CASE REPORT

Malignant rhabdoid tumour of the oesophagus: a case report

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Malignant extrarenal rhabdoid tumour (MERT) of the gastrointestinal tract is rarely reported in the literature. This report describes the clinical and pathological features of a malignant rhabdoid tumour of the oesophagus in a Chinese man. Ivor-Lewis oesophagectomy had been performed. The tumour behaved aggressively and the patient died from disseminated malignancy one year later. This is only the second case report of a malignant rhabdoid tumour affecting the oesophagus.

Rhabdoid tumour was originally reported in the kidneys of young children by Beckwith *et al* in 1978.¹ Malignant extrarenal rhabdoid tumours (MERTs) are rarely reported in the world literature.

Only about 100 patients with MERT have been reported so far, and the tumours occur predominantly in adults. The tumour has been reported in soft tissue, brain, heart, breast, liver, pancreas, uterus, bladder, prostate, oesophagus, colon, small bowel, thymus, salivary gland, and mucosal surface. To the best of our knowledge, this is only the second documented case report of MERT affecting the oesophagus. The patient died from disseminated malignancy one year after Ivor-Lewis oesophagectomy.

CASE REPORT

In August 2000, a 49 year old Chinese man presented to us with a two month history of progressive dysphagia. Physical examination of the patient was unremarkable except for the stigmata of chronic liver disease. He was a chronic smoker (two packs of cigarettes each day) and drinker (three units of alcohol each day). He had no significant past medical history. His blood tests, including complete blood picture, clotting profiles, renal and liver function tests, were all normal. Oesophagogastrroduodenoscopy revealed a malignant looking tumour located in the mid-oesophagus. A $2.7 \times 3.8 \times 6$ cm oval mass was seen in his mid-oesophagus on computerised helical tomography (CT) scan of his oesophagus. There were small lymph nodes (< 1 cm) seen in the precarinal and right paratracheal regions. No metastasis to the lung or other visceral organs was detected in the CT scans. Biopsy indicated the diagnosis of malignant rhabdoid tumour. We carried out Ivor-Lewis oesophagectomy for the patient. No liver metastasis or coeliac lymphadenopathy was observed. The patient made an uneventful postoperative recovery. However, he developed left cervical lymphadenopathy three months later. This was compatible with metastatic rhabdoid tumour on cytological examination of the material obtained by fine needle aspiration. The patient sought a second opinion from a practitioner of alternative medicine after the tumour recurrence. A subsequent CT scan of the abdomen and pelvis showed that he had right parahilar and retroperitoneal lymphadenopathy. He then received palliative chemotherapy in China but no

Figure 1 The tumour is composed of sheets of loosely cohesive highly pleomorphic tumour cells with vesicular nuclei and prominent nucleoli. Occasional paranuclear hyaline globules are present (arrow). Haematoxylin and eosin stain; original magnification, $\times 40$.

detailed information was available to us. The patient finally died from disseminated malignancy one year after the operation.

PATHOLOGICAL FINDINGS

A fleshy light brownish polypoid tumour measuring 5×2.8 cm was identified in the middle portion of the resected oesophagus. The cut surface showed whitish and brownish firm tissue infiltrating into the muscularis propria. Multiple lymph nodes were identified within the adventitial fatty tissue. Microscopically, the tumour infiltrated into the muscularis propria, sparing the adventitia. It demonstrated typical features of rhabdoid tumour; namely, sheets of discohesive polygonal anaplastic cells with highly pleomorphic, multilobated vesicular nuclei, with one to multiple prominent nucleoli and a large amount of pale eosinophilic cytoplasm that contained hyaline globules (fig 1). Mucin stain was negative in these globules. There was no perineural or lymphovascular permeation. The resection margins were clear of tumour cells and none of the seven lymph nodes showed evidence of metastasis. Immunohistochemical staining of tumour cells was positive for vimentin and epithelial membrane antigen, whereas it was negative for cytokeratin, smooth muscle actin, desmin, melanoma markers (S100 protein and HMB 45), lymphocyte common antigen, CD31, and CD34. Electron microscopy revealed poorly defined cell junctions between the tumour cells. There were prominent multiple nucleoli. Paranuclear aggregates of intermediate filaments were found,

Abbreviations: CT, computerised tomography; MERT, malignant extrarenal rhabdoid tumour

Figure 2 Electron microscopy shows whorls of intermediate filaments in the perinuclear region (arrows), characteristic of rhabdoid tumour. Original magnification, $\times 5400$.

corresponding to the hyaline globules on light microscopy (fig 2). The overall features were compatible with malignant rhabdoid tumour.

DISCUSSION

The histogenesis of MERT is unclear. However, these tumours share similar histological, immunohistochemical, and ultrastructural features to renal rhabdoid tumours. MERTs have been observed in pure or composite form in various anatomical sites. The composite form is characterised by the presence of both a rhabdoid component and other distinctive pathological components, such as transitional cell carcinoma, adenocarcinoma, renal cell carcinoma, various sarcomas, or malignant melanoma. It is debatable whether MERT represents a specific entity or is just a common phenotype shared among a diverse group of poorly differentiated neoplasms. The lack of uniform immunophenotypes, the lack of absolutely predictable ultrastructural features, and the heterogeneity of cytogenetic abnormalities do not favour the notion of a specific entity. However, Ota and colleagues² suggested that the rhabdoid tumour cells were derived from a primitive pluripotential cell that had the potential for a wide range of differentiation and accounted for the phenotypic heterogeneity observed in MERTs. There have been discussions concerning the molecular pathology of rhabdoid tumour and its histogenesis in recently published literature. Schofield and colleagues³ reported that there was deletion or mutation of a gene at chromosome 22q11–12 in most renal rhabdoid tumours. They suggested that the involved gene might be a tumour suppressor gene, the inactivation of which might be involved in the genesis of renal rhabdoid tumours. Douglass *et al* also reported a similar genetic abnormality in a subgroup of MERTs.⁴ They found that monosomy of chromosome 22 was consistently seen in MERT affecting the central nervous system. However, rhabdoid tumours in other extrarenal locations failed to show a similar cytogenetic aberration. Hence, the role of the inactivation or mutation of this putative tumour suppressor gene in the histogenesis of MERT remains to be elucidated.

Take home messages

- We describe a case of malignant extrarenal rhabdoid tumour (MERT) of the oesophagus in a Chinese man
- To the best of our knowledge, this is only the second case report of a malignant rhabdoid tumour affecting the oesophagus
- As seems to be the case with most MERTs, the tumour behaved aggressively and the patient died from disseminated malignancy one year later

"It is debatable whether malignant extrarenal rhabdoid tumour represents a specific entity or is just a common phenotype shared among a diverse group of poorly differentiated neoplasms"

Sixteen documented case reports of MERT affecting the gastrointestinal tract have been published in the world literature. The management of these tumours is usually surgical resection if the tumour is operable. Beneficial results of adjuvant treatment have been reported for MERTs in the skin⁵ and central nervous system,⁶ but not in the liver.⁷ The role of adjuvant treatment for MERT in the gastrointestinal tract is unknown. Most of the reports indicate that MERTs are highly aggressive tumours. Wick and colleagues⁸ reported that less than 50% of patients survived for five years without tumour recurrence, regardless of the treatment given. MERT in the gastrointestinal tract has a poor prognosis because most patients die within one year of surgery. Our patient's tumour did pursue an aggressive clinical course and he died one year after the operation.

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