

## 58th ANNUAL DIAGNOSTIC SLIDE SESSION 2017.

### CASE 2017-10

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#### Clinical History:

14-year-old boy with a history of myelodysplastic syndrome (MDS) status post bone marrow transplant. There was a strong family history of neoplastic disease including a paternal aunt who died at 26 years old due to lymphoma, a paternal grandmother and great-grandmother with breast cancer, and a maternal great aunt with ovarian cancer. Other medical history included posterior reversible encephalopathy syndrome (PRES) complicated by seizures, developmental delay, iron overload secondary to transfusions, gastrointestinal graft vs host disease, and repeated infections including CMV colitis, enterococcus sepsis, and pneumocystis pneumonia. A month prior to death, the patient was admitted to the hospital with a gastrointestinal hemorrhage, acute respiratory distress, renal failure, and disseminated intravascular coagulopathy. His family elected for comfort care. A complete autopsy was performed.

#### Autopsy findings:

In addition to extensive evidence of medical therapy and terminal medical complications, the general autopsy showed an irregular scale-like pigmentation of the skin with a yellowish cast overall. The eyelashes and eyebrows appeared diminished. The fingernails and toenails were abnormal. Internal exam demonstrated focal greenish discoloration of the left ventricular papillary muscle of the heart as well as patchy hemorrhage and peripheral nodularity of the lungs.

On neuropathological examination, the fresh brain weighed 770 g. The cerebellum was diffusely hypoplastic with significant vermian atrophy. Coronal sections revealed that the neocortex was unremarkable for prominent vasculature, hemorrhages, or mass lesions. There were multiple distinct cavitory lesions filled with firm, yellow-tan material within the thalami. There was a cavum septum pellucidum. The midbrain, basis pontis, and medulla were grossly unremarkable. Gross examination of the eyes showed no significant abnormalities.

#### Material submitted:

1. Gross photographs of the brainstem and cerebellum
2. H&E sections of cerebellum

#### Points for discussion:

1. Diagnosis and subtyping
2. Underlying pathogenesis