49th ANNUAL DIAGNOSTIC SLIDE SESSION 2008

CASE 2008-8

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Clinical History:

The patient was a 22-year-old female with neurofibromatosis type 1 diagnosed at 6 months of age. She had a past medical history of multiple cervical and thoracic plexiform neurofibromas, cutaneous neurofibromas and café au lait spots. In addition, she carried a history of optic glioma, diagnosed at 2.5 years age, which was treated with external beam radiation during childhood. She subsequently developed growth hormone deficiency (empty sella by MRI) and blindness, requiring eventual enucleation in 2005. For several years, she maintained stable intracranial lesions including the optic glioma, multifocal non-enhancing foci in basal ganglia and thalamus and small areas of cerebellar enhancement.

She presented to the emergency department with recent onset of seizure and 2 month history of nausea and vomiting following meals. An MRI showed new nodular enhancement of dural surfaces of the cerebrum, brainstem and spinal cord. An EEG showed nonspecific generalized abnormalities somewhat more prominent over the right hemisphere. There were no epileptiform abnormalities.

Two days after admission, her mental status changed from having periods of unresponsiveness and lethargy to obtundation. A lumbar puncture was performed and showed numerous mononuclear cells in the cerebrospinal fluid. She then underwent right frontal craniotomy with biopsy of the right frontal lobe and dura (http://dpig.upmc.com/DSS/home.aspx?u=Yeaney [username= trestle password= trestleuser]). On post-op day 2, MRI revealed acute right frontal and temporal infarcts involving both the anterior cerebral and middle cerebral arterial vascular territories, and an acute left middle cerebral territory infarct. MR angiogram of the head demonstrated diffuse narrowing of the middle and posterior cerebral arteries compatible with severe vasculitic change. The aortic arch, bilateral carotid and vertebral arteries appeared to be of small caliber. The patient was treated with high-dose steroids for a clinical diagnosis of vasculitis. Unfortunately, she died a few days later, approximately 1 week following admission.

Necropsy findings:

The fresh brain weighed 1520 grams. Leptomeninges showed focal areas of cloudiness. There was severe thickening of the right internal carotid wall. Other than a suprasellar/ hypothalamic mass, sections of cerebrum and cerebellum showed no masses. Right anterior cerebral artery and bilateral middle cerebral artery territories revealed loss of gray-white demarcation.

Material submitted:

H&E section of cerebrum and cerebellum
Unstained section of cerebrum and cerebellum

Points for discussion:
1. Differential diagnosis; 2. Diagnosis; 3. Pathogenesis