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***Case Report** / Olgu Sunumu*

A new technique for neonatal Jeune syndrome: External thoracic expansion

Neonatal Jeune sendromu için yeni bir teknik: Eksternal torasik ekspansiyon

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ABSTRACT

Jeune syndrome is a rare form of skeletal dysplasia characterized by a narrow, bell-shaped chest (thoracic cage), and typical phalangeal and pelvic bone deformities. Chest expansion is impaired by the short, horizontally positioned ribs, resulting in alveolar hypoventilation and eventually neonatal-infantile death in most cases. External distraction with sternoplasty is a new technique for the treatment of Jeune syndrome, which was firstly used by our team on a newborn by placing a sliding finger fixator which was designed for ulnar lengthening. We believe that this approach can be life-saving in neonates with improved and widespread usage.

Keywords: Jeune syndrome, sternoplasty, thoracic expansion.

ÖZ

Jeune sendromu nadir görülen bir iskelet displazisi olup, dar, çan şeklinde toraks (göğüs kafesi) ve falanks ve pelvis kemiklerinde tipik deformiteler ile karakterizedir. Yenidoğan döneminde dar ve yatay yerleşimli kaburgalar nedeni ile akciğer ekspansiyonu sağlanamaz; bu da, birçok olguda alveolar hipoventilasyon ve nihayetinde yenidoğan ve bebek ölümlerine neden olur. Sternoplasti ile eksternal distraksiyon, ulnar uzatma için tasarlanan kaydırılan parmak fiksatör ile ilk kez ekibimiz tarafından bir yenidoğanda kullanılan, Jeune sendromu tedavisinde yeni bir tekniktir. Bu yaklaşımın geliştirilip yaygınlaştırılarak uygulanması ile yenidoğanlarda hayat kurtarıcı olabileceği kanısındayız.

Anahtar sözcükler: Jeune sendromu, sternoplasti, toraks ekspansiyonu.

Jeune syndrome, which was first defined by Jeune et al.[1] in 1955, is a rare form of skeletal dysplasia with an incidence of 1 in every 100,000 to 130,000 live births. Jeune syndrome, also known as asphyxiating thoracic dystrophy, is an autosomal recessive skeletal dysplasia, newly reported a DYNC2H1 mutation, affecting the formation of endochondral bone *in utero*, characterized by a small and narrow chest (thoracic cage), phalangeal, and pelvic deformities, and typical radiographic signs.[2,3] Chest expansion is impaired by the short, horizontally positioned ribs, resulting in alveolar hypoventilation. This is the most common and important clinical feature of the syndrome. Respiratory symptoms widely vary from respiratory failure, leading to neonatal-infantile death in 60 to 80% of cases, to a mild phenotype without respiratory symptoms.[4] Pathological studies have shown that the lung alveolar growth is normal in children with Jeune syndrome.[4] Some infants with Jeune syndrome require mechanical ventilation and cannot be weaned, whereas others develop progressive respiratory failure and, after attacks of pneumonia, may require intubation and ventilatory support. Without a surgical intervention, these neonates die of respiratory failure, as their lungs become increasingly damaged by long-term ventilation.[5]

Herein, we present the first report of external distraction, a new technique for Jeune syndrome, performed by our team on a newborn.

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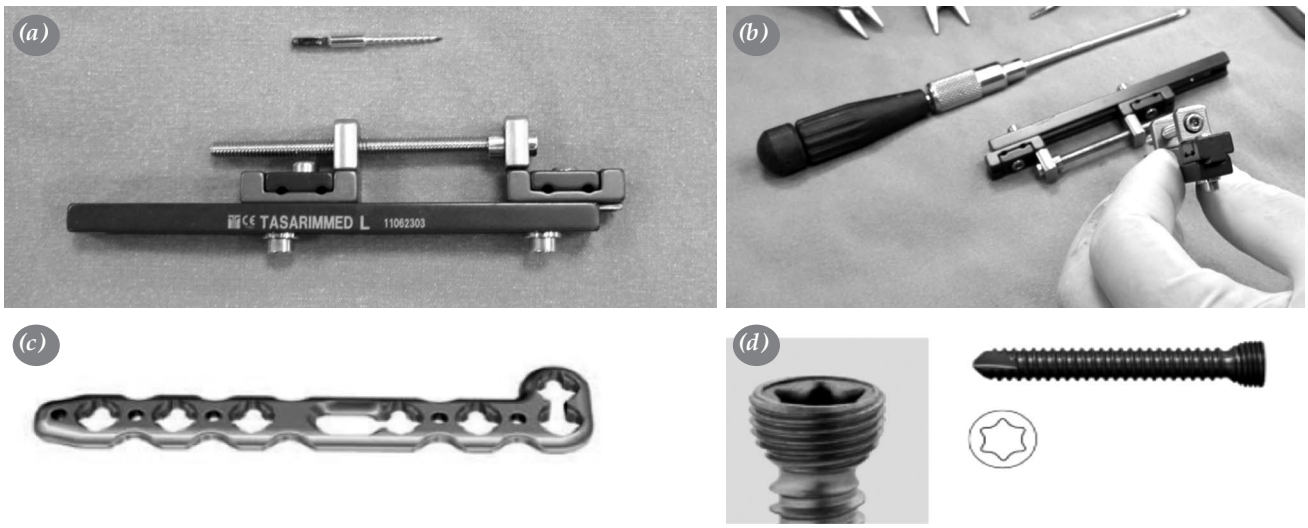

Figure 1. (a, b) Finger fixator. **(c)** VA-LCP Dorsal Distal Radius L-Plate 2.4. **(d)** VA Locking Screw, Stardrive, 2.4 mm, self-tapping.

CASE REPORT

A 23-day-old term newborn was diagnosed with the lethal form of Jeune syndrome and intubated for decreased oxygen saturation just after birth. The only option and chance of survival for our patient was to perform surgery that would allow expansion of his thoracic cage and operation was decided on Day 23. We decided to place a sliding finger fixator (Model

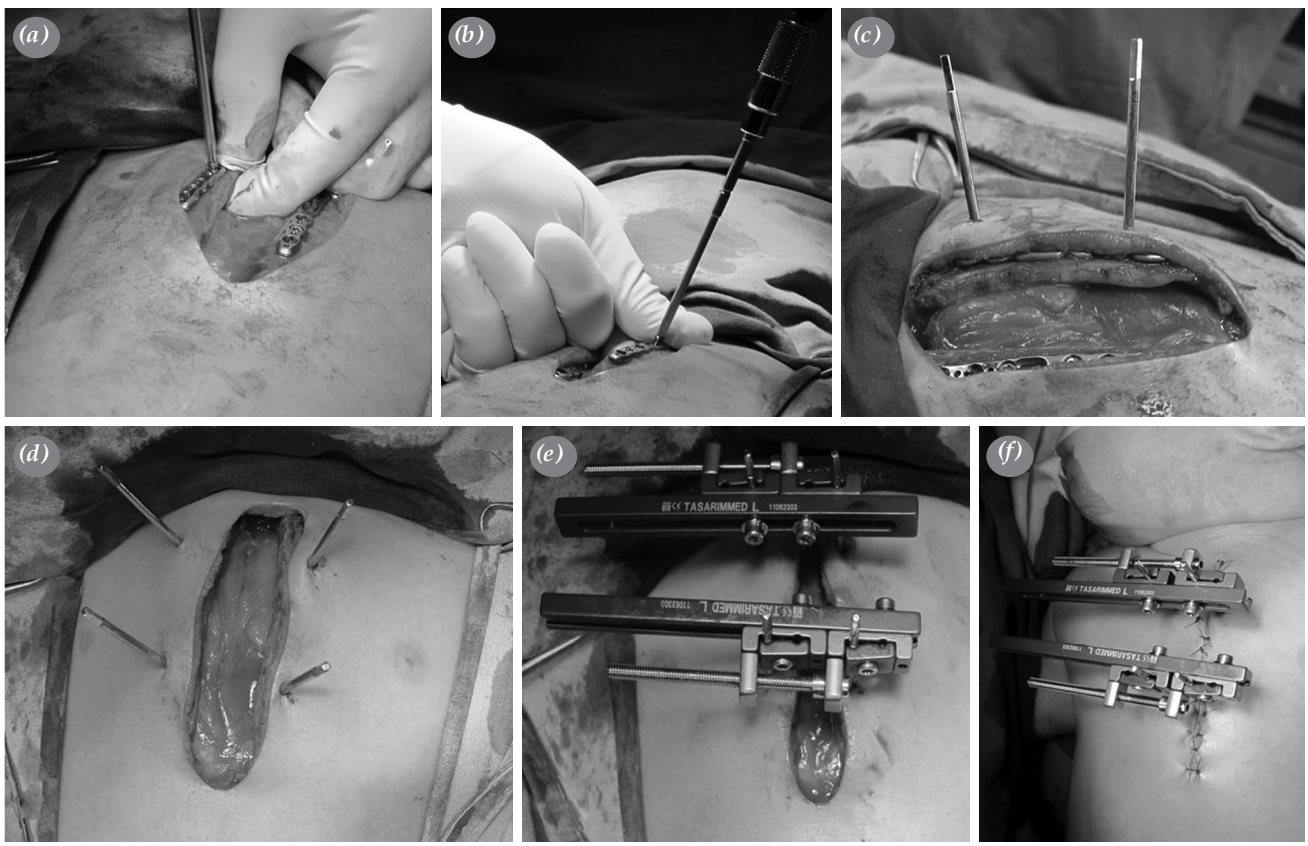

Figure 2. (a, b) Radius plates and screws placed at costal cartilages of sternum connections for fixing. **(c, d)** Fixation holes at each ends of the plates, to introduce four rods. **(e, f)** Two parallel linear external fixators placed on rods.

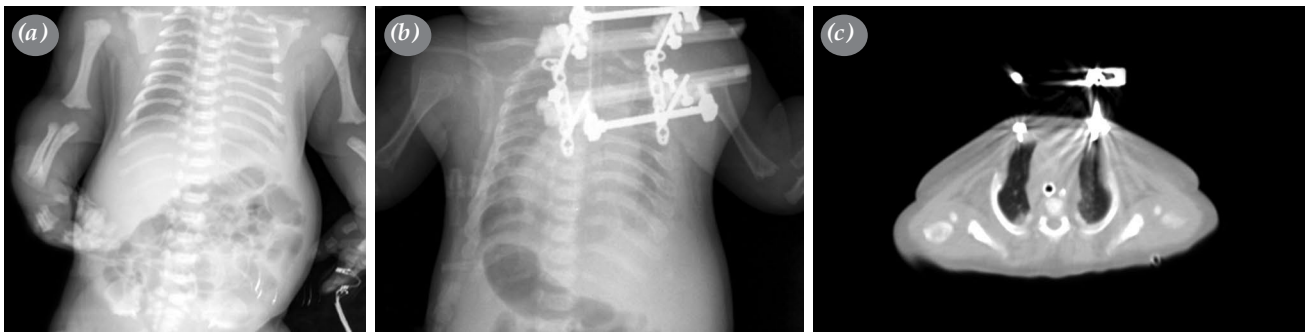

Figure 3. (a) Preoperative chest X-ray of the patient. **(b)** Chest X-ray immediately after the distractor placement. **(c)** Thoracic computed tomography scan showing sternal width before extracting fixator.

11062303 L, TASARIMMED Medical, Istanbul, Turkey) which was originally designed for metacarpal or ulnar lengthening in the practice of orthopedic surgery (Figure 1).

Immediately after sternotomy, costal cartilages of the sternal connections were fixed with longitudinally placed 2-mm-thick distal and radius plates and screws. Temporary fixation holes at each ends of the plates were used to introduce four rods to apply the two parallel linear external fixators (Figure 2). Following surgery, oxygen saturation was increased. On Day 5 postoperatively, distraction was initiated by using two ulnar distractors which was planned to be 1-mm each day, allowing adequate wound healing and bone formation between the two sternal edges.

On postoperative Day 35, 30-mm distraction was achieved between sides of the sternum. External fixator was extracted at 10 weeks under local anesthesia without any complication and the patient was followed with a ceased mechanical ventilation support for six months. Thoracic computed tomography (CT) scan showed sternal regeneration and measured to be in 7-cm width and the lungs were expanded (Figure 3). Despite all efforts, the patient died from disseminated vascular thrombosis and nosocomial sepsis.

A written informed consent was obtained from the parents and/or legal guardians of the patient.

DISCUSSION

There is a few number of techniques in the treatment of Jeune syndrome, such as sternal spreader, sternal split rib graft or prosthetic material, and lateral thoracic expansion.[6] We used the new technique, distraction osteogenesis with two external distractors, and our system is angularly stable and more controllable, allowing millimetric adjustments with no need to a second operation for fixator extraction. This technique is usually used on hard bony parts; however, we had to use it in cartilaginous parts for the first time, and a metal plate was placed to support fixing.

Distraction osteogenesis technique was used for lengthening the long bones of the body, we thought it would be beneficial for the infant. Premature consolidation during the distraction process in young children or neonates must be prevented, as bone healing in neonates occurs faster than in adults. Regeneration of the sternum on CT showed that sternal wall protected the intrathoracic structures and lungs had the capability of expansion. Thoracic expansion through sternoplasty and distraction with an external fixator may increase the predicted length of life and may yield promising results for decreased mortality. Jeune syndrome presenting with respiratory failure in neonates is fatal without a surgical intervention to expand the thoracic cage. If it is possible to create an enough cavity for lung expansion, some treatment techniques, such as extracorporeal membrane oxygenation, may be useful to maintain an effective ventilation.

In conclusion, our case is the youngest newborn (23 days) with Jeune syndrome in whom distraction osteogenesis was used to distract both sternum and ribs, applying a sliding finger fixator. Although this fixator is commonly used in orthopedic surgeries, it was our own idea to use it as a thoracic wall extractor for cartilaginous structures, which was never done before. We believe that this controlled thoracic expansion technique may offer hope to these neonates with severe thoracic dystrophy and insufficiency syndrome.

Declaration of conflicting interests

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