An Unusual Cause of Spontaneous Pneumothorax: The Mounier-Kuhn Syndrome

Abstract:
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We present the case of a 54-year-old woman referred to our service with an unusual presentation of an under-diagnosed condition. A life-long non-smoker, she was referred to respiratory services by our emergency department with a left sided pneumothorax, progressive dyspnoea on exertion, and recurrent chest infections. Subsequent investigation yielded findings consistent with Mounier-Kuhn syndrome (Tracheobronchomegaly), a condition characterised by marked dilatation of the proximal airways, recurrent chest infection, and consequent emphysema and bronchiectasis. Although rarely diagnosed, some degree of Mounier-Kuhn syndrome may occur in up to 1 in 500 adults.

Figure 1: Admission chest x-ray, demonstrating an apical left-sided pneumothorax and background chronic inflammatory changes. Note the calibre of the proximal airways.

Figure 2: CT of upper thorax, demonstrating marked tracheomegaly and severe paraseptal emphysema.

Discussion
Tracheobronchomegaly (TBM) was first reported by Czyhlarz in 1897, but is named after Mounier-Kuhn, who described the radiographic and bronchoscopic features of the syndrome in 1932. TBM would appear to be significantly under-diagnosed - a review of 500 bronchographies found a 1% prevalence. Its aetiology remains uncertain, although there are occasional reports of a link with connective tissue disease. Histology shows loss of main airway smooth muscle and cartilage, and associated tracheal diverticulosis. Dilatation of the trachea and proximal bronchi causes impaired secretion clearance, inefficient cough, persistent airway inflammation, and subsequent distal bronchiectasis and/or emphysema.

The majority of patients present with recurrent chest infections, and copious purulent mucus production, while others...
present with wheeze or progressive exertional dyspnoea. Mounier-Kuhn syndrome has been described in virtually all age groups, with reported diagnoses in neonates and a 79 year old woman but generally presents in the third or fourth decade of life. Prognosis is unpredictable; some patients may remain relatively asymptomatic, others may progress to respiratory failure. Radiological diagnosis can be established with plain chest radiograph alone. Woodring et al published radiographic measurements of the trachea and proximal bronchi, which can be used to diagnose TBM may be required to confirm airway measurements, and may additionally demonstrate associated pathology such as diverticulae and bronchiectasis.

Bronchoscopy may show dilated proximal airways with associated dynamic collapse, purulent secretions, and tracheal diverticulae. Recent data would suggest that bronchoscopy with confocal microscopy may aid diagnosis by permitting real-time analysis of the bronchial mucosal microstructure, demonstrating a deficiency of elastin fibres within the bronchial wall. Lung function testing will generally show an obstructive pattern, with increased total lung capacity and residual volume consistent with air-trapping. Asymptomatic patients may require no specific treatment. Otherwise, the mainstay of therapy is the prevention and/or treatment of infectious complications, using methods such as effective clearance of secretions and rotational antibiotics. Tracheal stenting can improve symptoms in patients with recurrent infections, severe dyspnoea, and exercise limitation, but can be associated with significant complications. Tracheobronchoplasty using a polypropylene mesh may improve symptoms, quality of life scores, and exercise tolerance in selected patients. Lung transplantation has been performed in at least two cases of Mounier-Kuhn syndrome, one of who unfortunately died in the post-operative period.

We are aware of only one other case presenting initially with spontaneous pneumothorax; hence the literature provides little guidance regarding ideal management in this situation. However, the clinical setting could be considered analogous to the development of secondary spontaneous pneumothorax in chronic obstructive pulmonary disease, whereby recurrence is common following conservative treatment, and surgical management is frequently necessary subsequently required surgical pleurectomy following recurrence of her pneumothorax on two separate occasions.

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References

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