Siblings with Acquired Tracheocele: Possible Hereditary Etiopathogenesis?

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Abstract

Tracheocele is a herniation of the tracheal mucosa through a defect in the tracheal wall. Familiar and/or genetical factors have never been taken into consideration in the etiopathogenesis of acquired tracheocele. Acquired tracheocele occurred in a brother and a sister from India. CT scan of the neck showed an air cyst next to the right tracheal wall. They underwent surgical excision through a transverse cervical incision. Histopathology of the lesion revealed a fibrous sac lined by a pseudostratified columnar respiratory epithelium in both patients. They had no recurrence of tracheocele at 12 months follow up. Genetic counseling did not evidence any significative associated abnormalities. No previous cases of familiar acquired tracheocele have been reported in the literature. Given the rarity of the lesion occurring in more than one member of a family, hereditary factors could be supposed as contributing factors in the etiopathogenesis of acquired tracheocele.

Keywords Tracheocele · Acquired tracheocele · Trachea · Tracheal lesion

Introduction

Tracheocele is a rare lesion. It can be congenital or acquired. Tracheocele is a herniation of the tracheal mucosa through a defect in the tracheal wall. It was described first by Rokitsky in 1838 in a cadaveric study [1]. Radiologically, it appears as a cystic lesion filled with air or with fluid and air. It arises at the junction between the membranous and the cartilaginous portion of the trachea. They are more common on the right side, probably due to the absence of the esophagus on this side, giving mechanical support only on the left side.

About 30 cases of tracheocele are reported in the English literature. They are sporadic cases. At our knowledge there

are no previous reported cases of acquired tracheocele in patients of the same family.

Case Report

Two cases of acquired tracheocele occurring in a brother and a sister from India were referred to our hospital. They used to live together until their teens and denied excessive exposure to environmental pollution. There had no history of previous surgery on the neck, positive pressure ventilation or smoking habit. They didn't play wind instruments or glass blow. Their parents were not consanguineous.

Patient 1

A 45-year-old man presented with a latero-cervical right-sided palpable mass with recurrent local infections since 4 years. He had no difficulty in swallowing, dysphonia or cough. Routine hematological blood tests were unremarkable. He was unemployed, and before he had worked as news vendor. Flexible laryngoscopy revealed normal vocal cord function. Flexible tracheoscopy was performed and revealed the site of the pathway of the tracheocele in the posterolateral wall of the trachea on the right side, adjacent to the

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pars membranacea. A computed tomography (CT) scan of the neck was suggestive of a tracheocele of $2.6 \times 1.9 \times 4.0$ cm in the right side, with focal thinning of the postero-lateral tracheal wall (Fig. 1), with anterior displacement of the thyroidal right lobe and lateral displacement of the common carotid artery.

Patient 2

A 51-year-old female nurse presented with persistent cough since some months. Routine hematological blood tests were unremarkable. She had no difficult in swallowing or dysphonia. Flexible laryngoscopy revealed normal vocal cord function. A CT scan of the neck and thorax was suggestive of a cystic lesion filled with air of 1.2 cm in larger dimension next to the trachea on the right side, at a line passing through the first thoracic vertebra (Fig. 2).

Surgery

We performed an elective surgical excision of the lesion under general anesthesia in both patients. Surgery was

Fig. 1 Preoperative computed tomography image with contrast showing tracheocele. **a** Axial section. The arrow indicates the tracheocele. **b** Sagittal reconstruction image, showing a very thin pathway (arrow) between trachea and tracheocele

Fig. 2 Preoperative computed tomography image with contrast showing tracheocele on the postero-lateral side of the trachea on the right side. Arrows indicate the lesion. **a** Axial section. **b** Coronal reconstruction image. **c** Sagittal reconstruction image

conducted with the aid of intraoperative inferior laryngeal nerve monitoring in *patient 2*. A transverse cervical incision was made. A subplatysmal flap was elevated. Strap muscles were separated by the *linea alba*. The thyroid gland and trachea were identified. The right thyroidal lobe was elevated (Fig. 3a). The tracheocele expanded behind the right thyroidal lobe, which was elevate and the tracheocele exposed. The

Fig. 3 Intraoperative findings. **a** Transverse cervical incision. **b** The tracheocele is evidenced after elevation of the right thyroidal lobe. The tracheocele appears as a translucent lesion. **c** Specimen

tracheocele appeared as a capsulated translucent bluish cystic lesion (Fig. 3b). The parathyroid glands were identified and preserved. The tracheocele was dissected from the common carotid artery. In *patient 1* the pathway of the tracheocele run in the lateral and posterior wall of the trachea between the first tracheal ring and the cricoid cartilage, very near to the entry point of the recurrent laryngeal nerve. In *patient 2* the tracheocele was localized in the lateral wall of the trachea between the second and third tracheal rings, adherent with esophagus. The lesion was dissected and excised (Fig. 3c). The right recurrent laryngeal nerve was lying stretched on the surface of the lesion in *patient 1*, while it run below the tracheocele in *patient 2*. It was preserved in both cases. The tracheocele was dissected from the surrounding tissue and excised. Rigid tracheoscopy was performed and revealed the site of the pathway of the tracheocele. The neck of the tracheocele was sutured with 3-0 Vicryl and closure was reinforced with muscles fragment sutured to the tracheal wall. A suction drain was put in, and the flap was repositioned and sutured in two planes.

In the post-operative period *patient 1* had transient right vocal cord paralysis in paramedian position without dysphonia. Recovery was without any complications in *patient 2*. The drain was removed after 2 days in both patients. Perioperative antibiotics were given.

Histopathology of the lesion revealed a fibrous sac lined by a pseudostratified columnar respiratory epithelium in both patients (Fig. 4).

They had no recurrence of tracheocele at 12 months follow up.

Genetic counseling did not evidence any significative associated abnormalities. In particular, other defects of the upper and inferior airways and superior digestive tract were excluded by anamnesis, fiberoptic endoscopy and CT scan. Possible connective tissue disorders (i.e. Ehlers-Danlos

syndrome, Marfan syndrome) were excluded by anamnesis and physical examination. The geneticist gave no indication to perform further genetic tests.

Discussion

Patients with acquired tracheocele may present with chronic cough [2], pulmonary infections, persistent or intermittent dysphonia [3], dysphagia or haemoptysis [4]. The development of symptoms in adulthood leads to consider a diagnosis of an acquired tracheocele, rather than congenital.

The clinical diagnosis is based on the presence of a right sided neck swelling associated with radiologic and endoscopic features. CT scan of the neck and thorax shows a paratracheal air cyst. Endoscopic examination can reveal the communication between the tracheal lumen and the tracheocele. However, the connecting pathway between the tracheocele and the tracheal lumen is not always easy to detect both at the radiologic and endoscopic examination. Manual compression on the cervical mass during the endoscopic examination could help to visualize air bubbles.

Authors [5] have distinguished between tracheocele and tracheal diverticulum. A tracheocele is usually a single sac with a large inlet, while tracheal diverticula, also known as “trachiectasis” or “multiple diverticular tracheomalacia”, are multiple herniations with narrow inlets of the membranous portion of the trachea. Moreover, tracheocele’s internal wall is composed just by a linear pseudostratified columnar respiratory epithelium. In contrast, the wall of tracheal diverticulum is also composed by cartilage, smooth muscle and mucous glands.

The etiopathogenesis is unknown. A multifactorial origin has been proposed. The presence of a weaker area in the tracheal wall associated with elevated pressure in the

Fig. 4 *Patient 1* **a** Cystic lesion containing air with focally fibrotic and hemorrhagic wall (H&E staining, 40× magnification) **b** In detail the cyst is lined by respiratory epithelium (H&E staining, 100× magnification). *Patient 2* **c** Cyst containing air with thick wall (H&E staining, 40× magnification) **d** In detail the cyst is lined by respiratory epithelium and the wall includes chronic inflammation (H&E staining, 100× magnification)

tracheal lumen are the hypothesized contributing factors. Elevated pressure in the tracheal lumen can be caused by playing wind instruments, glassblowing, positive pressure ventilation and chronic cough. Iatrogenic tracheocele occurring after surgery has also been reported [6, 7]. One case of voluminous tracheocele has been described in association with Mounier-Kuhn syndrome [8]. The syndrome is characterized by a marked dilatation of trachea and bronchi associated with thinning or atrophy of elastic tissue.

In the cases reported, both a brother and a sister developed an acquired tracheocele. They did not have other risk factors such as previous tracheal surgery or significant occupational history. No previous cases of familial acquired tracheocele have been reported in the literature. Given the rarity of the lesion occurring in siblings, possible hereditary etiopathogenesis with a phenotype characterized by weakness in the tracheal wall could be supposed. However, it could also happen just by chance and future studies are aimed in order to confirm this etiopathogenetic hypothesis.

Surgical treatment is indicated if the lesion is symptomatic. If left untreated, it can lead to aspiration pneumonia, dysphagia, vocal cord paralysis or recurrent infections.

The tracheocele can arise under the inferior laryngeal nerve causing a dislocation of the nerve, but dysphonia is not always present. Therefore, careful dissection of the tracheocele and identification of the inferior laryngeal nerve is mandatory before excision. Intraoperative nerve monitoring could be of aid in order to preserve the nerve.

Conclusion

Given the rarity of the lesion occurring in more components of a family, hereditary factors responsible for weakness in the tracheal wall could be supposed as contributing factors. Further genetical and histopathological studies are aimed in order to confirm this hypothesis.

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Compliance with Ethical Standards

Conflicts of interests The authors declares that they have no conflict of interest to disclose.

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