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Clinical usefulness

Respiratory support in the absence of abdominal muscles: A case study of ventilatory management in prune belly syndrome

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Educational aims:

The reader will be able to:

- Understand the importance of abdominal muscles in respiration.
- Identify the challenges in respiratory management in prune belly syndrome.
- Apply currently available respiratory techniques in prune belly syndrome.
- Develop a plan for respiratory management in prune belly syndrome.

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ABSTRACT

Prune belly syndrome (PBS) results in a total lack of abdominal musculature. Abdominal muscles have an important function during inspiration and expiration. This puts the patient at risk for respiratory complications since they have a very limited ability to cough up secretions. Patients in an intensive care unit (ICU) with PBS who receive mechanical ventilation are at even greater risk for respiratory complications. We review the function of the abdominal muscles in breathing and delineate why they are important in the ICU. We include an illustrative case of a long-term ventilated patient with PBS and offer respiratory management options.

INTRODUCTION BACKGROUND

Prune-belly or Eagle-Barrett syndrome (PBS) is a congenital disorder characterized by the clinical triad of abdominal muscle deficiency, severe urinary tract abnormalities and, in males, bilateral cryptorchidism. The term prune-belly refers to the wrinkled appearance of the abdominal wall, due to partial aplasia or hypoplasia of the abdominal muscles. Contrary to this name, PBS can be a multisystem disease affecting the cardiopulmonary, gastrointestinal, central nervous and musculoskeletal tract in 75% of cases [1]. There is a wide range of symptoms associated with PBS, resulting in a large variation in its clinical presentation.

PBS primarily occurs in males (95–97%), although there are rare case reports of this disorder in females. It affects 3.8 per 100,000 live male births [2,3].

The exact etiology of PBS is still unknown, although some genetic mutations have been identified, including the X-linked Filamin A gene [4]. Nowadays there are 2 main theories. Firstly, the obstructive theory, which states that bladder or urethral obstruction early in gestation causes bladder distention, ureteral dilatation, hydronephrosis and secondary wasting or atrophy of the abdominal wall muscles. At birth, the obstruction at the bladder outlet or the urethral obstruction may have been resolved, so that

Abbreviations: PBS, prune belly syndrome; ICU, intensive care unit; NICU, neonatal intensive care unit; NAVA, neurally adjusted ventilatory support; PEEP, positive end-expiratory pressure; EPAP, expiratory positive airway pressure; IPAP, Inspiratory positive airway pressure; FEV, forced expiratory volume; FEV1, forced expiratory volume in 1 second.

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no mechanical obstacle can be identified after birth. Another more widely accepted theory states that the underlying defect in PBS is an abnormal mesoderm development. This would affect embryogenesis of the musculature of the abdominal wall, the mesonephric musculature and paramesonephric ducts and urinary organs [3,5,6]. Despite advances in urologic care for children with PBS, this condition continues to be associated with high mortality, likely related to prematurity and associated pulmonary complications. Mortality early in life is frequently due to the pulmonary hypoplasia; mortality at a later stage in life can usually be attributed to renal failure from to the effects of renal dysplasia [7].

Due to the scarcity and complex nature of the disease, no best practice guidelines exist for the management of respiratory insufficiency in PBS patients admitted to the ICU. We present a patient with PBS with prolonged dependence on mechanical ventilation and accordingly we review the available evidence focusing on respiratory pathophysiology and evolving therapies.

CASE PRESENTATION

We present the clinical course of a neonate with PBS who was admitted in a tertiary referral hospital because of respiratory insufficiency. He was born prematurely at 28 weeks and 3 days of gestation.

Because of respiratory distress syndrome and lung hypoplasia the patient was intubated, ventilated and administered surfactant shortly after birth. He was admitted to the Neonatal Intensive Care (NICU) for further treatment. Physical examination at birth showed the typical appearance of a wrinkled abdomen with the absence of abdominal musculature, bilateral cryptorchidism, non-rigid club feet, severe scoliosis and a deformed thoracic cage. Based on this clinical data the diagnosis of PBS was established. This diagnosis was confirmed by renal ultrasound showing bilateral hydronephrosis, megaureters and a megabladder. Respiratory course during his stay at the NICU was complicated by frequent ventilator associated pneumonia, atelectasis and cytomegalovirus infection. Several extubation attempts failed and a tracheostomy was eventually performed. Hereafter the patient showed favorable progress with stable parameters on ventilatory support. Neurally Adjusted Ventilatory Assist (NAVA) was used to improve patient-ventilator synchrony, titrating respiratory support to the observed effort and drive. Frequent mucus plugging occurred resulting in atelectasis, even with high PEEP settings (10 cm H_2O). We installed an active humidification system and performed regular bronchoscopy for secretion clearance. Furthermore, we continued extensive physiotherapy with manually assisted cough, forced expiration, and intrapulmonary percussive ventilation. Together with our team of physiotherapists we developed a corset with a separate abdominal binder. We were gradually able to prolong the amount of inspiratory effort transitioning to a system suitable for subacute care facilities (Respironics V60), initially with high pressures (IPAP 38 cm H_2O and EPAP 16 cm H_2O). The patient was transferred to a long-term rehabilitation facility 8 months after birth. An abdominal wall reconstruction is planned at 18+ months of age.

DISCUSSION

In patients with PBS, multiple elements often co-exist that impact upon the respiratory system. First, the lack of abdominal musculature severely impacts the respiratory pump function. During inspiration, the diaphragm normally contracts pushing the abdominal contents downwards. A normal abdominal muscle tone will limit the descent of the diaphragm, resulting in an increased intra-abdominal pressure, outward movement of the lower ribs and lateral chest wall, thus increasing lung volume [8,9]. Decreased abdominal muscle tone will limit the increase in abdominal pressure with decreased lung volumes resulting [10]. The combination of lung hypoplasia and the suboptimal functioning of the inspiratory muscles are the main reasons why patients with PBS may need inspiratory respiratory support. An abdominal binder can move the diaphragm upwards to promote a more beneficial length-tension relationship for the diaphragm.

Expiratory function

Whereas inspiration is an active process, expiration is passive during quiet tidal breathing in healthy subjects. During expiration, the diaphragm relaxes and the abdominal pressure pushes the diaphragm upwards, resulting in a decreasing lung volume and thus expiration. However, the abdominal muscles can contract out of phase with the diaphragm to increase intra-abdominal pressure during expiration, actively promoting expiration. The transversus abdominis muscle is the first one that is active during expiration, and is frequently recruited. An active expiration pushes the diaphragm upwards, placing the diaphragm in an advantageous mechanical position, assisting the next inspiration [11]. This way, the (increase in) work of breathing is split between the inspiratory and expiratory muscles [12]. The central nervous system controls the inspiratory and expiratory muscles, resulting in thoracoabdominal synchrony.

Coughing and forced expiration

Classically, coughing involves three phases: inspiration, contraction of expiratory muscles against a closed glottis and opening of the glottis. The rise in intrathoracic pressures causes compression of the airways during the last phase [13]. Lost abdominal muscle activity results in a decrease in maximal expiratory force and a reduced ability to cough, clear secretions and protect the airway. Ineffective coughing due to the combined effect of pulmonary hypoplasia, absent abdominal musculature, relative flat diaphragm and chest deformity makes PBS patients prone for pulmonary complications including atelectasis and recurrent pneumonia [14,15].

In a recent publication Vivier et al. [16] found that clinical assessment of cough strength was the only predictor of extubation outcome. Patients who failed extubation were significantly less likely to have an effective cough than others (15% vs 42%).

This finding emphasizes the negative impact of abdominal wall muscle hypoplasia as seen in patients with PBS. For these reasons, in patients with PBS, assisted expiration and assisted coughing may be needed to replace the role of the abdominal muscles in these processes.

Activity during increased loading

The activity of the respiratory muscles change profoundly during increased load on the respiratory system. The precise details of these changes are described by Aliverti et al. [11]. In summary, as the workload increases, the abdominal muscles relax more during inspiration and contract more during expiration, effectively decreasing the chest wall volume at end-expiration. The work of breathing during heavy loads is thus split between the diaphragm, rib cage muscles and abdominal muscles, with the latter two acting fully out of phase of each other. At maximally applied workloads, the pressures generated by rib cage and abdominal muscles is greater than those generated by the diaphragm. Even small increases in applied inspiratory loads will result in abdominal muscle activity, and the transversus abdominis muscle will be recruited first [12]. As a result, patients with PBS may have difficulty coping with increased loads on the respiratory system (e.g.

during respiratory infection), lacking the potential to maximize the function of the diaphragm in this setting.

In severe PBS, lung hypoplasia due to oligohydramnios is present as well, further leading to respiratory insufficiency [5]. Furthermore, thoracic cage deformities, scoliosis and paradoxical movement of the abdomen during respiration may predispose affected patients to restrictive lung disease [14]. These elements explain their susceptibility to recurrent respiratory infections in childhood. Spirometry and lung volume measurements of children aged 7–19 years old demonstrated a mild reduction in forced vital capacity (FVC) and forced expiratory volume in the first second (FEV1) but a normal FVC/FEV1 ratio is suggestive of a restrictive process [14,15].

Physiotherapy

The optimal respiratory management of the PBS remains challenging. Chest physiotherapy has an important role in the treatment and prevention of respiratory complications in PBS [17]. Techniques like abdominal binding, manually assisted cough, mechanical in- and exsufflation and intrapulmonary percussive ventilation (IPPV) have been used successfully in patients with PBS [17,18]. Manually assisted coughing can replace the expiratory muscle function of the absent abdominal muscles by increasing the pressure below the diaphragm. It consists of a sharp inward and upward application of pressure to the upper abdomen just below the diaphragm by a physiotherapist and results in improved cough expiratory flows. A perforation of the small bowel after a manual cough maneuver has been documented [19]. Highlighting the importance of prudence when applying this technique especially in the absence of abdominal muscles.

Mechanically assisted coughing by an insufflation-exsufflation device (i.e. cough-assist) causes a rapid change in the pressure gradient from positive to negative, in a way that expiratory flow is increased and a cough initiated. It can be applied by a face mask or via a tracheal tube or tracheostomy. Its usage has already been demonstrated to be successful in patients with PBS [17,18].

Abdominal binder

The use of an abdominal binder or corset has been successful in some severe cases of PBS in the management of respiratory complications [17]. Due to the loss of abdominal muscle tone, abdominal contents are pushed out and the diaphragm is pulled down by the weight of the abdomen, especially in the upright position. The use of an abdominal binder pushes the diaphragm more cephalad into the chest by augmenting the intra-abdominal pressure. Thus, it increases resting length and appositional zone along the abdominal wall and thereby exerts a greater force on the ribs and improves expiratory forces and coughing.

Pectoralis major training

Another technique to enhance expiratory function could consist in training of the pectoralis major muscle. This technique, currently only described in tetraplegic patients [20], is based on the fact that in addition to the pressure generated by the abdominal muscles, the clavicular portion of the pectoralis major muscle causes dynamic compression of the airways in patients with restrictive lung function due to spinal cord trauma [12]. An increase in strength of this muscle will allow patients to cough at lower lung volumes and clear bronchial secretions from peripheral airways. This would, theoretically, reduce the incidence of bronchopulmonary complications.

Neurally adjusted ventilator assist

Neurally adjusted ventilator assist (NAVA) is a relatively new modality of ventilation. With this ventilation technique, the electromyography signal of the diaphragm is used to deliver an appropriate ventilator assistance, improving patient-ventilator synchrony [21]. In theory, this could be an advantage in patients who lack abdominal musculature and in whom the function of the diaphragm is already impaired [22]. Despite its promising theoretical advantages, no data currently exist on the use of NAVA compared with other ventilator modalities in patients with PBS.

Abdominal wall reconstruction

An abdominoplasty can be performed by an extraperitoneal plication of the facial folds. Over the past decades several innovations of this technique have been developed [3]. The goal is to improve the abdominal contour, strengthen the abdominal wall and promote a more normal function of the abdominal musculature. This could theoretically result in improvement of respiratory function. A recent modification of this technique by Franco et al. [23], using laparoscopic guidance, may improve morbidity and decrease complications compared to an open surgical procedure. A more invasive technique consists of a surgical intervention in which the abdominal wall defect is covered by muscles of the antero-lateral thigh including the rectus femoris, sartorius, gracilis, and tensor fascia lata. This muscle transposition technique, first described by Ger in 1983, attempts to replace the missing actions of the antero-lateral abdominal wall musculature [24]. This reconstruction of the abdominal wall provides firm support for an effective Vasalva manoeuver, which improves chest expectoration, enhances respiratory function and prevents the loss of spinal stability which counteracts the development of kyphoscoliosis [24,25], ultimately improving lung function. Unfortunately, long term outcome results are unknown.

CONCLUSION

Patients with PBS frequently have a combination of lung hypoplasia and the absence of abdominal wall musculature, resulting in the need for prolonged respiratory support. Respiratory management is the cornerstone in survival of these patients in their early life. A combination of respiratory support techniques and therapy are available, assisting both inspiratory and expiratory muscles. These include physiotherapy, abdominal wall binding and ultimately surgery to improve core stability.

DECLARATIONS

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DIRECTIONS FOR FUTURE RESEARCH

Most literature on prune belly syndrome has primarily focused on the renal and urogenital aspects of the syndrome. Currently, there are no guidelines nor consensus regarding the respiratory management of patients with PBS. Standardized treatment is further confounded by the rarity and broad spectrum of this syndrome. Furthermore, there is a lack of outcome data in these

patients. Significant research can be done focussing on standardisation and analysing outcome data and the results of implanting the more recent advances in (surgical) treatment.

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