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CASE REPORT

Synchronous Embryonal Rhabdomyosarcoma (NOS) of the Mid-oesophagus and Stomach

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Introduction

Oesophageal rhabdomyosarcomas (RMS) are rare tumours with embryonal variant being even rarer. Wolfensberger et al. in 1894 was the first to report a primary oesophageal rhabdomyosarcoma and demonstrated cross striations on electron microscopy.[1] Most of the oesophageal RMS have been reported in the lower oesophagus with regional nodal metastasis. We present a rare case of primary synchronous, mid-oesophageal and gastric, embryonal, not otherwise specified (NOS) variant of RMS presenting as polypoidal mid-oesophageal and gastric masses with paraesophageal and perigastric lymph nodal metastasis.

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Case Report

A 61-year-old male, normotensive, normoglycemic, patient presented with complaints of dysphagia to solids, loss of appetite and weight loss since last 4 months. An upper gastrointestinal endoscopy was done which showed an ulcerated, polypoidal lesion, situated 25–29 cm in the mid-oesophagus. The proximal oesophagus was distended. A contrast-enhanced chest computed tomography (CT) revealed a mid-oesophageal polypoidal filling defect expanding the lumen at the D7–D10 vertebral level. Similar nodular filling defect was also observed in the fundic part of the stomach. The aorta, pulmonary veins and gastroesophageal junction were normal. However, the CT revealed enlarged paraesophageal, celiac lymph nodes with multiple sub-centimetre lesions in segment II, VI and VII of liver. A positron emission tomography was done which revealed metabolically active lesion in the mid-oesophagus, paraesophageal, cardiac part of the stomach, and perigastric lymph nodal region. A biopsy from the mid-oesophagus was performed which revealed hyperplastic and ulcerated squamous mucosa with underlying submucosa showing a tumour with spindle cell morphology having moderate nucleo-cytoplasmic ratio, arranged in a haphazard pattern with nests at placed. The nuclei had vesicular chromatin with tiny conspicuous nucleoli. Due to its location and histomorphology, a diagnosis of carcinosarcoma was offered with cautionary note for extensive histologic sampling to exclude a carcinoma. The patient was given two cycles of cisplatin-based

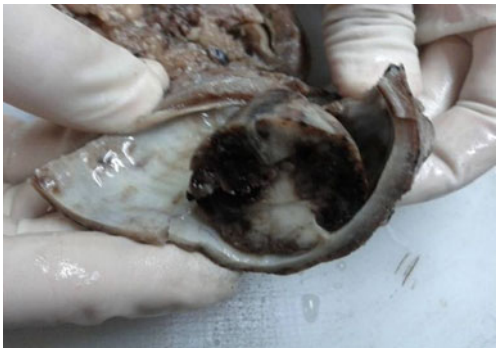

Fig. 1 Oesophagectomy specimen showing a polypoidal growth obliterating the entire lumen of the esophagus

chemotherapy with adjuvant radiotherapy but there was no decrease in size on contrast CT scan. Hence, a surgery involving an oesophagectomy with proximal gastrectomy was done with regional lymph node clearance. The proximal end of the oesophagus was anastomosed to the gastric fundus which was pulled up in the mediastinum. The specimen consisted of oesophagus which was 16 cm in length with a diameter ranging from 2.5 to 3.5 cm. Upon opening the specimen a grey brown polypoidal mass measuring 8.5×3.5×2.5 cm with a stalk was seen to be attached from the oesophageal submucosa [Fig. 1]. It was 5.8 cm from the proximal cut margin and 7 cm from the gastroesophageal junction. The cut surface was fleshy grey brown in appearance. The tumour was sampled extensively. The cardiac part of the stomach showed a submucosal nodular mass measuring 1.5×1×0.5 cm.The cut surface was similar to the oesophageal tumour. Multiple enlarged paraesophageal and perigastric lymph nodes were removed and sampled. On microscopy of the tumour, surface was covered by a stratified squamous epithelium with no dysplasia. The submucosa of the oesophagus and the cardiac part of stomach showed a high-grade tumour with round to spindle cells having moderate pink cytoplasm, vesicular nuclei with tiny nucleoli, arranged in interlacing fascicles with strap cells [Fig. 2a, b]. Also seen were scattered tumour giant cells and cells with anaplastic nuclear morphology. There were hypo- and hypercellular areas within the tumour with admixed myxoid and hyalinised areas. Perivascular condensation of tumour cells was observed in the less cellular areas. Many of the individual cells on light microscopy showed cytoplasmic cross striations [Fig. 2c, d]. The tumour showed numerous lymphovascular emboli at its periphery. Overall histologic features were of an embryonal rhabdomyosarcoma, not otherwise specified type. The regional lymph nodes showed metastasis with extracapsular extension. On retrospect, the small biopsy from oesophagus was reviewed and immunohistochemistry (IHC) tests were done simultaneously on punch biopsy from oesophagus, tumour sections from resected specimen from mid-oesophagus and gastric tumour. The primary antibodies used and the dilutions and sources are listed in Table 1. On IHC, the tumour cells expressed desmin, Myo D1 and myogenin [Fig. 3]. The tumour cells were negative for pancytokeratin, CK5, p-63 and smooth muscle antigen. The histomorphology with supportive immunohistochemistry hence proved it to be an embryonal rhabdomyosarcoma (NOS) type.

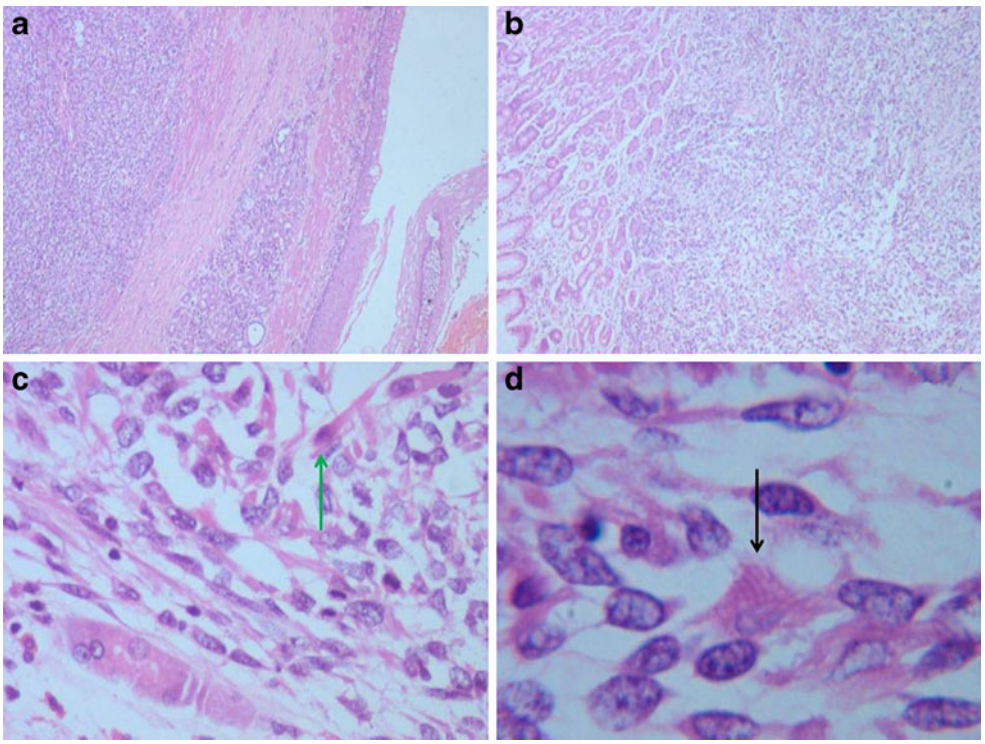

Fig. 2 **a** Section from the oesophagus showing a submucosal tumour composed of spindle cells and arranged in vague fascicles.(H & E, ×100) **b** Section from stomach showing a submucosal tumour composed of spindle cells and arranged in vague fascicles. (H & E, ×100) **c** Sections showing strap cells (*green arrow*). (H & E, ×400) **d** Sections showing strap cells with cross striations. (H & E, ×1,000)

Table 1 Antibodies, clones, dilutions and sources

Antibody (clones)	Dilution	Company
Desmin (D-ER-11)	1:100	Dako, (Denmark)
MyoD1 (5.8A)	1:40	Dako, (Denmark)
Myogenin (LO26)	1:40	Novocastra, (Newcastle)
Smooth muscle Actin (mAb 1A4)	1:200	DAKO, (Denmark)
Cytokeratins (AE1/AE3)	1:400	Dako, (Denmark)
CK5 (mAb clone D5/16B4)	1:100	Dako, (Denmark)
p-63 (mAb clone 4A4)	1:400	Dako, (Denmark)

After 1 month of surgery, the patient was started on ifosfamide and etoposide (IE) alternating with vincristine, cyclophosphamide and adriamycin. After 9 months of initial diagnosis and post surgery three cycles of chemotherapy, the patient developed a right supraclavicular lymph node which showed metastasis of RMS. A positron emission tomography-computed tomography (PET-CT) at this stage showed an overall progression of disease.

Discussion

Primary oesophageal sarcomas are rare, accounting for about 0.5 % of oesophageal cancers which include rhabdomyosarcomas, leiomyosarcomas, synovial sarcomas, osteosarcomas, fibrosarcomas and malignant nerve sheath tumours.[2] In literature, previously, only 15 cases of primary RMS of the oesophagus have been reported so far, with predominance in the geriatric age group. Of those, only one case had embryonal RMS histology.[3] Synchronous rhabdomyosarcomas in the oesophagus and stomach are exceedingly rare with previously only one reported case having a pleomorphic RMS histology by Templeton et al. in 1961.[4] Previously, only two cases of primary oesophageal RMS were diagnosed on cytology, both of which had a pleomorphic histology. One out of the two samples was an oesophageal brushings and the other one being imprint cytology. Both had pleomorphic cytomorphology in common with intracytoplasmic cross striations. [2,5]

To the best of our knowledge and extensive literature search, ours is the first case of synchronous oesophageal and gastric rhabdomyosarcoma with embryonal (NOS) histomorphology and the 16th undisputed case of RMS of the oesophagus.

Primary oesophageal rhabdomyosarcomas are rare aggressive tumours causing death within one year of its diagnosis. The proposed histogenesis is from the undifferentiated mesenchymal cells which, during embryonal evolution, becomes localised in the foregut. Most of the oesophageal rhabdomyosarcomas have been reported from the lower one third of the oesophagus, with pleomorphic histology, being the most common. The histologic diagnosis of oesophageal rhabdomyosarcomas is quite challenging especially on small biopsies which can mimic as a carcinosarcoma with rhabdomyoblastic differentiation or sarcomatoid variant of squamous cell carcinoma. The surface ulceration with extensive myofibroblastic/

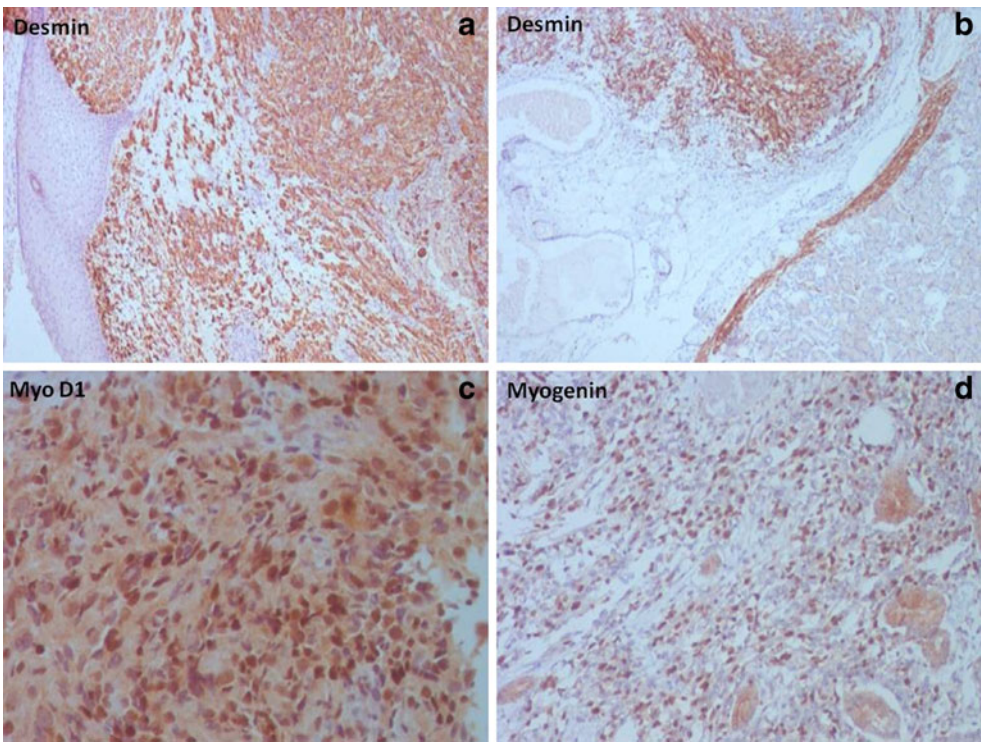

Fig 3 **a** Tumour cells in submucosa of the esophagus exhibiting immunopositivity for desmin. (DAB, ×100) **b** Tumour cells in submucosa of stomach exhibiting immunopositivity for desmin. (DAB, ×100) **c** Tumour cells exhibiting nuclear immunopositivity for Myo D1. (DAB, ×200) **d** Tumour cells exhibiting nuclear immunopositivity for myogenin. (DAB, ×100)

fibroblastic response can masquerade the biopsy, giving it an appearance of a desmoplastic stromal response [2].

Rhabdomyosarcomas of the mid-oesophagus and stomach are extremely rare and needs to be distinguished from other sarcomas, simply due to its metastatic aggressive behaviour similar to a carcinoma and most importantly due to its poor outcome with survival rates of less than 12 months. On light microscopy, careful and diligent search for cross striations should be made. The other most important differential to primary oesophageal rhabdomyosarcomas are carcinosarcomas with extensive rhabdomyosarcomatous differentiation described by Guarino et al. [6] These two entities can be differentiated by extensive sampling for carcinomatous areas along with the use of complimentary IHC studies with pancytokeratin, CK5, p-63 and high molecular weight cytokeratin which are negative in RMS and positive in carcinosarcomas.

Ours is a unique case, presenting as synchronous growths in the mid-oesophagus and the stomach with regional lymph node metastasis having a deceiving clinical presentation similar to a carcinoma and a morphology mimicking a carcinosarcoma. It was with the help of ancillary IHC studies and a retrospect diligent search for cross striations which led to a final conclusive and a rare diagnosis of embryonal RMS (NOS) type.

Conclusion

The pathologist needs to be aware of this entity, inspite of its rarity and due, its aggressive behaviour of regional nodal metastasis similar to a carcinoma as the clinical implications and the outcome can significantly be altered by early diagnosis.

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