Paediatric Respiratory Reviews 10 (2009) 3–6

Contents lists available at ScienceDirect

Paediatric Respiratory Reviews

Mini-Symposium: Medical and surgical aspects of chest wall deformities

Pectus excavatum: Pathophysiology and clinical characteristics

Anastassios C. Koumbourlis *

Division of Pediatric Pulmonary Medicine, Schneider Children's Hospital, North Shore-Long Island Jewish Health System, Albert Einstein College of Medicine, 865 Northern Boulevard, Great Neck, NY 11021, USA

ARTICLE INFO

Keywords:
Pectus excavatum
Chest wall deformity
Exercise intolerance
Lung function

SUMMARY

Pectus Excavatum is the most common congenital abnormality of the chest wall. In the majority of the cases the condition is idiopathic. Affected patients tend to have lung volumes that are mildly decreased but within the normal range and they are often associated with mild air-trapping. Many patients show evidence of lower airway obstruction. Exercise intolerance is the most common symptom associated with pectus excavatum, and it is now believed to be due to cardiovascular rather than pulmonary causes. The psychological effect of the deformity often exceeds its actual physical effect. Several surgical techniques are available for the repair of the deformity, although the need for it is still considered controversial by many. The current article provides an in depth review of the pathophysiology and clinical characteristics of pectus excavatum, as well as an overview of the treatment options in order to help the practitioners caring of affected patients in their evaluation.

INTRODUCTION

Pectus excavatum is the most common congenital deformity of the chest wall, accounting for 90% of all congenital chest wall abnormalities.[1] The condition has been associated with symptoms such as exercise intolerance and chest pain on exertion, as well as with the presence of restrictive lung defect on pulmonary function testing, although most patients tend to be asymptomatic. Various procedures have been successfully developed over the years for its repair, but there is no consensus among physicians as to whether or not the deformity produces symptoms that are severe enough to justify a surgical procedure. As a result the procedure is often denied by health insurers on the grounds that repair of the pectus is a 'cosmetic' procedure and not for an actual medical problem.

The aim of this article is to review the available evidence on the pathophysiology of idiopathic pectus excavatum, especially with regards to its effect on lung growth and cardiopulmonary function, as well as the current recommendations for its evaluation.

EPIDEMIOLOGY

The prevalence of pectus excavatum is estimated at approximately 1 in 300 live births.[1] Although the deformity affects both sexes, it is markedly more common among males, in a ratio of 5:1 or higher.[2] In 15–40% of cases there is a close relative on either side of the family with the same deformity, suggesting that the condition is hereditary.[3] However, neither the mode of transmission nor the responsible gene for the deformity is known. There is no known racial predisposition for the deformity,[1] although there appears to be a significantly higher prevalence among Caucasians. In a large series of patients with idiopathic pectus excavatum evaluated in a major teaching hospital in the USA, 89% were Caucasians, 9% were Hispanic and only 2% were Asian.[4] Whether this reflects actual genetic differences among races or different perceptions of the deformity among different racial groups is not clear.

PATHOGENESIS

The exact causes of pectus excavatum are not known and the majority of cases are considered idiopathic. The deformity appears to be caused by abnormal growth of the cartilages in the costochondral region, and the fact that its prevalence is higher among patients with conditions such as Marfan syndrome raises the possibility that the underlying mechanism may relate to defects in collagen formation. Pectus excavatum could be the result of lung hypoplasia (e.g. congenital diaphragmatic hernia) or lung agenesis. Acquired pectus can be seen in cases of severe chronic upper or lower airway obstruction requiring the generation of very high negative intrathoracic pressure by the use of the accessory respiratory muscles.[5]

PATHOPHYSIOLOGY

The rib cage provides the physical protection for the organs of the thoracic cavity and the surface on which the respiratory

* Tel.: +1 516 622 5280; fax: +1 516 622 5285.
E-mail address: akoumbou@LIJ.edu.

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doi:10.1016/j.prrv.2008.12.002

muscles work, and with which it constitutes the so-called 'respiratory pump'. Its expansion by the respiratory muscles creates the negative pleural pressure that is the driving force for actual breathing. Thus, any structural or functional abnormality of the chest wall has the potential of affecting significantly the breathing but also the individual organs of the thoracic cavity.

Effects on lung growth and airway function

In several studies of pulmonary function in patients with pectus excavatum the lung volumes have been reported to be decreased, suggesting the presence of a restrictive lung defect.[6–12] The restriction was assumed to be the result of the irregular, narrow shape of the rib cage, preventing normal growth of the lung. However, more recent studies showed that although lung volumes tend to be somewhat decreased, they remain within the range of normal[4,9] (Table 1). Most importantly, the pattern of lung function appeared to vary widely among affected patients. According to a recent study, normal lung volumes and airway function were found in 54% of patients, whereas 41% had an obstructive pattern and only 5% had a restrictive pattern.[4] The normal pattern was found predominantly among younger patients (< 9 years of age). In contrast, the prevalence of the obstructive and restrictive patterns increased with age[4] (Table 2). Considering that rapid lung growth and development take place in the early years of life, it can be speculated that the pectus excavatum has actually very little effect on lung growth.

A rather surprising finding is the increase in residual volume (RV) and the ratio RV/total lung capacity (TLC), which suggest the presence of air-trapping. Interestingly, air-trapping was found not only among patients with obstructive pattern but among those with normal or restrictive pattern as well.[4,9] Considering that an increase in residual volume is by definition accompanied by a decrease in the vital capacity, it is possible that the decreased forced vital capacity (FVC) often found in patients with pectus excavatum, may be due to air-trapping and not, as is usually assumed, to impaired lung growth.

Effects on cardiovascular function

The sternal depression that characterizes the deformity decreases significantly the anteroposterior dimensions of the rib cage. This decrease impedes the ability of the heart adequately to expand, and this in turn limits its ability to increase the stroke volume in order to meet increased metabolic demands (e.g. during exercise). The cardiovascular function may be further complicated by the displacement of the heart and possible rotation of the great vessels.[13–16]

Table 1
Clinical characteristics and lung function in patients with idiopathic pectus excavatum

Clinical characteristics (% of patients)	
Male:female	71:29
Family history	17
Sternal rotation	27
Scoliosis	22
Decreased exercise tolerance	20
Bronchial hyperresponsiveness	18
Lung function indices (% predicted normal)	
TLC	95 ± 15
RV/TLC	127 ± 46
FEV_1	93 ± 19
FEV_1/FVC	86 ± 9
FEF_{25-75}	87 ± 26
PEFR	98 ± 25

Adapted from Koumbourlis and Stolar[4].
TLC, total lung capacity; RV, residual volume; FEV_1, forced expiratory volume in first second; FVC, forced vital capacity; FEF_{25-75}: forced expiratory flow between 25–75% of FVC; PEFR, peak expiratory flow rate.

Table 2
Percentage of patients and type of lung function abnormalities according to age

Pattern of lung function	5–9 years (%)	10–14 years (%)	15–19 years (%)
Normal	67	42	50
Obstructive	33	47	46
Restrictive	0	11	4

Adapted from Koumbourlis and Stolar[4].

Effects on the spine

The thoracic cage is formed by the sternum (anteriorly), the thoracic vertebrae (posteriorly) and the ribs which are connected to the sternum and to the vertebrae with joints that allow their relatively free movement during the respiratory cycle. The depression of the sternum which occurs in pectus excavatum limits the movement of the ribs (especially of the lower ones), thus preventing the expansion of the lower thoracic cross-sectional area and creating the impression that the chest moves only 'vertically'. In addition, due to the asymmetric depression of the sternum, the ribs exert pressure on the spine that may eventually result in displacement of the vertebral bodies and the development of scoliosis.[17]

Effects on the respiratory muscles

The rib cage is covered on the outside by the internal and external intercostal muscles, and is separated from the abdominal cavity by the diaphragm. Although the pectus excavatum does not affect directly any of the respiratory muscle groups, the distortion of the rib cage (and possibly the protuberance of the abdomen) places them at a mechanical disadvantage, as suggested by the significant decrease in the maximal inspiratory and expiratory pressures (MIP and MEP) found in patients with pectus excavatum.[18] This functional impairment of respiratory muscle strength could explain the high prevalence of elevated RV/TLC among patients with pectus excavatum. Although the decrease in respiratory muscle strength is usually not severe, its effects may be much more profound considering that the decreased chest wall compliance would require a higher than usual pressure to achieve the same degree of lung inflation. Thus, it is possible that the decreased lung volumes that have been reported by various investigators are the result of impaired chest wall mechanics and not of impaired lung growth.

CLINICAL PRESENTATION

Pectus excavatum consists of a prominent but variable indentation primarily of the lower sternum that usually creates an asymmetric appearance of the two hemithoraces. Although it is usually present at birth, it may not be fully recognized until the child goes through the periods of rapid somatic growth, especially during puberty. When the body achieves its adult height the deformity tends to stabilize, although further worsening can occur if the patient has developed other complications (e.g. scoliosis).[3]

Patients with pectus excavatum have usually very characteristic appearance. They tend to be tall and thin with a narrow and often asymmetric chest due to the inward displacement of the sternum. The displacement tends to be rightward, although a leftward rotation can also be observed.[4] When standing, patients tend to have a characteristic posture consisting of 'slouching' and tilting of the shoulders to the side of the sternal rotation, and with their arms 'hanging' in the front as opposed to the side of the trunk. The leg on the opposite side to the tilt of the shoulders tends to be bent forward, while the patient leans on the other leg. Seen from

the back, there is often evidence of scoliosis.[4] Patients with mild to moderate pectus excavatum can straighten their back keeping their feet together, but in the more severe cases the slouching of the shoulders prevents such straightening. Despite their generally thin body habitus, patients have a protuberant abdomen. During inspiration the sternal depression becomes even more prominent, while the rib cage appears to move vertically 'upward' and not in the usual outward/upward motion of a normal chest.[3,4]

Idiopathic pectus excavatum during infancy and early childhood is rarely associated with specific symptoms. Even in older children, the first (and often the only) problem tends to be not an organic but a psychological one, caused by self-awareness of the deformity and/or by being teased by other children. As a result many affected children tend to avoid participation in sports and other physical activities, something that may have significant effects on their physical and social development. The problem usually intensifies during the teenage years when 'body image' becomes very significant and when the rapid somatic growth often causes considerable worsening of the deformity. Older children and adolescents seldom have any symptoms at rest but they may experience symptoms on exertion. The most common among these are easy fatigability, dyspnoea, wheezing, chest 'tightness' and, occasionally, sharp pain in the lower chest. Tachycardias and/or palpitations are common complaints as well. To what extent these effects are the direct effect of the pectus excavatum remains unclear.[4,9]

Exercise intolerance seems to be the most consistent of the symptoms associated with pectus excavatum, but its exact cause(s) is unclear and controversial.[13–16,19,20] In general, exercise intolerance is the result either of inability to increase the minute ventilation and/or inability to increase the cardiac output in order to meet the increased metabolic demands. The theory that the exercise intolerance in pectus excavatum is due to 'small' lungs does not appear to be true because in the vast majority of patients the lung volumes are within the normal range and thus patients should be able to produce the necessary increase in tidal volume.[4,9] In contrast, the heart is subjected to direct pressure from the depression of the sternum, which may limit its ability fully to expand, especially during maximal exercise when the cardiac output needs to increase four-fold. It is also possible that exercise intolerance may be due to the decreased respiratory muscle strength.[18] Because of their reluctance to participate in physical activities, patients with pectus excavatum are prone to physical deconditioning which undoubtedly contributes to exercise intolerance. However, some investigators dispute whether the physical deconditioning is a cause of exercise intolerance.[15]

DIAGNOSIS

Pectus excavatum is usually easily diagnosed due to the very characteristic appearance of the chest wall. However, neither the severity of the condition nor the presence of other associated abnormalities can be determined on the basis of the physical appearance alone. Thus, several tests are often necessary for the thorough evaluation of the affected patient, especially if surgical repair is seriously considered. The evaluation usually consists of the following.

1. Pulmonary evaluation

Evaluation of lung function is one of the easiest and most useful tests both for the initial assessment as well as for the long term follow-up. The evaluation should include:

- Body plethysmography for the measurement of the lung volumes in order to determine whether there is volume loss and/or air-trapping.
- Spirometry for the evaluation of the lower airway function. Because of the high prevalence of lower airway obstruction and airway hyperresponsiveness among patients with pectus excavatum,[4] it is recommended that maximal expiratory flow–volume curves are obtained before and after administration of a bronchodilator.
- MIP and MEP for the assessment of respiratory muscle strength. These may be significantly decreased despite the presence of normal lung volumes and/or normal airway function.

2. Radiographic evaluation

Plain chest radiographs can easily confirm the presence of the pectus deformity (especially in the lateral view) but offer very little additional information over the clinical examination and thus they are not routinely recommended. Computed tomography (CT) of the chest is currently used to provide an objective assessment of the severity of the deformity. The severity is assessed on the basis of the 'Haller index', which is the ratio of the transverse diameter of the thorax to the anteroposterior diameter at its narrower point.[3,21] In normal individuals this index is approximately 2.5. Although in very severe cases the index may be as high as 10–12, an index of 4 or more is usually considered severe enough to warrant repair.[3] Despite its accuracy in quantitating the anatomical abnormality, the Haller index can not accurately predict the functional impairment that patients may experience, and in this respect, it could be questioned whether or not it is justified to expose the patient to the high radiation of the CT scan. A better rationale for the use of the CT scan is to provide the surgeon with knowledge of the patient's thoracic anatomy. In the era of minimally invasive surgical repair, knowledge of the exact location of the various intrathoracic organs has direct implications for the type of repair and equipment that are going to be used. Thus, the CT scan is increasingly becoming part of the standard of care at least for patients who are to undergo surgical repair.

3. Cardiovascular evaluation

An electrocardiogram (ECG) and an echocardiogram are routinely recommended, primarily to rule out any other abnormalities. The ECG is usually normal but it may show right-axis deviation and depression of the ST segment. The echocardiogram is recommended in order to rule out the presence of mitral valve prolapse (due to the high association between pectus excavatum and Marfan syndrome). It may also show compression of the right ventricle and of the main pulmonary artery or even evidence of decreased stroke volume and decreased cardiac output. With the exception of mitral valve prolapse, most of the other findings are the result more of the displacement and not the actual dysfunction of the heart or great vessels.[3] In this respect, it is rarely recommended that patients undergo cardiac catheterization and angiography.

4. Cardiopulmonary exercise testing

From a functional standpoint, a progressive cardiopulmonary exercise test is probably the most useful evaluation because it provides a quantitative assessment of the cardiopulmonary limitations and helps determine whether these limitations are primarily due to cardiovascular or pulmonary dysfunction.[13–16,22]

TREATMENT

There are currently several techniques for the repair of the pectus excavatum (for a detailed description see the accompanying article in this mini-symposium by Keunzler and Stolar), but despite their generally good results (regardless of the technique used),

many physicians still argue that repair is not necessary. Their arguments focus on the facts that: (1) the cardiopulmonary abnormalities and limitations tend to be relatively mild; (2) surgical repair does not necessarily improve significantly (and often not at all) these abnormalities; and (3) it is not known whether and to what extent these abnormalities will deteriorate without repair.

The main argument in favour of repair is that the pectus excavatum is a deformity whose significance can not and should not be assessed on the basis of the presence of specific individual symptoms but on the basis of the overall physiological and psychological effect it has on the patient. In this respect repair should be considered to be 'reconstructive', aiming to restore patients to the body shape they would have had under normal circumstances. An analogy is often made with breast reconstruction in women who undergo mastectomies. In both cases, the reconstruction may not result in a measurable clinical improvement but it usually improves tremendously the patients' quality of life. From a purely clinical standpoint, the main (although not entirely proven) argument in favour of repair is the prevention of future deterioration, whereas in the short run it may improve exercise tolerance. It is possible that the latter to some extent may be due to the fact that patients gain in self-confidence and then may actually participate more actively in exercise.

The 'optimal' age for repair of pectus excavatum has changed dramatically over the years. On the assumption that the pectus impedes the normal growth of the lungs, repair was initially advocated at an early age (even in infancy). The results of this approach were often disastrous because the immature cartilages could not support the repair and many patients developed worse deformities, including asphyxiating thoracic dystrophy.[23] It was then assumed that the optimal age for repair would be in late childhood (7–10 years of age) when cartilages are stronger and lung growth is expected to continue for several more years. However, more recent studies have shown the effects of the pectus excavatum on lung growth usually to be quite small. Thus, many surgeons now decide to postpone repair until after the pubertal growth spurt which often significantly worsens the deformity; performed at this time repair can be more definitive and long lasting.[3]

POSTOPERATIVE EFFECTS OF THE SURGICAL REPAIR

Considering the conflicting data regarding the actual pathophysiological effects of pectus excavatum, it is not surprising that the effects of its repair are not much clearer. There appears to be consensus that the reconstructive benefit of the repair is generally successful regardless of the procedure used (modified Ravitch or Nuss procedure). There is also substantial evidence that the procedures are generally safe with low intraoperative and postoperative morbidities.[9] There is much less agreement on whether, when and by how much there is improvement in the lung and/or cardiovascular function.[6–16] To a large extent the conflicting results can be explained on the basis of differences in the patient populations studied (e.g. younger versus older patients); the different procedures used for the surgical repair; and differences in the time of the postoperative evaluation, especially in relation to the age of the patient (e.g. no significant effect should be expected on lung volumes in patients operated on after their lung growth and development are complete). Nevertheless, in reviewing the literature, it could reasonably be concluded that the overall effect of the procedure on the cardiopulmonary function is very modest and probably clinically insignificant (even in studies that reported statistically significant changes). There is still no information either on the natural history of unrepaired pectus excavatum or on the long term effect of the repair when patients reach middle or older age.

CONCLUSION

Pectus excavatum is a common congenital abnormality of the thorax with psychosocial effects that often outweigh its effects on cardiopulmonary function. The deformity may worsen slowly over time without causing any obvious symptoms. Thus, it is advisable for patients to undergo evaluations of their pulmonary and cardiovascular function on a routine basis (e.g. every 1–2 years for those with minimal or no symptoms) in order to detect abnormalities before they become clinically significant. At this point it appears that the reconstruction of the deformity may be the most tangible beneficial effect that the patient may experience. However, unless the pectus excavatum is associated with other serious abnormalities that necessitate early intervention, it is advisable to postpone the repair until or after puberty.

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