Eur Spine J
DOI 10.1007/s00586-017-5116-5

CASE REPORT

Atlantoaxial dislocation and os odontoideum in two identical twins: perspectives on etiology

Xiangsheng Tang[1] · Mingsheng Tan[1] · Ping Yi[1] · Feng Yang[1] · Qingying Hao[1]

Received: 8 March 2017 / Revised: 10 April 2017 / Accepted: 30 April 2017

Abstract
Purpose There are two theories about the origin of os odontoideum: traumatic or congenital. However, most studies favor the hypothesis of traumatic theory. To emphasize the congenital theory, we report a pair of identical twins both with atlantoaxial dislocation and os odontoideum, which is believed to be a congenital defect.
Methods We present two 14-year-old identical twins with atlantoaxial dislocation and os odontoideum. Neither of the twins had history of trauma in head nor cervical spine. We reviewed and compared the cervical radiographs of the identical twins. Posterior atlantoaxial reduction, pedicle screw fixation and atlantoaxial fusion were performed for the two twins.
Results Radiological examination showed the identical twins had typical atlantoaxial dislocation and os odontoideum. The twins had high similarity in the appearance of atlantoaxial dislocation and os odontoideum. The etiology of the os odontoideum in the twins is believed to be congenital. Both the twins had improvement in neurological function after surgery.
Conclusion Although a great number of cases with os odontoideum have been reported to be traumatic, there are some cases believed to be congenital.

Keywords Os odontoideum · Atlantoaxial dislocation · Identical twins · Congenital · Traumatic

✉ Mingsheng Tan
zrtanms@sina.com

[1] Department of Spinal Surgery, China-Japan Friendship Hospital, Beijing 100029, China

Introduction

Os odontoideum is an anatomic anomaly consisting of a shortened odontoid process and a smooth ossicle of bone [1, 2]. Os odontoideum is one of the reasons causing atlantoaxial dislocation. There has been a debate about the origin of os odontoideum. The traumatic hypothesis considers it an acquired pathology resulting from avascular necrosis caused by odontoid fracture [3–5]. Conversely, the congenital hypothesis considers it a segmental defect, which represented a failed fusion of odontoid and axis vertebral body [6–8]. However, at the present time, there are few published articles supporting the congenital hypothesis. To emphasize the congenital theory, we present a pair of identical twins both with atlantoaxial dislocation and os odontoideum, which is believed to be a congenital defect. To improve the accuracy of atlantoaxial pedicle screw placement, a novel personalized image-based drill guide template was used during the surgery.

Case presentation

Case 1

History and examination

Twin A, a 14-year-old girl, experienced weakness in lower limbs for 2 years. There was no history of trauma in head or cervical spine. 20 days ago, after falling down accidently, she got incomplete paraplegia and was unable to walk. She also had neck and shoulder pain, and upper extremity paresthesia.

Twin A underwent radiographic workup including flexion/extension lateral radiographs, computed

Published online: 15 May 2017

tomography scan (CT), and magnetic resonance imaging (MRI). Lateral radiographs showed a widened distance between the anterior arch of atlas and odontoid. Based on CT images, reconstruction of the atlas vertebra showed typical atlantoaxial dislocation and os odontoideum. There were apparently shortened odontoid process and smooth ossicle of bone. Reconstruction of CT images also showed congenital incomplete union of C1 posterior arch. Cervical MRI showed marked spinal cord compression and associated myelomalacia. There was thickening atlantoaxial ligament. No bone erosion of odontoid was observed. Testing for rheumatoid arthritis was negative (Fig. 1).

Twin A otherwise previously was entirely fit and well. There were not any features suggestive of connective tissue disorder or pertinent past medical history. There was no family history of Down's syndrome or other handicapping disorders.

Surgical treatment

Because of the severe neurologic deficit resulted from atlantoaxial dislocation, twin A underwent posterior atlantoaxial reduction and internal fixation. Atlantoaxial pedicle screw fixation [9] and fusion was applied, as it could preserve the range of motion of cervical spine at the most extent.

To improve the accuracy of screw insertion, a personalized image-based drill guide template was used for atlantoaxial pedicle screw placement [10]. The drill guide template was created in Mimics v17.0 (Materialise, Leuven, Belgium) and 3-matic v9.0 (Materialise) softwares, and was converted into physical model using rapid prototyping technique. The template had two location holes and guide rods. Atlantoaxial pedicle screw trajectories can be achieved by drilling through location holes and parallel to guide rods. If actual screw trajectory deviates from ideal trajectory, surgeons can adjust the drill direction based on guidance of the template (Fig. 2). During the surgery, after complete removal of the soft tissue overlying atlantoaxial posterior bony structure, a good fit between the posterior bony surface and the drill guide template was achieved. Then the ideal atlantoaxial pedicle screw trajectories could be drilled based on the location hole and guide rod of the template. Posterior arch of C1 was resected for complete

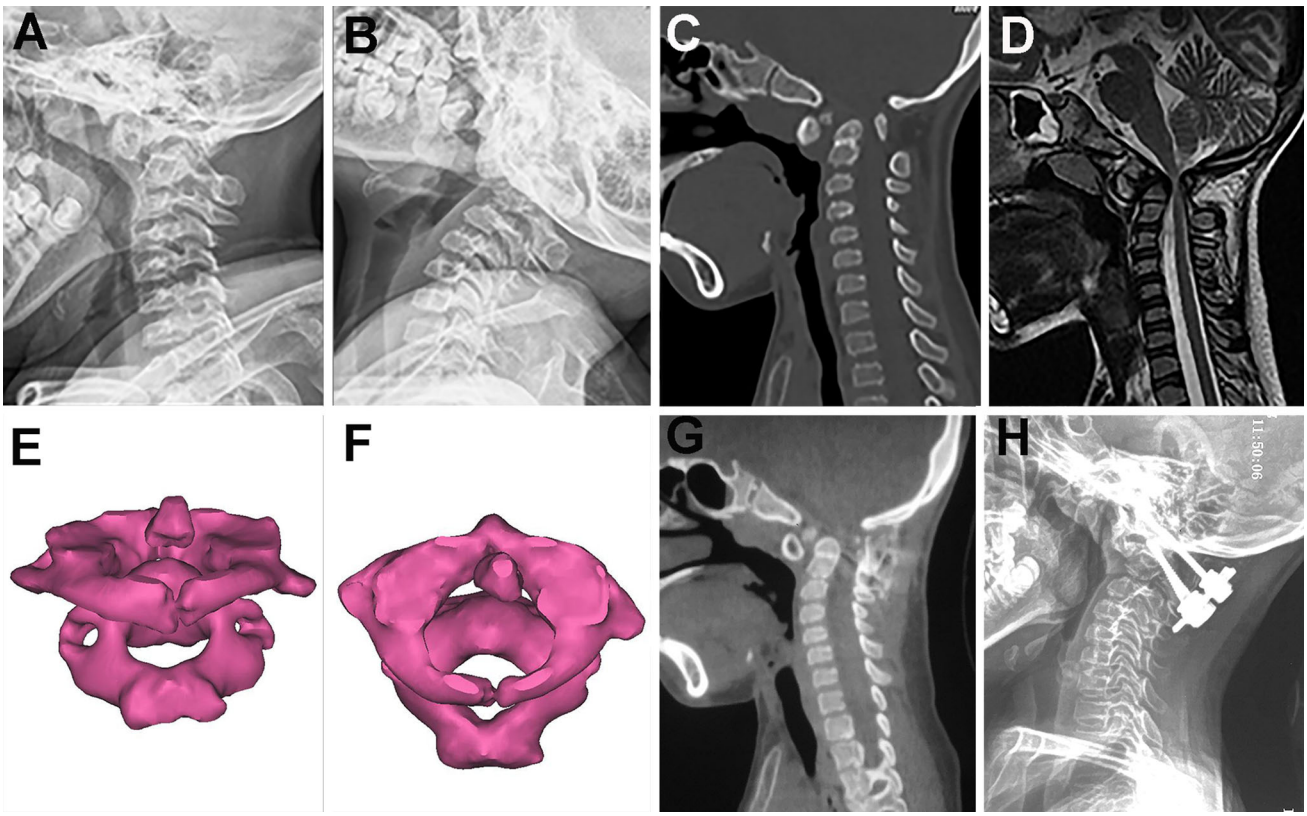

Fig. 1 Pre- and postoperative radiographs of twin A. **a–c** Flexion/extension lateral radiographs and sagittal CT reconstruction shows atlantoaxial dislocation. **d** Sagittal MRI shows atlantoaxial dislocation and marked spinal cord compression and associated myelomalacia. **e**, **f** Typical atlantoaxial dislocation, os odontoideum and congenital incomplete union of C1 posterior arch are presented in reconstructed atlantoaxial vertebrae models. **g** Postoperative sagittal CT reconstruction shows enlargement of spinal canal. **h** Postoperative lateral radiograph shows good positioning of atlantoaxial pedicle screws

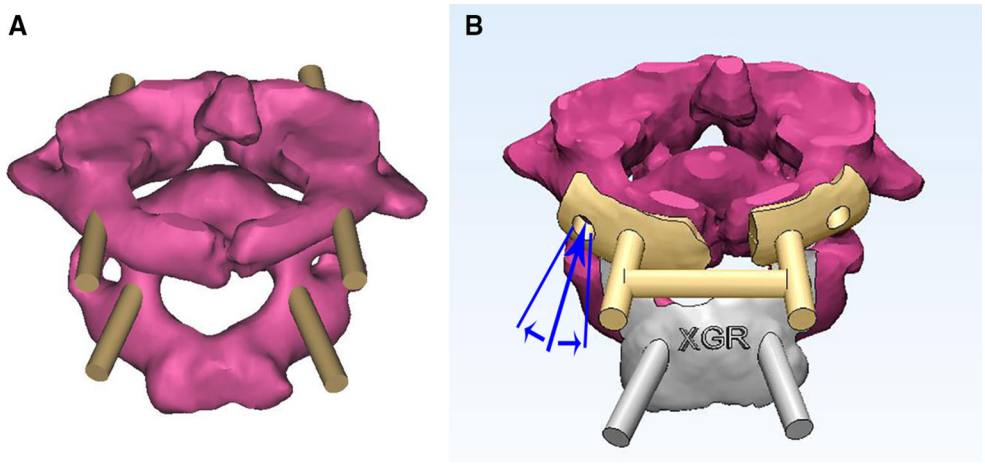

Fig. 2 **a** Reconstructed atlantoaxial vertebrae model and planned ideal pedicle screw trajectories of twin A. **b** The novel drill guide templates with two location holes and guide rods. By drilling through location hole and parallel to guide rod of the template, pedicle screw trajectory can be acquired. If actual screw trajectory deviates from planned ideal trajectory, surgeons can adjust the drill direction based on guidance of the template

neurological decompression. The C2 laminae was decorticated with a Rongeur, and an iliac crest graft was implanted on the posterior rim of C1 and C2.

At 10 days postoperatively, twin A could stand on floor and walk slowly. She was placed in a cervical collar for the next 12 weeks. At 3 months postoperatively, there was significant improvement in neurological function. Postoperative radiograph showed good bone graft fusion, and no screw loosening or fragmentation were found (Fig. 1).

Case 2

History and examination

After the diagnosis of atlantoaxial dislocation and os odontoideum in twin A, parents of twin A stated that the girl had an identical twin sister (twin B). We advised to perform radiological examination for twin B. Radiological examination showed twin B had atlantoaxial dislocation and os odontoideum just like twin A. The only difference is that twin B had a normal posterior arch without incomplete union in C1 (Fig. 3). After physical examination, twin B did not have obvious neurologic deficit, except that she complained of intermittent tingling and numbness in neck. As to previous medical history, twin B was otherwise previously were entirely fit and well.

Surgical treatment

Considering the instability of atlantoaxial joint, twin B underwent posterior atlantoaxial reduction and pedicle screw fixation. To preserve the range of motion of cervical spine at the most extent, twin B also underwent atlantoaxial fusion. Personalized image-based drill guide template was also used to improve the accuracy of screw insertion [10]. As her neurological function was relative good, posterior arch of atlas was not resected. After the C1 posterior arch and the C2 laminae were decorticated partly, an iliac crest graft was modified and implanted.

After the surgery, tingling and numbness in neck of twin B disappeared. She was also placed in a cervical collar for 12 weeks. Three months after surgery, radiological examination showed good bone graft fusion without internal fixation failure (Fig. 3).

Discussion

The origin of os odontoideum heretofore has been explained by two different theories: traumatic or congenital. To the present time, most studies favor the hypothesis of traumatic theory for os odontoideum [3, 11–14]. According to the traumatic hypothesis, os odontoideum is an acquired pathology resulting from avascular necrosis caused by odontoid fracture.

Freilberger et al. [14] reported a 25.5-month-old baby who had normal radiographs after a fall. However, new radiographs obtained 4 months later showed absence of the caudal portion of odontoid and atlantoaxial instability. Stillwell and Fielding [13] documented a 7-year-old boy with normal radiographs at the time of a fall. Subsequent radiographs showed an os odontoideum. Hukda et al. [12] hypothesized that traumatic event in infancy was the etiology of os odontoideum. Upper portion of the dens could be sheared off by the tautness of transverse ligament. Since it is cartilaginous, the fracture cannot be detected until apical segment is ossified [12]. With respect to os odontoideum in identical twins, Verska et al. [15] reported one

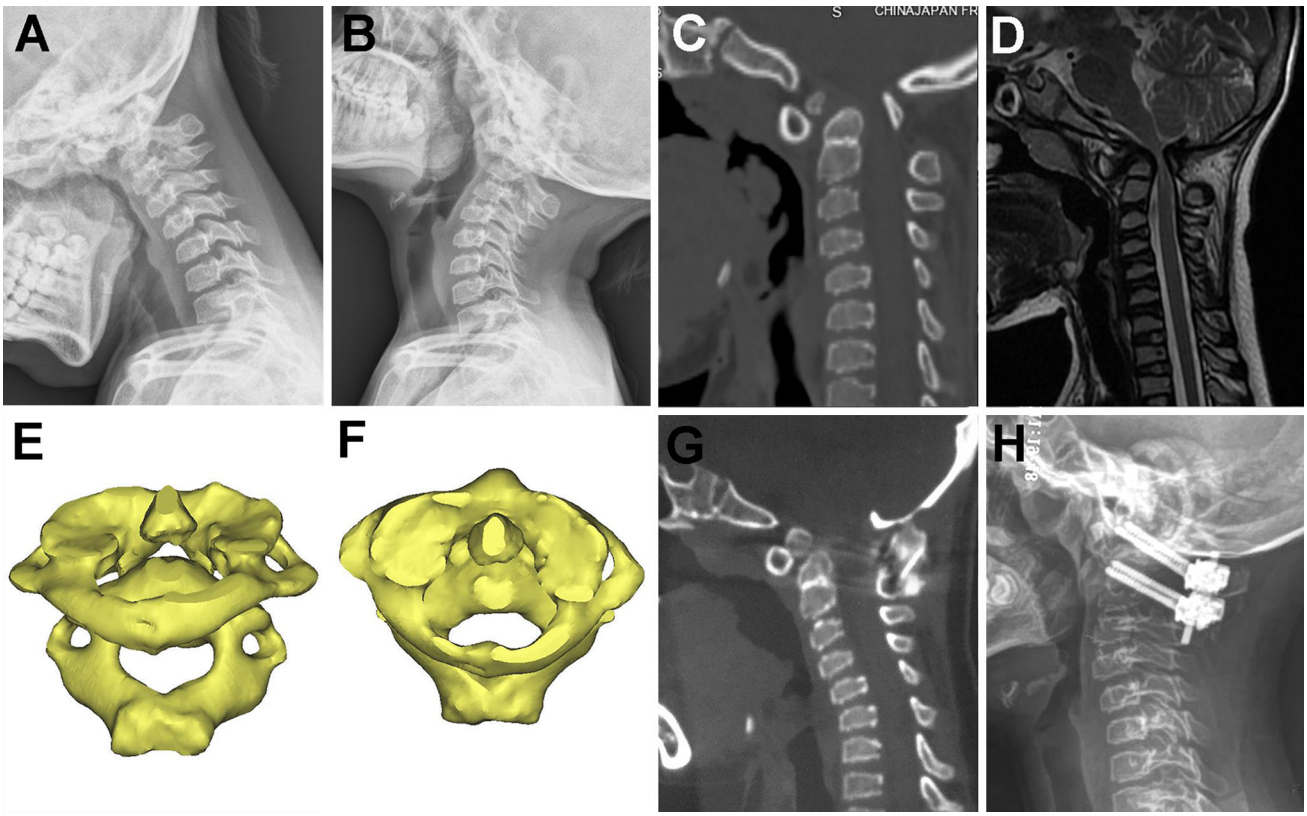

Fig. 3 Pre- and postoperative radiographs of twin B. **a**–**c** Flexion/extension lateral radiographs and sagittal CT reconstruction show atlantoaxial dislocation. **d** Sagittal MRI shows atlantoaxial dislocation and marked spinal cord compression and associated myelomalacia. **e**, **f** Typical atlantoaxial dislocation and os odontoideum are presented in reconstructed atlantoaxial vertebrae models. **g** Postoperative sagittal CT reconstruction shows reduction of atlantoaxial dislocation. **h** Postoperative lateral radiograph shows good positioning of atlantoaxial pedicle screws

twin had os odontoideum after trauma, while the other twin had normal cervical spine and had no history of trauma.

While the traumatic theory has been widely supported, it is clear from some studies [16, 17] that the os odontoideum may result from the congenital theory. The identification of identical twins with os odontoideum is helpful to study the origin of this disorder. Os odontoideum in identical twins without a history of trauma may be explained by the congenital theory. In these cases, the identical twins usually have nearly identical morphology of os odontoideum.

Kirlew et al. [16] first reported familial os odontoideum in a context suggesting a genetic etiology. In this report, os odontoideum was discovered in both female twins without prior trauma. Radiographs showed that both twins had partial fusion of the posterior elements of C2 and C3. And both twins also had overgrowth of anterior arch in C1, and stress-related concavity of spinous process in C2. Kirlew et al. believed the os odontoideum in the twins was associated with congenital etiology because of the consanguinuity and the similar appearances of the defect. To accommodate the congenital etiology, the authors proposed the origin of os odontoideum to be an incomplete migration of the axis centrum rather than incomplete fusion.

In the current report, both the identical twins had typical atlantoaxial dislocation and os odontoideum. Neither of the twins had history of trauma in head or cervical spine. The twins had high similarity in the appearance of atlantoaxial dislocation and os odontoideum, which was showed clearly in the reconstructed atlantoaxial vertebrae model. Besides, the twins did not have an underlying predisposition to hypermobility that may lead to os odontoideum, for example, Down's syndrome. There was no family history of Down's syndrome or other handicapping disorders. Therefore, we believe the os odontoideum in the twins is associated with the congenital etiology.

Another report supporting the congenital etiology was provided by Morgan et al. [17]. In this report, a 16-year-old boy underwent radiological examination after a motor vehicle accident, and os odontoideum was discovered. After further investigation, os odontoideum was also discovered in the boy's father and paternal grandmother. Then a family tree could be plotted. Klippel–Feil syndrome was

also discovered in these three individuals with os odontoideum.

To further analyze the gene expression profiles of os odontoideum patients, Straus et al. [18] performed a study based on genetic analysis. They compared the gene expression profiles of congenital os odontoideum, traumatic os odontoideum and the controls. A pair of identical twins both with os odontoideum, two nontwin patients with os odontoideum and four normal subjects were included in this study. A number of genes with significantly increased expression were detected in the twins as compared with the normal subjects. Six genes with significantly different expression profiles were identified between the twins and the nontwin patients. This study also demonstrated trends in gene expression profiles between os odontoideum patients and the normal subjects. Lots of these genes are associated with bone morphogenesis and maintenance.

In conclusion, although a great number of cases with os odontoideum have been reported to be traumatic, there are some cases believed to be congenital. Still some other cases with os odontoideum may have combined traumatic and congenital origins. It will be informative to further define each type and examine their relative prevalence. Investigation of the relevance of this distinction is appropriate to clinical evaluation and treatment.

Compliance with ethical standards

Source of funding No funds were received in support of this work. No benefits in any form have been or will be received from a commercial party related directly or indirectly to the subject of this manuscript.

Conflict of interest None of the authors has any potential conflict of interest.

Ethical approval The study protocol was approved by the local Medical Ethics Committee.

Informed consent Informed consent was obtained from all individual participants included in the study.

References

1. Matsui H, Imada K, Tsuji H (1997) Radiographic classification of os odontoideum and its clinical significance. Spine 22:1706–1709
2. Spierings EL, Braakman R (1982) The management of os odontoideum. Analysis of 37 cases. J Bone Jt Surg Br 64:422–428
3. Fielding JW, Griffin PP (1974) Os odontoideum: an acquired lesion. J Bone Jt Surg Am 56:187–190
4. Wang S, Wang C (2012) Acquired os odontoideum: a case report and literature review. Child's Nerv Syst 28:315–319
5. Zygourakis CC, Cahill KS, Proctor MR (2011) Delayed development of os odontoideum after traumatic cervical injury: support for a vascular etiology. J Neurosurg Pediatr 7:201–204
6. Currarino G (2002) Segmentation defect in the midodontoid process and its possible relationship to the congenital type of os odontoideum. Pediatr Radiol 32:34–40
7. Flemming C, Hodson CJ (1955) Os odontoideum; a congenital abnormality of the axis; case report. J Bone Jt Surg Br 37-B:622–623
8. Ellies DL, Krumlauf R (2006) Bone formation: the nuclear matrix reloaded. Cell 125:840–842. doi:10.1016/j.cell.2006.05.022
9. Tan M, Wang H, Wang Y et al (2003) Morphometric evaluation of screw fixation in atlas via posterior arch and lateral mass. Spine 28:888–895. doi:10.1097/01.brs.0000058719.48596.cc
10. Jiang L, Dong L, Tan M et al (2016) Accuracy assessment of atlantoaxial pedicle screws assisted by a novel drill guide template. Arch Orthop Trauma Surg 136:1483–1490. doi:10.1007/s00402-016-2530-9
11. Schuler T, Kurz L, Thompson D et al (1991) Natural history of os odontoideum. J Pediatr Orthop 11:222–225
12. Hukuda S, Ota H, Okabe N et al (1980) Traumatic atlantoaxial dislocation causing os odontoideum in infants. Spine 5:207–210
13. Stillwell W, Fielding J (1978) Acquired os odontoideum. A case report. Clin Orthop Relat Res (135):71–73
14. Freiberger RH, Wilson PD Jr, Nicholas JA (1965) Acquired absence of the odontoid process: a case report. J Bone Jt Surg Am 47:1231–1236
15. Verska J, Anderson P (1997) Os odontoideum. A case report of one identical twin. Spine 22:706–709
16. Kirlew K, Hathout G, Reiter S et al (1993) Os odontoideum in identical twins: perspectives on etiology. Skelet Radiol 22:525–527
17. Morgan M, Onofrio B, Bender C (1989) Familial os odontoideum. Case report. J Neurosurg 70:636–639
18. Straus D, Xu S, Traynelis V (2014) Os odontoideum in identical twins: comparative gene expression analysis. Surg Neurol Int 5:37