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CASE IMAGE

Pediatric basilar invagination: Unveiling a rare complication of inflammatory bowel disease unclassified

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This case report details the presentation of an 8-year-old boy with atlanto-axial rotatory dislocation (AARD) and basilar invagination, an exceedingly rare complication of inflammatory bowel disease unclassified (IBDU). Basilar invagination, an anomaly of the craniovertebral junction, results in the dens of the axis protruding into the foramen magnum.[1] It may be associated with concurrent abnormalities such as atlanto-axial dislocation, Chiari malformation, atlas occipitalization, Klippel-Feil syndrome, and atlanto-occipital hypoplasia, among others. To the best of our knowledge, this represents the first documented case reporting of basilar invagination as a life-threatening complication of IBD in children.

An 8-year-old boy was referred for rehabilitation treatment due to sudden, painless torticollis that appeared 2 months earlier without any apparent cause. There was no history of accidents, trauma, or upper respiratory tract infections reported. Initially the patient underwent evaluation at a local hospital. Neurological and otorhinolaryngological assessments yielded normal results. Brain and cervical spine magnetic resonance imaging (MRI), along with lumbar puncture procedures, did not reveal any pathologies. Psychological factors were considered, possibly related to the ongoing divorce of his parents. Consequently, the patient began psychotherapy and was prescribed muscle relaxants (guaifenesin) and non-steroidal anti-inflammatory drugs (NSAIDs) and received a referral for rehabilitation.

A physical examination conducted by a rehabilitation specialist revealed a head tilt to the left with rotation to the right, resembling a "cock-robin" position. Neck mobility was significantly restricted in both the frontal and transverse planes. Rotation and lateroflexion to either side were limited to 5 degrees, anteflexion to 30 degrees, and extension was completely restricted. The patient's mouth opening was limited to 2 cm. Palpation revealed a slight deviation of the transverse processes of the atlas to the right, bilateral spasms in the levator scapulae and trapezius muscles, as well as spasms in the right scalene and sternocleidomastoid muscles.

Eighteen months before the onset of torticollis, the patient was diagnosed with IBDU, manifested by recurrent sacroileitis, diarrhea, and weight loss. When the torticollis manifested, the patient was undergoing treatment with mesalazine and oral corticosteroids. In addition, his regimen had recently been augmented with budesonide and methotrexate to alleviate diarrhea and prevent further weight loss. Despite these measures, IBDU control remained suboptimal. The patient reported soft stools 2 to 3 times daily, along with elevated markers of inflammation, including C-reactive protein (CRP 20 mg/L) and erythrocyte sedimentation rate (ESR 40/70), as well as increased calprotectin levels in the stool (829 mg/kg). Nevertheless, no medical specialist who had evaluated

the patient previously considered the torticollis symptoms to be related to IBDU.

The patient was admitted to the rehabilitation department where he underwent physical therapy. This therapy, administered twice daily, included mobilization, soft tissue and muscle relaxation techniques, and muscle coordination training. In addition, the patient received a controlled multisensory environment therapeutic approach (Snoezelen) and aqua therapy thrice weekly. The patient was also prescribed NSAIDs (ibuprofen, 200 mg taken orally three times a day). Despite these rehabilitation efforts, the patient's symptoms persisted. Consequently, a detailed reanalysis of the cervical spine MRI was conducted. This examination revealed signs of spondylarthritis, notably signal alteration in the bone marrow near the occipital condyles and the adjacent part of the C1 articulation on both sides, more pronounced on the right. In addition, reactive edema in the surrounding tissues was observed. Inflammatory changes were also localized in the facet joints at levels T1, T2, and T3, accompanied by edema in the surrounding soft tissues.

It was only after these findings that a rheumatologist interpreted them as an extraintestinal manifestation of IBDU, leading to the initiation of biological treatment: adalimumab, 20 mg, administered subcutaneously every 14 days, and methotrexate, 15 mg, given subcutaneously once a week. A follow-up MRI conducted 3 months later revealed a partial reduction in the inflammatory signs. However, the study also showed progression in the fluid collection anterior to the dens of the axis, an increased gap between the dens of the axis and atlas, and signs of BI (Figure 1A). Computed tomography (CT) scans revealed a leftward rotational dislocation of the C1-C2 articulation and a ventral displacement of the C1 articular surface (Figure 1E). In addition, an upward displacement of the dens of the axis into the foramen magnum, indicative of basilar invagination, was detected (Figure 1D). As a result, physical therapy was immediately discontinued, and the patient was advised to rest, avoiding any traction or manipulation of the cervical spine. Surgical stabilization was subsequently performed using the Virage spinal fixation system on the C0-C1-C2 segments and a posterior fusion of C0-C2 with an autologous bone graft from the iliac crest (Figure 1C).

Six months after the surgery, and a year into the biological treatment, MRI revealed a significant reduction in edema and a normal alignment of the C1-C2 joint, with no signs of dislocation (Figure 1B). One-year post-stabilization, the metal implant was removed. The patient is currently in good condition, with the IBDU effectively controlled by the biological treatment. The head position is normal. However, movement in the cervical spine remains slightly limited due to the bone graft.

To the best of our knowledge, this is the first reported case of basilar invagination as a complication

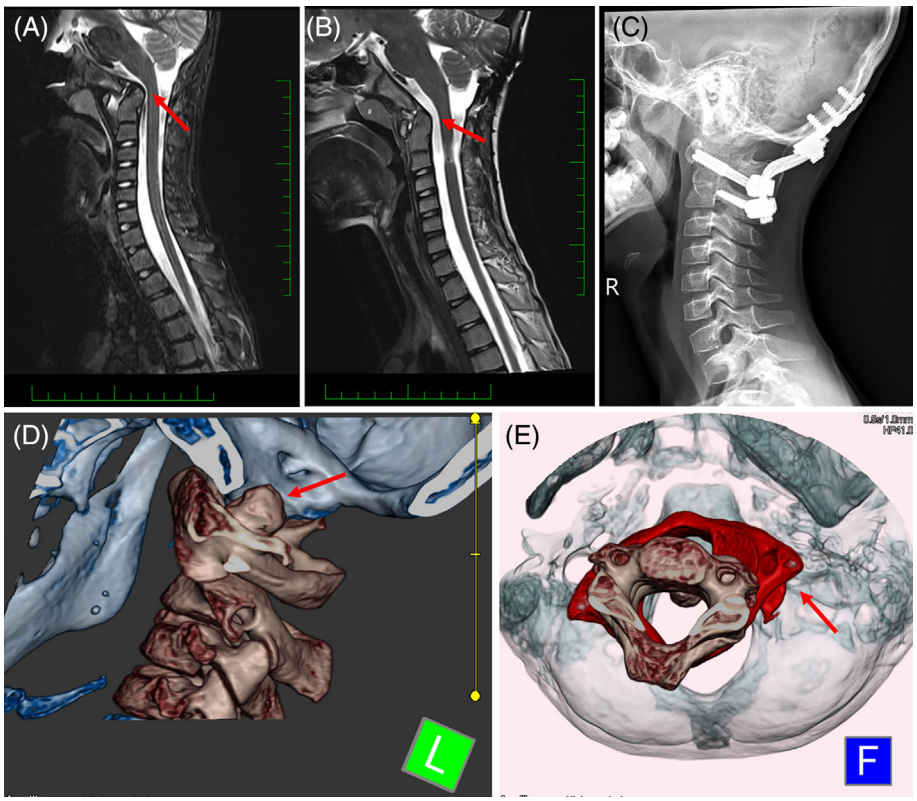

FIGURE 1 (A) Cervical spine magnetic resonance imaging (MRI) before treatment showing basilar invagination of the dens of the axis (red arrow). (B) Cervical spine MRI after the surgery, and a year into the biological treatment, MRI revealed a significant reduction in edema and a normal alignment of the C1-C2 joint, with no signs of dislocation (red arrow). (C) Surgical stabilization of the atlanto-occipital joint and the cervical spine. (D) Computed tomography (CT) scan before treatment revealing atlanto-axial rotatory dislocation of the atlas (red arrow). (E) CT reconstruction of the upper cervical spine before surgery demonstrating cranialization of the dens of the axis into the foramen magnum (red arrow).

TABLE 1 Previous reports of atlanto-axial dislocation associated with inflammatory bowel disease in children.

Authors	Age (years)	Sex	Duration of symptoms (days)	Associated disease	Treatment	Correction
Mahajan et al., 2001	9	M	7	Crohn's disease	Traction	Complete
Tauchi et al., 2013	17	M	385	Ulcerative colitis	C1-2 fusion	Partial
Bourghli et al., 2023	7	F	330	Crohn's disease	C1-2 fusion	Complete
Present study	8	M	196	IBDU	C0-C2 fusion	Complete

Note: F = female, M = Male.
Abbreviation: IBDU = Inflammatory Bowel Disease Unclassified.

of IBDU in a child. Basilar invagination has been documented as a complication of autoimmune diseases such as rheumatoid arthritis.[2] Synovitis and pannus formation in the cervical spine joints and ligaments can result in atlanto-axial instability. Ongoing erosions of the atlanto-axial joint may lead to the collapse of the C1 lateral masses, causing the cranial migration of the dens of the axis into the foramen magnum, resulting in basilar invagination.[3] We postulate a similar pathophysiological process in our patient, as IBD can induce arthritis, chronic inflammation, and erosive changes in the ligaments, thereby promoting joint instability, ligamentous laxity, and increasing the likelihood of torsion-related injury.[4]

Atlanto-axial dislocation has been reported in three pediatric patients with IBD.[5–7] Notably, none of these cases exhibited evidence of basilar invagination. A comparison between pediatric cases is presented in Table 1. The consideration of AARD and basilar invagination becomes crucial when a pediatric patient with IBD presents with torticollis and neck pain. Prolonged external orthotic immobilization is recommended to stabilize symptoms and prevent further invagination.[8] However, in our patient's case, rehabilitation was initially advised in this life-threatening condition with a risk of brainstem injury, cervical myelopathy, and sudden death.[3] Fortunately, a skilled therapist performed physical therapy, incorporating mobilization techniques, soft tissue and muscle relaxation techniques, and muscle coordination training. The therapist, recognizing the lack of improvement, insisted on further investigation, including imaging methods, which ultimately revealed the true cause, that is, AARD with basilar invagination.

This case underscores the critical importance of thorough examination and imaging methods in pediatric cases of torticollis associated with IBDU or similar conditions, preventing potential life-threatening consequences such as AARD and basilar invagination. It serves as a vital reminder for clinicians to exercise caution and pursue a comprehensive understanding of underlying conditions before embarking on rehabilitation interventions.

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DISCLOSURES
The authors declare no conflicts of interest.

ETHICS STATEMENT
The research proposal was approved by the ethics committee of The Second Medical Faculty, Charles University and University Hospital Motol, Prague, Czech Republic. Reference Number: EK-1247/23.

CONSENT FOR PUBLICATION
The patient's parents provided written informed consent for the publication of this anonymous case report.

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