Tracheocoele in a Duchenne muscular dystrophy patient. Case report

Un caso di tracheocele in paziente affetto da distrofia muscolare di Duchenne

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Key words

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Parole chiave

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Summary

Tracheocoele, a congenital or acquired lesion, is rarely detected radiologically and even more rarely diagnosed clinically. This tracheal lesion is characterised by the presence of a single cystic lesion filled with air or a mixture of liquid and air, of extremely variable size, occurring in almost all cases, in a *locus minoris resistentiae* situated in the right posterolateral portion of the trachea. The rare case is described of a voluminous tracheocoele located in the left paratracheal region, extending from the cricoid to sternal notch, manifesting clinically, 3 months prior to evaluation in our hospital in a 27-year-old male suffering from Duchenne muscular dystrophy since the age of 5 years. For l0 years, the patient had been treated with intermittent positive pressure ventilation via nasal mask, due to progressive deterioration of respiratory function. Diagnosis of tracheocoele, initially made at computed axial tomography scan, was confirmed by flexible laryngotracheoscopy under local anaesthesia. Due to severe comorbidity associated with the clinical picture described, the absence of a significant set of symptoms, and the problems concerning anaesthesiological management of the patient, palliative treatment was the only choice. This consisted in cervical compression bandaging during assisted nasal ventilation. Close follow-up was performed in order to monitor any progression of the lesion or onset of related complications. This is the second case of tracheocoele originating in the left paratracheal region reported in the literature, and the first associated with Duchenne muscular dystrophy and prolonged use of a positive pressure respirator. The aetiopathogenic mechanisms that may have determined the formation of this rare lesion are then taken into consideration.

Riassunto

Il tracheocele, nelle sue varianti congenita e acquisita, rappresenta una lesione di raro riscontro radiologico e di ancor più rara diagnosi clinica. Questa lesione tracheale è caratterizzata dalla presenza di un'unica neoformazione cistica a contenuto aereo o misto liquido-aereo, di dimensioni estremamente variabili e ad origine, nella quasi totalità dei casi descritti, da un "locus minoris resistentiae" localizzato a livello della porzione postero-laterale destra della trachea. Viene di seguito riportato un raro caso di voluminoso tracheocele localizzato in regione paratracheale sinistra, esteso dalla cricoide al giugulo, manifestatosi clinicamente 3 mesi prima della valutazione presso il nostro Ospedale in un ragazzo di 27 anni affetto dall'età di 5 da distrofia muscolare di Duchenne. Il paziente era trattato da 10 anni mediante respirazione assistita a pressione positiva intermittente con apposita maschera nasale a causa del progressivo deterioramento della sua funzione respiratoria. La diagnosi di tracheocele, posta inizialmente mediante esame TC, venne confermata da una laringo-tracheoscopia con strumento flessibile in anestesia locale. L'elevata comorbidità correlata al quadro clinico descritto, l'assenza di un corredo sintomatologico importante ed i problemi di gestione anestesiologica del paziente imposero un trattamento palliativo con medicazioni compressive cervicali da eseguirsi durante l'utilizzo del respiratore nasale. Uno stretto follow-up venne inoltre applicato con l'intento di monitorare l'eventuale progressione della lesione o l'insorgenza di complicanze ad essa correlate. È questo il secondo caso di tracheocele ad origine paratracheale sinistra riportato in letteratura, il primo associato a distrofia muscolare di Duchenne e ad utilizzo prolungato di respiratore a pressione positiva. Vengono quindi presi in esame i possibili meccanismi eziopatogenetici responsabili della formazione di questa rara lesione.

Introduction

Tracheocoele is a lesion that is rarely detected radiologically and even more rarely diagnosed clinically. There is no clear distinction, in the literature, between the definition of this nosologic entity and that of tracheal diverticulum; use of these two terms may even be ambiguous even if some Authors have suggested a size limit of 2 cm for diverticula [5]. The tracheocoele comprises a single cystic lesion of extremely variable dimensions, containing air or a mixture of liquid and air and generally arising at the junction between the cartilaginous and membranaceous portions of the trachea on the right, where it receives no posterior support from the oesophagus [13]. Cases of protrusion through the ligaments between

individual rings have also been reported in the literature, although this location is more appropriately to be attributed to tracheal diverticula in the strictest sense [3 4].

Rokitansky was the first to describe this expansive lesion affecting the trachea, during a post-mortem study in 1838 [12]. To our knowledge, only one other case of tracheocoele originating, like that described here, in the left paratracheal region, has been reported in the international English language literature since then [1]. In all other cases, the expansive lesions have always been situated in the right, probably due to the close anatomical relationship between the trachea and the oesophagus mentioned above.

The case is reported of a patient suffering from Duchenne muscular dystrophy (DMD), and, therefore, undergoing prolonged periods of positive pressure assisted ventilation, who complained of the appearance of a voluminous left laterocervical tracheocoele. This association, never before described, is analysed speculatively, in an attempt to establish possible aetiopathogenic correlations.

Fig. 1. Clinical picture upon presentation in our Department: sequelae of DMD are evident, with postural changes and nasal mask for intermittent positive pressure ventilation. The tracheocoele, 12 cm in cranio-caudal length, extends from the left lateral edge of the cricoid to the sternal notch, laterally displacing the laryngo-tracheal axis to the right and rotating it clockwise.

Clinical case

A 27-year-old male was referred to our Department in October 2001 on account of the appearance, 3 months earlier, of a progressively expanding left paratracheal swelling. The patient had suffered from DMD since the age of 5 and had been bedridden since he was 12. His general condition was compatible with that of an advanced dystrophic disease, characterised by diffuse muscular hypotrophy, significant ankylosis of the main joints, postural contractures with limbs flexed, complex torsion of the spine, with hyperextension and left rotation of the cervical spine. The deformity of the rib cage and progressive functional deterioration of the respiratory muscles, which first appeared at 17 years of age, had required the use of an intermittent positive pressure nasal respirator, initially only at night but, later, also during part of the day (Fig. 1).

The presence of the paratracheal swelling, on the left, had been noticed, for the first time, during a session of assisted ventilation and had rapidly increased in size over the following weeks. When the patient was evaluated at our Hospital, the lesion appeared taut and pliable upon palpation, yielded partially to manual compression, and was hyperphonetic upon percussion. Measuring 12 cm in length, it extended from the left lateral edge of the cricoid to the sternal notch, laterally dislodging the laryngotracheal axis to the right. The patient did not complain of coughing, dysphonia, dyspnoea, dysphagia or any other signs or symptoms that might have appeared recently and been attributed to the lesion.

Computed tomography (CT) scan of the neck, even though limited due to the patient's contracted posture, revealed a left paratracheal air-filled lesion, measuring 3x5x12 cm and extending from the crico-tracheal junction to a plane passing through the carina, where it came into contact with the anonymous artery (Fig. 2). The mass dislodged the larynx and trachea laterally to the right, simultaneously determining a clockwise rotation of the latter, without, however, causing any significant compression. No liquid/air level was detected inside the lesion, which appeared to be of cystic origin, containing only air, and was surrounded by a thin wall, with no cartilaginous component. The connection with the laryngotracheal lumen was radiologically hypothesised to be at the junction between the *pars membranacea* and the first tracheal ring, on the left (Fig. 3).

Endoscopy was, therefore, performed under local anaesthesia, using a flexible videoendoscope (Pentax 15 T2, Asahi Optical Corporation, Ltd, Tokyo, Ja-

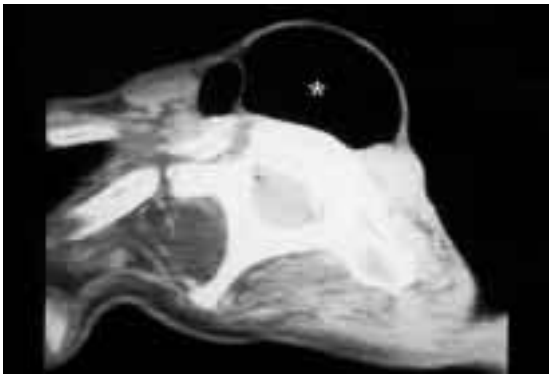

Fig. 2. CT reveals left paratracheal air-filled lesion, 3x5x12 cm in size, extending from crico-tracheal junction to a plane passing through the carina. Asterisk denotes tracheocoele lumen, trachea is dislodged to right, even if not significantly compressed.

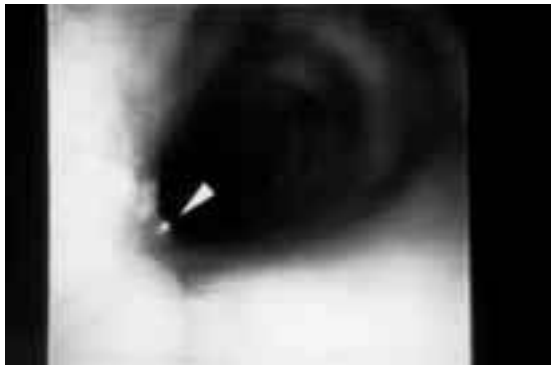

Fig. 4. Laryngotracheoscopy with flexible videoendoscope, during manual compression of tracheocoele *ab extrinseco*, reveals tiny air bubbles (arrow) at left junction between *pars membranacea* and first tracheal cartilagineous ring. Note clockwise rotation of trachea, not significantly compressed by cystic lesion.

pan). The procedure was particularly difficult, due to the poor compliance of the patient, heavy salivation and the unfavourable anatomo-morphological condition, with a marked angle between the neck and chest cage. Vocal cord motility and laryngotracheal lumen patency both appeared within normal limits. Initial endotracheal inspection failed to reveal a connecting pathway with the tracheocoele, however, following manual compression *ab extrinseco*, several small air bubbles were visible, situated in the angle formed on the left where the *pars membranacea* meets the first tracheal ring, as previously hypothesised on the basis of radiological evidence (Fig. 4).
The only treatment possible for a lesion of this type and extension would have been surgical excision under general anaesthesia, with a left cervicotomic approach. The critical condition of the patient, as well as, the difficulties to be overcome with the intra- and post-operative anaesthesiological management, associated with the total lack of symptoms, led us to exclude any surgical treatment of the lesion. A solution of a palliative type was, therefore, programmed with cervical compression bandaging during assisted ventilation in order to limit further increase in the size of the lesion. It was further recommended that the patient's clinical condition be observed at follow-up, to monitor local symptoms and possible complications due to infection. The patient's clinical symptomatic picture, when last evaluated in our Department in April 2002, was fundamentally unchanged.

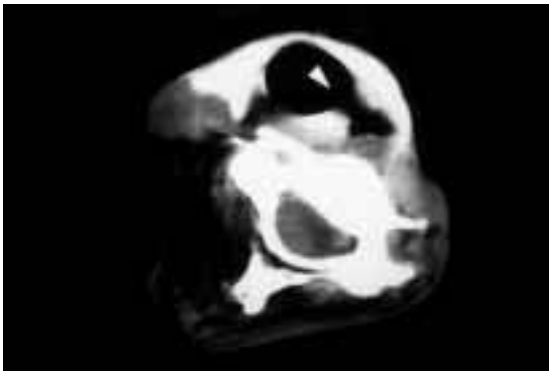

Fig. 3. Connection between tracheocoele and airway lumen of laryngo-tracheal axis (white arrow) is identified at left junction between *pars membranacea* and first tracheal ring.

Discussion

The aetiology of the tracheocoele has not yet been completely defined: the distinction between the congenital and acquired forms hypothesised by several Authors [11 14] is not, in fact, always easy to apply in clinical practice [2]. Upon surgical excision, instead, the tracheocoele wall is reported to be histologically devoid of muscular and cartilaginous tissue in the acquired variant, these being, vice versa, pathognomonic features of the congenital form [7]. The internal epithelium would, instead, always be of the pseudostratified respiratory type, occasionally associated with signs of chronic inflammation [8 15].
The symptoms correlated with this type of lesion may vary considerably and include paroxysmal attacks of coughing caused by the leakin of the liquid component of the tracheocoele into the airways, with or without recurrent bronchopulmonary infections [1], dysphonia and/or dysphagia due to recurrent straining of the lower laryngeal nerve [8], or the simple presence of an expansive cervical lesion, as in the case

described here. Incidental findings of tracheocoeles both during radiological exams [5] and autopsies [9], in totally asymptomatic subjects, have also been reported in the literature.

The sudden appearance of a tracheocoele, in an adult, would seem to indicate an acquired pathogenesis, even if it is almost impossible to ascertain the chronological development of a lesion of this type without prior detailed radiological documentation [13]. Some Authors who consider the distinction between congenital and acquired lesions to be purely theoretic have proposed a multifactorial origin for the tracheocoele [2]. The presence of an area with a weaker tracheal wall and acquired factors such as a chronic cough or particular manoeuvres, iatrogenic (positive pressure-assisted ventilation) and not (playing wind instruments, glassblowing), which cause elevated pressure in the tracheobronchial tree, can trigger the formation of a tracheocoele. In the case reported here, several predisposing factors may be identified to explain the formation of such a voluminous lesion in an anomalous site. DMD, in fact, involving the neck muscles (as seen in CT images in Figs. 2, 3), may have reduced the physiological mechanical containment carried out by the prelaryngeal muscles along the laryngotracheal axis. The abnormal posture of the patient, in cervical hyperextension with head rotated leftwards, may also have played an important part in the unusual location of the tracheocoele: in this position, in fact, the trachea would be thrust forward, with the sternocleidomastoid muscle, on the left, exercising less compression. Lastly, the prolonged use of intermittent positive pressure-assisted ventilation could have been the causal element responsible for the presence of increased endotracheal pressure, due to the fact that, in this case, the air does not reach the lungs due to the negative pressure created by the inspiratory muscles, but by means of the positive pressure produced by the mechanical respirator itself. The latter would, therefore, have played a decisive role in the genesis and rapid expansion of the tracheocoele.

Radiological and endoscopic detection of the communicating pathway between the tracheocoele and the airway was particularly arduous in the case described. In particular, it could not be detected during flexible laryngotracheoscopy, under local anaesthesia, until partial drainage, determined by manual compression, caused air bubbles to appear in the tracheal lumen itself. On the other hand, cases of surgical excision have been reported in which the tracheocoele pathway was not clearly identified, even after excision and ensuing anatomopathological examination [6 10].

The rapid development of a lesion of the size reported here would suggest the presence of a valve mechanism between the airway and the tracheocoele itself. This would also explain why the lesion could not be effectively reduced manually and why the pathway connecting it to the trachea was difficult to detect endoscopically. On the other hand, it is not clear why a cystic structure, presumably sheathed in respiratory epithelium, presented no accumulation of liquid inside, in spite of the large size and the fact that it had virtually taken over the main airway. An analogous case, with regard to size, radiological and clinical aspects was, however, also reported by Mathur et al. [10].

Even if the treatment of choice for tracheocoele is surgical excision, this option was not considered feasible in the case described here. The elevated comorbidity index, the risk of surgical complications and the absence of significant symptoms, in fact, all warranted palliative medical treatment, comprising medium-compression bandaging during assisted ventilation, in order to avoid further dilation of the tracheocoele. It was, however, made clear to the patient, his relatives and his personal physician that surgery would be necessary if the lesion continued to increase in size, or if fever, coughing, dysphonia, dysphagia, dyspnoea or antibiotic-resistant respiratory infections appeared.

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