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Case Report Open Access

The Diagnosis of Type I Osteogenesis Imperfecta in A Patient with Acute Spinal Burst Fracture after Low-Energy Trauma and Its Operative Treatment: A Case Report

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Abstract

Objective: To report a unique case of operatively and nonoperatively treated burst fractures of thoracolumbar spine in a single patient with type I osteogenesis imperfecta (OI), who previously had not been diagnosed as OI.

Summary of background data: OI is a genetic disorder that causes bone fragility. Although spinal fractures may commonly occur in these subgroup of patients, there have been only a few case report of thoracolumbar burst fractures. And to our knowledge, there has not been reported case of operative treatment for acute burst fracture of thoracolumbar spine in a patient with OI, especially in type I.

Methods: We report a case of 25-year-old woman who suffered unstable burst fracture of L1 after minor fall from a chair. The patient was treated operatively (anterior corpectomy, instrumentation and interbody fusion). She suffered stable burst fracture on T11 after a slip down in 10 months after the surgery, and was treated nonoperatively.

Results: The patient was diagnosed as type I OI, based on the relatively minor nature of the trauma and blue sclerae and histories of multiple fractures of her and her father. Successful bony union and good spinal alignment were achieved.

Conclusion: Anterior corpectomy, instrumentation and interbody fusion may be a feasible option for treatment of unstable burst fractures, even though the patients have any pathologic cause on their fractures. And any patient manifesting severe injuries that are caused by relatively minor trauma may have an underlying pathologic cause, including OI. They should be evaluated thoroughly to determine other cause and to be treated appropriately.

Keywords: Osteogenesis imperfecta; Thoracolumbar spine; Unstable burst fracture; Operative treatment

Introduction

Osteogenesis imperfecta (OI) is a genetic disorder caused by a defect in the synthesis of type I collagen. It is characterized by bone fragility, reduced bone mass, and other connective tissue problems [1]. Its clinical manifestations vary widely from multiple fractures during the fetal period, which leads to perinatal death, to being nearly asymptomatic, with a mild predisposition to fractures [1,2].

Previously reported spinal complications of OI include scoliosis, cervical spondyloptosis, and basilar invagination [3-7]. And spinal fractures also seem to commonly occur in these patients. However, to our knowledge, there has not been reported case of operative treatment for acute burst fracture of thoracolumbar spine in a patient with OI, especially in type I. Most of reported cases were treated nonoperatively [8-10] and only a report regarding surgical treatment for acute spinal fractures was for cervical spinal fracture [11].

We diagnosed a patient, who suffered an unstable burst fracture of the thoracolumbar spine after a minor injury, as type I OI and performed surgical treatment. And we also treated a stable burst fracture of the patient nonoperatively, which occurred in the adjacent vertebra after a minor injury during the follow-up period. We report this rare case with a review of the relevant literatures.

Case Report

The 25-year-old female patient visited the emergency room of the hospital mainly complaining about back pain after a fall from a chair she had been standing. She had midline tenderness over the thoracolumbar spine junction. Neurologic examination was normal. Blue sclera was found in her eyes (Figure 1A). She had relatively short stature. But there was no other abnormal finding on appearance. She had past history of fractures on the left humerus (two times) and the right humerus (once) at her childhood. And her father also had blue sclera (Figure 1B) and a history of multiple fractures with minor trauma.

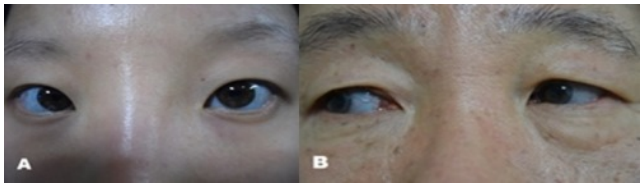

Figure 1: Blue sclerae were observed in the patient's A). and her father's B). eyes.

The plain radiographs demonstrated a fracture at L1with about 70% and 50% loss of height at anterior and middle column, respectively (Figures 2A and 2B).

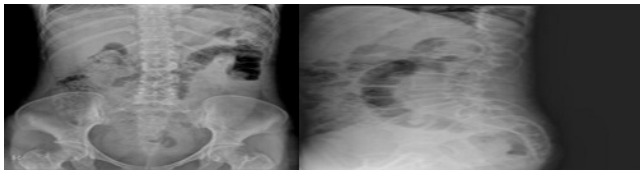

Figure 2: Plain AP A). and lateral B). radiographs of lumbar spine demonstrated a fracture at L1 with about 70% and 50% loss of height at anterior and middle column, respectively.

The kyphotic angle, measured based on Cobb's method, was 19° at T12-L2. Computed tomography (CT) scan demonstrated fracture of posterior cortex of L1 with retropulsion of the bony fragments, compromising about 55% of diameter of the spinal canal (Figures 3A and 3B).

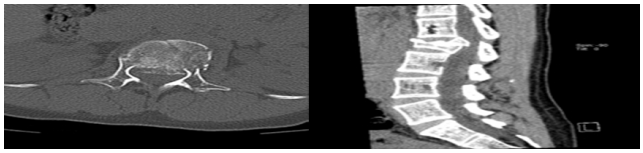

Figure 3: An axial view A). and a sagittal view B). of the CT scans show retropulsion of the bony fragments, compromising about 55% of diameter of the spinal canal.

The magnetic resonance images did not demonstrate any damage on the posterior ligamentous complex (Figures 4A and 4B).

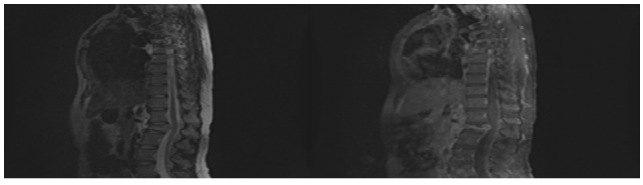

Figure 4: In the MR images, injury of posterior ligamentous complex was not observed A). T2WI B). T1WI with Gadolinium enhancement.

The patient was diagnosed as type I OI based on clinical findings including the relatively minor nature of trauma, blue sclerae, and multiple histories of fractures. For the spinal fracture, we diagnosed it as unstable burst fracture and performed anterior corpectomy, instrumentation and anterior interbody fusion using a mesh cage filled with autogenous vertebral bone and rib bone graft (Figures 5A and 5B). The patient was discharged 2 weeks after the surgery with thoracolumbosacral orthosis (TLSO) applied. In the CT scans, taken 7 months after the surgery, a stable bony union was observed (Figures 6A and 6B), and the patient had no symptom on her back.

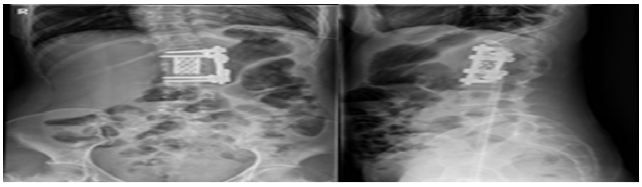

Figure 5: Plain AP A). and lateral B). radiographs, Corpectomy of L1 with anterior interbody fusion using Mesh Cages and rib bone graft was performed.

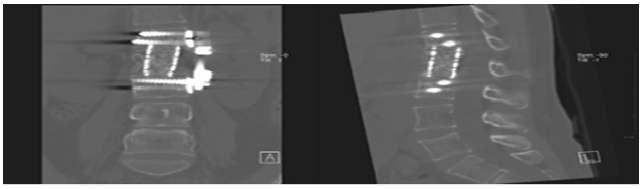

Figure 6: Stable bony unions were observed in the coronal A). and the sagittal B). images of the CT scan that was taken in 7 months after the operation.

After 10 months of the surgery, the patient revisited the emergency room of the hospital for back pain after a fall on the icy road. The plain radiographs and CT scans demonstrated that the previously treated area maintained stable bony union. However, a fracture of T11 with about 15% loss of height and minimal involvement of the spinal canal was noted (Figures 7A and 7B). The patient was diagnosed as stable burst fracture. She was treated nonoperatively with bed rest for a week, and discharged with TLSO applied.

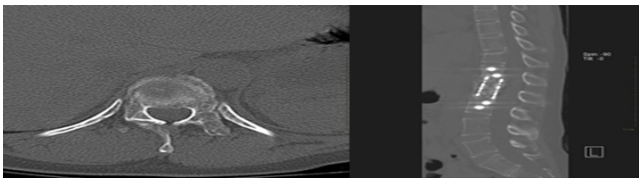

Figure 7: An axial A). and a sagittal view B). of the CT scans demonstrated a fracture of T11 vertebra with loss of about 15% of the height and minimal involvement of the spinal canal .

In the dual energy x-ray absorptiometry, the average bone mineral density of her lumbar vertebrae was 0.647 g/cm^2 and the T-score was -3.6. Her serum calcium, serum phosphorus and serum alkaline

phosphatase level were 8.8 mg/dL, 4.5 mg/dL and 58 IU/L, respectively. Serum 25-hydroxyvitamin D3 level was 5.0 ng/mL, blood osteocalcin level was 38.8 ng/dL, and urinary N-telopeptide level was 53 nM/mM creatinine. Oral administration of alendronate and vitamin D complex was prescribed for the treatment of OI and vitamin D deficiency. The patient is currently under regular outpatient follow-up without any symptoms until 2 years after the surgery (Figures 8A and 8B).

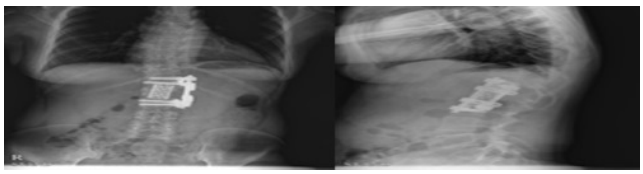

Figure 8: Plain AP A). and lateral B). radiographs were taken at 2 years after the operation. The T11 vertebra is in well healed state, although a slight reduction in height is observed. And the positions of the instruments and the spinal alignment are well maintained.

Discussion

OI is the most common of the genetic connective tissue disorder that primarily affect bone. Prevalence is estimated to 1 per 20,000 people, and type I OI is known to be the most common type, with a prevalence of 1 per 30,000 people [12]. Most patients with a clinical diagnosis of OI have a mutation in one of the two genes that encode the chains of collagen type 1 (COL1A1 and COL1A2). However, the diagnosis of OI cannot be excluded even when the genetic test is negative, because in some individuals no such mutations are detectable. Therefore, a diagnosis mostly depends on distinguished clinical findings and family history [2].

In 1979, Silence et al. [13] classified OI into 4 subtypes based on clinical manifestations and genotypes. Later additional subtypes were recognized, and currently 11 subtypes are known [14]. Type I, mild deforming OI, is characterized by normal or mild short stature, a mild to moderate increased fracture rate, and blue sclerae. This type of OI could be simply regarded as osteoporosis, because it is difficult to diagnose given that it presents only minor clinical symptoms without causing major skeletal deformities [15]. The cases of OI that have been reported are mostly type II or III, accompanied by severe clinical manifestations, and there is a relatively small number of cases that precisely match type I.

In this case, it was possible to diagnose the patient as type I OI without genetic testing, considering the spinal fractures, past history of humeral fractures, blue sclerae, and the family history. Particularly, in this case, the unstable burst fracture occurred after a relatively minor trauma, whereas unstable burst fractures mostly occur after high energy trauma such as traffic accidents and falls. The cause of this unexpected severe injury can be regarded as bone fragility due to OI.

It has been reported that spinal complications commonly occur in the patients with OI and these can lead to spinal deformities [2]. Many literatures regarding spinal complication in OI patients and its treatment have been presented [3-7]. However, most of them has predominantly focused on the treatment for scoliosis, kyphosis or basilar invagination. To our knowledge, there has not been reported case of operative treatment for acute burst fracture of thoracolumbar spine, especially in a patient with type I OI. Most of reported cases of spinal fractures were treated nonoperatively [8-10], and only a report regarding surgical treatment for acute spinal fracture was for cervical spinal fracture [11].

There is no established guideline in surgical treatment of spinal complications in OI patients. However, the data from other studies demonstrate that spinal instrumentation and fusion can be accomplished [3,4,11]. General surgical indications for thoracolumbar vertebral burst fracture includes a neurologic deficit, 50% or more of canal compromise, 50% or more of height loss of vertebral body or 30 degrees or more of kyphosis [16]. We recommend following these indications for patients with OI, but frequent radiologic and neurologic examination should be done with patients undergoing conservative treatment.

In our case, successful arthrodesis and good spinal alignment were achieved with anterior corpectomy, instrumentation and anterior interbody fusion using mesh cage and autogenous vertebral and rib bone graft. We chose an anterior approach because direct decompression of dorsally displaced vertebral body fragment is most reliably accomplished via an anterior approach [16]. We carefully think that this technique may be a feasible option for treatment of unstable thoracolumbar burst fractures in OI patients, although the possibility of intraoperative fractures and instrumentation failure should be still kept in mind.

Bisphosphonates are commonly prescribed in patients with OI. Both oral and intravenous form have been shown to increase BMD in both children and adults, but the effects on fracture incidence remain unclear [17,18]. Teriparatide and RANKL inhibitor have similar results and further investigation is also needed [18]. Nutritional support including calcium and vitamin D is recommended to potentiate bisphosphonate therapies [19].

Because it presents only minor clinical symptoms, type I OI is very difficult to diagnose, especially in patients who present the fracture(s) with definite history of trauma. However, because lifelong and multidisciplinary treatments are crucial for these patients, considering that OI can lead to complications such as re-fracture or deformity, proper and early diagnosis should be made. Therefore, it should be emphasized that any patient manifesting severe injuries than it is expected to considering actual injury mechanism, as in this study, should be evaluated thoroughly to determine other cause of pathological fractures, including OI.

We diagnosed a patient with the unstable burst fractures of the thoracolumbar spine after a minor injury as type I OI, performed surgical treatment, and treated the stable burst fracture nonoperatively, which occurred in the adjacent vertebra after a minor injury during the follow-up period. Under the experience from this case, we think that anterior corpectomy, instrumentation and interbody fusion may be a feasible option for treatment of unstable burst fractures, even though the patients have any pathologic cause on their fractures. And any patient manifesting severe injuries that are caused by relatively minor trauma may have an underlying pathologic cause, including OI. They should be evaluated thoroughly to determine other cause and to be treated appropriately.

Conflict of Interest

The authors report no conflict of interest concerning the materials and methods used in this study.

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