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Case report: C1/2 rotational instability progressing to extreme subaxial hyperkyphosis in an adolescent with severe catatonia

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ABSTRACT

Introduction: Autism spectrum disorder (ASD) is characterized by deficits in social communication, repetitive behaviors, and can be accompanied by a spectrum of psychiatric symptoms, such as schizophrenia and catatonia. Rarely, these symptoms, if left untreated, can result in spinal deformities.
Research question and case description: This case report details the treatment of a 16-year-old male ASD patient with catatonic schizophrenia and mutism, presenting with neck pain, left-rotated torticollis, and fever. MRI revealed atlantoaxial rotational instability and spinal cord compression from a dislocated dens axis. After inconclusive biopsies, empirical antibiotics, hard collar and halo fixation treatment, persistent instability necessitated C1/2 fusion. The ongoing catatonia was addressed with electroconvulsive therapy. Concurrently, he developed severe subaxial hyperkyphosis. The report examines the decision-making between conservative and surgical management for an adolescent with significant psychiatric comorbidity and progressive spinal symptoms against a backdrop of uncertain etiology.
Materials and methods: A case report and review of the literature.
Results: Posterior C1-C7 stabilization was successfully executed, effectively restoring cervical sagittal alignment, which was maintained throughout a two-year follow-up. Concurrently, the catatonia resolved.
Discussion and conclusion: To our knowledge, this is the third reported case of severe cervical deformity associated with fixed posture in a psychiatric patient. This case report emphasizes the critical importance of multidisciplinary collaboration in managing the interplay between neuropsychiatric disorders and severe spinal deformities. It showcases the practicality and efficacy of surgical intervention for persistent cervical deformity in pediatric schizophrenia patients, highlighting the necessity for a comprehensive risk-benefit analysis.

1. Introduction

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by deficits in social communication and interaction, along with restricted, repetitive patterns of behavior, interests, or activities (American Psychiatric Association and Association, 2013). ASD patients may experience psychiatric symptoms and motor abnormalities, including schizophrenic symptoms and catatonia (Chen et al., 2000; Pappa and Dazzan, 2009; Hauptman et al., 2023). While very rare, repetitive movements or fixed postures have previously been reported to cause spinal deformities, if left untreated (Hacioğlu et al., 2011; Saito et al., 2013). Here, we report an adolescent patient with ASD who experienced an episode of catatonic schizophrenia, presenting with atlantoaxial instability, followed by a sustained fixed posture eventually resulting in severe subaxial hyperkyphosis.

Abbreviations: ASD, Autism spectrum disorder; MRI, Magnetic Resonance Imaging; ECT, Electroconvulsive therapy; ACDF, Anterior cervical discectomy and fusion; TcMEPs, Transcranial motor evoked potentials; DSM-V, Diagnostic and Statistical Manual of Mental Disorders Fifth Edition.

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2. Case presentation

A 16-year-old male patient with ASD, hospitalized at our pediatric psychiatry department for a catatonic episode with mutism and refusal to eat, presented to our pediatric spine unit with fever, neck pain, and left-rotated torticollis. No sensorimotor symptoms were present. His history included two years of anxiety, compulsive behavior, and psychotic symptoms. Inflammatory markers and thyroid antibodies were elevated, raising suspicion of Hashimoto's encephalitis. Therefore, he underwent plasmapheresis and received high-dose intravenous steroids. Magnetic Resonance Imaging (MRI) revealed progressive atlantoaxial instability, elongation of the transverse ligament of the atlas (C1), cranialization of the dens axis, and myelon compression (Fig. 1).

Given the prior steroids and plasmapheresis, an infectious etiology was considered, and a transoral dens biopsy was performed. In the same procedure, to address the atlantoaxial instability, repositioning was performed, and halo fixation was installed and left in place for six weeks (Fig. 2). Two of three biopsies yielded upper airway pathogens; however, these were considered contamination. Blood cultures were negative. Following the removal of the external fixation, C1/2 instability persisted. After interdisciplinary consultation, three days after halo removal, he received a second biopsy with an additional three weeks of halo fixation and a two-week course of amoxicillin/clavulanic acid. The second biopsy revealed no pathogens. Three weeks later, while the root cause remained unknown, the patient underwent definitive C1/2 fusion due to the continued C1/2 instability (Fig. 3 B).

Three months later, radiographs showed stabilization of the C1/2 joint with early indications of fusion (Fig. 3 C–D). Concurrently, the patient's catatonia slightly improved through weekly electroconvulsive therapy (ECT) sessions and oral antipsychotic medication with clozapine. Inflammatory markers progressively decreased. However, the patient consistently maintained his neck in a hyper-flexed position, and intensive physiotherapy yielded only modest improvements in this regard. Despite some overall progress, the patient developed a severe subaxial hyperkyphosis. Radiographs confirmed a stable C1/2 joint, but with a C2–C7 Cobb angle of 85° (Fig. 3 D and Fig. 4A–C).

Given the risks associated with the severe kyphosis, we considered renewed halo-vest installation, collar immobilization, halo-gravity-traction and fusion, multi-level anterior release with anterior cervical discectomy and fusion (ACDF), either with or without posterior fusion, and direct posterior fusion. The chosen approach was C1-C7 posterior fusion, due to the rapid development of kyphosis and expected flexibility under anesthesia. Consent was also obtained for the option of anterior release and ACDF. The patient was positioned prone, with the head secured in a Gardner-Wells fixator. Transcranial motor evoked potentials (TcMEPs) were monitored. Repositioning from 90° to 21° kyphosis was achieved using halo traction with a 5kg weight for 30 minutes. Therefore, no anterior release was necessary. Ponte osteotomies at the C2-3, C3-4, and C4-5 levels were performed. Massa lateralis screws were placed at C3-5, and pedicle screws at C7, achieving firm purchase. The final alignment achieved was 7° lordosis between C2 and C7. No alterations in TcMEPs occurred. A cross-link was added after repositioning, and the osteotomies were augmented with autologous cancellous bone to promote posterior fusion (Fig. 5).

Six months postoperatively, cervical sagittal alignment was stable without adjacent segment instability, even though the catatonic episodes and fixed posture of both the body and neck initially remained. Two years later, the catatonic symptoms had notably decreased, and psychiatric care transitioned to an outpatient setting. The patient had no spine-related symptoms, and radiographs demonstrated consistent cervical sagittal alignment with unchanged Cobb angles and robust posterior fusion (Fig. 6).

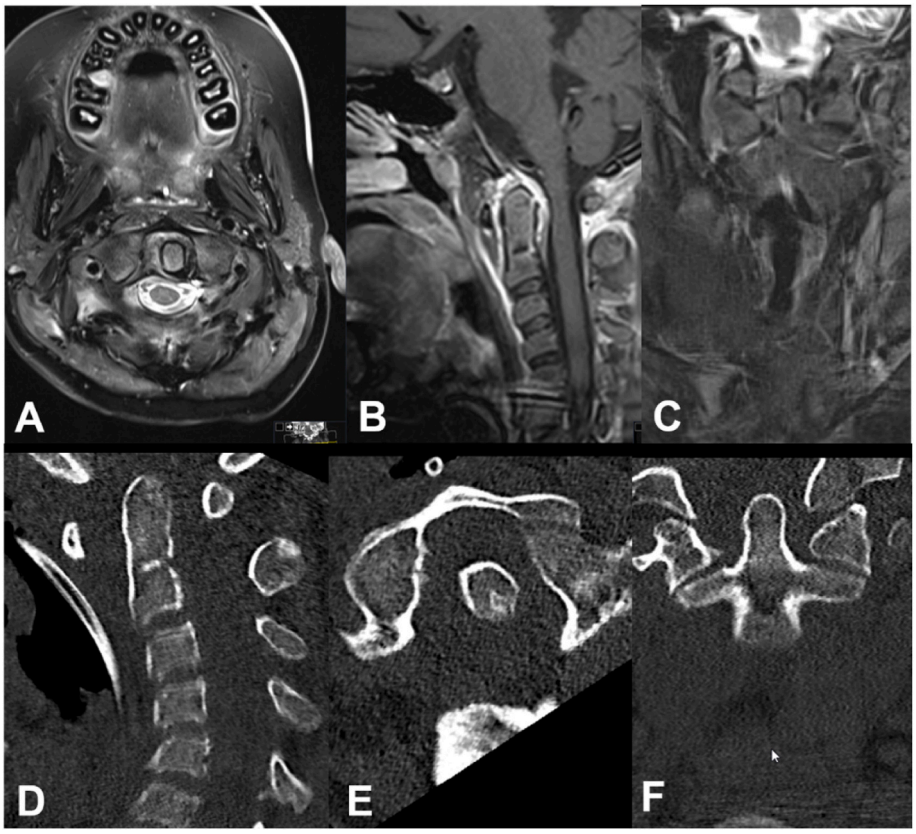

Fig. 1. A axial B sagittal C frontal T2-weighted MRI view of the dens axis at initial presentation, exhibiting dens cranialization, elongation of the transverse ligament of the atlas, and perifocal edema D sagittal E axial F coronal CT.

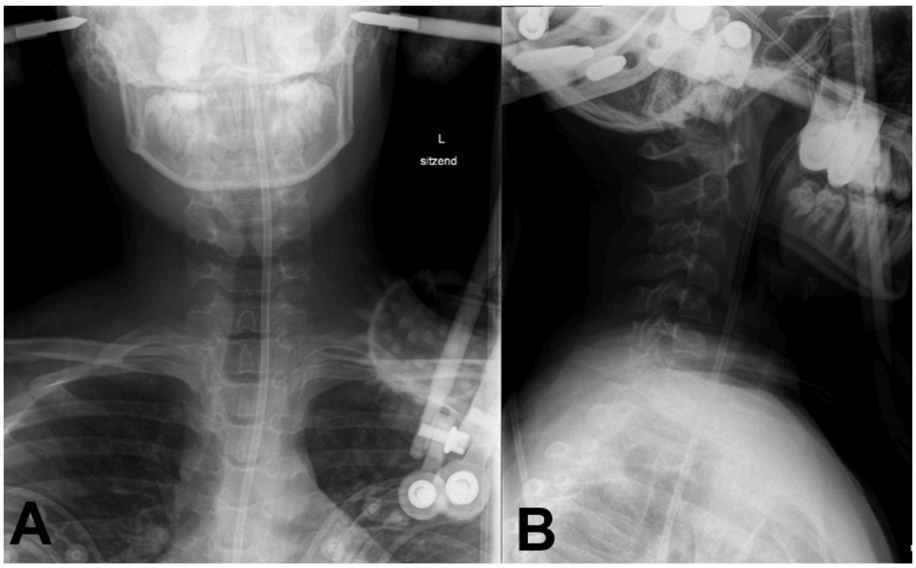

Fig. 2. Sitting lateral and ap radiographs following halo fixation.

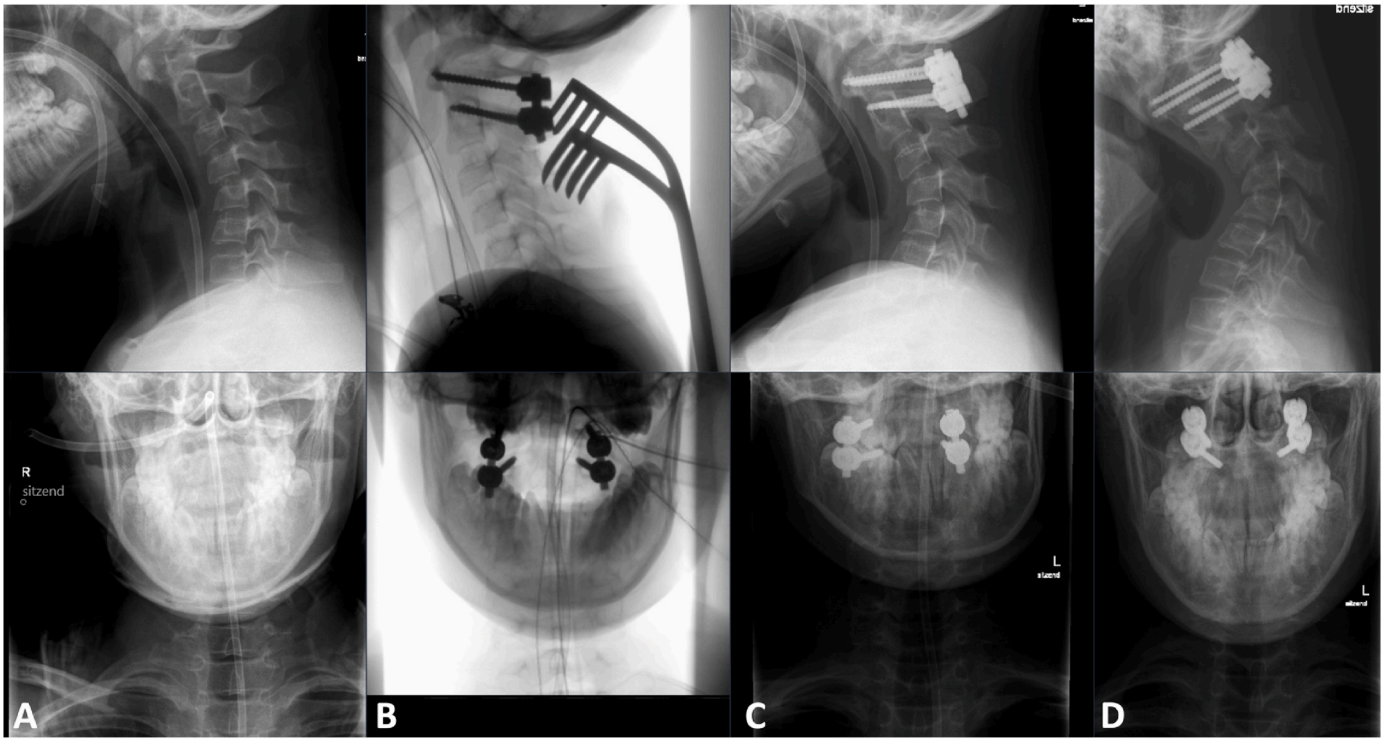

Fig. 3. A Sitting lateral and ap radiographs of the cervical spine with persistent atlantoaxial instability after repeated halo fixation B intraoperative fluoroscopy after C1/2 stabilization C radiographs one month postoperative and D four months postoperative show progressive cervical kyphosis.

3. Literature review and discussion

Cervical hyperkyphosis secondary to psychiatric disorders is exceptionally rare. In this case, a patient with ASD and concurrent catatonic schizophrenia developed severe cervical hyperkyphosis due to a persistently hyper-flexed neck posture, requiring surgical intervention.

The precise relationship between the catatonic episodes and the psychiatric history is unclear. The patient met all criteria for catatonic schizophrenia as per the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-V) (American Psychiatric Association and Association, 2013), which may relate to the history of ASD. There is a well-documented link between ASD and catatonic schizophrenia (Waris et al., 2013; Ghaziuddin et al., 2021), and distinguishing between causative and consequential relationships in these conditions can be difficult (Saito et al., 2011). Alternatively, the episode might be attributable to suspected autoimmune encephalitis, which is associated with catatonia and neuronal dysfunction (Flanagan et al., 2023). There are reports of Hashimoto's encephalitis presenting with catatonia in psychiatric patients, leading to potential misdiagnoses where catatonic symptoms are attributed to psychiatric disorders (Tsai et al., 2021; Johnson et al., 2022). However, given the patient's extensive psychiatric history and the lack of response to plasmapheresis, this hypothesis appears less probable.

The inflammation and atlantoaxial instability may have resulted

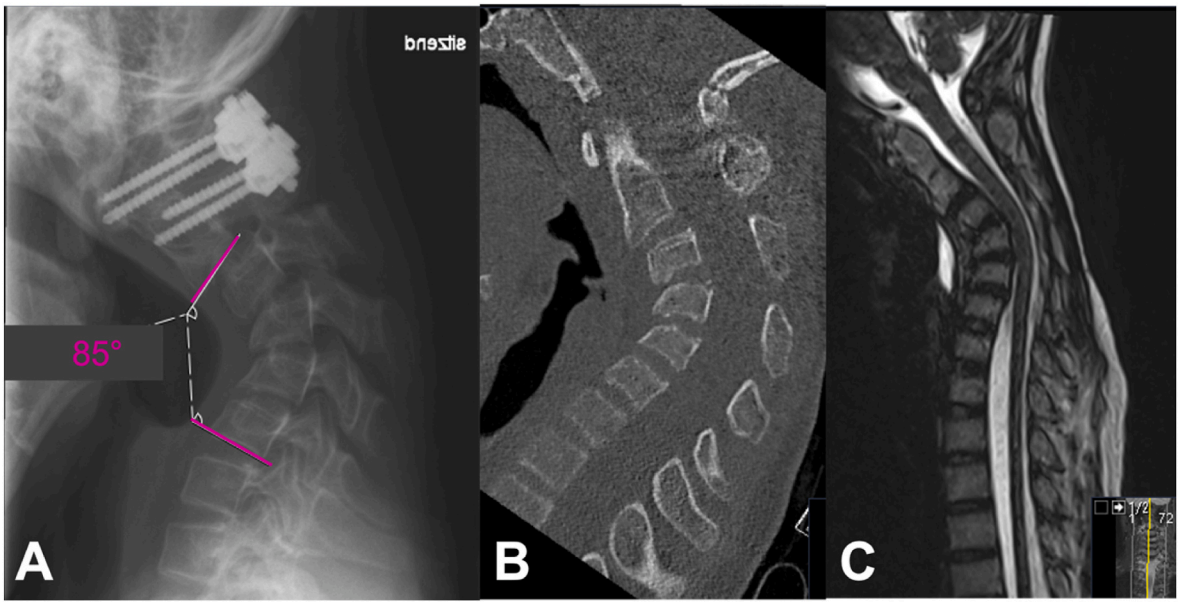

Fig. 4. A Lateral plain sitting radiograph of the cervical spine with a C2-C7 Cobb angle of 85° B sagittal CT view C sagittal T2-weighted MRI view (all imaging after 8 weeks of halo fixation).

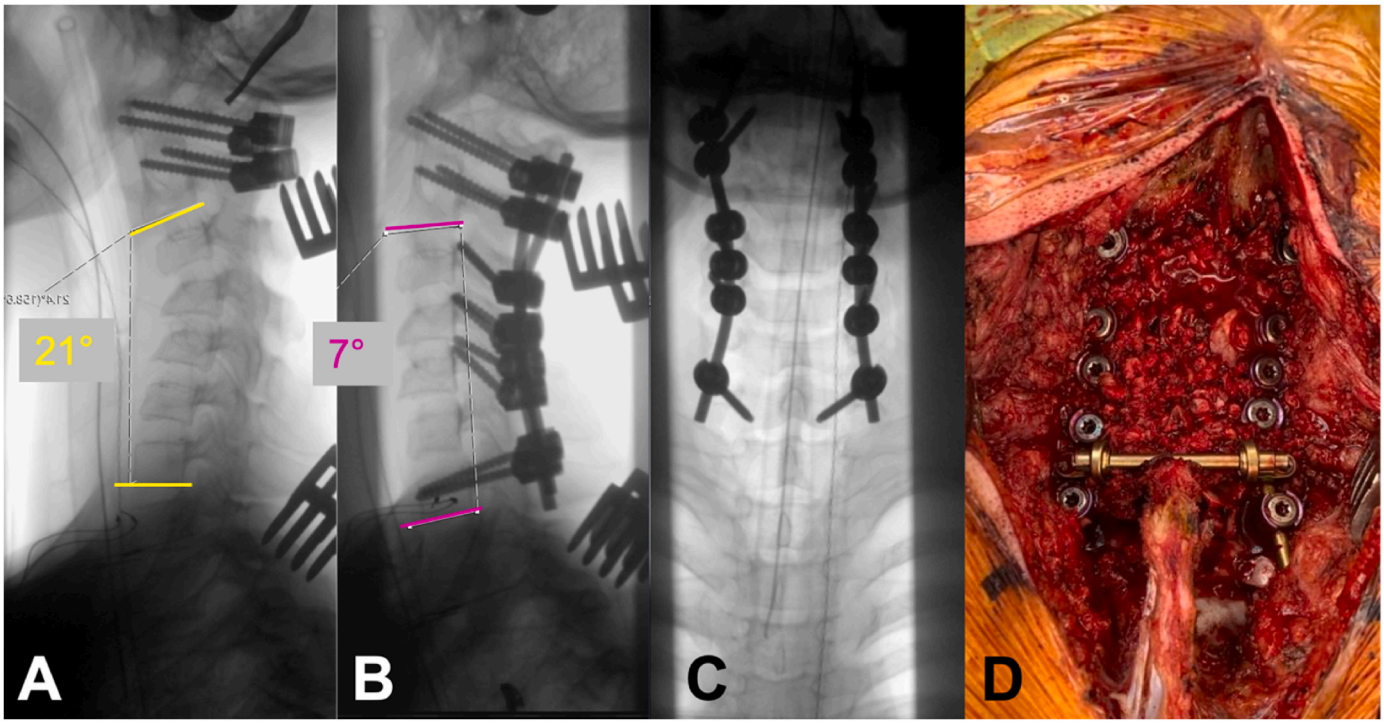

Fig. 5. A intraoperative lateral view of the cervical spine with C2-C7 Cobb angle correction to 21° kyphosis B after dorsal stabilization with correction to 7° lordosis C ap view D surgical field after completed posterior stabilization.

from infectious or autoimmune processes, triggered by either suspected Hashimoto's encephalitis or its treatment, namely plasmapheresis and high-dose intravenous steroids. Plasmapheresis can temporarily impair the immune system, leading to bacterial colonization and post-infectious autoimmune reactions in the dens axis region (Lu et al., 2019). The detection of upper respiratory pathogens in the patient's dens biopsies supported the hypothesis of an infectious etiology. Cases of infectious and autoimmune processes causing atlantoaxial instability are well documented (Dagtekin et al., 2011; Kerolus et al., 2015), with infectious etiologies such as Grisel syndrome being a known complication following head and neck infections and otolaryngological procedures. However, it is also plausible that contamination from transoral access was responsible for the positive bacterial cultures.

Though no definitive guidelines exist, pediatric inflammatory atlantoaxial instability is typically managed conservatively (Kerolus et al., 2015), with surgery reserved for refractory cases (Clark et al., 1988; Pilge et al., 2011; Yamazaki et al., 2008). Considering the patient's age, comorbidities, and uncertain etiology, an initial conservative approach was justified. In retrospect, surgery might have been a better option following the initial halo-fixation. The ultimately performed

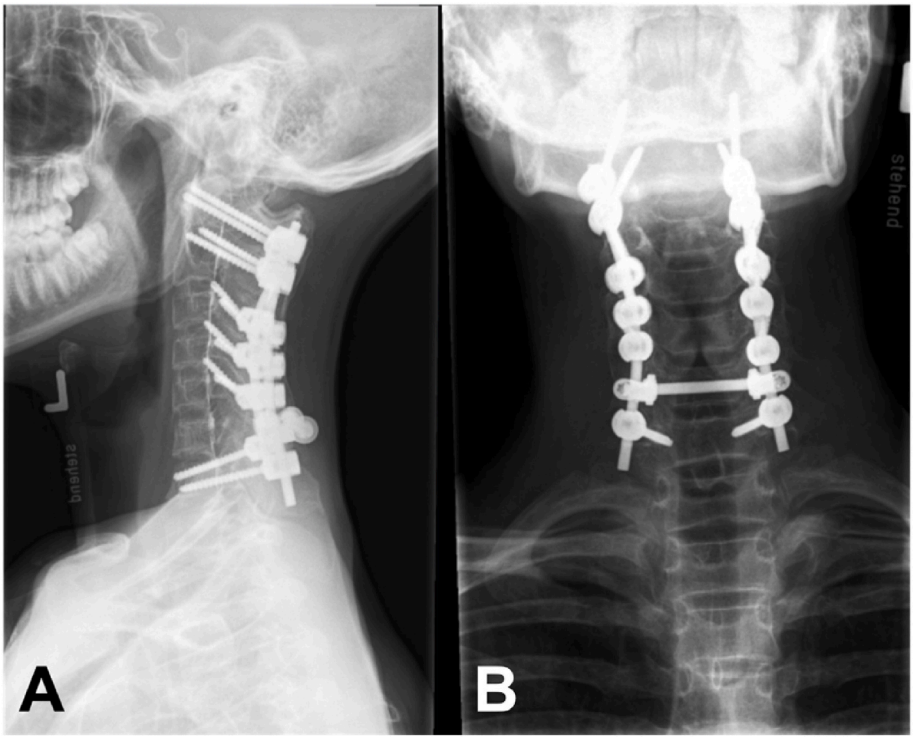

Fig. 6. Sitting ap and lateral plain radiographs two years after posterior C2-7 stabilization.

C1/2 fusion is a viable treatment for atlantoaxial instability in pediatric and adolescent patients (Abou-Madawi et al., 2021), achieving stabilization and early signs of fusion within three months.

Although there was a slight improvement in the catatonic state and related posture, they both likely played a role in the later development of severe cervical hyperkyphosis. While rare, complications such as contractures and joint deformities arising from postural disorders in psychiatric conditions have been documented (Frykman et al., 1983; Mashimo et al., 1995; Srivastava et al., 2008; Devi et al., 2011). However, cases comparable to this are exceedingly scarce.

On June 4, 2023, a comprehensive literature review was conducted across PubMed, Google Scholar, and Medline. The search terms employed included 'deformity', 'hyperkyphosis', 'psychosis', 'schizophrenia', and 'catatonia', ensuring a broad retrieval of related, cited, and citing publications. This search was unrestricted by publication date or language. The literature reveals two reported cases of cervical deformity associated with schizophrenic or catatonic symptoms (Table 1).

Hacioğlu et al. described a schizophrenia patient who, consistently keeping her neck bent forward for 2 years as reported by her daughter, developed severe cervical kyphosis and degenerative changes at C5-6 and C6-7, attributed to persistent neck flexion posture rather than organic causes. After failed physiotherapy, these changes were deemed severe and irreversible, leaving anti-inflammatory medication as the only viable treatment option (Hacioğlu et al., 2011). Saito et al. described an 18-year-old schizophrenic patient who developed cervical hyperkyphosis with a C2-C7 Cobb angle of 100° from permanently sitting with a hyper-flexed neck against a wall. They hypothesize that the hyperkyphosis resulted from the posture caused by the psychiatric symptoms, rather than the schizophrenia in and of itself (Saito et al., 2011, 2013). This case bears similarity, supporting the notion that fixed postures due to psychiatric conditions can lead to severe cervical deformity. However, in our case, this was compounded by prolonged treatment for atlantoaxial instability. The exact interplay between schizophrenia, catatonia, and postural disorders remains uncertain. Two factors in schizophrenia – neuromotor dysfunction with rigidity, increased muscle tone, and dyskinetic movements (Chen et al., 2000; Northoff, 2004; Edwards and Bhatia, 2012), and impaired proprioception with a diminished pain response (Merskey et al., 1962; Singh et al., 2006; Bonnot et al., 2009) – may predispose individuals to such deformities. Under physiological conditions, pain from sustained postures prompts adjustment, a protective mechanism potentially impaired in schizophrenia (Hacioğlu et al., 2011).

As Law et al. proposed, early surgical intervention for cervical spinal deformity can achieve stabilization and correction, prevent neurological complications, and enhance quality of life (Law, 1959). While the management of cervical spinal deformity is not well-defined, progressive myelopathy is considered an absolute indication for surgery (Ames

Table 1
Existing cases with cervical deformity associated with psychiatric symptoms.

Author	Year	Age	Sex	Psychiatric diagnosis	Clinical presentation of the spine	Spinal deformíty	Intervention/ management	Outcome
Hacioğlu	2011	39	Female	Schizophrenia	Continuously flexed neck posture, no neurological symptoms	Severe cervical kyphosis, degenerative changes at C5-6 and C6-7 levels	Physiotherapy	Persistent psychiatric and spinal symptoms
Saito	2013	18	Female	Schizophrenia	Fixed flexion deformity of the neck measuring greater than 100°, hyperreflexia in all extremities but no further neurologic deficits	C2-C7 Cobb-angle 109° at rest; 78° with manual traction, suggesting a rigid cervical deformity	Posterior C2-C7 fusion combined with an anterior release	Resolution of psychiatric symptoms, stable residual kyphosis of 32° at C2-C7

et al., 2013). Severe pain, increasing kyphosis, and symptoms like difficulty swallowing or maintaining a forward gaze may also warrant intervention (Tundo et al., 2019). In psychiatric patients, surgical decisions are more challenging due to the risk of exacerbating existing conditions through added stress (Menendez et al., 2014). Considering the degree of hyperkyphosis, myelopathy risks, and the impact on the patient's social interactions, we opted for multi-level fixation.

Anterior, posterior, or combined approaches are employed in cervical deformity correction, with the choice guided by deformity flexibility and facet ankylosis. A fixed deformity without facet ankylosis requires an anterior approach, while facet ankylosis warrants both anterior and posterior interventions. For flexible kyphosis, correctable with extension imaging or traction, either a posterior or anterior approach can be effective (Steinmetz et al., 2007). In this case, the patient underwent direct posterior fusion. Upon intraoperative traction, flexibility was confirmed with the kyphosis reducing to a 30° C2-C7 Cobb angle on intraoperative radiographs (Fig. 4A). The choice of a posterior approach was supported by the existing access, and that adding the stabilization to the existing "anchor" of C1/2 was logical, rather than using an anterior plate/cage-system from C2-C7. Crossing the cervicothoracic junction (Th2) was considered but deemed unnecessary due to the patient's young age, overall spinal flexibility, and the absence of deformities in the thoracic and lumbar regions. At the two-year follow-up, no significant adjacent segment instability was noted.

Retrospectively, the treatment choice appears correct by the positive clinical and radiographic outcomes. Due to the rarity of such cases and alternative treatment options, the decision was reached following comprehensive interdisciplinary discussions, with the patient's family fully involved. A similar case by Saito et al. (2013) was identified, where they performed a posterior C2-C7 fusion combined with an anterior release due to preoperative facet ankylosis. Their case showed a similar outcome after two years, with stable residual kyphosis of 32° at C2-C7 (Saito et al., 2013).

4. Conclusion

This case is the third known instance of severe cervical deformity associated with fixed posture in a psychiatric patient, set against an uncertain etiology. It emphasizes the critical need for multidisciplinary collaboration to manage the complex relationship between neuropsychiatric disorders and significant spinal deformities. The case effectively demonstrates the practicality and success of surgical intervention in treating persistent cervical deformity in pediatric schizophrenia patients, highlighting the importance of a comprehensive risk-benefit analysis.

Informed consent

Written informed consent for the submission of this paper was obtained from the patient.

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Declaration of generative AI in scientific writing:

During the preparation of this work the author(s) used the large language modelGPT-4 (OpenAI Inc, San Francisco, USA) for spell and grammar checking of the final manuscript. After using this tool, the author(s) reviewed and edited the content as needed and take full responsibility for the content of the publication.

Ethical approval

The patient provided consent for the writing and publication of this case.

Author contributions

All authors significantly contributed to the development of this article and have approved the final version for publication.

Declaration of competing interests

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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