Primary Esophageal Extra-Gonadal Yolk Sac Tumor Metastasized to the Liver

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Published: 05 May 2023

Introduction: Extra-gonadal germ cell tumors (EGCTs) usually arise from midline structures such as the retroperitoneum, mediastinum, and sacrococcygeal region. EGCTs originating from the gastrointestinal system such as the stomach and esophagus are rarely reported. No reported case of primary esophageal yolk sac tumor (YST) has been published yet in the literature.

Case Presentation: A 62-year-old male presented with difficulty swallowing and feeling of food stuck in the middle of his chest for two months. There were associated right upper-quadrant abdominal pain, early satiety, and weight loss (25 pounds) in the last three months. Abdominal CT demonstrated abnormal thickening in the distal esophagus and metastatic disease in the liver adjacent to the distal esophagus. Biopsy of the liver lesions showed poorly differentiated carcinoma with features consistent with YST (positive isochromosome 12p FISH). EGD showed partially obstructing tumor in the lower third of the esophagus. Biopsy of the esophageal mass also showed findings consistent with the YST. PET scan showed increased activity in the lower esophagus but did not identify testicular activity. Blood tests showed AFP of 12,752, HCG of 11, and LDH of 1039. Brain MRI and testicular ultrasound findings were unremarkable. Eventually, he was diagnosed with stage IIIc M1b (liver metastasis) primary esophageal YSK. The patient was started on a chemotherapy regimen with etoposide, ifosfamide, and cisplatin.

Discussion: Primary gastrointestinal germ cell tumors have been very rarely reported in the literature. To the best of our knowledge, our case is the first extra-gonadal yolk sac tumor that originated from the esophagus and metastasized to the liver.