

## 63<sup>rd</sup> ANNUAL DIAGNOSTIC SLIDE SESSION 2022

### CASE 2022-7

#### Submitted by:

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#### Clinical History:

11-year-old female presented to the medical autopsy service with a past medical history of global developmental delay, seizure disorder, cerebral palsy, and nephrotic syndrome. A sibling of the decedent died at a young age with similar clinical history. The feeding tube-dependent decedent's recent hospitalizations were remarkable for infection and multisystem complications, including small bowel perforation. She eventually developed a thrombus within the inferior vena cava and experienced cardiac arrest.

#### Autopsy findings:

External physical exam findings post-mortem included microcephaly, dysmorphic facies, malrotation of the extremities, and rigidly dorsiflexed feet. The brain weight was 470 grams, fresh. Aside from the overall smallness of the brain, there were no obvious gyral abnormalities of the cerebrum. The cerebellum was small and markedly firm to palpation. Additional gross neuropathologic findings included slightly thickened leptomeninges, and mildly dilated ventricles. Additional autopsy findings included the surgical absence of one kidney, an enlarged contralateral kidney, a thrombus within the caval vasculature, and abdominal cavity adhesions.

#### Material submitted:

1. Images of cerebrum and cerebellum
2. One H&E slide of cerebellum

#### Points for discussion:

1. Neuropathologic findings
2. Genetics