Creating Breathing Room: Columbus Children’s Surgical Approach to a Rare Disease

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Figure 1

Figure 2
Jeune syndrome, also known as asphyxiating thoracic dystrophy, was first described by Jeune et al., in 1954 and 1955, in a pair of siblings. This genetically transmitted disease involves multiple deformities, but the major one relates to the chest wall. The ribs are broad, short, and horizontal, with very irregular junctions between the cartilage and the sternum. This results in a bell-shaped, very rigid chest cavity, which does not grow well with time. As a result, these children slowly and progressively run out of room to breathe and, therefore, asphyxiate. Problems may begin as episodes of pneumonia requiring more and more time in the hospital, increasing dependence on the ventilator, and ultimately the inability to ventilate even with maximal support.

The disease is rare. As a result, it fits into the category of an orphan disease, because there are only a very few groups of individuals around the country willing to work with these patients. Furthermore, the condition is chronic and progressive, with outcomes that, to date, have not been particularly good.

Our initial entry into this field came as a result of an inquiry from a patient activist group. When they first contacted us, they stated that they had not found anyone willing to operate on a Jeune syndrome patient prior to total ventilator dependency and deterioration on the ventilator. They wondered if we would be willing to undertake surgical intervention at an earlier point in the disease process, with the hope of allowing additional lung growth by expanding the dimensions of the chest.

When we made our initial assessment, there were a number of operative approaches that had been undertaken, but the most common one was to divide the sternum and hold it apart with prosthetic material. Our opinion was that, instead of expanding the sternum, an approach to the side of the chest would eventually allow for a bilateral expansion, thus doubling the total enlargement possibility, despite the fact that this would be a two-stage procedure. We also tried to devise an enlargement procedure that would actually create more chest wall. This led to the concept of sliding out the rib from the underlying periosteum, which could then regenerate more rib.

Taking into account the above considerations, a collaborative effort was undertaken between thoracic surgery and plastic surgery, and the following concept evolved. Ribs number four through nine are approached. After elevating the skin, subcutaneous tissue and muscles overlying the chest wall, the ribs are separated from their underlying rib beds as well as the remainder of the chest wall, the intercostal muscles, and parietal pleura. The ribs are then divided in a staggered fashion (Fig. 1). The underlying chest wall with the rib bed is also divided in a staggered fashion in the opposite direction from the rib divisions. The long ends of the fifth and sixth ribs are then pried apart and put together to expand the diameter of the chest (Fig. 2). This is done also with the seventh and the eighth ribs. The divided fourth and ninth ribs allow for expansion. The two expanded rib segments are held together with titanium plates to stabilize the enlargement. The plates act as an internal cast to maintain the expansion while the ribs heal. The periosteal bridges are then brought together underneath the ribs to create new areas for calcification and chest wall stability for the ultimate enlargement. Our general approach has been to enlarge one side at a time, with 6-12 months between procedures.
Since 1993, we have performed 14 procedures on 10 patients, expanding the chest wall in this fashion, or a variation of this operation. We have been very pleased with our results in patients older than 3 years. Of the seven patients 3 years or older, we have attained significant clinical improvement and, in some cases, measurable increase in lung capacity. Clinical improvement can be seen either by decreases in the amount of hospitalization required for respiratory difficulties, decrease in oxygen requirements, or decreasing ventilator support.

Working with Dr. Robert Castile of our Pulmonology Division and Drs. Frederick Long and Brent Adler in Radiology, we have also been able to objectively quantify both the functional and anatomical changes produced by our chest expansion procedures. Dr. Castile has developed innovative new methods for measuring standard adult-type pulmonary function tests in infants, including fractional lung volumes. He and his team have been able to measure the changes in lung volume produced by chest expansion in our last seven patients. Drs. Castile and Long have also developed a new technique for controlling breathing and degree of lung inflation during CT imaging that has markedly improved our ability to define the anatomy of the lungs and rib cage in infants. This new method has provided useful pre-operative information and anatomically documented the post-operative changes in chest size.

High-resolution CT images of the lungs have demonstrated unsuspected alterations in lung and airway anatomy in some of the children with Jeune syndrome, suggesting that in some patients, the problem may be more complicated than just a thoracic cavity that is too small. Our experience to-date indicates that the lungs, when confined within a small thorax, may grow abnormally. Expanding the chest of a child whose lungs have grown abnormally may result in less functional improvement than one might otherwise expect. The feedback provided by these objective assessments has enabled us to better understand the clinical spectrum of this syndrome and the impact of our surgical procedure.

Unlike the older children, results in the group under 1 year of age have been disappointing. Two patients who underwent the thoracic expansion procedure ultimately went on to die of their disease without significant benefit from the procedure. While their mortalities were late and unrelated to the operation, it is clear that they did not benefit from the procedure. Both of these infants, it turned out, had severe underlying lung tissue disease. Both had significant tracheomalacia or bronchomalacia, and their clinical courses were consistent with progressive air trapping and ventilatory difficulty based on the airway disease. In those cases, it was clear that simply enlarging the size of the chest did not benefit them. It is likely that airway disease, not simply chest size, was the rate-limiting step for their ventilation.

Based on this experience, we have not recommended the procedure in infants with underlying lung disease. A recent infant, referred from another city, was operated on with the knowledge that the underlying lung was normal by CT scan until one month prior to the procedure, when atelectasis developed in the right upper lobe. This procedure was done only recently, and results are yet to be evaluated.

CURRENT RECOMMENDATIONS

We remain enthusiastic about the procedure for children 3 years of age or older, and are recommending a staged thoracic expansion procedure for these patients. We are not recommending the procedure for infants under the age of 1 year, except in special circumstances and after determining that they have otherwise normal lungs.

Despite the fact that we have one of the largest series of Jeune syndrome patients in the world, we nonetheless consider that we have
only early experience and a limited understanding of the long-term outlook. As time goes on, the durability of the repairs will need to be evaluated. Also, the ability to make any expansion beyond the initial procedure on any given side is also unknown, although that would remain a theoretical possibility.

We have seen one problem on multiple occasions in long-term follow-up of these patients since 1993—fracture of the titanium struts. In the older patients, these strut fractures have tended to be late, and have not altered the course of the improvement. In only one case, the fracture happened in such a way that a sharp piece of the titanium was irritating the underside of the skin and, in that case, we simply removed the offending fragment. Otherwise, the strut fractures have not been important clinically, and have not required therapy. On the other hand, in the infant patients, strut fractures have been important and have tended to detract from the original degree of expansion obtained. It is surprising that the soft infant rib tends to prevail over what should be strong titanium metal. At the moment, our thinking is that it is the repeated and continual flexion of the metal strut that results in its fatigue and ultimate strut fracture. Perhaps a more flexible metal such as flexible steel or plastic would be a suitable material, although this remains speculative.

Long-term outlook for these patients remains unclear. The stability of the repair seems reasonable at this point, but it has not been tested over decades. The hypothesis that expansion of the thoracic cavity will result in expansion of the underlying lung remains a hypothesis, and we have yet to document this if, in fact, this is the case. It is unlikely that the Jeune syndrome is purely a matter of thoracic cage malformation, and a more pervasive abnormality is possible. Varying degrees of underlying pulmonary malformation may exist, and this could result in varying clinical results. We are careful to emphasize to all parents that this is largely uncharted territory. However, this procedure does seem to hold out hope for extension of longevity and improved quality of life in the older age-group children. We continue to offer the procedure to appropriate patients and hope, with further experience, we can add even more to the useful lifetime of these patients.

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