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Rigid occipitocervical instrumented fusion for atlantoaxial instability in a 18-month-old toddler with brachytelephalangic chondrodysplasia punctata: a case report

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Abstract

Study Design. Case report

Objective. We report here on an 18-month-old boy with brachytelephalangic chondrodysplasia punctata(BCDP), whose atlantoaxial instabilitywas successfully managed with occipitocervical instrumented fusion(OCF) using screw and rod instrumentations.

Summary of Background Data.Recently, there have been a number of reports on BCDP with early onset of cervical myelopathy. Surgical OCF is a vital intervention to salvage affected individualsfrom the life-threatening morbidity. Despite recent advancement of instrumentation techniques, however, rigid OCF is technically demanding in very young children with small and fragile osseous elements. To our best knowledge, this is the first report on application of the instrumentation technique to a toddler patient withBCDP.

Methods. A 16-month-old boy with BCDP presented with tetraplegia and swallow obstacle. Hypoplasia of the odontoid process and atlantoaxial instability were present in lateral radiographs. T2 weighted MR images revealed a high signal region in the spinal cord at the C1-2 and C7-T1 levels. Cervical CT showed the pedicles and lateral masses in the cervical spine were small and immature but the laminae were comparatively thick.

Results. One week prior to surgery, the patient was fitted with a Halo-body jacket. We performed plate-rod placement with occipital cortical screws and C2/C3 interlaminar screws, and added an autogenous bone graft using the right 8 and 9 ribs. Rigid fixation of the occipito-cervical spine was completed successfully without major complications. Postoperative halo-body jacket immobilization was continued for 3 months, after which Aspen collar was fitted. CT confirmed occipitocervical bone fusion at 6 months after surgery. Mild clinical improvements in motor power of the affected muscles and swallowing were witnessed at one yearpostoperatively.

Conclusions. Rigid fixation using screw, rod, and occipital plate instrumentation was successful in a 18-month-old toddler with BCDP and atlantoaxial instability. Bone fusion was achieved atpostoperative 6 months.

Key Words: brachytelephalangic chondrodysplasia punctata; toddler; instrumented fusion; atlantoaxial instability; rigid fixation; occipitocervical fusion; Halo-body jacket; interlaminar screw

Level of Evidence:5

Introduction

Brachytelephalangic chondrodysplasia punctata (BCDP) constitutes a large subgroup of CDP as heterogeneous skeletal dysplasias with punctate calcifications in the cartilage and nasomaxillary hypoplasia [1]. Recently, there have been a number of reports on BCDP with early onset of cervical myelopathy. Despite recent advancement of instrumentation techniques, however, rigid occipito-cervical fusion (OCF) is technically demanding in the very young pediatric populationwith small and fragile osseous elements [2-6]. We report here on an 18-month-old boy with BCDP, whose atlantoaxial instability was successfully managed with OCF using screw, plate and rod instrumentations. To our best knowledge, this is the first report on application of the instrumentation technique to a toddler patient withBCDP.

Materials and Methods

Informed consent was obtained from the patients. The male infantwas referred to our hospital for tetraplegia and swallow obstacleat the age of 16months. The patient could not crawl, roll over, stand, hold objects, or elevate the upper limbs to grasp objects.

Imaging data

The atlantoaxial distance between the anterior surface of the odontoid process and the posterior surface of the anterior arch of the atlas was 11 mm. (Fig. 1A). Neck extension under fluoroscopic imaging showed no reduction of the instability (Fig. 1B). However, the atlantoaxial instability became spontaneously reduced in MRI of a supine position under general anesthesia. (Fig. 1C). The posterior arch of the atlas and odontoid process were hypoplasic (Fig. 2A). Although the pedicles and lateral mass of each cervical spine were small and immature, the laminae were comparatively thick and large (Fig. 2B, C, D, and E).

Halo-body jacket immobilization

One week prior to surgery, the patient was fitted with a halo vest(Lil' Angel® Halo, Ossur, Reykjavik, Iceland) under general anesthesia and placed in a supine position.Ten scalp pins were inserted by probing with the fingers (Fig.3A). Intraoperative CT confirmed the insertion position of the scalp pins (Fig. 3B).Reduction of the atlantoaxial joint was successful with the halo vest(Fig.3C).

Surgical technique

One week after halo-body jacket immobilization, the patient was placed in a prone position under general anesthesia and the halo-body jacket was connected to a May field using a special adaptor(Fig.4). The MOUNTAINEER® OCT Spinal System (DePuy Synthes Spine, MA, USA) was adopted for the placement of C2, 3 intralaminar screws (diameter 3.5 mm, length 20 mm) and occipital screws(4.5 mm diameter, length 8 mm and 10 mm, 1 each) and plate with rods along with an autogenous bone graft from the right 8 and 9 ribsand artificial bone (ReFit®)(Fig. 5A, B, C, and D).A navigation system was not used for this surgery, because the presence of cartilage, which cannot be detected by CT, can interfere with accurate navigation. Cancellous laminar bone was observed under direct vision, and bone interior was assessed with a Lenke probe, ball-tip probe, and a tap, in that sequence. Four laminar screws were inserted using free hand technique. The length of occipital screws was determined by measuring thickness after drilling through the occipital bone; occipital screws were then inserted. After instrument implantation, a transverse skin incision was made immediately above the left center of the 8th rib in prone position, and bone harvesting was performed.Operative time was 245 min and blood loss volume was 30 mL.

Results (postsurgical outcome)

No major post-surgical complications were encountered. Halo-body jacket immobilization was continued for 3 months.MRI at 5 months after surgeryindicated that the C1/2 spinal cord compression was relieved with decreased T2signaling(Fig. 5E).CT at 6 months confirmed occipito-cervical bony fusion (Fig. 6A, B, C,D, E, F). The patient showed mild neurological improvement at one year; he was able to shake a toy, kick a blanket with his feet, orally ingest small amounts of food, and interact more actively with his parents.

Discussion

From the technical viewpoint, it is important to bring to attention the following aspects of our experience with OCF fora toddler patient: 1) a halo-body jacket (10 scalp pins) was tolerated for 3 months, 2) rigid internal fixation by screw, rod, and occipital plate was feasible, and 3) bone fusion by this technique was achieved at 6 months postoperatively.

Surgical treatment for upper cervical instability in skeletal dysplasias is challenging, particularly in very young children because of their small, fragile osseous elements. The outcomes of internal rigid fixation in skeletal dysplasia patients of less than 2 years are summarized in Table 1.Bone union was confirmed by Couture et al., while in the report by Savage et al., bone union was not obtained despite successful rigid fixation [7-10]. Sankaran et al. described successful posterior fixation using sublaminar wires over C4 and C5+C6 laminae and contoured luque rods overlayed with a morselized rib graft in a 10-week-old patient with congenital subaxial cervical instability[11]. On the other hand, a recent meta-analysis indicated that cervical spine screw fixation provided a significantly higher bone fusion rate and lower incidence of complications in pediatric cervical spine disease than did wiring [12].To the best of our knowledge, this is the first report of successful OCF on a toddler (i.e., less than 2years old) with BCDP.

In our experience, limited rotation is observed in about 50 to 60% of occipito-cervical fixation subjects and 30 to 40% of atlantoaxial fixation subjects (unpublished data). Adjacent intervertebral disorders were not observed after 10 to 15 years of surgery; however, further follow-up is warranted. Limitation of range of motion between O and C3 may occur, and should be about 30° for extension, 17° for lateral bending, and 55° for rotation in a healthy adult; [13] in addition, the occurrence of an adjacent intervertebral disorder in the middle or lower cervical vertebrae is likely. Toyama et al. [14] reported that 4 out of 12 children (mean age at surgery: 9.8 years old) who underwent C1-C2 posterior fusion showed postoperative cervical malalignment (kyphosis or swan-neck deformity) after a mean follow-up of 6.2 years. We expect a similar ratio of cervical disorders, even though our surgery involves C0-C3 fusion.

Conclusion

Rigid OCF with screw, rod, and occipital plate instrumentation was completed successfully for atlantoaxial instability in an 18-month-old toddler with BCDP.

Reference

1. Irving MD, Chitty LS, Mansour S, et al. Chondrodysplasia punctata:

a clinical diagnostic and radiological review. Clin Dysmorphol. 2008; 17:229-41.

2. Superti-Furga A, Unger S. Nosology Group of the International Skeletal Dysplasia Society.Nosology and classification of genetic skeletal disorders: 2006 revision. Am J Med Genet A. 2007; 143:1-18.

3. Ain MC, Chaichana KL, Schkrohowsky JG. Retrospective study of cervical arthrodesis in patients with various types of skeletal dysplasia. Spine(PhilaPa1976). 2006;31: E169-74.

4. Koop SE, Winter RB, Lonstein JE. The surgical treatment of instability of the upper part of the cervical spine in children and adolescents. J Bone Joint Surg Am. 1984;66: 403-11.

5. Miyoshi K, Nakamura K, Haga N, et al. Surgical treatment for atlantoaxial subluxation with myelopathy in spondyloepiphyseal dysplasia congenita. Spine (Phila Pa 1976). 2004;29: E488-91.

6. Svensson O, Aaro S. Cervical instability in skeletal dysplasia. Report of 6 surgically fused cases. Acta Orthop Scand. 1988;59:66-70.

7.Gluf WM1, Brockmeyer DL. Atlantoaxial transarticular screw fixation: a review of surgical indications, fusion rate, complications, and lessons learned in 67 pediatric patients. J Neurosurg Spine 2005; 2:164–169.

8. Anderson RC, Ragel BT, Mocco J, et al. Selection of a rigid internal fixation construct for stabilization at the craniovertebral junction in pediatric patients. J Neurosurg 2007;107 (1 Suppl):36–42.

9. Couture D, Avery N, Brockmeyer DL. Occipitocervical instrumentation in the pediatric population using a custom loop construct: initial results and long-term follow-up experience. Clinical article. J Neurosurg Pediatr 2010; 5:285–291.

10.Savage JG, Fulkerson DH, Sen AN, et al. Fixation with C-2 laminar screws in occipitocervical or C1–2 constructs in children 5 years of age or younger: a series of 18 patients. J Neurosurg Pediatrics 2014; 14:87–93.

11.Sankaran R, Shah R, Menon S, et al. Congenital subaxial cervical subluxation presenting as a bilateral Erb's palsy: surgical management, rehabilitation, and outcome. Childs Nerv Syst 2016; 32:849-855.

12. Hwang SW, Gressot LV, Rangel-Castilla L, et al. Outcomes of instrumented fusion in the pediatric cervical spine. J Neurosurg Spine. 2012; 17:397-409.

Figure Legends

Fig.1: Maximum flexion (A) and extension (B) radiographs and preoperative sagittal MRI (C). Atlantoaxial distance was 11mm. Spinal cord accommodation was 2mm. The instabilitywasnot reduced by neck extension. Cervical MRI of a supine position under general anesthesia revealed spontaneous reduction of the atlantoaxial instability

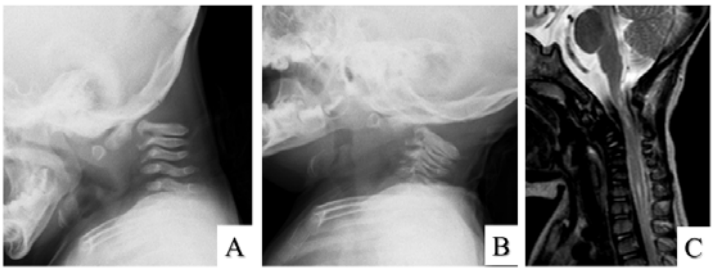

Fig.2: Cervical plain CT in the sagittal view (A), axial view of C1 (B), C2 (C), and C3 (D), and a 3D model (E). The posterior arch of the atlas and odontoid process were hyperplasic. The pedicles and lateral mass of each cervical vertebra were small and immature, but the laminae were comparatively thick and large.

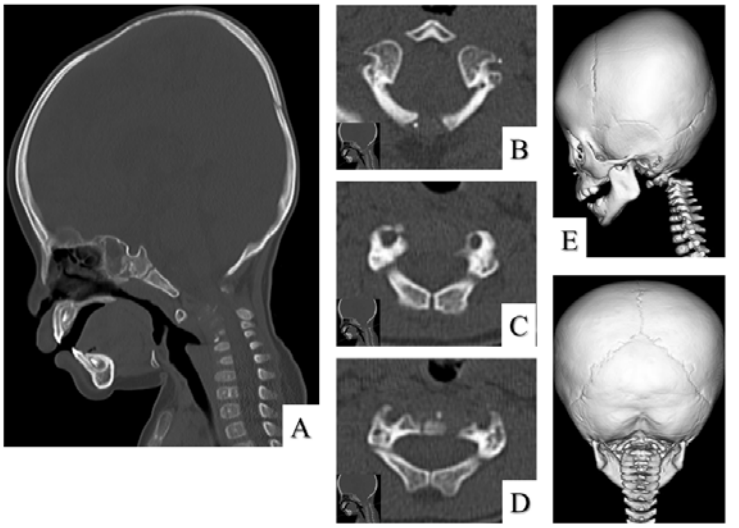

Fig.3: Halo-body jacket immobilization. Ten scalppins were placed by finger palpation(A). Intraoperative CT confirmed the insertion positions of the scalp pins (B). Reduction of the atlantoaxial joint was achieved by setting the halo-body jacket in a supine position under general anesthesia (C).

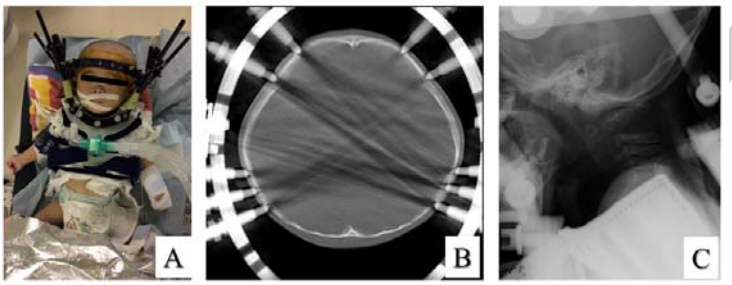

Fig.4: The patient was placed in a prone position under general anesthesia and the halo-body jacket was connected to a May field using a special adaptor.

Fig.5: Plate-rod placement with occipital cortical screws and C2/C3 interlaminar screws. Addition of an autogenous bone graft from the right 8 and 9 ribs combined with a hydroxyapatite/collagen composite(A). Anteroposterior (B),lateral extension (C), and lateral flexion (D) radiographsand sagittal MRI (E) showing after occipitocervical fusion.

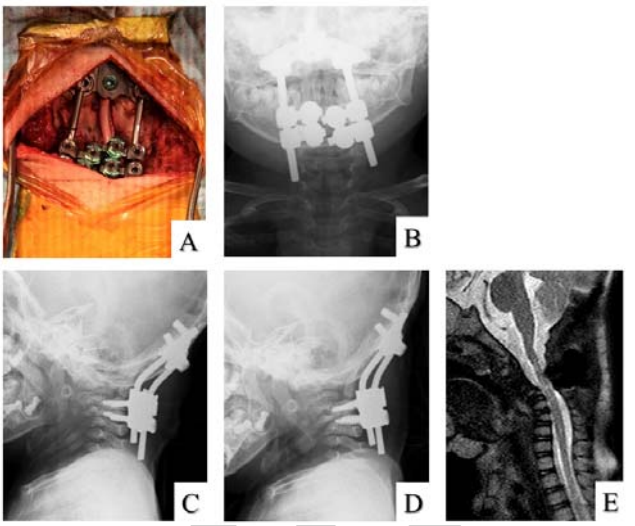

Fig.6: Plate-rod placement with occipital cortical screws and C2/C3 interlaminar screws. Cervical CT at postoperative 6 months. C2 (A), C3 (B) and occipital bone (C) axial views, sagittal view (D) (E) and coronal view (F). Bone fusion was achieved despite some screw loosening.

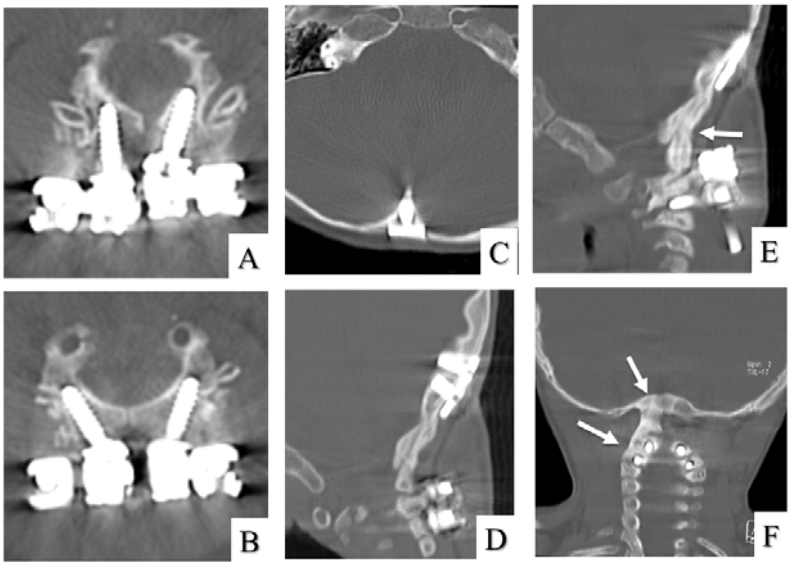

Table 1. Literature review of occipitocervical fusion in infants and toddler less than 2 years'old

Author	Year	Age at surgery (yrs)	Diagnosis	Surgery target	Fusion location	Surgical procedures	Bone union
Anderson RC et al. [13]	2007	1.3	ND	Atlanto-occipital instability	Oc-C2	Occipital screws,C2 pars screws	ND
Couture D et al.[14]	2010	1.2	Post-traumatic	Atlanto-occipital instability	Oc-C2	Occipital screws,bilateral pars screws	Yes
Savage JG et al.[15]	2014	0.8	Chondrodysplasia punctata	Congenital instability	Oc-C4	Occipital screws,unilateral translaminar screw	No

ND: not determined