

## 59th ANNUAL DIAGNOSTIC SLIDE SESSION 2018.

### CASE 2018 - 6

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#### **Clinical History:**

A twenty-year-old male presents for esophagogastroduodenoscopy for persistent dysphagia with a specific chief complaint of “I’m unable to chew because I can’t open my mouth and my teeth hurt.” He has a history of hypertension, obstructive sleep apnea, and a congenital musculoskeletal disorder. During the procedure, the patient was noted to have airway instability with subsequent desaturation. Resuscitative efforts were unsuccessful and the patient expired.

#### **Autopsy findings:**

General autopsy revealed a thin man of short stature weighing 75 lbs. He had various musculoskeletal abnormalities including scoliosis, pes planus, and asymmetrical muscular atrophy. The brain weighed 1,560 grams. Gross examination revealed an enlarged brainstem with a markedly stenotic aqueduct. The medulla appeared ovoid rather than the normal “papilionaceous” shape. Internal architecture of the brainstem appeared distorted. The dentate nucleus of the cerebellum was difficult to delineate grossly. The remainder of the cerebrum and cerebellum appeared normal.

#### **Material submitted:**

H&E-stained section of the medulla

#### **Points for discussion:**

1. Differential diagnosis and ancillary studies
2. Pathogenesis