Unusual presentation of more common disease/injury

Germ cell tumour of the trachea

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Summary

A 28-year-old man presented with stridor and dyspnoea. Imaging showed a tracheal mass with severe narrowing of the subglottic airway. Histopathology was consistent with non-seminomatous germ cell tumour. The patient underwent cricotracheal resection and reconstruction of the trachea with tracheostomy. Subsequent positron emission tomography demonstrated new right upper lobe nodules. Postoperative chemotherapy was initiated using the VIP regimen (etoposide, ifosfamide and cisplatin). After four cycles of chemotherapy, CT of the thorax showed interval resolution of most of the pulmonary nodules. Thoracoscopy with right upper and lower lobe wedge resections was performed to remove the residual disease. The patient is currently disease-free and undergoing continued surveillance to assess for clinical, biochemical or radiographical evidence of disease recurrence.

BACKGROUND

This case highlights a unique presentation of a non-seminomatous germ cell tumour (NSGCT) and emphasises how a multidisciplinary, patient-centred approach to treat this rare and life-threatening disease can yield a satisfactory result.

CASE PRESENTATION

A 28-year-old man presented with stridor and dyspnoea over a 2-week period. CT of the neck showed a 4 cm × 3.2 cm lobulated cystic mass centred at the right tracheoesophageal groove with severe narrowing of the subglottic airway (figure 1A,B). The patient underwent rigid bronchoscopy with mechanical and laser debulking. Histopathology was consistent with 'teratoma' showing mature elements including squamous epithelium (figure 1C), ductules lined by columnar epithelium and goblet cells and a focus with formation of acini. However, admixed with these mature components were foci of immature tissue comprised of cartilage (figure 1D), spindle cell stroma with mitosis, and clusters of partially crushed small basophilic cells with neuroepithelium. Some of the mature ductal structures lined by columnar cells and goblet cells display a few interspersed cells positive for the CD56 (figure 2) and synaptophysin (figure 3) immunostains, in keeping with neuroendocrine origin. Doppler ultrasound of the scrotum and CT of the thorax, abdomen and pelvis were negative for masses and metastasis, respectively. Baseline α -fetoprotein (AFP) was 16.2 ng/ml (reference range <8 ng/ml) and β -human chorionic gonadotropin (B-HCG) was 92 mIU/ml (reference range <5 mIU/ml). Definitive cricotracheal resection and reconstruction of the trachea with tracheostomy was performed. Subsequent positron emission tomography (PET) demonstrated new right upper lobe nodules. Postoperative chemotherapy was initiated using the VIP regimen (etoposide, ifosfamide and cisplatin). After four cycles of chemotherapy, the AFP decreased to <3 ng/ml and B-HCG to <2 mIU/ml. CT thorax showed interval resolution of

most of the pulmonary nodules. Thoracoscopy with right upper and lower lobe wedge resections were performed to remove the residual disease. Postoperative PET/CT scan and repeat tracheal biopsies showed no evidence of locally recurrent or metastatic disease.

OUTCOME AND FOLLOW-UP

The patient is currently disease-free and undergoing continued surveillance to assess for clinical, biochemical or radiographical evidence of disease recurrence.

DISCUSSION

GCTs arise primarily in the gonads. A small subset, 2–5%, arise in extragonadal sites¹; most commonly mediastinum, followed by head/neck, retroperitoneum, central nervous system and liver.² Extragonadal tumours arise due to migration and misplacement of primordial cells during embryogenesis.³ The precise mechanism of tumour formation is unknown.⁴ A recent hypothesis proposes that teratoma formation is provoked by DNA methylation modifications on very small embryonic-like stem cells.⁵ A teratoma containing poorly differentiated cells is classified as immature.⁶ A teratoma originating in the testis typically represents benign histology and is not associated with increased serum levels of AFP or B-HCG. In the current WHO classification, the terms 'immature' and 'mature' teratoma have been incorporated into a single subclassification of 'teratoma'. Both mature and immature elements were present in the pathological specimen obtained from the tracheal biopsy, hence using nomenclature from male GCT, the tumour would more properly be termed as NSGCT with both mature and immature features. Poor prognostic features for extragonadal teratomas include mediastinal location, non-pulmonary visceral metastases or any of the following: AFP (>10 000 ng/l), B-HCG (>50 000 IU/l) and lactate dehydrogenase or LDH (>10× upper limit of normal range).⁷ Airway-obstructing tumours require aggressive surgical intervention, followed by cisplatin-based chemotherapy due to the propensity

Figure 1 (A, B) CT of the neck showed a 4 cm × 3.2 cm lobulated cystic tracheal mass with severe narrowing of the subglottic airway. (C) Histopathology showed mature elements including squamous epithelium, ductules lined by columnar epithelium and goblet cells and a focus with formation of acini. H&E ×100. (D) Foci of immature tissue comprising cartilage admixed with the previously described mature components. H&E ×40.

for distant metastases.⁸ The standard of care calls for surgical resection of any residual disease after systemic chemotherapy. The standard first-line chemotherapy regimen is BEP (bleomycin, etoposide and cisplatin). In this case, a regimen generally reserved for salvage therapy, VIP, was used to avoid bleomycin-related pulmonary toxicity. BEP and VIP have comparable overall and progression-free survival rates, but different toxicity profiles, which

should be considered for each individual patient.⁹ Long-term survival may be achieved in more 90% of patients with NSGCT, if the patient has good prognostic features at diagnosis and completely resectable disease.¹⁰ Metastatic or surgically unresectable disease is generally considered incurable. Routine surveillance consists of serial symptom assessment, physical examination, monitoring tumour markers and radiological imaging. This case

Figure 2 CD56 positivity. CD56 stain ×100.

Figure 3 Synaptophysin positivity. Synaptophysin immunostain ×40.

emphasises how a multidisciplinary, patient-centred approach to treating this rare and life-threatening disease can yield a satisfactory result.

Learning points

- Recognise non-seminomatous germ cell tumour (NSGCT) of the trachea as an exceptional diagnosis scarcely reported in current medical literature.
- Manage NSGCT of the trachea with combined-modality therapy.

Competing interests None.

Patient consent Obtained.

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Please cite this article as follows (you will need to access the article online to obtain the date of publication).

Gamalski S, Munoz J, Diaz-Kuan A, Wollner I. Germ cell tumour of the trachea. *BMJ Case Reports* 2012;10.1136/bcr-03-2012-5991, Published XXX

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