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Comprehensive Treatment of a Patient with Osteogenesis Imperfecta and Congenital Hemivertebra: A Case Report

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Case Report

Keywords: Osteogenesis imperfecta, Congenital scoliosis, Hemivertebra, Halo-gravity traction, Surgical correction

Posted Date: November 1st, 2026

DOI: https://doi.org/10.21203/rs.3.rs-11199521/v1

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Additional Declarations: No competing interests reported.

Abstract

Osteogenesis imperfecta (OI) is a rare hereditary connective tissue disorder characterized by extreme bone fragility, making the surgical management of associated severe scoliosis exceptionally challenging due to the high risk of instrumentation failure. The coexistence of OI and a congenital hemivertebra in a single patient is exceedingly rare and further complicates spinal deformity correction. We report the case of a 17-year-old boy with OI who presented with progressive, severe thoracolumbar scoliosis and worsening lumbar pain. Imaging revealed major Cobb angles of 85° and 95°, a fully segmented L3 hemivertebra, and markedly reduced bone mineral density with a total lumbar Z-score of –5.8. To optimize surgical safety and outcomes, a comprehensive, multidisciplinary treatment strategy was implemented. This included preoperative halo-gravity traction to improve spinal flexibility and alignment, alongside bisphosphonate therapy to enhance bone mass. The patient underwent a posterior L3 vertebrectomy and spinal fusion from T4 to S1. Postoperative radiographs demonstrated satisfactory deformity correction. Although asymptomatic pedicle screw breakage was observed at the 6-month follow-up—highlighting the immense biomechanical stress posed by the inherent bone fragility in OI—the patient maintained stable spinal alignment without significant loss of correction at 12 months. This case underscores that while meticulous perioperative optimization (including traction and pharmacological therapies) combined with advanced surgical planning can achieve successful deformity correction in patients with OI and congenital scoliosis, continuous long-term monitoring remains vital due to the persistent risk of hardware fatigue.

Trial registration

Not applicable. This study is a case report and not a clinical trial.

Introduction

Osteogenesis imperfecta (OI) is a rare hereditary connective tissue disorder, most commonly caused by pathogenic variants in the genes encoding type I collagen [1, 2]. In addition to bone fragility, patients present with multisystem manifestations, including short stature, blue sclerae, and cardiovascular abnormalities[2]. The prevalence of scoliosis among individuals with OI is high, ranging from 39% to 78% [3]. A coronal Cobb angle greater than 70° is generally considered severe and often warrants surgical correction to prevent pulmonary compromise [4].

However, bone fragility in OI poses significant challenges for spinal surgery, particularly due to the increased risk of instrumentation failure [5]. To minimize complications, perioperative management should include preoperative traction, pharmacologic treatment for osteoporosis, and careful surgical planning. While scoliosis in OI is well-documented, its coexistence with a structural congenital anomaly—a hemivertebra—is exceedingly rare. This report describes a complex case where systemic bone fragility and focal structural instability synergistically drove a severe 95° deformity.

Case report

The patient was a 17-year-old boy with a history of a healed right rib fracture associated with OI. At age 5, he was diagnosed with scoliosis and followed for 10 years without active treatment. Over the last three years, he developed worsening lumbar pain and left lower limb numbness. Physical examination revealed severe thoracolumbar kyphoscoliosis (Fig. 1). Neurologic examination was essentially normal. Standing radiographs revealed Cobb angles of 20° (T1–T6), 85° (T7–T12), and 95° (L1–L5), indicating an extremely rigid and severe deformity (Fig. 2a-c). Three-dimensional computed tomography (3D-CT) demonstrated a fully segmented L3 hemivertebra (Fig. 3a, b). Dual-energy X-ray absorptiometry (DEXA) showed profoundly reduced bone mineral density, with a total lumbar Z-score of − 5.8 (Fig. 4). The final diagnoses were severe congenital scoliosis secondary to an L3 hemivertebra, osteogenesis imperfecta, and osteoporosis.

To optimize the surgical window, the patient underwent four weeks of halo-gravity traction (HGT) and received bisphosphonate therapy preoperatively. Subsequently, scoliosis correction, L3 hemivertebra resection (vertebrectomy), and fusion with internal fixation from T4 to S1 were performed. Long-segment fusion was chosen to distribute the mechanical load across multiple anchors in the fragile bone.

Postoperative radiographs demonstrated Cobb angles of 5° (T1–T6), 45° (T7–T12), and 55° (L1–L5), indicating satisfactory correction (Fig. 2d-g). Additionally, the complete resection of the L3 hemivertebra was clearly demonstrated (Fig. 3c, d). At the 3-month follow-up, there was no significant loss of correction (Fig. 5a). However, radiographs at the 6-month follow-up revealed pedicle screw breakage at the L4 level (Fig. 5b). Despite this instrumentation failure, the patient remained asymptomatic, and the 12-month follow-up radiographs demonstrated stable spinal alignment without significant loss of correction, suggesting clinical success through biological fusion (Fig. 5c).

Discussion

The primary goal of scoliosis management in OI is to prevent curve progression, yet brace treatment is typically ineffective [3]. Surgical correction is often necessary for severe curves but carries substantial risks due to bone fragility and deformity rigidity [4, 5]. In our case, the coexistence of OI and a congenital hemivertebra created a unique “double-hit” biomechanical challenge. According to the natural history studies by McMaster et al., a single fully segmented lumbar hemivertebra typically leads to a curve of approximately 45° by skeletal maturity [6]. Our patient’s deformity progressed to 95°, suggesting that the underlying systemic bone fragility of OI synergistically exacerbated the structural instability caused by the hemivertebra.

The surgical risks in this scenario are multifaceted. First, the severity of the 95° curve increases the tension on the spinal cord during correction, potentially leading to neurological injury [7]. Second, altered vertebral anatomy in congenital scoliosis reduces the safety margins for pedicle screw insertion [8]. Third, the collagen defect in OI impairs bone healing and increases the risk of “stress shielding” at the bone-screw interface, leading to instrumentation failure [5, 9].

To mitigate these risks, multiple perioperative strategies were employed. Preoperatively, HGT was used to improve spinal flexibility and pulmonary function [10, 11]. Concurrently, bisphosphonate therapy was initiated to enhance bone mass and screw purchase [12]. Intraoperatively, a 3D-printed spine model and computer simulation guided the screw trajectories, while titanium rods were selected for their relative elasticity, which may be advantageous in fragile OI vertebrae [13]. Furthermore, intraoperative blood salvage and tranexamic acid were used to manage the risk of major blood loss associated with brittle bones [14].

In our case, the pedicle screw breakage at the 6-month follow-up is a critical clinical finding. It highlights that in OI patients, the competition between hardware fatigue and biological bone fusion is precarious. The delayed union characteristic of OI likely subjected the screws to prolonged mechanical stress, eventually leading to fatigue failure. However, the maintenance of stable alignment at 12 months indicates that the comprehensive multidisciplinary approach—combining structural resection and long-segment fusion—was robust enough to allow for eventual stabilization. Postoperative rehabilitation remains vital to improve core strength and support the fused segment [15].

Conclusion

The management of severe scoliosis in OI requires more than standard corrective techniques. The presence of a hemivertebra acts as a structural catalyst for extreme deformity. While multidisciplinary care involving HGT and pharmacological bone enhancement can achieve successful initial correction, the late complication of screw breakage underscores that biological fusion is a slow process in the OI population, necessitating meticulous long-term monitoring.

Declarations

Funding

The authors received no specific funding for this work.

Trial registration

Not applicable. This study is a case report and not a clinical trial.

Ethics approval and consent to participate

Ethics approval was waived by the institutional review board for this single-patient case report, in accordance with local policy. Written informed consent to participate was obtained from the patient and his legal guardian.

Consent for publication

Written informed consent for publication of clinical details and images was obtained from the patient and his legal guardian.

Author Contribution

(I) Conception and design: Longyu Chen, Daming Liu, Zixiang Wu;(II) Administrative support: Zixiang Wu, Quanchang Tan;(III) Provision of study materials or patients: Tao Jiang, Yibo Geng, Yuming Zhang, Siming Huang;(IV) Collection and assembly of data: Longyu Chen, Daming Liu, Yafei Feng;(V) Data analysis and interpretation: Longyu Chen, Daming Liu, Zixiang Wu, Quanchang Tan;(VI) Manuscript writing: Longyu Chen, Daming Liu;(VII) Final approval of manuscript: All authors.

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Figures

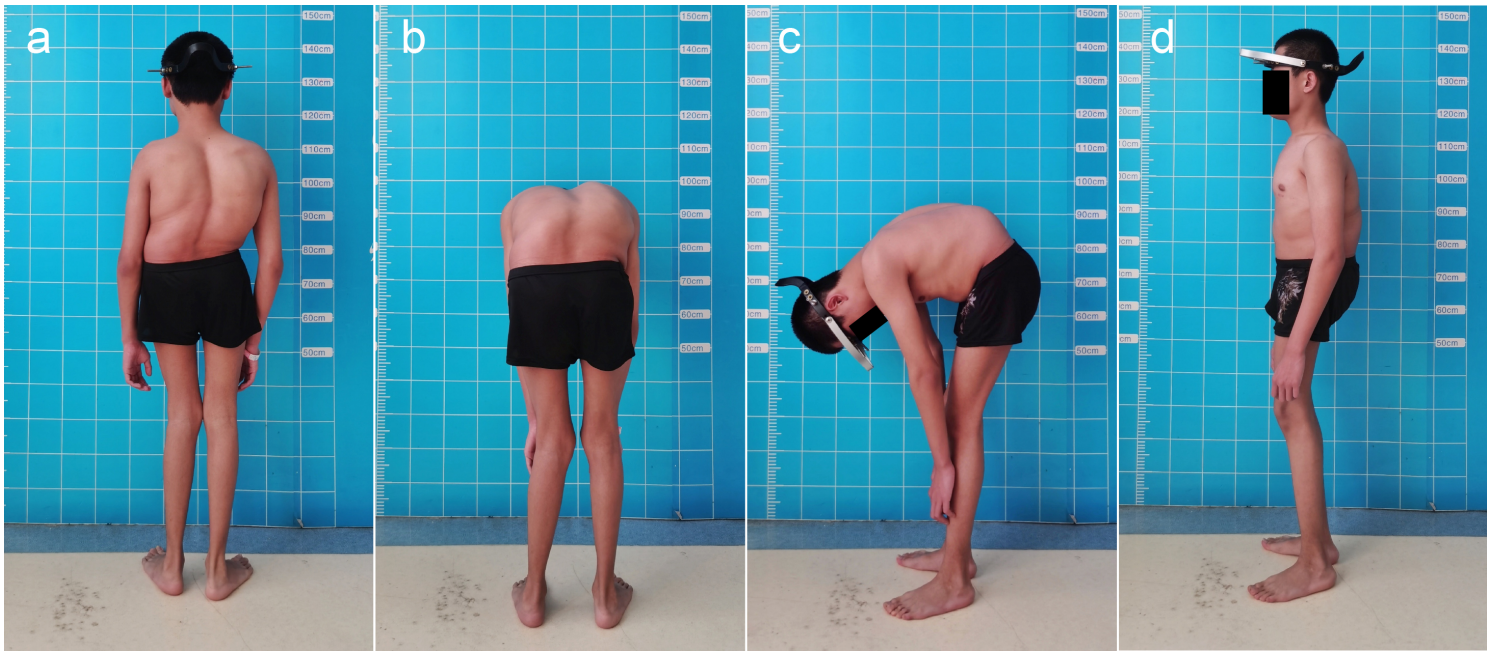


Figure 1

Preoperative clinical photographs.

The photographs demonstrate severe thoracolumbar kyphoscoliosis with significant trunk shift and asymmetry in both coronal and sagittal planes.

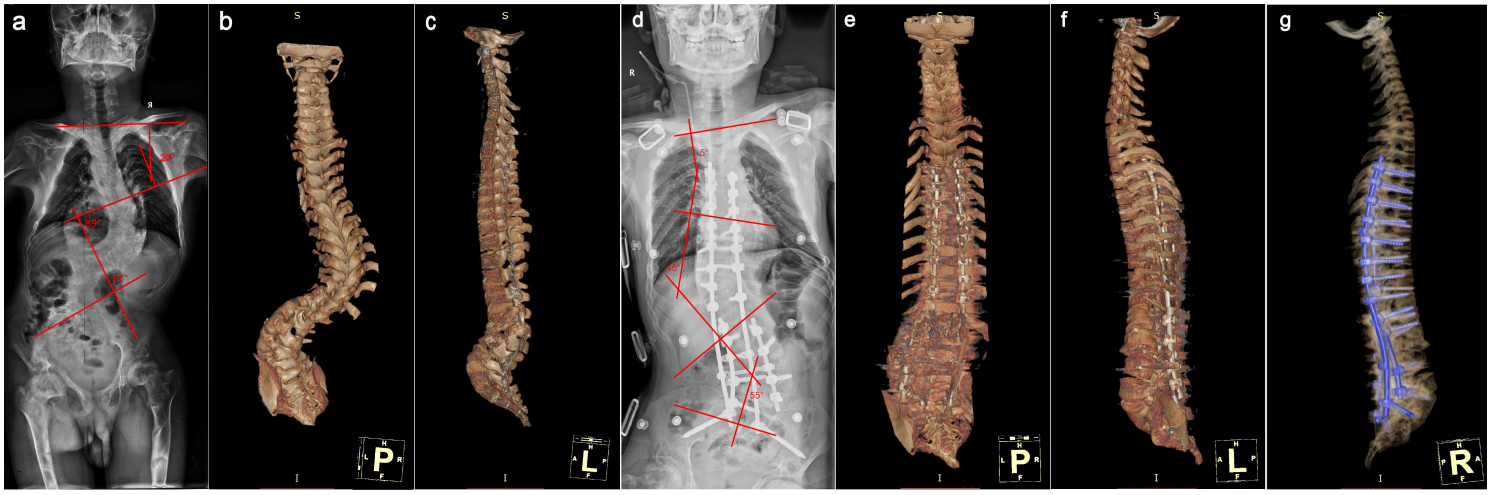


Figure 2

Preoperative and postoperative standing full-spine radiographs.

(a–c) Preoperative radiographs show severe spinal deformity with Cobb angles of 20° (T1–T6), 85° (T7–T12), and 95° (L1–L5).

(d–g) Postoperative radiographs demonstrate satisfactory correction of the deformity, with Cobb angles reduced to 5° (T1–T6), 45° (T7–T12), and 55° (L1–L5) following T4–S1 posterior spinal fusion.

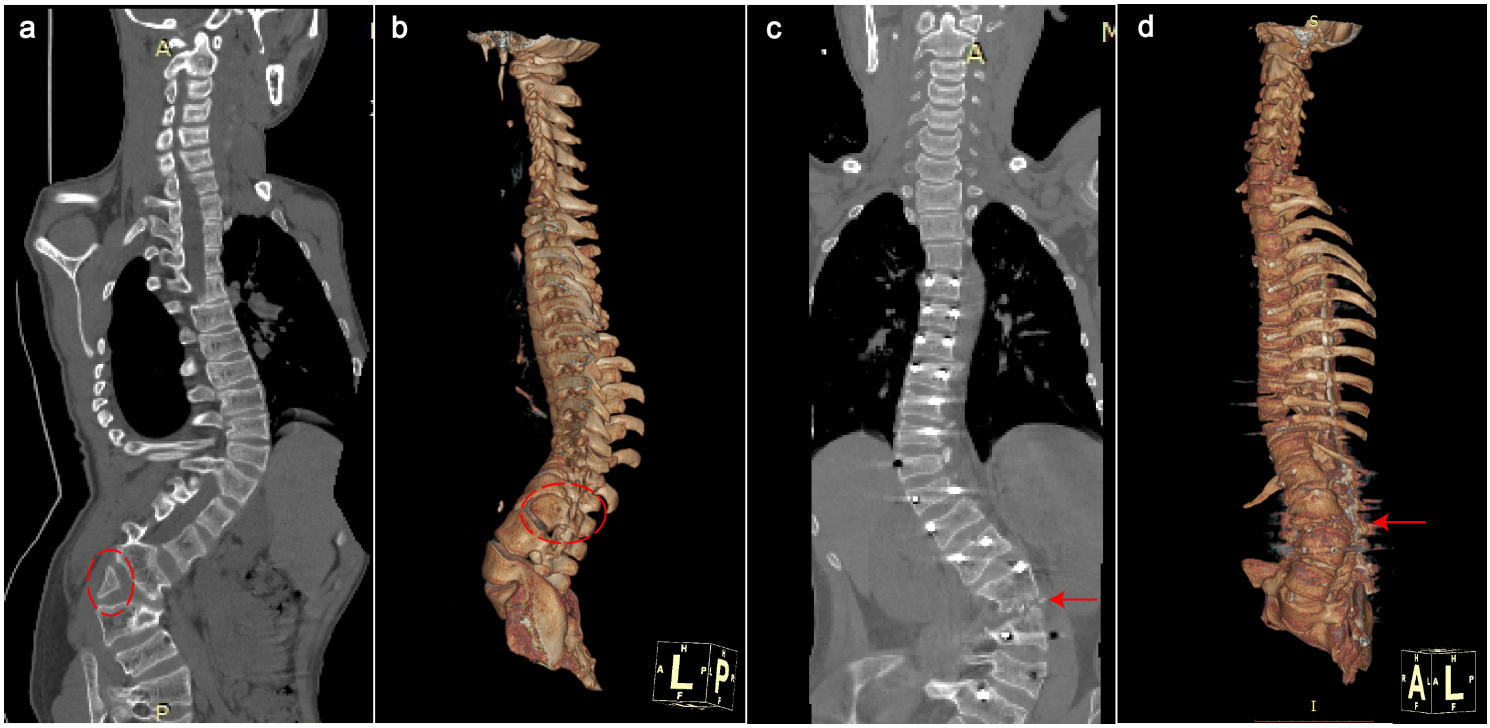


Figure 3

Three-dimensional computed tomography (3D-CT) of the L3 hemivertebra.

(a, b) Preoperative 3D-CT reconstructions confirm the presence of a fully segmented L3 hemivertebra, characterized by distinct intervertebral disc spaces on both its superior and inferior surfaces.

(c, d) Postoperative imaging clearly demonstrates the complete resection of the L3 hemivertebra (vertebrectomy) and the subsequent realignment of the lumbar spine.

Referring Physician:

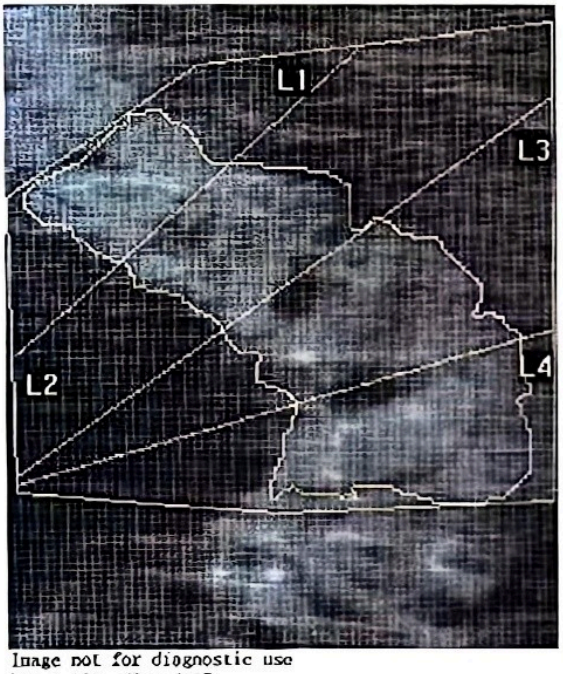


Image not for diagnostic use
k = 1.134, d0 = 44.5
126 x 119

Scan Information:

Scan Date: 12 June 2020 ID: A06122014
Scan Type: f Lumbar Spine
Analysis: 12 June 2020 10:35 Version 13.3:5
Lumbar Spine
Operator:
Model: Discovery Wi (S/N 83860)
Comment:

DXA Results Summary:

Region	Area (cm^2)	BMC (g)	BMD (g/cm^2)	T - score	PR (%)	Z - score	AM (%)
L1	9.44	3.44	0.364		35	-4.5	39
L2	13.31	2.01	0.151		14	-6.2	15
L3	16.98	3.32	0.196		17	-6.2	19
L4	16.83	7.40	0.440		40	-4.3	44
Total	56.55	16.18	0.286		26	-5.8	29

Total BMD CV 1.0%, ACF = 1.013, BCF = 1.026, TH = 6.397

Figure 4

Dual-energy X-ray absorptiometry (DEXA) scan.

The DEXA scan results indicate profound systemic osteoporosis, with markedly reduced bone mineral density. Specific Z-scores for L1–L4 are –4.5, –6.2, –6.2, and –4.3, respectively, with a total lumbar Z-score of –5.8.

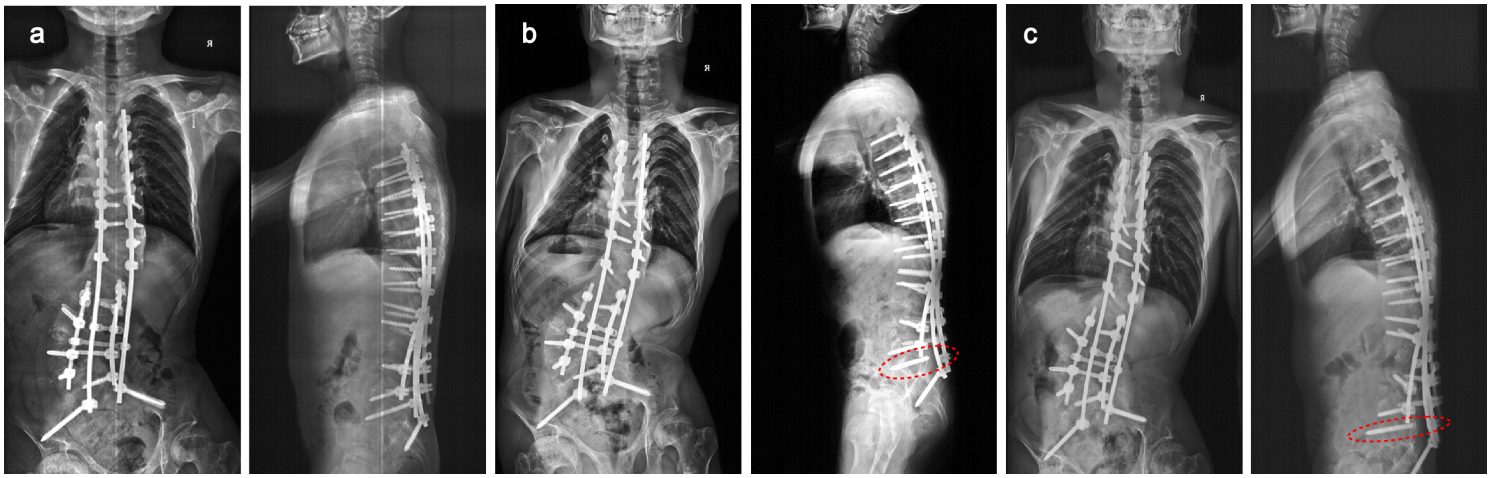


Figure 5

Radiographic follow-up demonstrating instrumentation status.

(a) Radiograph at the 3-month follow-up shows stable spinal alignment without evidence of hardware complications.

(b) Radiograph at the 6-month follow-up reveals asymptomatic pedicle screw breakage at the L4 level, likely due to fatigue failure in the context of inherent bone fragility.

(c) Radiograph at the 12-month follow-up demonstrates that despite the hardware failure, the spinal alignment remains stable with no significant loss of correction, indicating progress toward biological fusion.