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## **Surgical Treatment of Atlantoaxial Dysplasia and Scoliosis in Spondyloepiphyseal Dysplasia Congenita: A Case Report**

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### **Abstract**

#### **BACKGROUND**

Spondyloepiphyseal dysplasia congenita (SEDC) is a rare autosomal dominant hereditary disease caused by COL2A1 mutations. SEDC primarily involves the skeletal system, with typical clinical manifestations, including short stature, hip dysplasia, and spinal deformity. Due to the low incidence of SEDC, there are only a few case reports regarding the surgical treatment of SEDC complicated with spinal deformities.

#### **CASE SUMMARY**

We report a case of a 16-year-old male patient with SEDC. He presented with typical short stature, atlantoaxial dysplasia, scoliosis, and hip dysplasia. Cervical magnetic resonance imaging showed spinal canal stenosis at the atlas level and cervical spinal cord compression with myelopathy. The scoliosis was a right thoracic curve with a Cobb angle of 65°. He underwent atlantoaxial reduction, decompression, and internal fixation from C1-C2 to relieve cervical myelopathy. Three months after cervical surgery, posterior correction surgery for scoliosis was performed from T3 to L4. Scoliosis was corrected from 66° to 8° and remained stable at 2-year follow-up.

#### **CONCLUSION**

This is the first case report of a patient with SEDC who successfully underwent surgery for atlantoaxial dysplasia and scoliosis. The study provides an important reference for the surgical treatment of SEDC complicated with spinal deformities.

## **INTRODUCTION**

Spondyloepiphyseal dysplasia congenita (SEDC) is a rare autosomal dominant genetic disorder with an incidence of approximately 3/1000000 [1]. SEDC affects bone development, particularly of the spine and the long bones of the limbs [2]. SEDC is caused by mutations in the COL2A1 gene, which regulates the synthesis of type II collagen, a major component of cartilage and other types of connective tissues [3]. Mutations in COL2A1 disrupt the normal production and assembly of type II collagen, which lead to abnormal bone growth and development [3,4]. The symptoms of SEDC vary widely, including short stature, atlantoaxial dysplasia, abnormal curvature of the spine (scoliosis or kyphosis), hip dysplasia, joint pain, early onset arthritis, and vision disorders [5]. Some individuals with SEDC may also have cleft palate or other craniofacial abnormalities [5]. There is currently no cure for SEDC, but surgical treatment for skeletal deformities can help manage symptoms and improve quality of life [6]. Although a few cases of atlantoaxial and hip dysplasia have been reported, surgical treatment of SEDC with scoliosis is very rare [7-10]. Herein, we report the case of a patient with SEDC who successfully underwent surgery for atlantoaxial dysplasia and scoliosis.

## **CASE PRESENTATION**

### ***Chief complaints***

A 16-year-old male patient presented with a 5-year history of progressive scoliosis.

### ***History of present illness***

At the age of 11, the patient was diagnosed with scoliosis and was treated with a brace for more than 12 h daily and had regular check-ups. However, his scoliosis continued to deteriorate.

### *History of past illness*

The patient experienced difficulty walking and had challenges with physical activities since the age of 1 year. His growth and development were delayed compared to those of his peers. Radiography of the hip joints revealed bilateral hip dysplasia.

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### *Personal and family history*

The patient denied any family history of similar diseases or malignant tumors.

### *Physical examination*

Physical examination revealed that the patient was 128 cm in height, weighed 46 kg, and was unable to ambulate steadily. There was a right-sided thoracic spine curvature, resulting in a "razorback" deformity. The patient exhibited decreased proximal muscle strength in both lower limbs (grade IV), and bilateral Hoffman signs and Babinski signs were positive.

### *Laboratory examinations*

Ventilation function, electrocardiogram, echocardiogram, and other preoperative examination results were normal. Blood calcium, phosphorus, vitamin D, and alkaline phosphatase levels were normal for bone metabolism.

### *Imaging examinations*

During the initial outpatient examination, the spine X-ray indicated a leftward proximal thoracic curve of 32° and a rightward main thoracic curve of 65° (Figure 1A). Three months following atlantoaxial decompression and fixation surgery, and prior to scoliosis correction, the spine X-rays revealed the proximal thoracic curve was 36°, the

main thoracic curve was 65°, the thoracic kyphosis was 7°, and the sagittal vertical axis (SVA) was 12.5 cm (Figure 1B-C). On the bending X-rays, the Cobb angle of the proximal thoracic curve was 33° and the main thoracic curve was 46° (Figure 1D-E). Computed tomography (CT) three-dimensional (3D) reconstruction of the spine showed flattening of the vertebral body and widespread narrowing of the intervertebral space (Figure 2A-C). 3D CT of the cervical spine showed dysplasia of the odontoid process of the axis and the anterior and posterior arches of the atlas (Figure 3A-B). Spinal CT showed shorter pedicle screw lengths in the thoracic and lumbar vertebrae (2.94–5.17 cm), but the cancellous bone of the pedicles was well developed. A hip X-ray revealed dysplasia of bilateral hip joints and severe damage to the femoral heads. No obvious abnormality was found on the X-ray of bilateral knee joints and the full-length of both lower limbs (Figure 3C-E). Magnetic resonance imaging (MRI) of the spine indicated stenosis of the cervical spinal canal in the atlantoaxial region and myelopathy of the cervical spine (Figure 4C). The spinal MRI did not reveal other intraspinal abnormalities. The appearance of the patient before scoliosis surgery is shown in Figure 2D-E.

### **FINAL DIAGNOSIS**

Combined with the patient's medical history and gene detection results, a diagnosis of SEDC was made.

### **TREATMENT**

To avoid irreversible damage caused by cervical myelopathy, the patient was recommended to have atlantoaxial decompression and fixation surgery. Skull traction was implemented under anesthesia during the surgical procedure, and the atlantoaxial reduction was observed to be satisfactory. Consequently, one-stage posterior atlantoaxial reduction and fixation procedure was employed. During the procedure, lateral mass screws were placed on both sides of the atlas and pedicle screws were placed on both sides of the axis and fixed with connecting plates, and autogenous iliac

cancellous bone were used for bone graft fusion. Three months after the operation, the patient underwent posterior spinal fusion, internal fixation, and bone graft fusion from T3 to L4 to treat his deteriorating scoliosis. During the surgery, Smith–Petersen osteotomies were performed within the fusion range to fully release the spine, and pedicle screws were inserted between T3 and L4. Autogenous combined with allogeneic bone particles were used for Moe bone grafting. Intraoperative blood loss was 450 mL, and the duration of surgery was 3.6 h. No abnormality was identified during intraoperative spinal cord monitoring.

### **OUTCOME AND FOLLOW-UP**

The patient was successfully extubated and recovered smoothly after both surgeries. After the atlantoaxial decompression and fixation surgery, the compression of the cervical spinal cord was significantly relieved, and the instability of the atlantoaxial joint was improved (Figure 4). Before the corrective surgery of scoliosis, the patient's muscle strength of bilateral proximal lower limbs was significantly improved to grade V. The Hoffman signs and Babinski signs were negative. One week after corrective surgery for scoliosis, the spinal X-ray revealed significant correction of scoliosis with the main thoracic curve corrected from 65° to 10°, thoracic kyphosis corrected from 7° to 26°, SVA corrected from 12.5 cm to 3.2 cm, and considerable improvement in the back's unevenness (Figure 6A–D). At the postoperative 2-year follow-up, the patient reported no symptoms of discomfort, such as back pain and weakness of the lower extremities, the atlantoaxial joint reduction remained effective and spinal correction retained its stability (Figure 6E–H).

### **DISCUSSION**

SEDC is a rare genetic disorder that affects skeletal growth and development. Diagnosis of SEDC involves a combination of clinical examinations, radiographic imaging, and genetic testing [1,11]. The radiographic findings typically include flattened vertebrae, shortened long bones, and hip dysplasia. Due to the low incidence of SEDC and its

main clinical feature of short stature, it can be easily confused with relatively common skeletal dysplasia that also cause short stature, such as osteogenesis imperfecta, multiple epiphyseal dysplasia, and achondroplasia [12-14]. In addition, mutations in COL2A1 can cause various skeletal dysplasia, such as Kniest and Stickler dysplasia, which can also be easily confused with SEDC [15]. Therefore, it is sometimes difficult to distinguish SEDC from these diseases based only on clinical examination and imaging findings. However, genetic testing can confirm the diagnosis and identify the specific genetic mutation responsible for the disorder. Consequently, genetic testing has become an important tool for the diagnosis of SEDC. Barat-Houari *et al.* analyzed the phenotypes and genetic diagnoses of over 700 patients with type II collagenopathy and identified 415 different COL2A1 mutations [15]. Our patient presented with skeletal dysplasia in infancy; however, owing to the lack of genetic testing at that time, the diagnosis was not confirmed until the patient was 16 years of age. His genetic testing revealed a COL2A1 heterozygous mutation c. 2965C>T (p. Arg989Cys), which is the most common mutation associated with SEDC [15]. The spectrum of gene mutations in SEDC will be further expanded and may guide the treatment of SEDC [16]. Genetic testing should be recommended for patients with clinical and radiological evidence suggestive of SEDC. Early diagnosis is crucial for providing appropriate follow-up treatment recommendations and genetic counselling for families affected by SEDC.

Currently, there are no specific therapeutic interventions for COL2A1 mutations, and treatments primarily focus on managing skeletal deformities. Surgical treatment for SEDC aims to correct deformities and prevent complications, such as joint pain, stiffness, and spinal cord compression. Treating hip dysplasia in SEDC can be challenging owing to the severe deformity and small volume of femoral heads, and often concomitant knee and spinal deformities. However, developmental dysplasia of the hip is an important factor that affects the walking ability of patients with SEDC. Shetty *et al.* performed bilateral valgus-extension osteotomy with hybrid external fixation in eight adolescent patients with SEDC who were followed up for 2 years and found significant improvements in the Harris hip score and postoperative mobility [17].



Bayhan *et al.* reported that in children with SEDC, correcting the hip dysplasia by valgus hip osteotomy can maintain satisfactory walking function during 5 years of follow-up and improve hip mobility <sup>[10]</sup>. Bisht *et al.* used CT to reconstruct the 3D structure of the hip joint and visualize the anatomical structure of the femoral head and neck, which was helpful for selecting implant sizes <sup>[18]</sup>. Scheduled hip surgery can improve walking ability and delay the progression of hip joint osteoarthritis. However, our patient missed the opportunity for timely hip osteotomy surgery due to a lack of early diagnosis of SEDC. Even though the patient had severe bilateral hip joint damage, the presence of neurological damage due to atlantoaxial dysplasia makes it crucial to prioritize saving neurological function. Additionally, it was observed that the Cobb angle of scoliosis was still increasing three months after atlantoaxial decompression and fixation surgery. To avoid the potential need for more extensive surgery if scoliosis worsens, the decision was made to undergo scoliosis correction surgery before hip replacement surgery.

Among the various skeletal abnormalities associated with SEDC, atlantoaxial dysplasia is common and poses a serious threat to the patient's health. Liu *et al.* reported a case of SEDC complicated with atlantoaxial instability, spinal cord compression at the cranio-cervical junction, and quadriplegia. The patient underwent decompression of the cranio-cervical region and occipital-C4 fusion surgery, which improved neurological function after the operation <sup>[19]</sup>. Serhan *et al.* performed upper cervical spine fusion with autogenous iliac crest bone grafting in 20 children clinically diagnosed with SEDC who were followed up for 8 years and reported that the instability of the upper cervical spine was significantly improved, and the bone graft fusion remained stable without any non-union <sup>[20]</sup>. Miyoshi *et al.* analyzed seven patients with SEDC complicated with atlantoaxial subluxation who underwent reduction and fusion surgery and found that when the diameter of the atlantoaxial spinal canal was less than 10 mm, atlantoaxial plate removal and occipital-cervical fusion were recommended <sup>[9]</sup>. Kaissi *et al.* retrospectively analyzed 10 patients with SEDC and found that orthopedic treatment should begin with cervical spine evaluation to avoid severe neurological dysfunction or



death [21]. Multiple skeletal abnormalities were detected during the initial examination of our patient. However, atlantoaxial dysplasia exerted the greatest impact on the patient's quality of life. Therefore, atlantoaxial decompression and fixation surgery were recommended, resulting in improved muscle strength and the prevention of further spinal cord damage. From our perspective, surgery for atlantoaxial deformity should be performed promptly if atlantoaxial instability or cervical spinal cord compression is found.

Reports of successful scoliosis correction surgery in patients with SEDC are rare. Beighton *et al.* reported two cases of SEDC combined with scoliosis, both of which underwent posterior spinal fusion with Harrington instrumentation; however, the surgeries were unsuccessful [22]. Winter *et al.* performed posterior spinal fusion with Harrington instrumentation in two patients with SEDC; during the follow-up period, one patient maintained good correction for 7 years, while the other experienced internal fixation failure due to pseudoarthrosis [23]. Morita *et al.* used Luque rods to treat severe thoracolumbar kyphoscoliosis in a patient with SEDC. Although the correction was stable for 6 years postoperatively, scoliosis and kyphosis correction rates were only 10.5% and 25.9%, respectively [7]. With the advancement of pedicle screws, posterior spinal fusion is believed to achieve a good corrective effect [24]. In this study, a high scoliosis correction rate of 84.6% was achieved through one-stage posterior spinal fusion, and there was no internal fixation failure or loss of correction during the 2-year follow-up. The high correction rate may be due to the use of a pedicle screw. In addition, the appropriate timing of treatment is also an important factor, as our patient's preoperative curve angle was 65°, which was smaller than in the five previously reported cases [7,22,23]. Furthermore, the preoperative spinal CT revealed that although the vertebral body in SEDC patients is small and flat, the cancellous bone of the pedicles developed well, suggesting that pedicle screw technology is suitable for scoliosis correction in SEDC. The selection of the upper and lower instrumented vertebrae (UIV and LIV) was also a crucial aspect of the pre-operative planning. Before surgery, the patient had balanced shoulders. The bending X-ray revealed that the proximal thoracic

curve exceeded 25°, indicating the necessity for fusion [25]. Consequently, T3 was chosen as the UIV. As for the LIV, the center sacral vertical line (CSVL) passed between the pedicles of L4 in the bending X-ray. However, CSVL did not touch the pedicle of L3, so we opted to select L4 as the LIV. Our case suggests that a reasonable surgical plan, in combination with early treatment of scoliosis can achieve good results.

### **CONCLUSION**

This is the first case report of a patient with SEDC who successfully underwent surgeries for atlantoaxial dysplasia and scoliosis. Early diagnosis of SEDC is important for providing appropriate treatment recommendations, and genetic testing is useful in confirming the diagnosis. Orthopedic evaluation of SEDC should begin with the cervical spine, as atlantoaxial dysplasia can be particularly detrimental. Early surgical intervention for hip dysplasia and scoliosis can result in favorable treatment outcomes. The use of pedicle screws may enhance the efficacy of scoliosis correction in patients with SEDC.

### **ACKNOWLEDGEMENTS**

Thank the patient and his guardian for their participation.

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