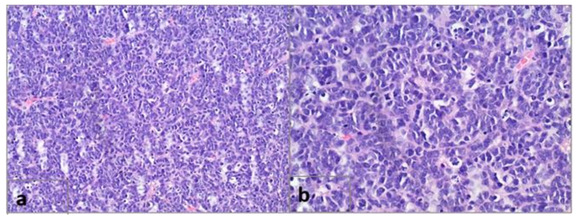

Figure 2: a- Highly cellular embryonal neoplasm with small cell phenotype, arranged in sheet like pattern (H-E; 10x) b- Tumor cells show prominent nuclear molding and brisk mitotic activity, and also scattered karyorrhexis. (H-E; 20x)

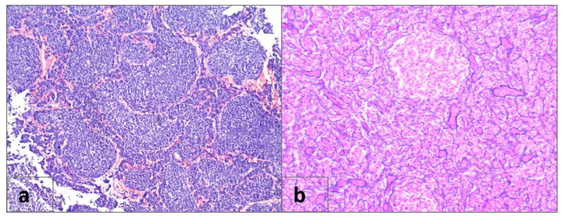

Figure 3: a- Tumor shows occasional areas of nodularity. (H-E; 10x) b-Tumor nodules lacking staining with reticulin. (Reticulin; 4x)

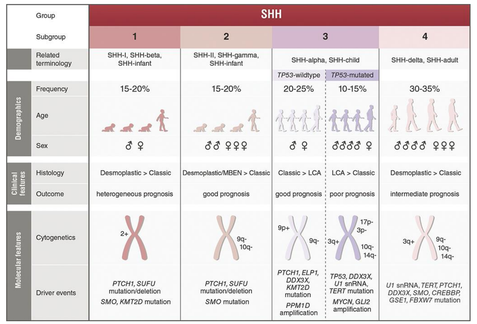

Figure 4. SHH activated medulloblastoma subgroups (WHO 5th ed)

Discussion

Adult MBs unlike pediatric are reported to be of two molecular subtypes, instead of four, with Group 3 occurring as an exception and Group 4 restricted to midline. Lateral location is reserved for WNT and SHH tumors, with the former mostly seen in CPA and the latter in the cerebellar hemisphere. Also, superior location abutting and/or reaching the tentorium is a specific imaging feature of SHH-subgroup MB seen in 48% of patients as opposed to 6% in other groups.

In a study done by Zhao et al., it was shown that there is a preponderance of SHH-type tumors in adult MB (62%), followed by group 4 tumors (28%) and WNT-activated tumors (10%), with an absence of group 3 cases, suggesting that this subgroup may be restricted to pediatric MB. There have been only 12 reported cases of extra-axial MB in the adult literature. There have been only 12 reported cases of extra-axial MB in the adult literature. Ten out of 12 reports so far published considered meningioma as a provisional diagnosis based on broad tentorial attachment, despite heterogeneous enhancement.

Conclusion

Adult MB is distinct from MB in children from a molecular and clinical perspective. Even in adults over 50 years old, medulloblastoma should be included in the differential diagnosis of posterior fossa tumor. SHH-activated TP53-wildtype MB represents the most frequent type of MB in adults. The majority of SHH-MB TP53-wildtype tumors show desmoplastic/nodular histological patterns, which may also be present in focal areas only, while MB with extensive nodularity is not observed in adults.

References

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