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CASE 2019-#6

Submitted by:
Julieann C. Lee[1] and Joanna J. Phillips[1,2]
1. Department of Pathology, University of California, San Francisco, CA
2. Department of Neurological Surgery, University of California, San Francisco, CA

University of California, San Francisco (UCSF)
Department of Pathology, Division of Neuropathology
505 Parnassus Avenue, #M551, Box# 0102
San Francisco, CA 94143

Clinical History:
The decedent was a 23-year-old female. Her head circumference was below average, and there were early developmental delays. MRI at 10 months of age demonstrated vermian hypoplasia and diminished myelination. She developed and gained skills in the first decade of life, with subsequent progressive motor regression beginning at age 10. By her late teens she was almost entirely wheelchair bound, and was fed primarily by a gastric tube. She became increasingly nonverbal, with dystonia and ballistic movements. Serial MRIs demonstrated progressive volume loss most notable in the periventricular white matter, thalamus, midbrain, and pons. There were bilateral T1 hyperintense signal abnormalities within the caudate, putamen, globus pallidus, thalamus, subthalamic nuclei, hippocampal cortex, red nucleus, substantia nigra, and cortical spinal tract. CT imaging did not show basal ganglia calcifications. Her peripheral iron and magnesium levels were unremarkable. Her eye exam did not reveal retinopathy, Kayser-Fleischer rings, or cataracts. She became anemic in the setting of menorrhagia, dehydrated, and lethargic with episodes of emesis, and began home hospice care. There has been no one else in the family with similar clinical findings.

Autopsy findings:
Autopsy findings included aspiration pneumonia and cachexia. Gross evaluation of the brain showed hydrocephalus ex vacuo with marked volume loss, particularly involving the white matter of the cerebrum, cerebellum, and brainstem. There was reduced volume of the cerebellum and brainstem, with severe hypoplasia/atrophy of the cerebellar vermis.

Material submitted:
H&E section including the right insular cortex, putamen, and globus pallidus.

Magnetic resonance image showing T1 hyperintense signal abnormalities.

Points for discussion:
1. Differential diagnosis
2. Immunohistochemistry, special stains, and molecular evaluation