Double non-contiguous fractures in a patient with spondylo-epiphyseal dysplasia with spinal ankylosis treated with open and percutaneous spinal fixation technique: a case report

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Abstract

Background: Patients with ankylosing spines are susceptible to developing spinal fractures even with minor trauma and can develop early or late neurological injuries. These fractures require early and aggressive surgical management to enable spinal stability and/or neural decompression. Being highly unstable by nature, they require relatively long segment instrumentation and fusion, which can increase paravertebral soft tissue damage and perioperative bleeding. The purpose of this report is to describe a rare case of traumatic double fractures at the cervico-thoracic and thoraco-lumbar transition zones in ankylosing spine with spondylo-epiphyseal dysplasia (SED) of unknown cause, which were successfully treated with a combined open and percutaneous spinal fusion procedure.

Case presentation: A 46-year-old woman who was diagnosed with non-contiguous fractures in cervico-thoracic and thoraco-lumbar junction zones among multiple injuries sustained in a traffic accident was treated with hybrid techniques for posterior instrumentation with an open approach using a computed tomography (CT)-based navigation system and percutaneous pedicle-screwing method. She regained mobility to pre-admission levels and started walking on crutches 3 months postoperatively. Genetic testing for the cause of SED revealed no mutation in the COL2A1 or TRPVR4 genes. The union of fractured spine was confirmed on CT scan 1 year postoperatively.

Conclusion: This is the first report of double spinal fractures in an ankylosing spine with genetically undetermined spondyloepiphyseal dysplasia. A long-segment posterior instrumentation procedure incorporating the invasive treatment of spinal fractures in ankylosing spondylitis or diffuse idiopathic hyperostosis was effective.

Keywords: Trauma, Spine, Spondylo-epiphyseal dysplasia, Ankylosing spine, Spinal fracture
Background
Ankylosing spondylitis (AS) or diffuse idiopathic skeletal hyperostosis (DISH) commonly accompanies ankylosing spines; spinal fractures frequently occur with minor trauma [1, 2] and are extremely unstable because of the long lever arms of the fused spinal column and several complications, including common early and late neurological symptoms [2]. Therefore, early surgery for neural decompression and spinal stability is recommended [3, 4]. A case with double fractures of an ankylosing spine with genetically undetermined spondyloepiphyseal dysplasia (SED) and spinal ankylosis was successfully treated with hybrid open and percutaneous spinal fusion.

Case presentation
A 46-year-old woman involved in a traffic accident was brought to our hospital. On arrival at the emergency room, her vital signs were stable and physical examination revealed no neurological deficit. Whole body computed tomography (CT) confirmed double spinal fractures at the cervicothoracic (C-T) and thoracolumbar (T-L) junctional zones in an ankylosing spine, with atlas to coccyx fusion, as well as traumatic hemopneumothorax and multiple rib fractures. A right thoracic curve with a Cobb angle of 15°, a sagittal kyphotic thoracic curve with a Cobb angle of 73°, sacral anteversion, and coccygeal retroversion were observed (Fig. 1). Magnetic resonance imaging of the spine revealed a mild dural sac indentation at the T-L junctional zone, but without spinal cord compression.

In infancy, the patient had progressive multiple joint contractures indicating arthrogryposis multiplex congenital, which was not genetically confirmed. Height and weight on admission were 130 cm and 40 kg, respectively. No visual or acoustic deficits were evident. Whole body roentgenograms revealed marked osteoarthritic changes in almost all joints and ankyloses in the knees and shoulders. She had no medical history of fractures and underwent left and right hip joint replacement surgeries at 32 and 39 years of age, respectively (Fig. 2). Respiratory distress from traumatic hemopneumothorax resolved within 8 days of hospitalization, and the patient subsequently underwent surgical posterior spinal fusion of both spinal fractures without bone grafting (Fig. 3).

The patient underwent tracheal intubation in the neutral cervical spine position using a monitor-integrated video laryngoscopy. She was then placed in the prone position taking care not to displace floating thoracic spine fragments. Spinal alignment in the prone position was checked using fluoroscopy and CT; a mattress was inserted between the body and four-post spine frame to equalize the space created by the thoracic spine kyphotic deformity, and C2–T4 and T9–L3 posterior fusion was performed. A C2 lamina screw (left side), pedicle screw (right side), cervical lateral mass screws (C3–C6), and thoracic pedicle screws (T2–T4) were inserted via an open approach. A C-arm three-dimensional navigation system (Brainlab Spinal Navigation, Munich, Germany) was used for placing C2 and thoracic pedicle screws; lateral mass screws were placed using a lateral fluoroscope. Contoured rods from C2 to T4 and a cross-link at the T1 level were placed. Pedicle screws were percutaneously placed from L1 to L3 using biplanar fluoroscopy.

Although identifying pedicle contours of T9–T11 was difficult because of spinal osteoporosis, screw placement was safely performed without a navigation system via the open approach because the diameters of the pedicles at these levels were large enough (> 6.0 mm on the preoperative CT...
scan). Pedicle screws were not placed along the left side of T9 because the pedicle was sclerosed and a rib fracture dislocation occurred while preparing the pilot hole. The 5.5-mm contoured rods were inserted through polyaxial heads of pedicle screws at the open thoracic incision and caudally passed subcutaneously through polyaxial heads of lumbar pedicle screws. Cross-links were placed between T12 and L1. Decompressive laminectomy or laminotomy was not performed because of the absence of cord compression. Surgical duration was 8 h, with approximately 1100 mL blood loss. The patient recovered well without postoperative complications and was discharged for rehabilitation 2 months postoperatively. She could walk and resume work, respectively, at 3 and 6 months postoperatively. One-year follow-up CT revealed a bony fusion of C-T and T-L fractures (Fig. 4). We suspected SED based on her medical history and whole body roentgenograms, but did not find COL2A1 and TRPV4 mutations; further genetic testing was not performed because no candidate gene for this phenotype was identified.

Discussion and conclusions
We report double noncontiguous spinal fractures in a patient with SED having spinal ankylosis. No similar cases have been reported, which necessitated adaptation of spinal fracture treatments in patients with AS or DISH.

1. Fracture characteristics
AS or DISH usually accompany ankylosing spines. Non-contiguous spinal fractures in patients with ankylosing spine, as in the present case, are not rare [1, 5]. Because of poor bone quality, the spine is brittle, osteoporotic, and stiff [4, 6, 7], increasing susceptibility to fracture even with low-energy trauma [8, 9]. Fracture pattern in patients with AS typically involves three columns [10], as in this case. Compared with previous reports, this study is unique because the fracture occurred in an ankylosing spine with SED. The cervical spinal fracture in our patient was observed at the C6–C7 segmental level, consistent with previous reports [1, 4, 11]. Spinal fractures in patients with AS frequently accompany hyperextension fractures [1, 11] and are classified into four patterns based on fracture excursion through intervertebral disc, vertebral body, or both [1]. However, coronal CT in our case demonstrated a gap between the left and right fracture lines at both cervical and thoracic vertebral bodies—a pattern not previously observed. Based on the accident and the difference of number of left and right rib fractures (4:1), we attribute the pattern of fracture lines to direct or indirect lateral external forces impacting the spine.

2. Etiology of ankylosing spine
Congenital SED is a rare form of skeletal systemic disease [12]. Previous studies have demonstrated that patients with congenital SED present with a variety of deformities in the spine, including instability of the atlanto-axial joint, progressive kyphoscoliosis, platyspondylitis, lordotic lumbar vertebrae, and pear-like shape to the corps vertebrae at the thoracolumbar interface [13, 14]. However, our patient had extremely rare features in the spine such as ankylosing spine and the absence of instability of the atlanto-axial joint with SED. To our knowledge, only two studies have reported ankylosing spines with SED [15, 16], wherein patients exhibited X-linked recessive inheritance and associated mutations of transport protein particle TRPPC2 [17], with onset later than congenital SED. This patient probably had another causative gene because of her sex and appearance of spinal ankylosing and multiple joint contractures in infancy. Because SED is a skeletal disorder mediated by COL2A1, we investigated for gene mutations [18] but found none. Moreover, no mutation of TRPV4, reportedly related to spondylo-metaphyseal dysplasia (Kozlowski type) and SED (Maroteaux type) [19], was observed. The genetic etiopathology in our patient remains unknown.
3. General problems treating ankylosing spinal fractures

Ankylosing spine creates high instability at fracture site because of long lever arms secondary to spinal column stiffness. Fractures frequently occur at the C-T and T-L transitional zones, which are subjected to extension force when patients are supine, resulting in delayed union [20, 21], epidural hemorrhage, and/or late-onset paralysis [22, 23]. Furthermore, a much higher mortality rate is reported in patients with cervical spinal fracture who have AS (> 30%) [1, 11] than in those without AS (18%) [11]. Therefore, early aggressive surgeries with posterior and/or anterior fixation are recommended. A disadvantage of treating this fracture type is the increase in the number of fused spinal segments (average, 5.6 segments) [4]; it is generally recommended to make multiple anchor points by extending the instrumentation over at least three vertebral levels above and below the fracture site [4, 24].

4. Specific devices used for treatment

Our patient had extensive thoracic kyphosis; therefore, we carefully monitored motor- and somatosensory-evoked potentials intraoperatively. To prevent intraoperative worsening of spinal alignment in the prone position, secondary to contact pressure disparities at the frontal truncal surface due to the rigid and kyphotic spine, we inserted cushioning materials into the space between the trunk and operating table. A CT-based navigation system was used for placing pedicle screws at C2 and the thoracic spine, considering the extreme difficulty in identifying anatomical landmarks for pedicle screw insertion. We made C2 anchors because we were concerned that cervical lateral mass screws had significantly lower resistance to pull-out forces than pedicle screws [25]. No new fracture was observed in our case at the thoracic spine without instrumentation between T4 and T9 partly due to her lower activity level secondary to ankyloses of all joints and spine. However, even with similar fracture patterns, non-skip cervicolumbar fusion would be favorable in patients with higher activity levels. Eventually, bony
fusion of three-column spinal fractures at C-T and T-L junctional zones using hybrid open and percutaneous spinal fusion techniques was successful, despite difficulties ensuing from SED and spinal ankyloses of idiopathic genetic etiopathogenesis.

Abbreviations
SED: spondylo-epiphyseal dysplasia; CT: computed tomography; AS: ankylosing spondylitis; DISH: diffuse idiopathic skeletal hyperostosis; CT: cervico-thoracic; T-L: thoraco-lumbar.

Authors’ contributions
TU, KK, and GK have been involved in the clinical management, data acquisition and interpretation of data of the patient. TM, TK, AI, SI and NH were involved in genetic testing for TRPV1 mutation. GN and MT performed genetic testing for COL2A1 mutation. TK, GN SI and NH were involved in image diagnosis of the patient. TU, KK and GK drafted the manuscript. All authors read and approved the final manuscript.

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Competing interests
The authors declare that they have no competing interests.

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References
1. Caron T, Bransford R, Nguyen Q, Agel J, Chapman J, Bellabarba C. Spine fractures in patients with ankylosing spinal disorders. Spine (Phila Pa 1976). 2010;35(11):E458–64.
2. Osgood C, Abbasy M, Mathews T. Multiple spine fractures in ankylosing spondylitis. J Trauma. 1975;15(2):163–6.
3. Broom MJ, Raycroft JF. Complications of fractures of the cervical spine in ankylosing spondylitis. Spine (Phila Pa 1976). 1988;13(7):763–6.
4. Kanter AS, Wang MY, Mummennari PV. A treatment algorithm for the management of cervical spine fractures and deformity in patients with ankylosing spondylitis. Neurosurg Focus. 2008;24(1):E11.
5. Olerud C, Frost A, Bring J. Spinal fractures in patients with ankylosing spondylitis. Eur Spine J. 1996;5:51–5.
6. Ball J. Enthesopathy of rheumatoid and ankylosing spondylitis. Ann Rheum Dis. 1971;30(3):213–23.
7. Carette S, Graham D, Little H, Rubenstein J, Rosen P. The natural disease course of ankylosing spondylitis. Arthritis Rheum. 1983;26(2):1186–90.
8. Einsiedel T, Schmelz A, Arand M, Wilke HJ, Gebhard F, Hartwig E, Kramer M, Neugebauer R, Kinzl L, Schultheiss M. Injuries of the cervical spine in patients with ankylosing spondylitis: experience at two trauma centers. J Neurosurg Spine. 2006;5(1):33–45.
9. Thumby K, Poller R, Trudinger B, Trudinger BL, Mathew KM. Spinal cord injury in patients with ankylosing spondylitis: a 10-year review. Spine (Phila Pa 1976). 2007;32(26):2989–95.
10. El Tecle NE, Abode-Iyamah KO, Hitchen PW, Dzahdaleh NS. Management of spinal fractures in patients with ankylosing spondylitis. Clin Neurol Neurosurg. 2015;139:177–82.
11. Murray GC, Persellin RH. Cervical fracture complicating ankylosing spondylitis: a report of eight cases and review of the literature. Am J Med. 1981;70(5):1033–41.
12. Spranger JW, Wiedemann HR. Dysplasia spondyloepiphyseal tarda congeinta. Helv Paediatr Acta. 1966;21:598–611.
13. Hensinger RN, Even GDM. Congenital anomalies of the spine. In: Rothman RH, Simoone FA, editors. The spine. 2nd ed. Philadelphia: WB Saunders; 1982. p. 299–304.
14. Tolo VT. Spinal deformity in skeletal dysplasia. In: Weinstein SL, editor. The pediatric spine, vol. 1. New York: Raven Press, 1994. p. 369–96.
15. Bos J, Rogge CW. A family with late spondyloepiphyseal dysplasia. Ned Tijdschr Geneesk. 1974;118(16):576–81.
16. Job-Deslandre C, Menkles CJ. Spondylo-epiphyseal dysplasia with ankylosing development: Apropos of a case. Rev Rheum Mal Osteoartic. 1991;58(9):635–6.
17. Savarirayan R, Thompson E, Gécz J. Spondyloepiphyseal dysplasia tarda (SEDL, MIM #313400). Eur J Hum Genet. 2003;11(9):639–42.
18. Spranger J, Winterpacht A, Zabel B. The type II collagenopathies: a spectrum of chondrodysplasias. Eur J Pediatr. 1994;153(2):66–71.
19. Nishimura G, Lautsch E, Savarirayan R, Shiba M, Spranger J, Zabel B, Ikegawa S, Superti-Furga A, Unger S. TRPV4-associated skeletal dysplasias. Am J Med Genet C Semin Med Genet. 2012;160C(3):190–204.
20. Deburbe A, Guigui P, Ouahes M, Barre E. Cervical pseudoarthrosis in ankylosing spondylitis. A case report. Neurosurg Spine. 2006;5(1):33–45.
21. Osterman G, Mandl J, Glintborg B, Riise T, Skovbro J, Ringsted K. Spinal cord injury in patients with ankylosing spondylitis: a 10-year review. Spine (Phila Pa 1976). 2007;32(26):2989–95.
22. El-Tecle NE, Abode-Iyamah KO, Hitchen PW, Dzahdaleh NS. Management of spinal fractures in patients with ankylosing spondylitis. Clin Neurol Neurosurg. 2015;139:177–82.
23. Murray GC, Persellin RH. Cervical fracture complicating ankylosing spondylitis: a report of eight cases and review of the literature. Am J Med. 1981;70(5):1033–41.
24. Spranger JW, Wiedemann HR. Dysplasia spondyloepiphyseal tarda congeinta. Helv Paediatr Acta. 1966;21:598–611.
25. Hensinger RN, Even GDM. Congenital anomalies of the spine. In: Rothman RH, Simoone FA, editors. The spine. 2nd ed. Philadelphia: WB Saunders; 1982. p. 299–304.
26. Tolo VT. Spinal deformity in skeletal dysplasia. In: Weinstein SL, editor. The pediatric spine, vol. 1. New York: Raven Press, 1994. p. 369–96.
27. Bos J, Rogge CW. A family with late spondyloepiphyseal dysplasia. Ned Tijdschr Geneesk. 1974;118(16):576–81.
28. Job-Deslandre C, Menkles CJ. Spondylo-epiphyseal dysplasia with ankylosing development: Apropos of a case. Rev Rheum Mal Osteoartic. 1991;58(9):635–6.
29. Savarirayan R, Thompson E, Gécz J. Spondyloepiphyseal dysplasia tarda (SEDL, MIM #313400). Eur J Hum Genet. 2003;11(9):639–42.
30. Spranger J, Winterpacht A, Zabel B. The type II collagenopathies: a spectrum of chondrodysplasias. Eur J Pediatr. 1994;153(2):66–71.
31. Nishimura G, Lautsch E, Savarirayan R, Shiba M, Spranger J, Zabel B, Ikegawa S, Superti-Furga A, Unger S. TRPV4-associated skeletal dysplasias. Am J Med Genet C Semin Med Genet. 2012;160C(3):190–204.
32. Deburbe A, Guigui P, Ouahes M, Barre E. Cervical pseudoarthrosis in ankylosing spondylitis. A case report. Neurosurg Spine. 2006;5(1):33–45.
33. Osterman G, Mandl J, Glintborg B, Riise T, Skovbro J, Ringsted K. Spinal cord injury in patients with ankylosing spondylitis: a 10-year review. Spine (Phila Pa 1976). 2007;32(26):2989–95.