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CASE REPORT

Surgical Treatment of severe Thoracic Lordoscoliosis in an Adult with Freeman–Sheldon Syndrome and Respiratory Dysfunction: A 2-year Postoperative Follow-Up

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ABSTRACT

Purpose: We present data of 2-year postoperative follow-up of surgical treatment for severe lordoscoliosis and respiratory dysfunction in a male patient with Freeman–Sheldon Syndrome (FSS).

Methods: He was diagnosed with FSS at birth. He presented to our institution at the age of 28 years for significant spinal deformity and associated respiratory distress. Physical examination revealed joint contractures of all four limbs; 110° scoliosis was observed at T4–L4, with a severe thoracic lordosis measuring 54°. Dyspnea, poor exercise tolerance, and intermittent back pain were documented; a compressed left main bronchus due to intrathoracic vertebral protrusion was identified. Anterior spinal release and fusion followed by posterior corrective fusion were performed at the age of 28 years.

Results: The patient's scoliosis was corrected to 21° and 40° at T2–T11 and T11–L4, respectively, and thoracic kyphosis now measured at 18° at T2–T11. There was a 190% improvement in the endothoracic hump ratio, a 118% increase in the anteroposterior chest diameter, and a 98% increase in exercise tolerance. Subjective dyspnea and back pain had improved; airway obstruction had been resolved. However, pulmonary function test results remained low. There were extensive and severe postoperative complications, including respiratory failure, prompting subsequent tracheostomy.

Conclusion: Corrective spinal surgery achieved adequate correction for severe thoracic hyperlordosis, and early mortality was averted. Although treatment proved successful in this case, there is no guarantee that success will be replicated in future cases. Therefore, we recommend early surgical intervention prior to progression of severe lordoscoliosis and respiratory failure.

Introduction

Severe spinal deformities can compromise pulmonary function and have deteriorative consequences that may lead to death [1]. Syndromic cases of scoliosis presenting with sagittal plane defects, particularly thoracic hyperlordosis, is associated with even greater deformity [2]. Such cases include Freeman–Sheldon Syndrome (FSS), an extremely rare genetic disorder that has seldom been described in literature in conjunction with lordoscoliosis

[3,4]. FSS is the most severe form of distal arthrogryposis syndrome presenting with multiple congenital joint contractures and skeletal deformities [5,6]. A clinical diagnosis can be made based on the presence of major contractures of 2 distal limbs, a whistling-face like appearance, microstomia, prominent nasolabial folds, and "H-shaped" dimpling of the chin [5-7].

Thoracic lordoscoliosis is a progressive and debilitating condition with the current incidence estimated at approximately 15% of the low back pain population aged ≥60 [8] and is associated with increased risk of postoperative complications, respiratory failure, and significant mortality [9]. In the case series presented by Winter et al., lordoscoliosis and its deleterious effects were observed in all patients with congenital scoliosis who died of cor pulmonale or respiratory failure [1]. Compromised respiratory function in such casescan be attributed to the anterior progression of the thoracic spine, causing thoracic cage deformity and impaired rib dynamics [10-12]. In addition, airway obstruction and reduction in the anteroposterior diameter of the chesthave been documented.Protrusion of vertebral bodies into the thorax associated with thoracic lordosis has been defined as an endothoracic vertebral hump, and intrusion severity can be measured using the endothoracic hump ratio (EHR) [12,13]. EHR is significantly associated with pulmonary distress, and patients may present with dyspnea at rest, hypoxia, and hypercapnia [14]. Because of its severe and progressive nature and the likelihood of lordoscoliosis-induced respiratory failure, early recognition and surgical treatment are crucialand early mortality may be avoided [1,4,15].

Nevertheless, surgical procedures for correcting lordoscoliosis are extremely complex, and potential postoperative complications are extensive, with some literature reporting the prevalence of postoperative complications to be as high as 23% [14,16]. Neurologic injury, extensive blood loss, pulmonary embolism, venous thromboembolisms, and subsequentdeath are common postoperative manifestations. Respiratory failure and difficulties in recovery from pulmonary compromise are also common, with reported incidence of 3.5%. However, there is general reluctance to operate on patients with a history of such severe dysfunction because of surgical complexities and high risk of complication-induced mortality [14]. Despite the high incidence of postoperative complications, surgical intervention in cases of severe progressive deformity is necessary to prolong life. There are very few cases reported in the literature detailing surgical and therapeutic management of patients with FSS with severe lordoscoliosis and poor respiratory capacity, and patient numbers are generally low. To the best of our knowledge, this study is one of the few studies reporting on clinical outcomes of a patient with FSS after lordoscoliosis corrective surgery, with accounts of perioperative and postoperative complications, spinal curve measurements, and Pulmonary Function Test (PFT) data over the course of 2 years. We report the case of a 28-year-old male patient diagnosed with FSS presenting with severe lordoscoliosis and respiratory distress and requiring prompting urgent surgery.

Case Overview

A male was born at 30 weeks of gestation via Cesarean section, weighing 3000 g; there was no relevant family medical history. Joint contractures were identified at birth, thereby prompting subsequent clinical diagnosis of FSS (Figure 1). His first unassisted gait was achieved at the age of 1 year 2 months, and there was no neurological impairment. He underwent treatment for multiple joint contractures via a tendon release for his left hand and right foot at the age of 9 and 11 years, respectively.

The patient first visited our institution at the age of 28years. He was referred to our institution for his significant spinal deformitiesand associated respiratory distress. Moderate intermittent back pain, extending from his scapula to his buttocks, with mild leg paresthesia was described. Physical examination revealed difficultyin breathing, with significant dyspnea and dysphonia at rest. The patient could walk short distances, advancing only 113m in 6 min (6-minute walk test) before needing to rest, with blood gas analysis

revealing deteriorating oxygen saturation from 94% to 90% at that time.

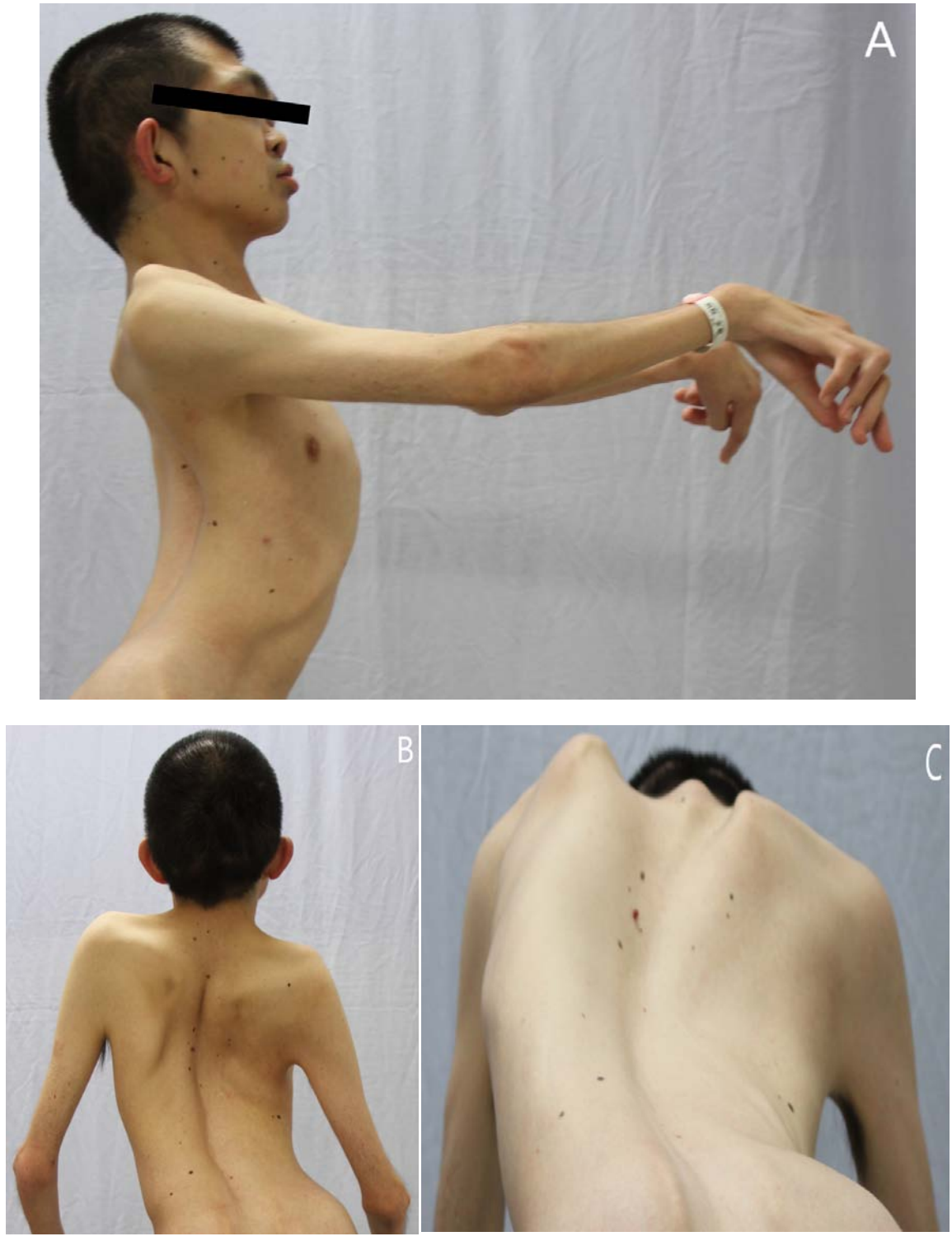

Figure 1: Clinical photograph of a 28-year old male with FSS.

A) Lateral image displaying joint contractures and thoracic hump.

B) Posterior image presenting severe scoliosis.

C) Posterior image with hip flexion emphasizing severe thoracic lordosis.

The patient was 156.6cm in height and significantly underweight at 39 kg, with a Body Mass Index (BMI) of 14.9. A slight mitral and tricuspid regurgitation was noted; however, no neurological impairments were identified. On physical examination, classic signs of FSS were observed, including multiple joint contractures of all four limbs and severe knee joint contracture. A knee extension lag of 35° and 25° was identified on the right and left leg, respectively, and hip flexion was 120° bilaterally.

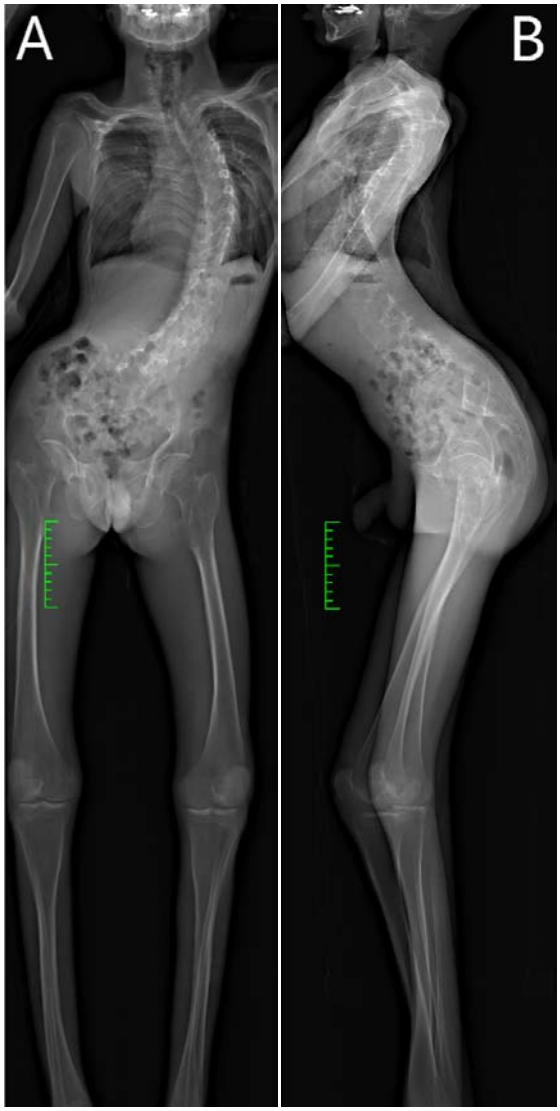

Figure 2: Preoperative chest X-ray radiographs of a 28-year-old male with FSS.
A) Coronal image displaying significant scoliosis present at T4–L4 measuring 110°.
B) Sagittal image presenting severe thoracic lordosis was also detected at T2–T12 measuring 54°, a kyphosis at T12–L2 measuring 11°, and a lordosis at L2–S1 measuring 29°.

A large scoliosis of 110° was identified at T4–L4. Overall, a severe lordotic sagittal curvature of the thoracic and lumbar segments measuring 83° resulted in such a deformity that the patient was physically unable to lie in the supine position (Figure 2). On a detailed examination, a severe thoracic lordosis was found at T1–T12 measuring 54°, kyphosis at T12–L2 measuring 11°, and lordosis at L2–S1 measuring 29°. In addition, an intrathoracic hump was observed, and CT imaging identified significant reduction in the anteroposterior diameter of the chest, with the distance at T4 and T10 measuring as little as 43 and 17 mm, respectively (Figure 3). Compression of the left main bronchus due to significant intrusion of the thoracic spine was also noted.

Because of the severity of the thoracic lordoscoliosis and likelihood of early mortality due to respiratory failure, a surgical approach was suggested after obtaining consent from the patient. First, halo-gravity traction was performed for a period of 3 weeks, followed by a two-staged surgical procedure. Anterior spinal release and fusion from T6–7 to T11–T12 with morselized bone grafts from the ribs was performed; partial resection of ribs 6–10 was performed using a convex side double thoracotomy approach. Subsequently, secondary posterior spinal fusion from T2 to L4 with pedicle screws and bone auto graft was performed. It was necessary to partially resent ribs 5–10 on the concave side; sub laminar wiring, hooks, and Smith-Peterson osteotomy from T6–T12 was also required. The total surgery time was 5 h and 55 min for the first surgery and 7 h for the second surgery. Total amount of blood lost by the patient was 8535mL, and a cell saver device was employed. Postoperatively, the patient's scoliosis was corrected to 21° and 40°at T2–T11 and T11–L4, respectively, and a thoracic kyphosis was constructed measuring 18° from T2–T11. Lordosis from T12–S1 measured 50°.Curve corrections were maintained at the 2-year follow-up (Figure 4). Preoperatively, a Coronal Decompensation (CD) of 66mm, positive sagittal imbalance with a Sagittal Vertebral Axis (SVA) of 87 mm, and pelvic incidence (PI) of 88° were documented. Postoperatively, CD, SVA, and PI measured 15 mm, 82 mm, and 86°, respectively.

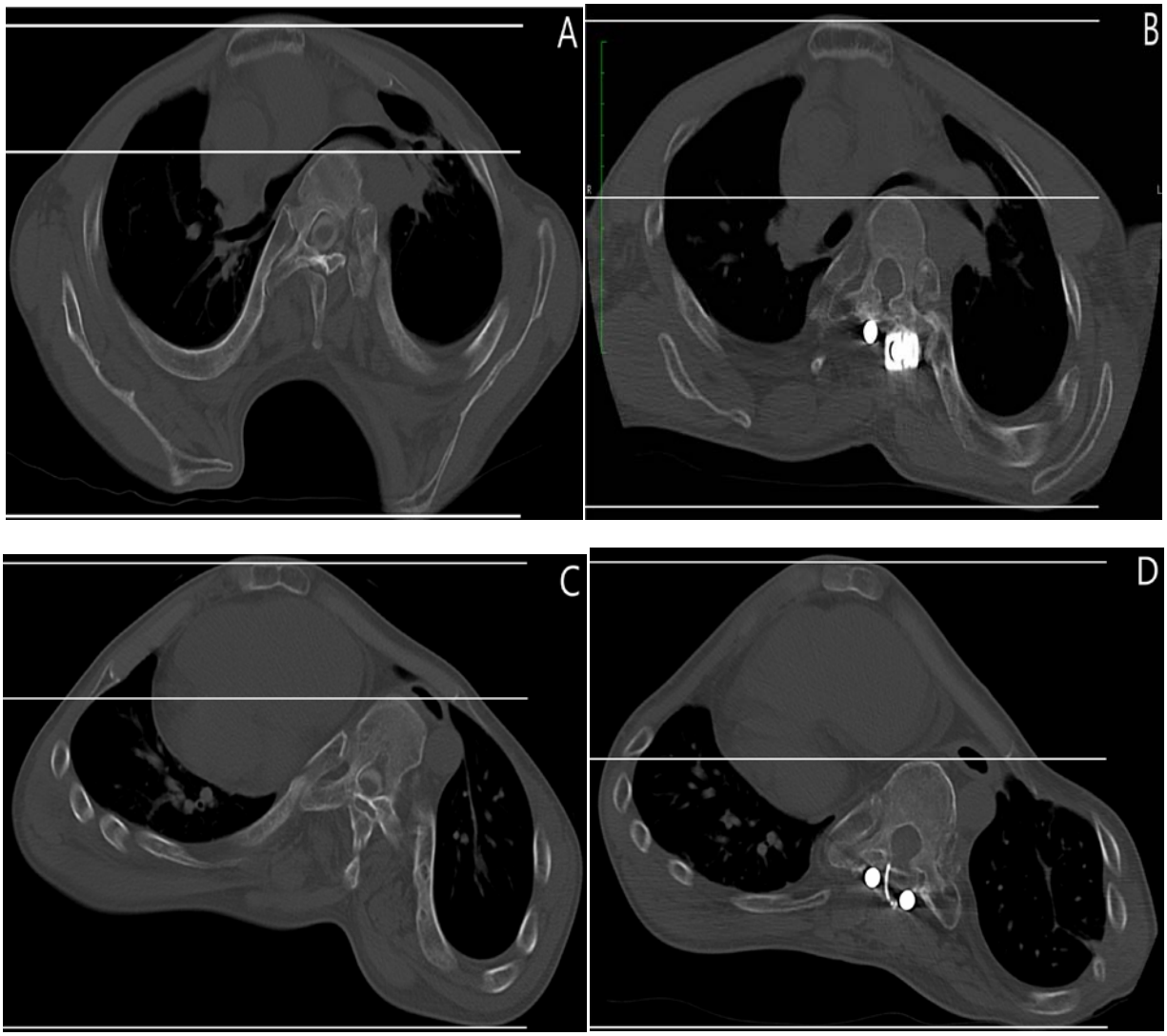

Figure 3:
Preoperative and postoperative computed tomography radiographs of the chest of 28-year-old male with FSS.
A) Preoperative EHR and anteroposterior diameter of the chest at T4 measuring 3.2 and 43mm, respectively. A compressed leftmain bronchus and a severe thoracic lordosis can also be observed.
B) Postoperative EHR and anteroposterior diameter of the chest at T4 measuring 1.1 and 46mm, respectively.
C) Preoperative EHR and anteroposterior diameter of the chest at T10 measuring 2.1 and 17mm, respectively.
D) Postoperative EHR and anteroposterior diameter of the chest at T10 measuring 1.1 and 37mm, respectively.

The patient suffered from extensive perioperative complications, including colitis, ileus, and empyema. The total postoperative recovery time in the Intensive Care Unit (ICU) was 35 days. The patient developed respiratory failure and required subsequent tracheostomy and mechanical ventilation immediately after the first operation, and respiratory assistance was weaned-off at postoperative 4 months. The tracheostomy was maintained at 2 years postoperatively because the otolaryngologist was still concerned about a future respiratory event after closure.

Postoperatively, the anteroposterior diameter of the chest significantly increased, measuring 46 and 37mm at T4 and T10, respectively, and the corrected thoracic kyphosis now allows the patient to lay in the supine position (Figure 3). EHR showed substantial improvement between preoperative and postoperative values, measuring 3.2 and 1.1 at T4 and 2.1 and 1.1 at T10, respectively. However, postoperative PFT results identified a reduction compared with preoperative outcomes. Preoperative results included Vital Capacity (VC), percentage vital capacity (%VC), and Forced Vital Capacity (FVC) of 0.87L, 21.1%, and 0.86L,

respectively. In addition, the patient had a forced expiratory volume per second (FEV1) of 0.62L and an FEV1/FVC ratio of 0.72. At the 2-yearpostoperative follow-up, VC, %VC, FVC, FEV1, and FVC/FEV1 ratio were 0.75L, 17.3%, 0.75L, 0.56L, and 0.75, respectively.

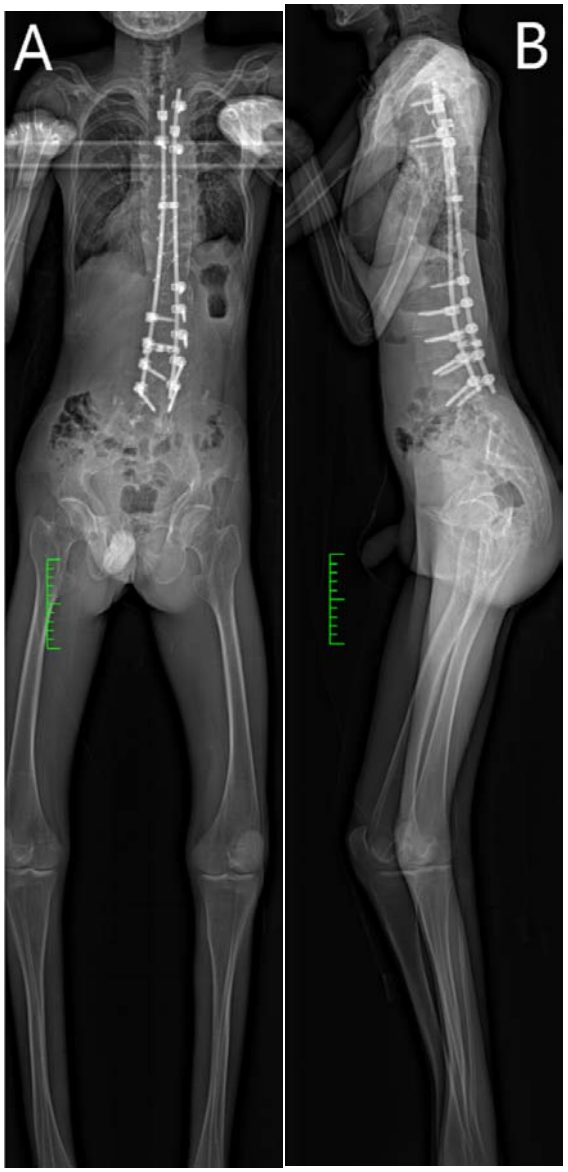

Figure 4: Two-year postoperative chest X-ray radiographs of 28-year-old male with FSS.
A) Coronal image presenting corrected scoliosis measuring 21° and 40° at T2–T11 and T11–L4, respectively.
B) Sagittal image displaying corrected thoracic kyphosis at T2–T11 measuring 18° and lordosis at T12–S1 measuring 50°.

The patient exhibited an increase in height from 156.6cm preoperatively to 162.0cm postoperatively. However, weight and BMI remain low at 38 kg and 14.7at the 2-year follow-up. The patient reported a significant decrease in low back pain, buttocks, and leg; he no longer reported paresthesia of his leg or lethargy. Subjective dyspnea substantially improved, and he can walk 224m. In addition, the patient reports an improvement in unassisted ADL's: being able to climb stairs, lay in the supine position, drive, and return to work1.5 years postoperatively. Quality of life was measured by a scoliosis-research society patient outcome questionnaire (SRS-22). Preoperative scores for the five domains, including function, pain, self-image, mental health, and satisfaction, with management of health were 2.6, 2.0, 4.0, 2.8, and 3.0, respectively, with a total mean score of 2.86. Postoperatively, SRS-22 results increased with mean scores for function, pain, self-image, and mental health as3.6, 4.2, 3.8, and 4.0, and the total mean score of 3.9.

Discussion

FSS is an extremely rare genetic disorder, characterized by multiple congenital joint contractures of the upper and lower limbs [6,17]. Its prevalence is unknown owing to uncertainties surrounding its diagnosis, although up until 1990, there have been only 65 reported cases, affecting both genders equally, without an ethnic preference [5,18]. Our patient was diagnosed with FSS at birth and presented with severe thoracic lordoscoliosis with associated respiratory dysfunction. A two-staged surgical procedure was necessary for correcting the spinal deformity and preventing further pulmonary decline and early mortality.

Scoliosis with severe thoracic lordosis is associated with far more deteriorative respiratory consequences than scoliosis without a sagittal plane deformity [2]. Our patient presented with significant respiratory distress that manifested because of intrusion of the thoracic spine. In addition to the thoracic cage deformity and impaired rib dynamics, anterior progression of the thoracic spine and the resulting endothoracic vertebral hump had obstructed the upper-airway, namely the left

main bronchus. Dyspnea at rest, severely reduced anteroposterior thoracic diameter, an extremely low %VC, and an obstructive FEV1/FVC ratio pointed toward an impending pulmonary function decline and early mortality. In the largest case series reported for lordoscoliosis to date, Dubousset et al. described 18 pediatric cases of thoracic lordosis where severe intrusion of the thoracic vertebrae caused airway obstruction and subsequent respiratory distress [12]. After restoration of normal thoracic kyphosis, we sawa 190% improvement in EHR at T4 and a 118% increase in anteroposterior chest diameter at T10, and the FEV1/FVC ratio no longer suggests an obstructive lung disease picture. Furthermore, exercise tolerance has improved by 98% and subjective dyspnea has also improved, a crucial development considering the difficulty of recovery of adults with scoliosis. The overall surgical results were good, and postoperative improvements are encouraging, suggesting that surgical intervention have helped in preventing respiratory failure and early mortality.

Corrective spinal surgery for adult patients with severe lordoscoliosis with pulmonary dysfunction remains a challenging and contentious area of surgery because of its complexity and high incidence of severe postoperative complications. In the case series presented by Bradford et al., postoperative complications were present in 66% patients after surgical treatment of thoracic lordosis [2]. Extensive blood loss and respiratory failure, prompting subsequent tracheostomy and mechanical ventilation, were significant consequences in the present case. In addition, there was no recovery of pulmonary function even after 2years, although the lack of pulmonary recovery can be accounted for by the presence of the tracheal stoma, which is still maintained. Nevertheless, corrective spinal surgery remains critical in severe cases, where lack of intervention will likely result in death. Therefore, a collaborative evaluation by the surgeon and patient, discussing the risks and benefits of treatment, is crucial. The decision must balance the likelihood of success against potential consequences from postoperative manifestations. In summary, a passive surgical approach in our case would have resulted in insufficient correction of the deformity while subjecting the patient to reduced postoperative complications. Comparatively, an aggressive approach could have achieved necessary corrections but with significant postoperative risks that may have also resulted in early mortality. The patient opted for an aggressive approach as he was aware of the severity of his spinal deformity and progressive respiratory dysfunction. Therefore, the objective in such surgical cases must not only focus on correction of the spinal deformity but also focus on monitoring and minimizing postoperative complications. Ultimately, the patient had overcome significant adversity, and surgical intervention achieved significant correction of the spinal curve deformity, averting likely mortality.

A two-stage anterior release and fusion followed by posterior corrective fusion was critical for correcting an incapacitating spinal deformity, re-establishing trunk stability, and prolonging life. An anterior–posterior surgical procedure is extremely complex and involves a double thoracotomy approach, making this procedure particularly challenging for this case because of the already compromised pulmonary function. Anterior release is crucial for achieving necessary curve alterations prior to posterior correction. In the case series presented by Winter et al., an anterior approach was necessary for intervertebral disk excision and posterior ligament resection, and subsequent correction was achieved by elongating and shortening the posterior and anterior columns, respectively [15]. This combined approach achieves spinal curve correction that would not have been attained using a posterior approach alone. Despite complexities and the potential for extensive postoperative complications, the procedure was necessary for correcting a severe thoracic lordosis and preventing inevitable respiratory failure and subsequent mortality.

In any condition, the principal treatment objective is always management of symptoms that will optimize the quality of life. This includes a focus on improving the overall independence by increasing the capacity to perform unassisted ADLs and independent ambulation [19]. Through surgical correction, our patient's severe

spinal deformity had been corrected, and early mortality had been prevented. Furthermore, the patient can now drive, has returned to work, and has experienced a significant improvement in his quality of life, which is reflected in his 2-year postoperative SRS-22 scores. Overall, surgery has improved the patient's independence, mobility, and social participation capacity, all of which are measures for successful treatment outcomes.

Conclusion

Surgical intervention for severe thoracic hyperlordosis remains a challenging field owing to surgical complexity and likelihood of severe postoperative complications. Extensive blood loss and respiratory failure, prompting subsequent tracheostomy, were significant consequences of this case. Nevertheless, with surgical intervention successful correction of spinal deformity was achieved and likely mortality was averted. Although treatment proved to be successful in this case, there is no guarantee that success in future cases can be replicated. Therefore, we recommend prompt and early surgical intervention for future patients prior to the progression of severe thoracic hyperlordosis and pulmonary failure.

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