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CASE REPORT

Isolated Idiopathic Cervical Dystonia with a Rare Type of Atlantoaxial Dislocation. A Case Report and a Short Literature Review

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Introduction: Dystonia is a prevalent movement disorder characterized by involuntary muscle contractions, and cervical dystonia is among the most common forms. This report presents a rare case of severe isolated cervical dystonia in a young male patient that progressed to type IV atlantoaxial rotatory subluxation, highlighting the clinical presentation and diagnostic challenges associated with this condition.

Case Description: A 27-year-old male patient presented with abnormal head posture and severe cervical pain. Clinical examinations revealed severe cervical muscle spasms and a unique head posture, as if he was looking at the tip of his right shoulder. Radiological evaluation revealed a type IV atlantoaxial rotatory subluxation. Despite initial attempt of close reduction under anesthesia, recurrence of the subluxation occurred, and surgical intervention was necessary.

Conclusion: This case underscores the importance of thorough clinical evaluation and imaging in the diagnosis of isolated cervical dystonia, particularly given its potential to present with atypical features. These findings suggest that early recognition and intervention may be critical for preventing complications and improving patient outcomes.

Keywords: dystonia, cervical dystonia, atlantoaxial rotatory subluxation, movement disorder, surgical intervention, clinical diagnosis

Introduction

Dystonia is a movement disorder characterized by prolonged or intermittent muscle contractions. Dystonic contractions cause abnormal, often repetitive, movements, postures, or both.[1] Dystonia is the third most common movement disorder after Parkinson's disease and essential tremors.[2] Dystonia can be classified into three main types based on the affected body region: generalized dystonia affecting the entire body; segmental dystonia affecting adjacent regions; and focal dystonia limited to a specific area, such as the neck, hand, or voice.

Dystonia is more prevalent in the cervical region than in other parts of the body including the eyes and upper limbs.[3] A recent estimate suggests that the incidence of cervical dystonia is 1.18 per 100,000 person-years. Cervical dystonia is often isolated without neurological abnormalities.[4] Cervical dystonia is often isolated without neurological abnormalities.[1] The severity of cervical dystonia varies from mild twisting and pulling to extremely powerful and painful torticollis. Despite its common occurrence, the specific pathophysiological mechanisms that lead to rapid symptom progression in isolated cervical dystonia, and particularly to severe complications such as atlantoaxial rotatory subluxation, are still not well understood. This report presents a severe form of isolated cervical dystonia in an adult patient, whose symptoms evolved over a short period, leading to a rare type of rotatory C1-C2 subluxation.

Case Description and Clinical Course

A 27-year-old male patient presented with a history of abnormal head posture for five weeks associated with severe posterior cervical pain. The patient denied any history of trauma, fall, or recent fever, and had no family history of similar complaints. On clinical examination, the patient's head was tilted and rotated to the right. The patient's head was positioned as if he was

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looking at the tip of his shoulder, requiring him to turn his upper torso to the left to maintain a relatively straight gaze while walking. He demonstrated a jerky horizontal head tremor with approximately two to three head oscillations, followed by rest for a few minutes. Severe spasms of the posterior cervical, trapezium, and sternocleidomastoid muscles were observed on the ipsilateral side. The cervical spine was fixed, and the head position could not be improved by either passive stretching or light touching of the chin. Cranial and peripheral nerve examinations were normal. Radiography revealed that the cervical spine was turned and tilted to the right (Figure 1A and B). CT scan of the coronal images showed asymmetry between the odontoid process and lateral mass of the atlas (Figure 2A). Sagittal views showed incongruency of the C1-C2 joint (Figure 2B), and in the axial cuts, showing no fracture of C1 or the Denis (Figure 2C). CT reconstruction showed type IV atlantoaxial rotatory subluxation according to the Fielding and Hawkins classification (Figure 3).[5] His blood test results, including complete blood count and inflammatory markers, were within the normal ranges. Due to atlantoaxial subluxation, the patient underwent a closed reduction trial in the operating theater. Fiber optic intubation was performed because of the patient's head and neck posture. Upon administration of muscle relaxants, a significant improvement in the head and neck posture was observed. The

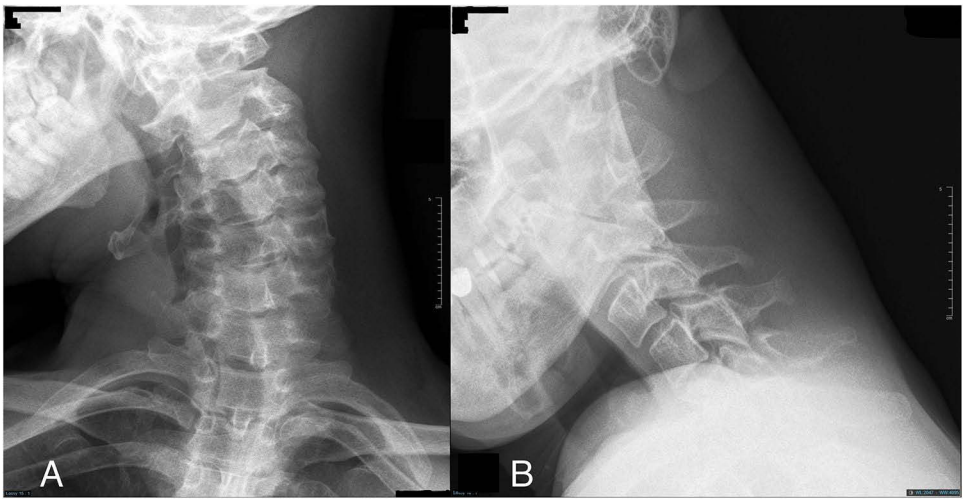

Figure 1 Cervical x-ray (**A**) Anteroposterior view. (**B**) Lateral view.

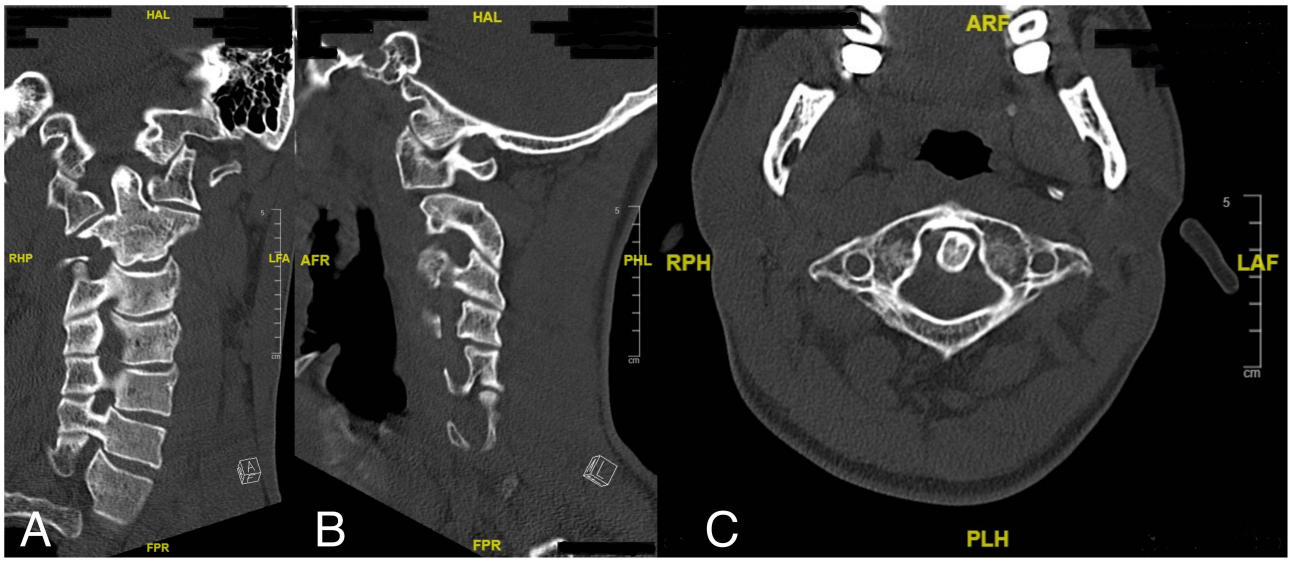

Figure 2 CT scan images. (**A**) Coronal view showing asymmetry between the odontoid process and lateral mass of the atlas. (**B**) Sagittal view showing incongruency of C1-C2 joint. (**C**) Axial view showing no fracture of C1 or tip of the Denis.

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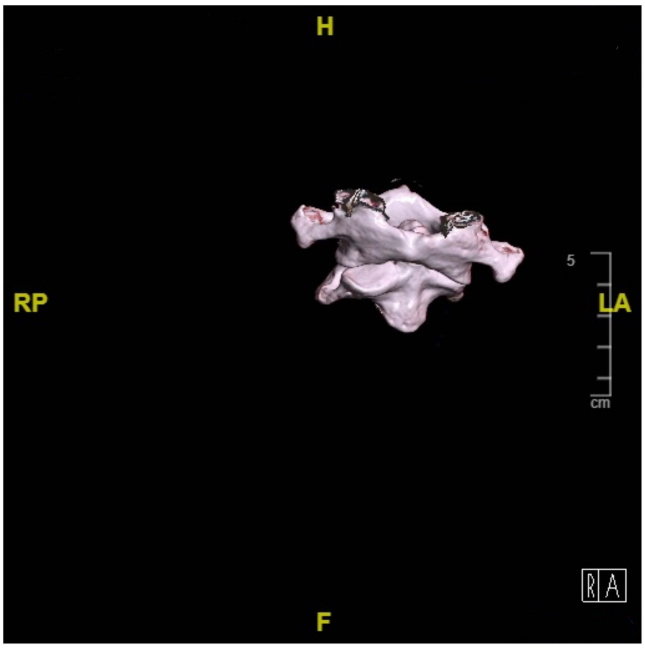

Figure 3 CT reconstruction Image showing Type IV Atlantoaxial subluxation.

surgeon applied gentle traction with a skull clamp and used their index finger to carefully press on the body of C2 through the oropharynx, which led to a corrective "popping" sound indicating reduction. The reduction was confirmed using an image intensifier (Figure 4A and B). The patient was subsequently placed for neck immobilization. On the first postoperative day, the patient exhibited spasm in the posterior cervical muscles with slight tilting of the head and neck. Analgesia and muscle relaxants were also administered. On the second day, severe spasm of the neck muscles with recurrence of the abnormal head and neck posture was noted. Repeated CT scan revealed recurrence of atlanto-axial subluxation. Multidisciplinary consultations were conducted with neurology, ophthalmology, and otolaryngology departments. Clinical and radiological evaluation did not reveal any other pathologies. The patient was diagnosed as having isolated cervical dystonia. Owing to the persistence of subluxation, the patient was advised about the potential need for surgical intervention. The patient was scheduled to undergo surgery. Under general anesthesia, close reduction was performed using a Mayfield skull clamp, and the reduction was confirmed intraoperatively using an image intensifier. The patient was positioned prone on a spine table and underwent

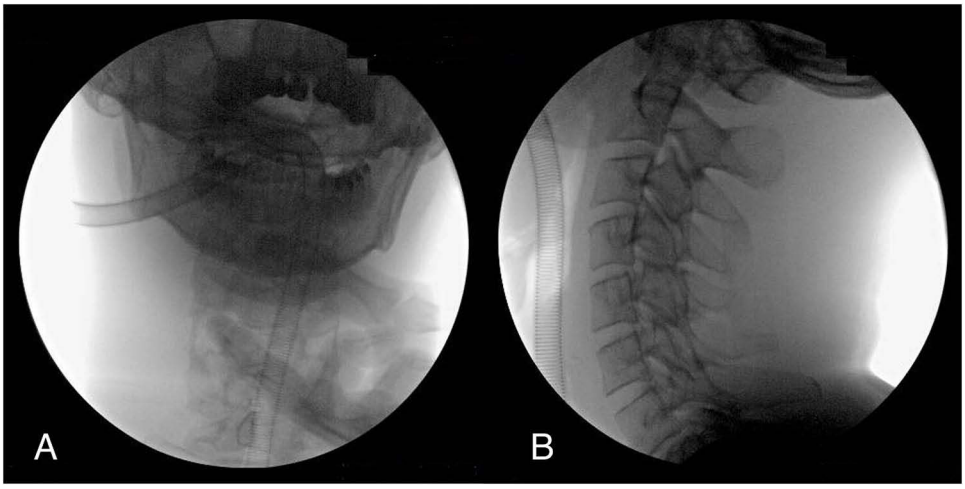

Figure 4 Intraoperative images showing reduced C1-C2 joint. (**A**) lateral view. (**B**) anteroposterior view.

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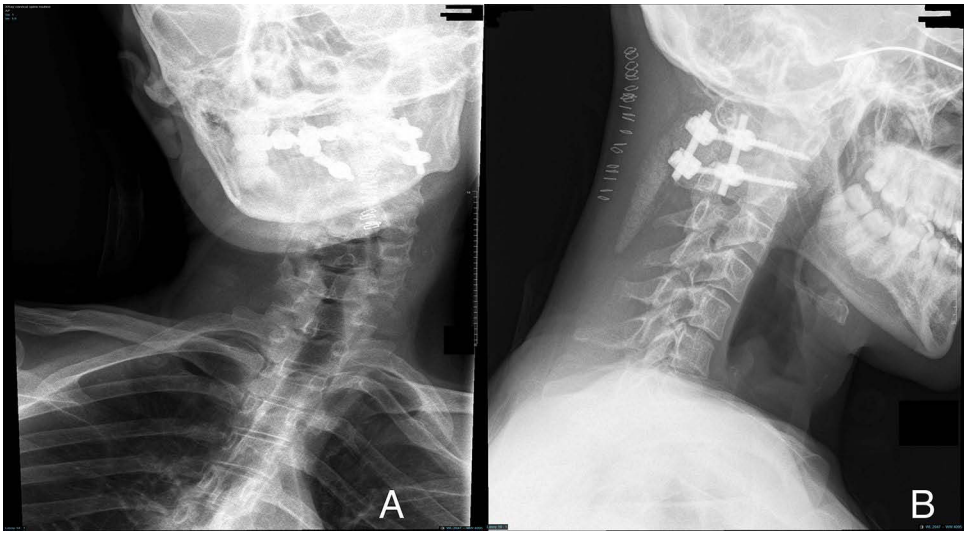

Figure 5 Post operative x-ray. (**A**) Anteroposterior view. (**B**) Lateral view.

posterior C1-C2 fixation (Figure 5A and B). Postoperatively, the patient remained neurologically intact and was immobilized on a rigid Philadelphia cervical collar. The patient was discharged on the fourth postoperative day and visited the outpatient clinic within 10 days with a clean surgical wound and improvement in his neck posture. Botulinum neurotoxin injections were scheduled on an outpatient basis.

Discussion

This report describes a young adult male patient with isolated cervical dystonia and type IV atlantoaxial rotatory subluxation, without compressive myelopathy. Atlantoaxial rotatory subluxation is often considered to mimic isolated cervical dystonia, given its potential etiologies such as trauma, inflammation, or infection.[6] However, in this patient, none of these causal factors could be identified.

Cases of idiopathic isolated cervical dystonia with atlantoaxial subluxation have been previously reported.[7–9] In previous reports, the patients were in the third and fifth decades of their age, with symptoms of cervical dystonia developed over three to six years before presenting with symptoms of compressive cervical myelopathy. In addition, in previously reported cases the head and neck posture was described as "cock robin" deformity in which the head is tilted to one side and turned in the other side. In the literature, the neck posture in rotatory atlantoaxial subluxation is commonly described as "Robin listening to the ground for worms" in which the head is in a forward position and the cervical spine is slightly flexed and rotated to either side. This metaphor is used in relation to atlantoaxial subluxation because the most common type is type II, and the head and neck are perceived as such. However, patients with isolated cervical dystonia have been observed to exhibit atypical head and neck position. The use of terms such as torticollis or spasmodic torticollis is no longer deemed accurate. In pediatric cases, the term torticollis is associated with a diverse range of conditions, ranging from benign to life-threatening, including infections and brain tumors. Recently, the Collum- caput concept has been introduced which relies on identification of those muscles acting on either the cervical spine, the head, or both that lead to a distinctive head and neck posture. Accordingly, 11 different subtypes has been identified in isolated cervical dystonia.[10] Based on both CT scan and MRI, it had been shown that in 20% of those patient presented with lateral flexion and rotation, the muscles acting on the Atlanto occipital joint are affected. Consequently, the clinical presentation can be variable.

Heterogeneities in clinical presentations, criteria, and mimics have led an expert panel to establish a consensus on the clinical features, terminology, and diagnostic criteria for isolated cervical dystonia.[6] This consensus provides clinicians with guidelines for accurate diagnosis, addressing the challenges posed by clinical heterogeneity, and aiding in distinguishing cervical dystonia from mimics. The gold standards for diagnosis are patient history, clinical features, and phenomenology. Two key clinical features are crucial for alleviating maneuvers and pain. Alleviating maneuvers, such as sensory tricks,

improve dystonic posture in less than half of patients.[11] Pain primarily in the neck and shoulders may extend to the head, upper back, and ipsilateral upper limbs. Head tremor, often overlooked, is present in 75% of cervical dystonia cases and manifests as involuntary head movements. These tremors can appear in various directions without affecting other body regions,[12] and may indicate early stages of cervical dystonia. Early intervention can potentially improve these tremors before full disorder manifestation.[13]

While dystonia is generally considered a neurodegenerative disorder, its course is usually perceived as progressive, similar to that of other movement disorders. Recently, perception of the progressive course of cervical dystonia has been challenged. A total of 100 patients with idiopathic cervical dystonia were followed up over a period of 17.5 ± 11.5 years, and two distinctive types were identified based on onset.[14] Type one characterised by a gradual onset over approximately four years, with a constant course of disease before they reach a plateau phase. In contrast, type two has a rapid onset over a few weeks. Despite the rapid progression of symptoms in type two, the majority of patients experienced substantial remission. The observed disparity in the onset and progression patterns underscores the critical importance of early diagnosis and individualized treatment strategies for each variant of the condition. This differentiation in disease progression patterns highlights the need for tailored therapeutic approaches and long-term monitoring to effectively manage both variants of this condition. The necessity for multidisciplinary consultations in complex cases of cervical dystonia underscores the need for integrated care models that are currently lacking, particularly in patients presenting with atypical symptoms and complications. Managing cervical dystonia requires a range of approaches, with botulinum toxin injection being the primary treatment method. Botulinum toxin injections have been established as a safe and effective treatment for cervical dystonia with significant improvements in patient outcomes. However, 5–25% of patients fail to respond to treatment.[15] In cases where botulinum toxin fails, other medical treatments such as anticholinergics and muscle relaxants may be considered, although their efficacy is less established. Surgical intervention can be beneficial for patients who do not respond to conservative treatment. Deep brain stimulation has emerged as an effective neurosurgical treatment for those cases of refractory cervical dystonia that is not responding to medical treatment, with a strong evidence base. A recent meta-analysis, which included 39 articles with a total of 208 patients who received deep brain stimulation for refractory cervical dystonia with a mean follow-up of 23 months, demonstrated significant improvement in the Toronto Western Spasmodic Torticollis Rating Scale, severity, disability, and pain when measured at short- and long-term follow-up.[16]

Owing to its nature, complications in the cervical spine have a major impact on the patient's quality of life. Complications, such as compressive myelopathy, vertebral subluxation, and fixed bony deformities, warrant surgical management. Finally, the primary aim of management is not to cure the underlying condition but rather to alleviate symptoms and reduce complications associated with the cervical spine.

Conclusion

Cervical dystonia, affecting the neck region, is the most prevalent form of dystonia, a common movement disorder. Accurate diagnosis relies heavily on comprehensive medical history and thorough clinical assessment. A collaborative approach involving multiple medical specialties is crucial for ruling out conditions that mimic cervical dystonia. Understanding the progression and manifestations of isolated cervical dystonia is key to implementing effective treatment strategies and enhancing patient outcomes.

Ethical Statement

This paper has been obtained from Institutional review board in Imam Abdulrahman bin Faisal University (IRB-2024-01-314).

Consent for Publication

The patient has given consent to participate as well as consent to publish the data.

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Disclosure

The author declares no conflict of interest.

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