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Torticollis Revealing Atlantoaxial Rotatory Subluxation in A Down Syndrome Patient

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Abstract | **Case Report**

Atlantoaxial rotatory subluxation (AARS) is a rare condition, more frequently observed in pediatric populations and individuals with predisposing factors such as congenital syndromes. Patients with Down syndrome (trisomy 21) are at increased risk of cervical spine instability due to ligamentous laxity and bony anomalies. We report the case of a 17-year-old male patient with a known history of Down syndrome and ventricular septal defect, who was hospitalized for infective endocarditis. During his hospital stay, he developed an acute torticollis. Cervical radiography, computed tomography (CT), and magnetic resonance imaging (MRI) revealed a Fielding and Hawkins type II atlantoaxial rotatory subluxation. No recent trauma or upper respiratory infection was reported. The patient was managed successfully with conservative medical treatment.

Keywords: Atlantoaxial – Rotatory – Subluxation – Down – Torticollis – Imaging.

INTRODUCTION

Atlantoaxial rotatory subluxation (AARS) is a rare cervical spine disorder characterized by abnormal rotation of C1 vertebra on C2, most often seen in children due to ligamentous laxity and immature bony anatomy [1,2]. While often associated with trauma or infection, it can also occur spontaneously, particularly in individuals with predisposing conditions.

Down syndrome is a known risk factor for atlantoaxial instability, attributed to ligamentous laxity, odontoid dysplasia, and hypotonia [3,4]. Although asymptomatic instability is relatively common in this population, symptomatic AARS remains rare and underdiagnosed [5].

We report a case of an atraumatic Fielding and Hawkins type II AARS in a 17-year-old patient with Down syndrome, diagnosed during hospitalization for infective endocarditis, and managed successfully with conservative treatment.

OBSERVATION

A 17-year-old male patient with a known history of Down syndrome and a congenital ventricular septal defect was admitted to our hospital for treatment of infective endocarditis. During his hospital stay, he developed an acute onset of atraumatic torticollis, characterized by painful neck stiffness and abnormal head posture without any reported recent trauma or upper respiratory tract infection. Physical examination revealed limited range of motion of the cervical spine with a characteristic "cock-robin" head position. Neurological examination was unremarkable, with no signs of spinal cord compression or radiculopathy.

A cervical spine radiograph was initially performed. It showed subtle asymmetry of the atlantoaxial joint with slight malalignment of C1 relative to C2. No fractures or gross bone abnormalities were noted *(Figure 1)*. The radiograph suggested possible atlantoaxial instability but was not definitive, warranting further imaging.

Subsequently, a cervical computed tomography (CT) scan with multiplanar reconstructions confirmed a rotatory anterior and left displacement of C1 relative to C2, with subluxation of the right inferior articular facet of C1 measuring 3.6 mm, consistent with a Fielding and Hawkins type II classification.

There was also an increased anterior atlanto-dens interval measuring up to 6 mm. The cervical spine demonstrated straightening without evidence of other alignment abnormalities *(Figure 2)*. Magnetic resonance imaging (MRI) was performed and showed C1–C2 subluxation involving the right inferior articular facet of C1, with rotatory displacement of this facet anterolaterally to the left relative to C2. Associated posterior displacement of the odontoid process compresses the transverse ligament, which appears thickened and stretched, without visible discontinuity. A focal T2 hyperintensity was noted in its left portion. These findings result in a reduction of the bony spinal canal diameter at this level, measuring 8 mm at its narrowest point, without abnormal spinal cord signal. *(Figure 3).* Given the absence of neurological deficits, the patient was managed conservatively with analgesics, cervical immobilization using a rigid collar, and close clinical follow-up.

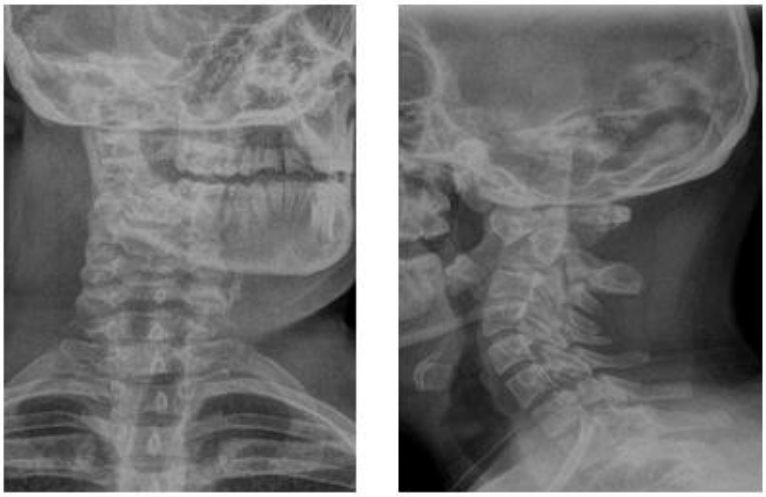

Figure 1: Cervical spine radiographs (anteroposterior and lateral views).

There is subtle asymmetry of the atlantoaxial joint with slight malalignment of C1 relative to C2. No fractures or gross bony abnormalities are observed.

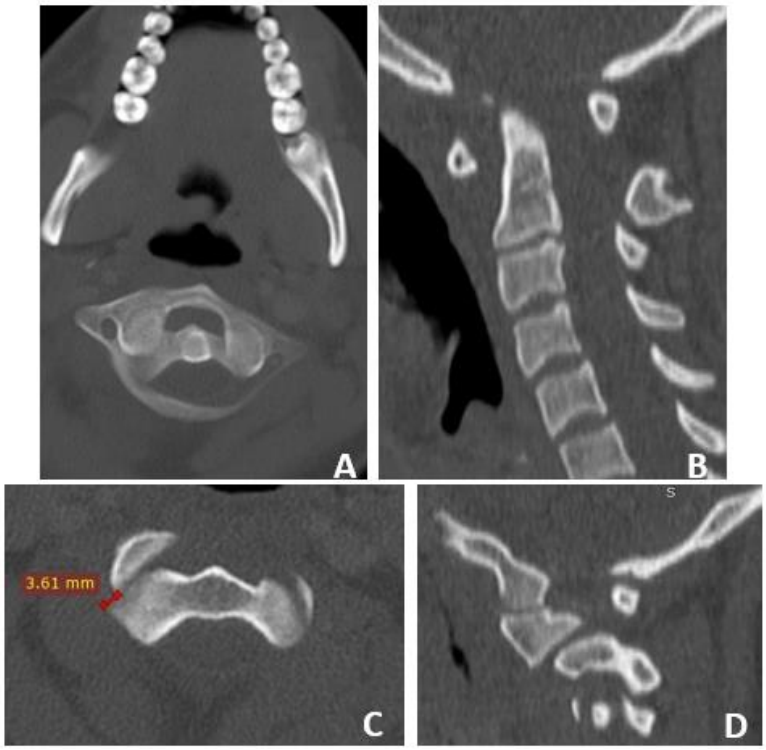

Figure 2: Axial (A, C) and sagittal (B, D) CT images showing a subluxation of the right inferior articular facet of C1 measuring 3.6 mm, consistent with a Fielding and Hawkins type II classification

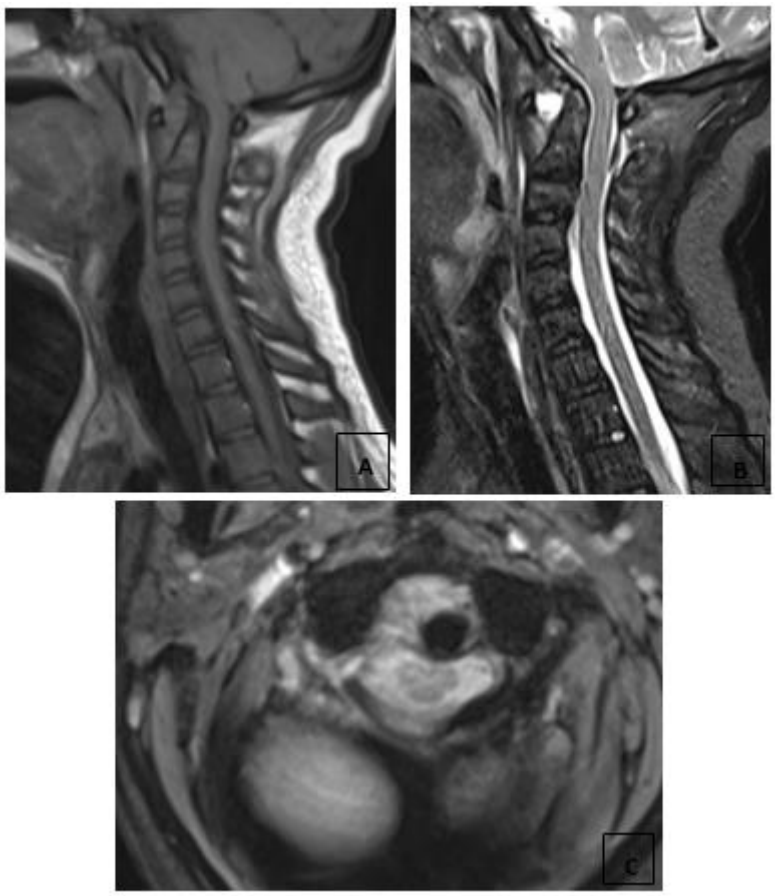

Figure 3: Cervical spine MRI in sagittal T1, sagittal T2, and axial T2-weighted images (A, B, C) showing C1–C2 posterior displacement of the odontoid process which compresses the transverse ligament, that appears thickened and stretched, along with a focal T2 hyperintensity on its left portion. The bony spinal canal is narrowed at this level (minimal diameter: 8 mm), without spinal cord signal abnormality

DISCUSSION

Atlantoaxial rotatory subluxation (AARS), also referred to as atlantoaxial rotatory fixation, is an uncommon but potentially serious condition presenting as acute torticollis (1,2). It may arise spontaneously or secondary to trauma or congenital malformations of the cervical spine [3]. The underlying mechanism often involves forced rotatory movements causing abnormal fixation or limited rotation of the C1–C2 joint, which may or may not be accompanied by dislocation [3]. Given the proximity of the atlantoaxial joint to the medullo-cervical junction and vertebral arteries, prompt recognition is crucial to avoid neurological complications.

AARS is classified by Fielding and Hawkins into four types based on the degree and direction of odontoid displacement relative to the atlas facet [6]. Type II, as observed in our patient, involves rotatory subluxation with anterior displacement of the atlas and an increased atlanto-dens interval, typically exceeding 3 mm.

This pathology predominantly affects children, who are predisposed due to unique anatomical and biomechanical features of the pediatric cervical spine. These include the horizontal orientation of the C1–C2 articular facets, greater rotational range (>45°), ligamentous laxity, and underdeveloped neck musculature [5,7,8]. Inflammatory conditions or minor trauma can trigger AARS in this susceptible population [9,10].

Patients with Down syndrome have an increased risk of atlantoaxial instability due to congenital ligamentous laxity, odontoid hypoplasia or dysplasia, hypotonia, and abnormal joint anatomy [3,4]. These factors contribute to greater joint mobility and predispose to both asymptomatic instability and, more rarely, symptomatic subluxation such as AARS. Although atlantoaxial instability is relatively common in Down syndrome, symptomatic AARS remains infrequently reported, underscoring the importance of clinical vigilance in these patients presenting with torticollis [11].

Imaging plays a pivotal role in the diagnosis and classification of AARS. While conventional radiographs can provide initial clues such as asymmetry or malalignment at the C1–C2 level, they have limitations due to overlapping structures and difficulties in

visualizing subtle subluxations [12]. In this context, CT scanning with multiplanar and 3D reconstructions is the gold standard, allowing precise evaluation of the rotational displacement, measurement of atlanto-dens intervals, and classification according to Fielding and Hawkins [6,13]. Dynamic CT may also assist in assessing reducibility and joint stability.

MRI complements CT by assessing soft tissues, including ligaments, joint capsules, and the spinal cord, and is essential for excluding spinal cord compression or edema [14]. MRI is particularly important in Down syndrome patients, who are at risk for both mechanical instability and neurological injury.

Timely diagnosis based on clinical suspicion and imaging allows for appropriate conservative management, which generally includes cervical immobilization with collars or traction and analgesia. Most pediatric cases, including ours, respond well to non-surgical treatment, with a favorable prognosis and low risk of neurological sequelae [2,9,15]. Surgery is reserved for persistent instability, neurological deficits, or recurrent subluxations.

CONCLUSION

In conclusion, atlantoaxial rotatory subluxation (AARS) is a rare but important differential diagnosis of acute torticollis, especially in pediatric patients with predisposing factors such as Down syndrome. The presence of ligamentous laxity and craniovertebral junction anomalies in these patients increases the risk of cervical instability. Accurate and timely diagnosis relies on cross-sectional imaging, particularly CT for classification and MRI for assessing soft tissue and neurological involvement. Early conservative treatment, as in our case, generally yields favorable outcomes. Clinicians and radiologists should maintain a high index of suspicion in trisomic patients presenting with neck pain or abnormal head posture, even in the absence of trauma.

Conflicts of interest

Authors declare no conflict of interest.

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