G35 Primitive Neuroectodermal Tumor (PNET) Masquerading as Non Accidental Head Trauma in an Infant: Lessons for Multiple Disciplines

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After attending this presentation, attendees will understand the process of evaluating intracranial pathology in an infant and how to identify spontaneous as well as traumatic causes of intracranial hemorrhage, specifically, hemorrhage resulting from a supratentorial primitive neuroectodermal tumor.

The ability to discriminate not only between nonaccidental and accidental head trauma, but also between spontaneous and traumatic etiologies of intracranial hemorrhage, while keeping an open mind to all of the possibilities during the descision-making process, is among the most challenging of intellectual tasks that confront the froensic pathologist.

The appropriate recognition and accurate documentation of fatal nonaccidental craniocerebral trauma in infants and young children are among the most important contributions to be made by a forensic pathologist. Equally crucial, however, is the ability to identify both accidental causes of head trauma and spontaneous (nontraumatic) etiologies of intracranial pathology.

A rare cause of spontaneous intracerebral, subarachnoid, and subdural hemorrhage in a 3-month-old child resulted from a supratentorial primitive neuroectodermal tumor (PNET) with glial differentiation, emphasizing that not all forms of intracranial pathology are inflicted, or even traumatic. Furthermore, because the various interpretations of the imaging studies created significant controversy and disagreement regarding the assessment of the disease process findings, the potential problems that exist with neuroradiologic interpretations of lesions in this age group are emphasized.

An 11-week-old male infant presented to the MUSC Pediatric Emergency Department (ED) with a 2-day-long history of lethargy, decreased oral intake and urine output, and favoring of the left side of his face. His mother stated that his left eye had appeared “abnormal” for the past few weeks. In the ED, he was found to have a bulging fontanelle and a fixed and dilated left pupil with a minimally reactive right pupil. Of note, prenatal history was unremarkable; he was born at 39 weeks gestation with 1- and 5-minute Apgar scores of 9 and 9. He had been essentially healthy up to his current presentation. A head CT scan demonstrated obstructive hydrocephalus and transtentorial herniation, prompting admission to the Pediatric Intensive Care Unit (PICU) with ventriculostomy placement for elevated intracranial pressure (ICP). However, he succumbed to refractory intracranial hypertension and was declared clinically brain dead on the 7th hospital day. Reports from imaging studies were notably discordant, with one interpreting the findings as “multiple trauma of varying ages”, the other as a “mass”. Autopsy revealed an extensively necrotic and hemorrhagic, supratentorial mass with resultant swelling, softening, and hemorrhage of the adjacent brain parenchyma bilateral thin-layered subdural hemorrhages accompanied by thin-layered subarachnoid hemorrhage, both overlying the cerebral convexities; subgaleal edema and hemorrhage confined to the ventriculostomy site; and anasarca with bilateral serous pleural effusions (15 mL each), and serous ascites (30 mL). There were no other injuries. The eyes were not examined. Microscopic examination of the subdural hemorrhage revealed predominantly red blood cells with rare fibroblasts, and sparse hemosiderin-laden macrophages, consistent with a recent origin (of ~6 days’ duration). Sections of the mass demonstrated sheets of spindled to epithelioid cells with large, eccentric nuclei, punctate, “salt and pepper” chromatin, and inconspicuous nucleoli; some cells contained eosinophilic, smooth, “glassy” cytoplasm. The mass was accompanied by scattered hematoidin pigment and extensive, confluent necrosis. There was no significant nuclear atypia or vascular endothelial proliferation. An immunohistochemical battery employing antibodies to glial fibrillary acid protein (GFAP), synaptophysin, chromogranin A, vimentin, smooth muscle actin (SMA), pancytokeratin (AE1/AE3), low-molecular weight cytokeratin (CM 5.2), CD99, and epithelial membrane antigen (EMA) showed the tumor cells to express only GFAP and none of the other antigens. The collective histopathologic features and immunohistochemical profile of the tumor were most compatible with a PNET manifesting purely glial differentiation.

Sound critical thinking and an open mind on the part of the forensic pathologist when confronted with significant intracranial pathology in an infant is of great importance. Although the impressions of the imaging studies and the presence of bilateral thin-layered subdural and subarachnoid hemorrhages appropriately raised the suspicion of inflicted) head trauma, the identification of a reasonable underlying etiology for the hemorrhage, the absence of injuries to suggest a pattern of repeated abuse, and the mother’s description of neurologic signs entirely consistent with increasing ICP collectively provided strong evidence of a nontraumatic etiology for this baby’s condition.

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