Pectus Excavatum, Rare or Underdiagnosed Pathology - A 11'year Review

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Abstract

Background: In Romania, there is no effective screening regarding Pectus Excavatum, there for we cannot estimate a real incidence of these pathologies in our country. I retrospectively studied a group of 39 patients, admitted in the “Marie Curie” Emergency Pediatrics Hospital of Bucharest, in a period of 11 years, with different congenital disorders of the thorax. 32 patients were admitted with Pectus Excavatum.

Methods: I included 39 patients in this study, aged from 0 to 24 years old. The diagnosis included all chest malformations, but only 31 had pectus excavatum, associated or not with other pathologies.

Results: The majority of the Pectus Excavatum patients were males with no symptoms. Minimally invasive surgery was the main choice with great results.

Conclusions: Most of the patient have no symptoms, there for they seek medical care for esthetic reasons, or, in the majority of cases, Pectus Excavatum is discovered by chance when patients seek medical care for other pathologies. Minimally invasive surgery is efficient, with small complication rate, proving to be the best option.

Keywords: Pectus Excavatum; Males; Nuss Technique; Lorenz Bar; Aesthetical; Social

Introduction

Pectus Excavatum represent the most common anterior chest malformation with an incidence of 1 per 300 new born males in most studies [2,3,6], without knowing the exact incidence in Romania. The symptoms are rare, there for patient seek medical care for other reasons or, in rare cases, for aesthetic purposes. As a conclusion, this pathology tends to be neglected, under diagnosed and sometimes mistreated.

When necessary, treatment is minimally invasive, thoracoscopic, with excellent results. In Romania, this type of treatment was introduced for the first time at the Pediatric Orthopedics Clinic from Marie Curie Hospital in Bucharest.

Background

In all scientific literature Pectus Excavatum is considered to be the most frequent chest wall malformation. The clinical presentation may vary, but in essence it is represented by the plugged sternum and it not involves the manubrium steni or the two first ribs [1]. How it occurs is still a mystery. It is usually associated with Marfan and Ehlers Danlos syndromes, or with scoliosis [4].

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One of the reasons for which Pectus Excavatum is so rare in our literature is because it has no symptoms in the majority of cases, and so the Romanian patients do not seek medical help for this pathology. When it is symptomatic, Pectus Excavatum comes with shortness of breath, chest pains, fatigue, dizziness and recurrent respiratory infections [9]. For these patients, it is recommended to have chest X Ray, CT Scan, MRI Scan, heart Ultrasound, EKG and functional lung tests [9,10]. The Haller Index will establish the proper treatment for each patient.

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Treatment can be either conservatory or surgical. There are many surgical techniques described in the literature, some of them being used still today in our country, but the gold standard is the Nuss procedure [5-8]. It is a minimally invasive technique that involves implanting a bar in the retrosternal space and twisting it at 180 degrees with an anterior sternal mobilization [3].

Unfortunately, in Romania there is no effective screening for Pectus Excavatum. Also, there is no correct medical education and so, patients with this pathology refuse to address the medical system unless severe symptoms are present. This tendency fades as younger generations of parents appear, more concerned with aesthetic aspects.

**Figure 3: Intra-operative images – Nuss Procedure. Source: Pediatric Orthopedics Clinic, Marie Curie Hospital Bucharest.**

Materials and Methods

This is a retrospective study in only one hospital over a period of 11 years (2004 - 2014). Data was gathered from the hospital’s charts and the clinic’s photo-video database. Because of the small number of patients, we analyzed the data using Microsoft Excel.

The cohort includes 39 patients with anterior chest wall deformities, out of which 31 had Pectus Excavatum in different clinical presentation.

We included in this study only the patients that were admitted and treated in Orthopedics Clinic of “Marie Curie” Emergency Pediatrics Hospital of Bucharest. There was no sufficient data regarding the patients that were evaluated in the Hospital Ambulatory and not admitted in the clinic, and so we cannot asses the exact number of patients with Pectus Excavatum that addressed Marie Curie Hospital.

Results

Single/Multiple Admissions

In the 10 year period we found a total of 55 charts. In these charts we discovered 31 charts with Pectus Excavatum, out of which there were 22 single admissions and 9 patients with multiple admissions.

<table>
<thead>
<tr>
<th>Total Patient Charts</th>
<th>55</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total patients</td>
<td>39</td>
</tr>
<tr>
<td>Pectus Excavatum</td>
<td>31</td>
</tr>
<tr>
<td>Single admission</td>
<td>22</td>
</tr>
<tr>
<td>Multiple admissions</td>
<td>9</td>
</tr>
</tbody>
</table>

Enrollment years

By far 2008 and 2013 were the best years of enrollment, with 10 and 9 patients. The rest of the years had only one, two or three cases per year. Without having a pattern concerning the presentation, we can only conclude that there are many more cases of Pectus Excavatum in our country waiting to be discovered.

Age

The minimal age was 7 years, the maximal age was 24 years, with an average of 16.3125 years.
Sex ratio

Our study coincide with the literature in regard to the sex ratio. We found 25 males and only 6 females over the studied period.
Clinical presentation

As mentioned, we discovered different clinical presentations, from simple, symmetrical Pectus Excavatum to asymmetrical or associated with other pathologies, such as Scoliosis and Marfan Syndrome.

Symptoms

As expected the majority had no symptoms. The ones that had symptoms presented shortness of breath, fatigue, and progressive loss of physical endurance.

<table>
<thead>
<tr>
<th>Symptoms</th>
<th>Number</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>No symptoms</td>
<td>25</td>
<td>80.66%</td>
</tr>
<tr>
<td>Shortness of breath</td>
<td>1</td>
<td>3.22%</td>
</tr>
<tr>
<td>Fatigue</td>
<td>4</td>
<td>12.90%</td>
</tr>
<tr>
<td>Progressive loss of physical</td>
<td>1</td>
<td>3.22%</td>
</tr>
<tr>
<td>endurance</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>31</td>
<td>100%</td>
</tr>
</tbody>
</table>

Respiratory tests

Despite the fact that the great majority of patients had no symptoms, the respiratory tests showed abnormalities in more than one third of the patients. 29.03% had mild respiratory restriction and 9.67% had moderate respiratory restrictions. No patient had severe respiratory restriction.
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Treatment

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77.4% had surgical treatment and only 22.6% were treated conservatory. This argument confirms once again that mostly severe cases with operatory indications address our medical system. The great majority on Pectus Excavatum, the ones that are with moderate or small malformation, usually avoids medical help for this condition.

7 cases needed re-interventions.

When dealing with such small number of cases, the learning curve is difficult to reach, and so we encountered 3 cases with recurrence and several postoperative complications, all resolved promptly, with great overall success.

In this study group, we found one open surgery, a Stratos Technique and 23 cases with minimally invasive surgery, the Nuss Technique.

Discussion and Conclusions

Pectus Excavatum is the most common anterior chest wall malformation. It varies in terms of clinical presentation, symptoms, functional tests and management. But all of them have something in common – they are real challenges of diagnoses and treatment.

Most of them have absolutely no symptoms, and there for, at least in Romania, they do not seek medical help. This poses a real problem in terms of real incidence of this pathology in our country.

As newer generations appear, more and more patients became aware of their esthetics and social aspects, and will start seeking medical advice for their condition.

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Our cohort is very small, only 31 cases, with an uncommon display over the 11 years we have studied, with one or two cases per year, and nine-ten cases over two years (2008 and 2013).

Age and sex ratio was similar with the literature, as was the symptoms and clinical presentation.

Surgical treatment is necessary in only a small percent of patients, as mentioned by Robert Kelly in a paper from 2008, only when two or more criteria are present from the total manifestations. The fact that 74% of our patients needed surgical treatment suggest that they are many more patients in our country with this condition, and only the severe ones seek medical advice.

There is a great need for effective screening, targeted treatment and management protocols for Pectus Excavatum because this pathology is rare, general practitioners are not used with it, and even professional orthopedic surgeons tend to neglect it.

When this patient reaches experienced clinics, the overall results are very good as minimally invasive surgery becomes standard in the treatment of Pectus Excavatum. In Romania, Marie Curie Hospital in Bucharest was the first hospital where Nuss procedure was performed with encouraging results.

**Bibliography**