Eur Spine J
DOI 10.1007/s00586-016-4783-y

CASE REPORT

Accidental or linked: separated odontoid process fused to the enlarged anterior arch of the atlas associated with atlantoaxial subluxation in a Kashin–Beck disease patient

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Received: 15 July 2016 / Revised: 9 September 2016 / Accepted: 15 September 2016

Abstract
Purpose KBD is an endemic disease affecting the epiphyseal growth plate and articular cartilage of multiple joints, resulting in extremities' deformation and skeletal dysplasia. More attention has been paid to the visible deformed extremities instead of inconspicuous spinal condition. There is a lack of reports concerning the spinal radiological features, especially for the atlantoaxial joint. The aim of this paper is to report a case of a Kashin–Beck disease (KBD) patient diagnosed with atlantoaxial subluxation, concomitant with separated odontoid process fused to the enlarged anterior arch of the atlas.
Methods We report the case of a 60-year-old woman with 54 years' history of KBD complaining of occipitocervical pain, decreasing motor strength and sensory function of both upper and lower extremities. Subsequent radiological examinations of lateral plain radiography, computed tomography scans and magnetic resonance imaging were performed to reveal these rare characteristics of atlantoaxial joint in this patient. Then, we review the associated articles to postulate whether this anomaly is accidental or linked in a KBD patient.
Results She had an extremely rare variant with three aspects of characteristics: atlantoaxial subluxation concurrent with severe spinal canal stenosis and spinal cord compression, odontoid process separating from the body of axis, and the enlarged anterior arch of the atlas fusion with odontoid process. Comparing with the congenital anomaly of atlantoaxial joint, we postulated that this aetiology of anomaly might be linked to the acquired form attributed to the histopathology of KBD, rather than an accidental event.
Conclusions The anomaly of atlantoaxial joint might occur in KBD patients. Larger numbers of KBD candidates with earlier symptoms are recommended for radiological examinations of atlantoaxial joint, especially for the adolescents. Spinal surgeons are suggested to involve the research of the spinal anatomy and variation for the prevention and earlier therapy for KBD patients.

Keywords Kashin–Beck disease · Odontoid process · Atlantoaxial subluxation · Fusion

Introduction

Kashin–Beck disease (KBD) is a chronic, endemic disease affecting people in a crescent geographical distribution through in Tibet, northern China, Mongolia, Siberia and North Korea [1]. It starts in childhood, and commonly attacks the epiphyseal growth plate and articular cartilage of multiple joints [2], resulting in skeletal dysplasia and bony deformities [3, 4]. Previous researches have focused on the clinical and dysfunctional features of upper and lower limbs of KBD patients [4–6]. Yet as of date, there is a lack of reports concerning their spinal radiological features, especially for the atlantoaxial joint. In this report, we present a 60-year-old woman with a long history of KBD.

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Published online: 21 September 2016

She was diagnosed with atlantoaxial subluxation, and radiological imaging showed that separated odontoid process from body of axis fused to the enlarged anterior arch of the atlas, concurrent with severe spinal canal stenosis and spinal cord compression. Is this abnormality accidental or linked for a KBD patient?

Case presentation

A 60-year-old female (height: 120 cm, weight: 31 kg, BMI: 21.53) was referred for hospitalisation with a 6-month history of occipitocervical pain, numbness and weakness of both upper and lower extremities that worsened rapidly in the recent 1 month. She came from an endemic area of KBD. Physical examinations revealed a short-trunk dwarfism with multiple enlarged and deformed joints with restricted range of motion, muscle atrophy, shortened fingers (Fig. 1). She was diagnosed with III degrees KBD, according to the Diagnostic Criteria of KBD in China (WS/T207-2010) [7]. The symptoms of multiple joints pain and deformities occurred and gradually progressed since she was 6 years old. She had slight tenderness of occipitocervical region and slight neck movement restriction. Her motor strength and sensory function of upper and lower extremities were remarkably deceased (myodynamia decreased to Grade 3–4 of 5), and the muscle tension increased. It was difficult to induce tendon reflex due to the severe deformity and limited movement of joints. The pathological reflexes, such as Hoffmann's sign and Babinski's sign, were positive. She had urinary and fecal incontinence. She did not remind of trauma of her neck. Lateral plain radiography on her cervical spine showed enlarged, rounded anterior arch of atlas. There is a bright gap between the posterior margin of anterior arch of atlas and anterior edge of axial body with the sclerotic margin (Fig. 2). The computed tomography (CT) scans demonstrated that enlarged anterior arch of atlas fused with the shortened odontoid process which was separated from the irregular body of axis. Meanwhile, the posterior arch of atlas migrated anterior to the imaginary line connecting the spinolaminar lines. The spinal canal was severely narrow in the level of atlantoaxial joint (Fig. 3). Cervical magnetic resonance imaging (MRI) revealed compression of the spinal cord with the appearance of swelling signal (Fig. 4). Fully understanding the high risk of operation, the patient refused to accept any kind of treatment and was discharged home.

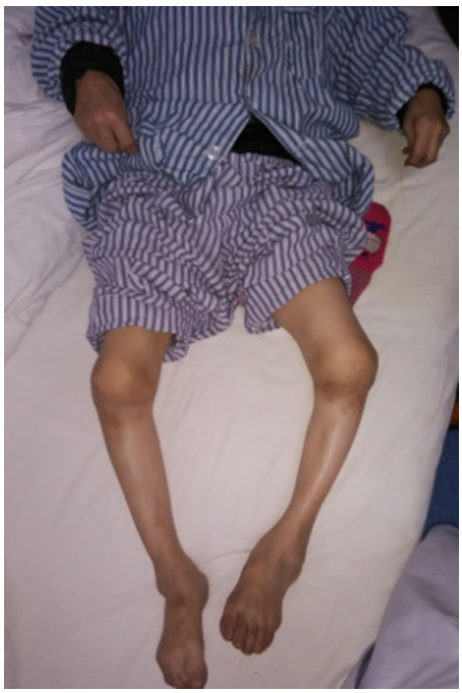

Fig. 1 Photographs of the KBD patient: she has a short-trunk dwarfism with enlarged and deformed wrist-, finger-, elbow-, knee-, ankle-, and toe-joints, with restricted range of motion and muscle atrophy

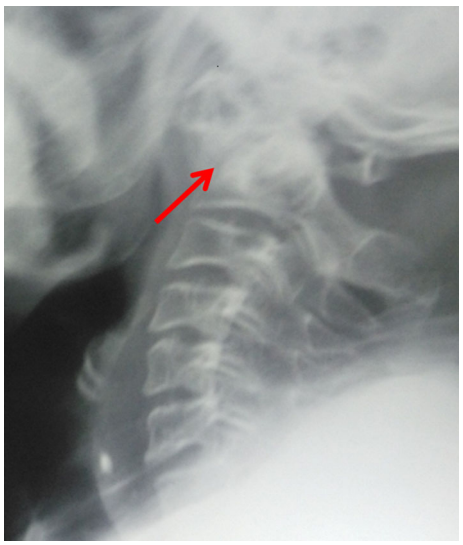

Fig. 2 Lateral plain radiograph on her cervical spine showed enlarged, rounded anterior arch of atlas and irregular morphometry of axial body. There is a bright gap between the odontoid process and body of axis (*red arrow*) with the sclerotic margin

Discussion

The KBD patients are featured with skeletal dysplasia and bony deformities. However, more attention has been paid to the visible deformed extremities instead of inconspicuous spinal condition. Here, we report a case of rare atlantoaxial disturbance in a KBD patient who was diagnosed as atlantoaxial subluxation [8] (Fig. 3a). It is also

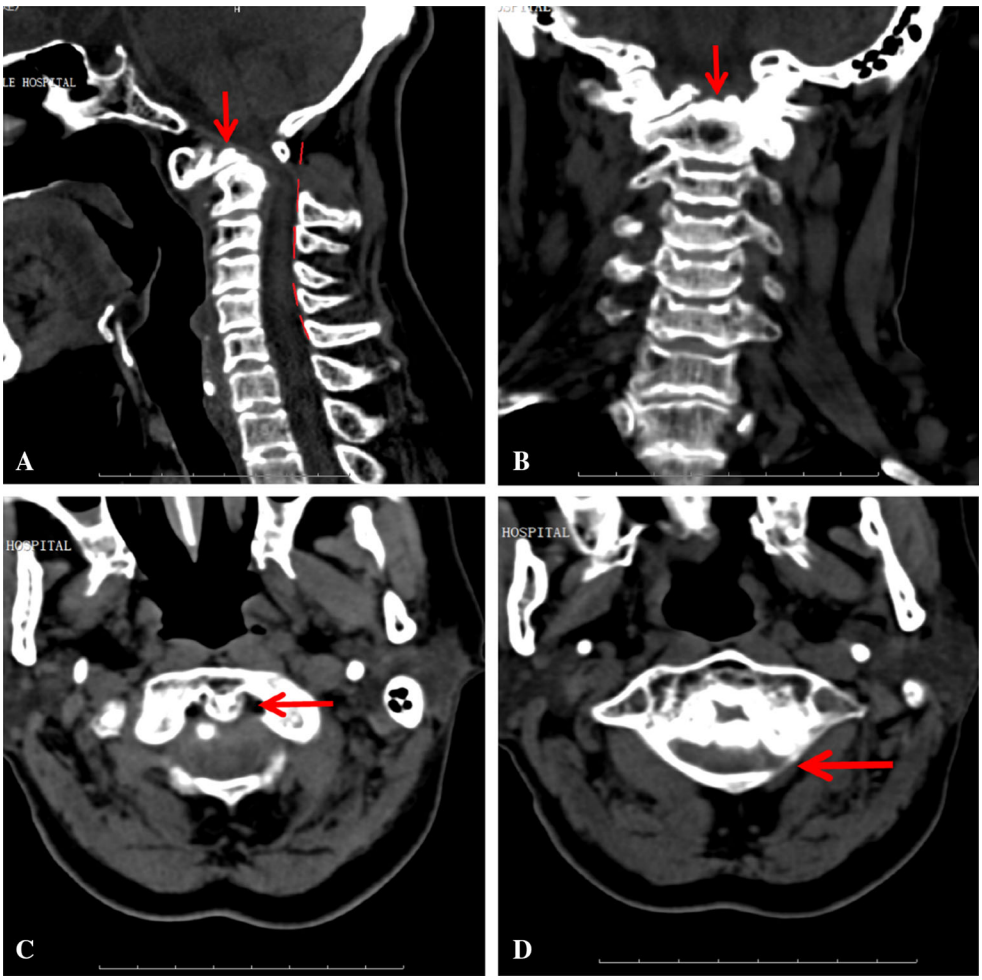

Fig. 3 **a** The sagittal view of computed tomography (CT) image showed enlarged anterior arch of atlas fused with the shortened odontoid process (*red arrow*) which separated from the body of C2. The posterior arch of atlas migrated anterior to the imaginary line (the *red incontinuous lines*) connecting the spinolaminar revealing the atlantoaxial subluxation [8]. **b** The coronal view of CT showed the defect of odontoid process cephalical to the body of the axis (*red arrow*) and the osteophytes formation in atlantoaxial joints. **c** The axial view of CT image revealed an atlas-odontoid process fusion (*red arrow*). **d** Another axial view showed severe spinal canal stenosis (*red arrow*)

demonstrated that odontoid process is separated from the body of vertebra dentata and fuses with enlarged anterior arch of atlas. This abnormality might be the contribution to the atlantoaxial subluxation. The anterior migration of the atlas and hypertrophy of the axial body results in severe cervical spinal canal stenosis at C1–2 level with ventral spinal cord compression, which is the direct reason for her symptoms of neck pain and neurological deficits. She had not recalled a history of trauma episode in neck, and the radiological appearance implicates that this disorder is not consistent with a recent fracture. Thus, the atlantoaxial subluxation may be spontaneous, not traumatic.

In all, she has an extremely rare variant with three aspects of characteristics: atlantoaxial subluxation, odontoid process separating from the body of axis, and the enlarged anterior arch of the atlas fusion with odontoid process. This kind of abnormality has never been reported in a KBD patient. Is this anomaly accidental or linked in a KBD patient? Does it stem from acquired form or congenital variant?

A clear understanding of the anatomy and developmental centers of the axis in childhood is of considerable value for enlightened clinical and radiographic disorders associated with odontoid process and atlas. The odontoid process fuses with the atlas originating from the first cervical sclerotome between the sixth and seventh weeks of gestation [9]. It includes single apical secondary ossification center and the two lateral primary ossification centers (Fig. 5) [10]. The epiphyseal growth plate, acting as synchondrosis, separates these ossification centers. The tip of the odontoid process lays out the atlas and then moves caudally to join the body of the axis by the age of 4 years. Synchondrosis among the odontoid process, body, and neural arches fuse at the age of 3–6 years [10]. The

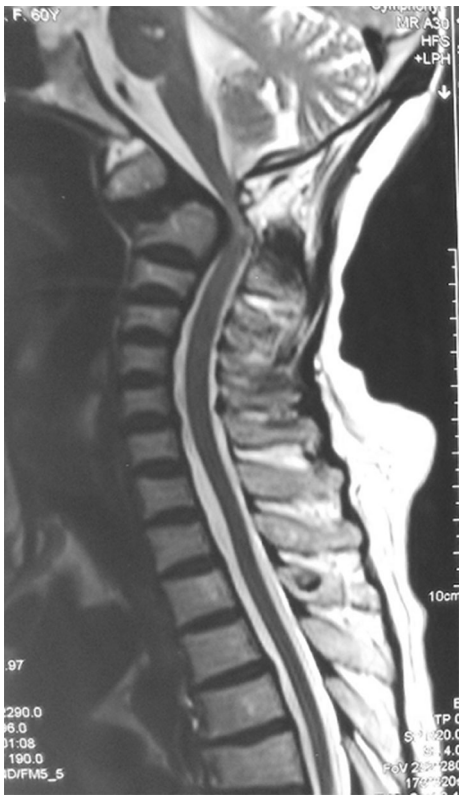

Fig. 4 Sagittal view of magnetic resonance imaging (MRI; T2) of cervical spine revealed enlarged anterior arch of atlas and compression of the spinal cord with the appearance of swelling signal. Subdental synchondrosis of the axis was not present

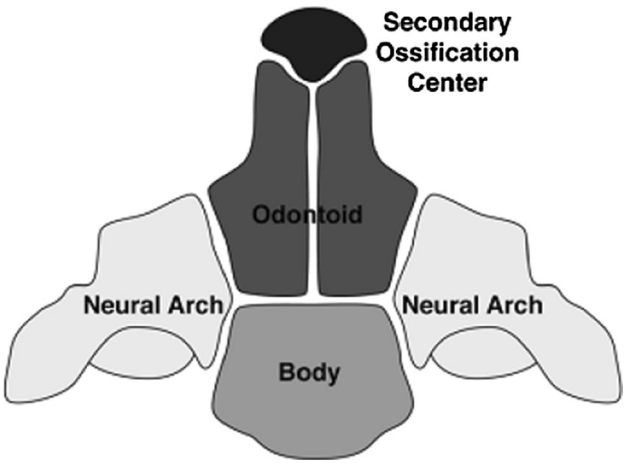

Fig. 5 Developmental centers of the axis including the two primary and single secondary ossification centers of the odontoid process. Referred to [10]

odontoid process fuses with the neural arches and the body by the age of 6 years [10].

Therefore, some anomalies may be attributed to the failure separation of ossification center of odontoid process with the anterior arch of atlas, or result from the failure fusion of odontoid process with future dens in the developing stage. The odontoid process has several anomalies described in previous literatures [10, 11], including aplasia, hypoplastic, os odontoideum, ossiculum terminale, dens bicornis, and fusion with the anterior arch of the atlas. Congenital fusion of the atlas and odontoid process is a extremely rare cervical anomaly [12], often associated with other cervical variants of varying degree. Tubbs et al. [13] reported the nonseparated odontoid process fused with the left hemi-arch of the atlas in an 8-year-old male. Gil et al. [14] presented fusion of the odontoid process with the anterior arch of the atlas and a bipartite atlas in an old man. This variant has been postulated to be a segmentation defect in the sclerotomes of the first cervical somite [15]. CT images showed the anterior arch cleft and the odontoid process were more or less hypoplastic in the above both cases. In our patient, the CT scan revealed that markedly shortened odontoid process fused with enlarged anterior arch of atlas, without fissure of the atlas arch. Therefore, our patient's disorder is different with the congenital fusion of odontoid process and atlas with cleft arches.

Os odontoideum is referred as variable size ossicle separated from odontoid tip with smooth circumferential cortical margins [10]. Its main aetiology is due to the congenital failure of fusion of ossification centers in the odontoid process or acquired occult fractures with subsequent avascular necrosis [16]. But the anomalous fusion of os odontoideum with the atlas is rarely reported. For this case, the separation of odontoid process is not referred to be this congenital disorder.

Therefore, any diseases that damage the development of epiphyseal plate of cartilage of in young and adolescent may lead to the abnormalities of craniovertebral junction, just as KBD. The most commonly affected lesions of KBD involve the epiphyseal growth plate and the articular cartilage of multiple joints [2, 17]. The deficiency of blood supply in the epiphyseal growth plate will lead to the disturbed endochondral ossification, or even earlier closure of the epiphyseal growth plate. This histopathology results in the growth retardation and the occurrence of severe joint deformities during development in childhood [17]. The clinical manifestations and classical radiographic abnormalities appear as early as 5 years of age. The symptoms of this patient occurred when she was 6 years old, therefore, her clinical and radiological demonstrations were severe. The skeletal dysplasia or short stature implicates not only the noticeable retarded growth of extremities, but also the axial skeleton abnormalities which have not been paid attention to until now.

As for our case, the aetiology of anomaly of atlantoaxial joint may be linked to the acquired form attributed to the

histopathology of KBD, rather than an accident event. The following observations may be the reasons. Firstly, the chondronecrosis of epiphyseal growth plate of the KBD patients often occurs in the age of 4–15 years, resulting in impaired endochondral ossification and chondrocyte differentiation. During these periods, the complete fusion between the secondary ossification center of the odontoid process, and the odontoid process fusion with the body and the neural arches of axis occur. The above overlapping periods implicate that the lesions of KBD may also affect the epiphyseal growth plate in the atlantoaxial joint, and then resulting in abnormality. Secondly, earlier closure of the epiphyseal growth plate and disturbed endochondral ossification induces the growth retardation, while the chondronecrosis induces a scar formation, bony enlargements and osteophytes which reflect the secondary repair and remodeling of the adjacent joints [6]. In our patient, the diameters of odontoid process were small and irregular, while the size of axis body and anterior arch of atlas is comparatively larger. The resemble morphometry of typical characteristics of limbs in KBD and this atlantoaxial anomaly in this case shows that KBD may explain this pathogenesis.

Here, we reported a case of KBD patient diagnosed with atlantoaxial subluxation concomitant with separated odontoid process fused to the enlarged anterior arch of the atlas. To our knowledge, this is the first report concerning the radiological features of atlantoaxial joint in KBD patient. The reason of this anomaly is postulated to link to the histopathology of KBD. This associated atlantoaxial disorder develops slowly and clinically silent until a traumatic instability or the severe spinal cord compression occurs which results in acute myelopathy and loses the surgical chance, just as this patient. Therefore, whatever this anomaly is linked or accidental, even acquired or congenital in KBD patients, lager numbers of KBD candidates should be recruited to explore the morphometry of atlantoaxial joint, especially for the adolescents. Besides, to improve the quality life and socio-economic status, we recommend spinal surgeons to involve the research of the spinal anatomy and variation for the prevention and earlier therapy for KBD.

Compliance with ethical standards

Conflict of interest The authors declare that they have no competing interests.

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