INTRODUCTION

- Menkes disease (MD) is rare with prevalence of 1/250,000.
- Mortality occurs by age 3 years in most.
- It is defined by copper metabolism and connective tissue (CT) derangements from reduced lysyl oxidase activity — a copper dependent enzyme important for collagen synthesis.
- The gastrointestinal (GI) tract relies of CT integrity and GI complications have been previously described.
- This is a case of a patient with MD who outlived his life-expectancy more than 7-fold presenting with massive hemorrhagic ascites due to volvulus of the transverse colon.

CASE DESCRIPTION

Patient Description
- 23-year old male with past medical history of MD which resulted in:
  - Severe intellectual disability
  - Seizure disorder
  - Neuromuscular dysfunction making him: tracheostomy and gastrostomy dependent

Outside Hospital Course
- Admitted after being found less responsive than usual.
- Found to have a urinary tract infection and pneumonia and required treatment with antibiotics and vasopressor support.
- Initially began to improve clinically.
- On day 5, he was noted pale and hypotensive with Hbg of 3.9 g/dl, decreased from 9.3 g/dl initially.
- Transfused 4U pRBC, 1U FFP, reintiated on vasopressors and air-transported to our facility.

Admission Data

Physical exam:
- General – Frail patient, diffuse muscle atrophy, extremity contractures, dysmorphic features, appeared younger than stated age.
- Cardiac – tachycardic rate, regular rhythm
- Respiratory – coarse ventilatory sounds
- Abdominal – taut and distended.

Diagnostic Procedure/Imaging

Abdominal Paracentesis
CT Abdomen/Pelvis

Fig1. Needle aspiration of grossly sanguineous fluid from abdomen
Fig2. CT of abdomen demonstrating a markedly dilated transverse colon and extensive intraperitoneal hemorrhagic products.

Case Outcome
- General surgery was emergently consulted however deemed him a poor surgical candidate.
- After family discussion, comfort care measures were initiated. He soon passed after vasopressors were discontinued.
- He later underwent an autopsy. Mucosa of the transverse colon was noted hemorrhagic with multiple perforations and twisted around a fibrous adhesion band (figure 3A,B).

DISCUSSION

- Hemorrhagic ascites from a transverse colon volvulus has not been previously reported in MD or otherwise.
- We suspect Menkes related CT abnormality may have led to the formation of the adhesion band facilitating volvulus formation, as he had no risk factors for intrabdominal adhesions.
- The uniqueness of this case is further supplemented by our patient’s age of presentation as MD patients rarely survive past 3 years of age.
- This case illustrates a novel gastrointestinal complication in MD and exemplifies the importance of maintaining vigilance for catastrophic hemorrhagic events in this population.