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Proximal junctional kyphosis after posterior spinal fusion for severe kyphoscoliosis in a patient with PIEZO2-deficient arthrogryposis syndrome

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ABSTRACT

Study Design: Case report

Objective: Describe the clinical and radiological outcomes of a patient with a piezo-type mechanosensitive ion channel component 2 (PIEZO2)-deficient arthrogryposis receiving surgery for severe kyphoscoliosis.

Summary of Background Data: Spinal deformity is a characteristic feature of arthrogryposis due to PIEZO2 gene deficiency, for which surgical correction is indicated when the deformity is progressive to avoid neurological deficits and respiratory impairment. However, there exist few reports on the surgical treatment of spinal deformity in PIEZO2-deficient arthrogryposis, and no therapeutic standards have been established.

Methods: We retrospectively reviewed a case of proximal junctional kyphosis after posterior spinal fusion for severe kyphoscoliosis in PIEZO2-deficient arthrogryposis.

Results: The patient was a 13-year-old girl with PIEZO2-deficient arthrogryposis who underwent posterior spinal fusion with an all-pedicle screw construct from T2 to L2 for a preoperative main thoracic curve Cobb angle of 78 degrees and thoracic kyphotic angle of 83 degrees. Postoperative Cobb angle of the main thoracic curve and thoracic kyphotic angle were improved at 11 and 34 degrees, respectively. Although revision surgery was required for neurological deficits from proximal junctional kyphosis, she could walk with a crutch and improvements in clinical questionnaire scores were noted at two years and three months after surgery.

Conclusions: Based on the present case, posterior spinal fusion represents a good treatment option for severe spinal deformity in PIEZO2-deficient arthrogryposis. Careful consideration of fusion level is needed to prevent proximal junctional kyphosis.

Key Words: Posterior spinal fusion; PIEZO2; arthrogryposis; kyphoscoliosis; radiological findings; proximal junctional kyphosis

Level of Evidence: 5

Key Points

●Spinal deformity is a characteristic feature of arthrogryposis due to a piezo-type mechanosensitive ion channel component 2 (PIEZO2) gene deficiency.

●We retrospectively reviewed a case of proximal junctional kyphosis after posterior spinal fusion for severe kyphoscoliosis in PIEZO2-deficient arthrogryposis.

●Posterior spinal fusion represents a good treatment option for severe spinal deformity in PIEZO2-deficient arthrogryposis, but careful consideration of fusion level is needed to prevent proximal junctional kyphosis.

Introduction

Arthrogryposis multiplex congenita (AMC) is a disease characterized by the contracture of two or more joints from birth that displays decreased muscle tone and muscle weakness [1].

A deficiency of the piezo-type mechanosensitive ion channel component 2 (PIEZO2) gene, which is expressed in dorsal root ganglion neurons and sensory endings, has been implicated as a cause of arthrogryposis syndrome [2-5]. Several reports have described severe scoliosis accompaniment of PIEZO2-deficient arthrogryposis [6-9]. However, none provided precise details on the sagittal deformity, which could influence activities of daily living, or spinal measurements such as Cobb angle.

Yamaguchi et al. have already reported on the overall clinical characteristics and course of the present PIEZO-deficient arthrogryposis patient [10]. To date, few studies exist on the surgical correction of the spinal deformity in PIEZO2-deficient arthrogryposis, and no treatment standards have been established. We herein describe the clinical and radiological outcomes of a patient with PIEZO2-deficient arthrogryposis who was surgically treated for severe kyphoscoliosis.

Case presentation

A 13-year-old girl was diagnosed as having PIEZO2-deficient arthrogryposis immediately after birth. She had bilateral foot deformities and had been walking with the aid of crutches since childhood. Scoliosis was first noted at roughly four years of age, and she exhibited severe scoliosis and gait disturbance at age 12. Her height was 131 cm, weight was 30 kg, and body mass index was 17.5 kg/m^2 at consultation. Her lumbar and total hip bone mineral densities were low at 0.815 g/cm^2 and 0.722 g/cm^2, respectively. Preoperative

Scoliosis Research Society-22 domain scores were 4.4, 4.8, 2.4, 4.0, and 3.9 for function, pain, self-image, mental health, and subtotal, respectively. We performed posterior spinal fusion by means of an all-pedicle screw construct from T2 to L2 for her preoperative main thoracic (MT) curve (T3-11) Cobb angle of 78 degrees and thoracic kyphotic angle (T1-12) of 83 degrees (Figure 1a and Figure 2). No obvious vertebral abnormalities were detected in the affected segment. Ponte osteotomy was not performed. Surgical time was 307 minutes and blood loss was 650 g. Her postoperative Cobb angle of the MT curve improved to 11 degrees and thoracic kyphotic angle was ameliorated at 34 degrees (Figure 1b and Figure 3).

Three months after surgery, proximal junctional kyphosis (PJK) (70 degrees) was detected in the absence of neurological deficits. Seven months after the operation, she began to exhibit progressive weakness in the lower extremities and gait was impossible. CT and MRI revealed spinal cord compression from increased thoracic kyphosis at the proximal adjacent fusion level (Figure 3 and Figure 4). Additional posterior spinal fusion was performed successfully from C6 to T2 (Figure 5). Surgical time was 238 minutes and blood loss was 100 g.

Two months after revision surgery, she could walk with a crutch. Her lower extremity weakness was fully improved at one year postoperatively. CT confirmed bone union nine months after surgery (Figure 6). Post-surgical Scoliosis Research Society-22 domain scores were 4.6, 5.0, 4.2, 4.2, 4.5, 3.5, and 4.4 for function, pain, self-image, mental health, subtotal, satisfaction, and total, respectively, at two years and three months after surgery. At that time, she had no complaints and reported improvements in posture, appetite, weight, and breathing. No radiological deterioration was noted. Her visual analogue scale result for low back pain was 0 (no pain). She could resume walking with crutches at two years and three months after surgery.

Discussion

In the present case of a patient with PIEZO2-deficient arthrogryposis receiving posterior spinal fusion for severe kyphoscoliosis, pedicle screw fixation remarkably improved radiological findings and achieved good clinical results without severe perioperative complications.

There are currently four searchable reports on PIEZO2-deficient arthrogryposis, in which 15 of 16 (93.8%) patients were accompanied with severe scoliosis [6-9]. However, no detailed descriptions on the sagittal deformity or spinal measurement results including Cobb angle were included and discussion on the spinal deformity was limited. Thus, we have employed previous literature on overall AMC patients for a basic clinical picture of PIEZO2-deficient arthrogryposis.

Roughly 2.5-69.6% of AMC patients are complicated with scoliosis [11-14]. Such cases are generally progressive, and orthodontic treatment is ineffective [1,13,14]. Accordingly, surgical correction of the spinal deformity in AMC is indicated to prevent neurological deficits and respiratory impairment. Spinal instrumentation in AMC is challenging due to the risk of insufficient correction and respiratory complications. Since the jaw is small and difficult to move, tracheal intubation and respiratory problems are a concern throughout the perioperative period. The scoliosis in AMC patients is often rigid and hard to correct, with reported correction rates of 32-52% [1,12-14]. In our case, the correction rates of the MT curve and thoracic kyphosis were 85.9% and 59.0%, respectively, without respiratory complications.

In AMC patients with a thoracic curve pattern, the selection of fusion levels and instrumentation is similar to that of idiopathic scoliosis [6]. We performed posterior correction fusion from T2 to L2 according to the indications for idiopathic scoliosis and

achieved adequate correction. However, three months after surgery, severe PJK occurred with progressive lower extremity muscle weakness. We had wanted to correct the patient's local kyphosis and improve her sagittal vertical axis, but in situ fusion was ultimately performed because of her rigid kyphotic deformity and low bone mineral density for additional posterior spinal fusion. Moreover, active correction was avoided out of concern for postoperative paralysis, although additional correction could have been gained with osteotomies plus cemented screws or osteotomies with or without anterior release.

The prevalence of PJK in AIS patients is 10-27%, with multivariate analysis excluding the level of the upper instrumented vertebra (UIV) as a significant risk factor [16,17]. On the other hand, PJK prevalence was 28% in a recent study of growing rod treatment for early-onset scoliosis, in which a UIV distal to T2 was identified as an independent risk factor [18]. We initially sought to preserve the T1-2 interspinous ligament during the first surgery, although PJK developed soon afterwards. Such an outcome might have been avoided if we had selected T1 as the UIV.

Conclusion

Based on the present case, posterior spinal fusion represents a good correction option for severe spinal deformity in PIEZO2-deficient arthrogryposis. Fusion level should be decided carefully to prevent PJK.

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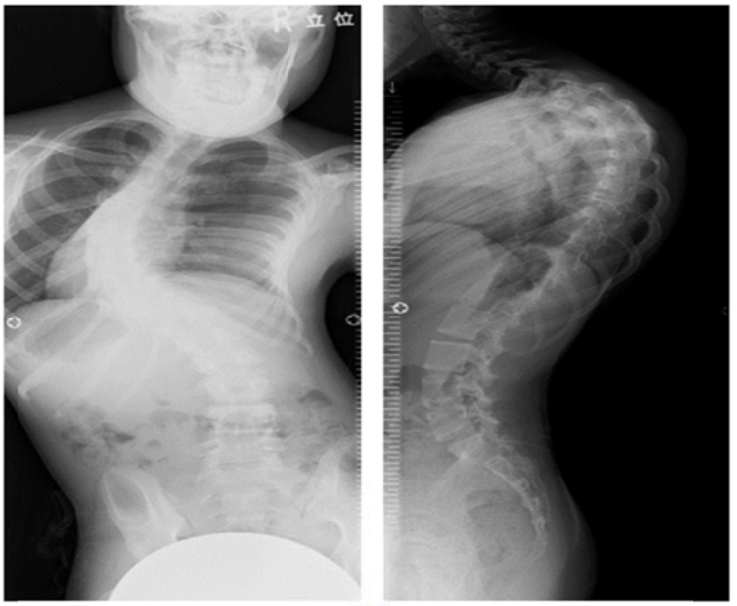
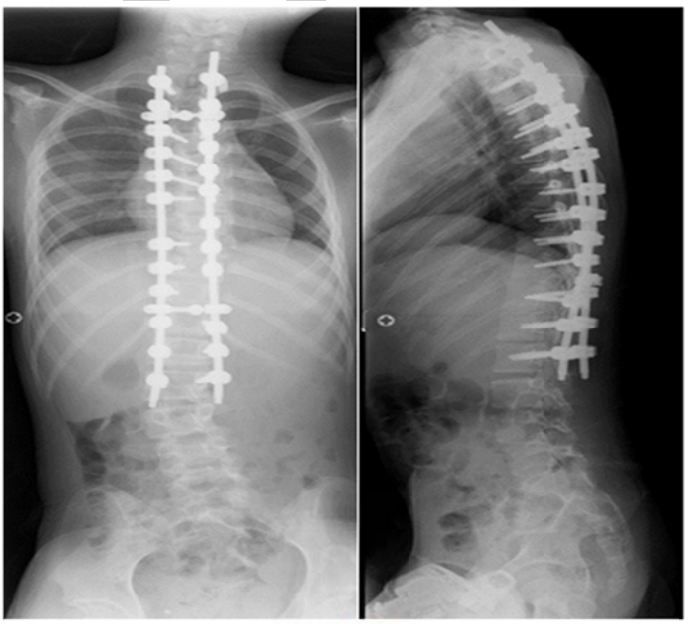
Figure 1. Case: a 13-year-old girl.

(a) Preoperative Cobb angle of the main thoracic curve was 78 degrees and thoracic kyphosis was 83 degrees. (b) We performed posterior spinal fusion with a pedicle screw construct from T2 to L2. Postoperative Cobb angle of the main thoracic curve improved to 11 degrees and thoracic kyphotic angle improved to 34 degrees.

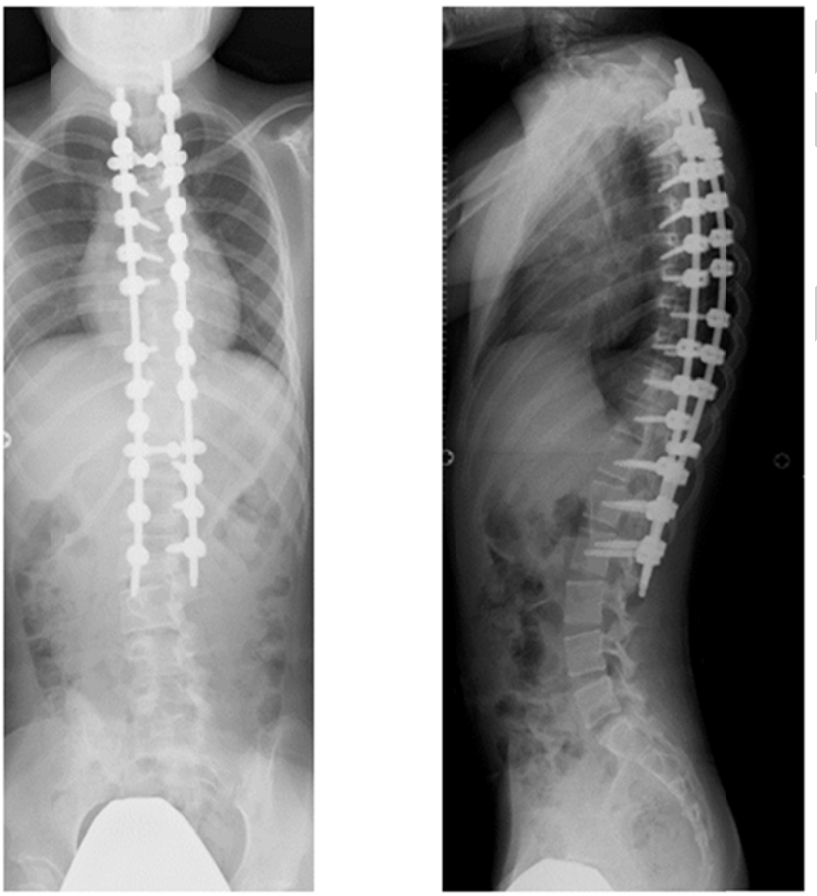


(a)



(b)

Figure 2. Postoperative radiographs showed proximal junctional kyphosis three months after surgery.



Radiograph taken at 3 months postoperatively. T5-11 Cobb angle: 12 degrees; T11-L3 Cobb angle: 11 degrees; T1-12 kyphotic angle: 71 degrees (hyperkyphosis); T12-S1 kyphotic angle: -60 degrees.

Figure 3. Postoperative CT demonstrated severe proximal junctional kyphosis.

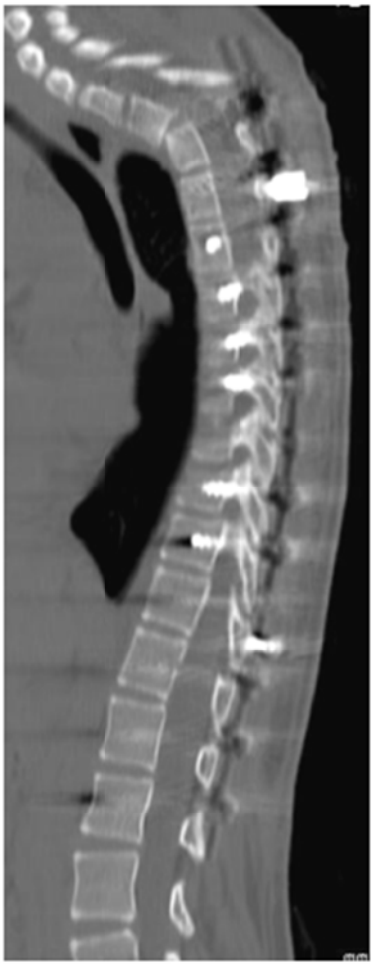


Figure 4. MRI revealed spinal cord compression from increased thoracic kyphosis at the proximal adjacent fusion level.

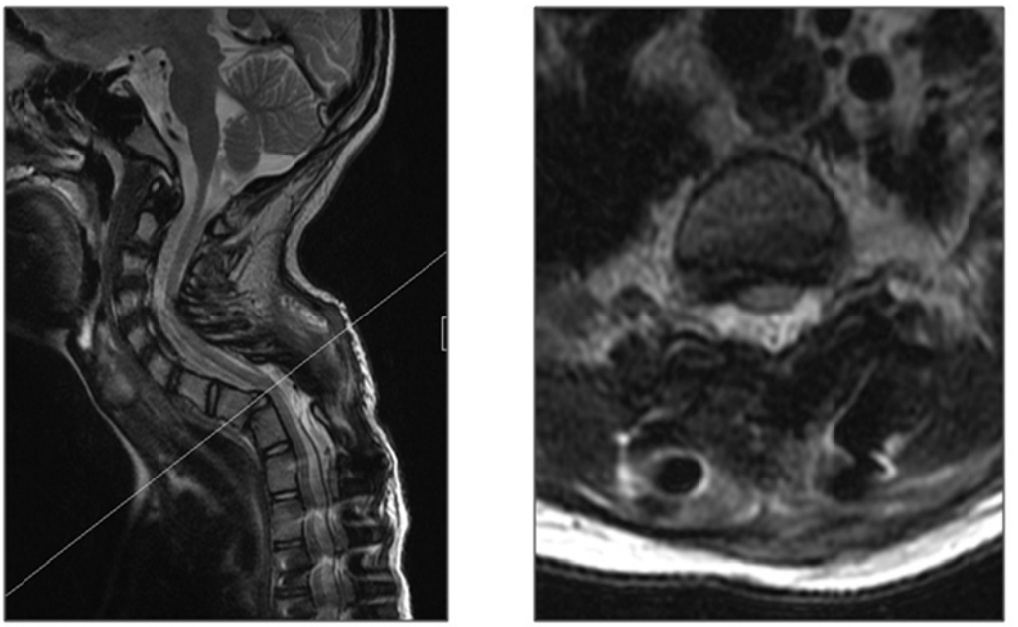
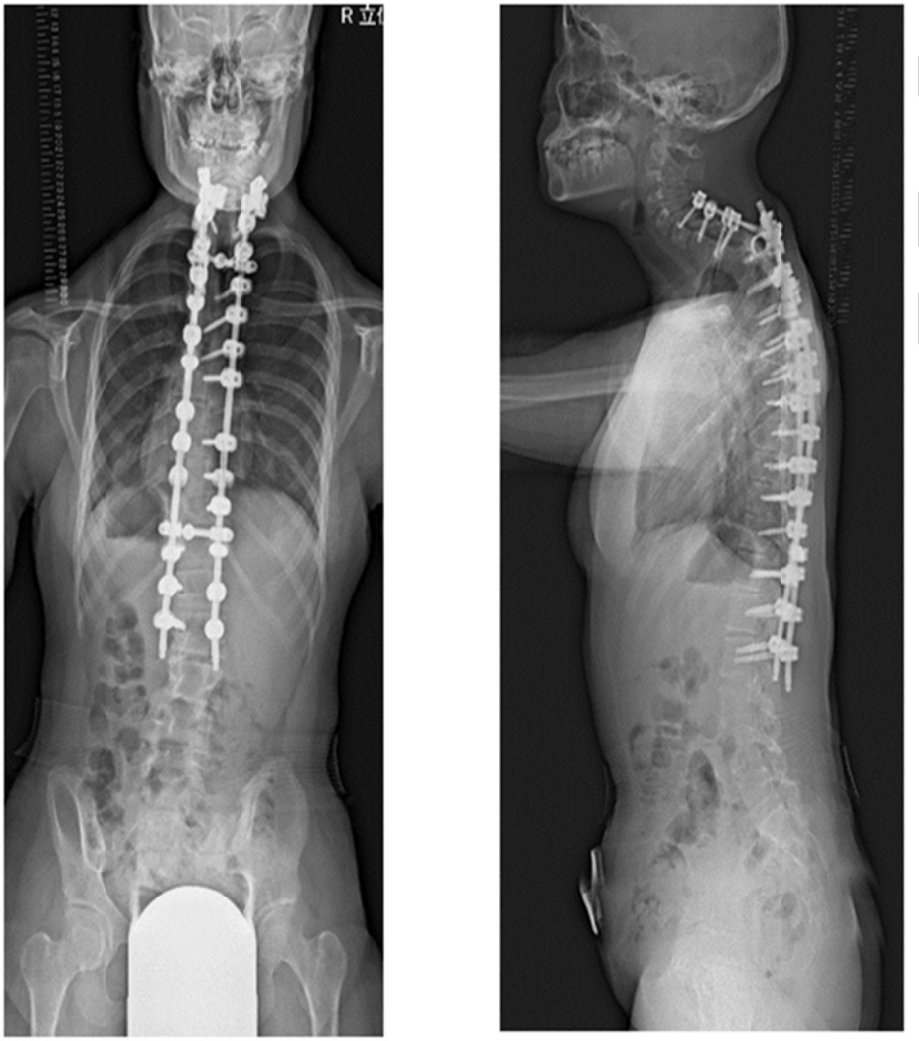


Figure 5. Additional posterior fusion from C6 to T2 was performed.



Radiograph taken at 3 months postoperatively. T5-11 Cobb angle: 12 degrees; T11-L3 Cobb angle: 11 degrees; T1-12 kyphotic angle: 71 degrees (hyperkyphosis); T12-S1 kyphotic angle: -60 degrees.

Figure 6. Postoperative computed tomography showed achievement of bone union at nine months after surgery.