CASE REPORT

Twelve-year-old boy with recurrent presentations of shortness of breath: an intraluminal tracheal tumour

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SUMMARY

A 12-year-old boy was admitted with symptoms of recurrent shortness of breath since 12 months along with mild intermittent retrosternal chest pain that had developed 3 weeks ago prior to admission. He was previously treated for asthma with no improvement. Initial posteroanterior chest X-ray (CXR) revealed no abnormality on two occasions. Lateral CXR showed a mass lesion in the distal trachea which was subsequently confirmed by a CT scan of the chest.

Micro-laryngobronchoscopy (MLB) was performed to identify the mass. It was then excised by KTP (potassium titanyl phosphate) laser. Histopathology and immunohistochemistry of the excised tissue confirmed it as an inflammatory myofibroblastic tumour (IMT). Follow-up MLB after 6 months revealed regrowth at the same site, though MLB performed earlier at 4 weeks postoperative period had ruled out any regrowth. Following re-excision, the lesion has shown no regrowth of the tumour in the past 1 year.

BACKGROUND

IMT is an extremely rare locally invasive benign tracheal tumour. Initial symptoms can be mistaken for asthma or other reactive airway diseases. One should consider the possibility of obstructive mass lesions such as IMT, carcinoid and lymphoma in the upper airways, if the patient is not improving in spite of adequate treatment of reactive airway diseases. Though IMT is mostly benign, it has also got malignant potential, so early diagnosis is important.

CASE PRESENTATION

A 12-year-old boy presented to the emergency department with symptoms of progressive shortness of breath, more so on exertion, since 12 months. He had associated dull, retrosternal chest pain that had developed 3 weeks ago. He had also become hoarser with loss of appetite during the last few weeks and had lost 4 kg of weight in the past 3 months.

Earlier, he had presented 3 weeks prior to this with similar symptoms. Chest X-ray (CXR) performed during this visit was reportedly normal. A presumptive diagnosis of asthma was made and he was discharged on salbutamol with advice to return if symptoms did not improve. He had a history of dyspraxia and bilateral grommet insertion. His vaccinations were up to date and had no known drug

allergy. His maternal uncle had died at 63 years of age from throat cancer.

On examination, he had no fever and had respiratory distress with shallow rapid breathing (respiratory rate 50 breaths/min). He preferred to sit upright and hold his neck in extension. Oxygen saturation was 97% in room air and chest auscultation revealed equal bilateral air entry along with minimal wheeze on both the sides. His blood pressure was 143/82 mm Hg with a heart rate of 102 bpm. Examination of his other systems was unremarkable. His weight (55 kg) plotted on the 91st centile.

INVESTIGATIONS

Full blood count, urea, electrolytes, liver function test and ECG were normal. Initial posteroanterior CXR (figure 1) was reportedly normal, but the lateral CXR (figure 2) showed a round radio-opaque defect in the distal trachea approximately 1.9 cm in diameter with mild pneumomediastinum. Subsequently, a non-contrast CT of the thorax confirmed a rounded lesion occupying the lumen of the distal trachea just above the carina (figures 3 and 4). The lesion appeared to be attached to the right posterolateral tracheal wall. Pneumomediastinum was also seen on the CT scan.

DIFFERENTIAL DIAGNOSIS

- ▶ Lymphoma
- ▶ Carcinoid tumour

Figure 1 Normally reported chest X-ray (posteroanterior view).

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Figure 2 Lateral chest X-ray showing (arrow) a mass lesion in the distal trachea.

- Papilloma
- Other neoplasms

TREATMENT

He underwent MLB soon after diagnosis. A mass was identified in the right lower trachea and was excised with KTP laser. Histopathology of the excised tissue confirmed it as a benign myofibroblastic tumour.

OUTCOME AND FOLLOW-UP

Postoperative recovery was uneventful. Repeat MLB performed at 4 weeks postoperative period showed no recurrence of the growth locally. However, a further follow-up MLB 6 months later showed a small area of recurrent myofibroblastic lesion in the lower right anterolateral aspect of the trachea which was re-excised with the KTP laser. He is currently being followed up and has not shown any further recurrences in the past 1 year.

DISCUSSION

Tracheal tumours are extremely rare in children. Almost two-thirds of them are benign in nature.¹ The 1998 literature review noted 38 cases over 30 years in children. IMT (also known as plasma cell granuloma, inflammatory pseudotumour, fibrous histiocytoma, fibroxanthoma and xanthogranuloma) includes lesions with a spectrum of manifestations. It can present at any age, but most patients are less than 35–40 years of age.² Though very rare, it is one of the most common primary lung tumours in the paediatric age group.³ Both

Figure 3 CT scanogram showing the level of lesion (horizontal line).

genders are equally affected. It presents either as solitary or multiple nodules and can rarely be locally invasive. IMT can also be found in other places such as the orbit, skull base, thyroid, liver, spine, spleen, lymph nodes and other organs. It is unclear whether these lesions are a primary inflammatory process or a low-grade malignancy with significant inflammatory responses.^{4 5}

The precise aetiology is unknown. About one-third of lesions develop after a respiratory infection.^{4 5} IMTs have also been reported in patients with multiorgan diseases such as in juvenile idiopathic arthritis, Sjogren syndrome and Epstein-Barr virus infections.⁶ Berardi *et al*⁴ reported that 70% of patients with subsequent diagnosis of IMT can be asymptomatic at presentation. The presentation may be non-specific with cough, wheeze, stridor, dyspnoea, hoarseness and haemoptysis. Initially, it is often mistaken for asthma or other reactive airway diseases.

CXR would be the first investigation of choice but is rarely diagnostic. CT is a better way of delineating the size, location and extent of tracheal tumour. Bronchoscopy then would be the next step to obtain a histology sample for the confirmation of diagnosis and to resect the tumour.

In our case, histology of the tumour showed predominant spindle cells associated with significant inflammatory cell infiltrates including plasma cells, lymphocytes and eosinophils. This was consistent with an IMT. IMT is a histological diagnosis with

Figure 4 CT scan showing a tumour in the distal trachea.

proliferation of spindle cells admixed with variably dense polymorphic infiltrates of mononuclear inflammatory cells as its characteristic features. The presence of local and vascular invasion along with nuclear pleomorphism is associated with malignancy and poor prognosis.⁷

Recently, gene fusion involving the anaplastic lymphoma kinase (ALK) gene at chromosome 2p23 has been described in IMTs. The resultant ALK protein overexpression in the myofibroblastic component of these tumours is detectable by immunohistochemistry. ALK protein expression detected by immunohistochemistry and ALK gene rearrangements detected by fluorescence in situ hybridisation (FISH) are useful in distinguishing IMTs from other malignant tumours.^{8–10} In our patient, ALK staining was negative.

Therefore, accurate microscopy and immunohistochemistry is indispensable in diagnosis. Repeat bronchoscopy is needed to detect tumour regrowth. Cases with secondary regrowth after primary resection have been reported. However, recurrent regrowth is very rare.^{11–13}

Complete surgical resection is curative for IMT.^{14 15} For tracheal and bronchial tumour resection, the choice should be a rigid bronchoscopy, as it permits tissue sampling concomitantly. However, IMTs in the lung can also be resected using video-assisted thoracoscopy or open thoracotomy, depending on the local expertise. Benign tumours are cured by resection only, whereas malignant ones require further postoperative radiotherapy.

It is recommended that patients diagnosed with IMT be monitored closely over the long term, as recurrences have been reported for a period as long as 11 years after resection.¹¹

Learning points

- ▶ Consider a tracheal tumour if a child presents with progressive shortness of breath.
- ▶ One should follow-up cases of tracheal tumours, even after resection, as there may be regrowth of the tumour.
- ▶ The diagnosis of paediatric tracheal tumour requires immunohistochemical and molecular genetic studies (anaplastic lymphoma kinase gene fusion).

Tumours that are less than 3 cm in diameter are associated with a decreased risk of recurrences after excision.¹³

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Competing interests None.

Patient consent Obtained.

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