



Case Report

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Coexistence of Chiari 1 Malformation and Marsh 3b Celiac Disease

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ABSTRACT

This case report presents the first simultaneous occurrence of Chiari malformation type 1 and celiac disease in a 27-year-old male patient. To the best of our knowledge, there are no previously published cases in the literature. This rare association suggests that nutrient malabsorption caused by celiac disease may exacerbate the symptomatic severity of Chiari malformation. We recommend screening for celiac disease in patients diagnosed with Chiari malformation who present with anaemia, vitamin deficiency or unexplained gastrointestinal complaints. Conversely, in the presence of atypical neurological symptoms in celiac disease, craniocervical magnetic resonance imaging should be performed and a multidisciplinary approach strategy should be considered in treatment.

KEYWORDS: Chiari Malformation Type I, Celiac disease, Malabsorption syndromes, Syringomyelia, Magnetic resonance imaging, Vitamin deficiency

INTRODUCTION

Chiari 1 malformation (CM1) is a craniovertebral junction anomaly characterized by the descent of the cerebellar tonsils below the level of the foramen magnum, and its etiopathogenesis is not fully understood (12).

Celiac disease is a chronic autoimmune enteropathy that causes villous atrophy in the small intestine and is triggered by the consumption of gluten-containing foods in genetically predisposed individuals (11). As a result of malabsorption syndromes, nutrient deficiencies such as iron, folate and vitamin B12 may occur, leading to systemic symptoms (8).

To the best of our knowledge, a search of the PubMed and Scopus databases using the English-language search terms "Chiari malformation AND celiac disease" and "Chiari AND malabsorption," without language restrictions, identified no case reports describing the coexistence of these two condi-

tions. Although neurological findings that can be seen in celiac disease, such as epilepsy, ataxia, peripheral neuropathy and myelopathy, have been extensively described in the literature (11), there is no clinical picture reported to occur simultaneously with Chiari malformation (CM). This indicates that the case we present is the first of its kind in the literature and highlights its unique contribution. This coexistence suggests potential connections between the pathophysiological mechanisms of both conditions and may offer new perspectives, particularly on the effect of malabsorption syndromes on neurological complications. Vitamin and mineral deficiencies caused by celiac disease may exacerbate the symptoms of CM1 and lead to a more complex clinical presentation (1,3).

CASE REPORT

A twenty-seven-year-old male patient presented to our clinic with long-standing numbness and pain in the right upper ex-

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tremity. Cervical magnetic resonance imaging (MRI) revealed loss of cervical lordosis, and both cerebellar tonsils had extended approximately 12 mm beyond the foramen magnum. There was expansion of the central spinal canal between C5 and T1, consistent with syringomyelia (Figure 1). The patient, whose findings were consistent with CM1, was operated on at an outside centre (Figure 2).

In the second postoperative month, the patient presented to the gastroenterology outpatient clinic with complaints of abdominal pain. Laboratory investigations revealed deficiencies in ferritin, folate, vitamin B12, vitamin D and iron. Ferritin: 7.4–14.1 µg/L (low; normal 30–400 µg/L), folate: 1.2–1.4 µg/L (low; normal 3.8–20 µg/L), vitamin B12: 211–343 ng/L (at the lower limit; normal 197–771 ng/L), 25-OH Vitamin D: 22–60 ng/mL (lower limit) and iron: 18.8 µg/dL (low; normal 45–158 µg/dL) were detected. Additionally, HbA1c, thyroid function tests, testosterone and cortisol levels were found to be within

normal limits. For diagnostic purposes, an esophagogastroduodenoscopy was performed and the mucosa of the second part of the duodenum was observed to have a scalloped appearance. The antrum mucosa was hyperemic and edematous; the pylorus was centralized and functioning. On retroflexion, the cardia and fundus mucosa were evaluated as normal, and the hiatus was observed to be mildly lax. Biopsies taken from the antrum and duodenum were reported to be compatible with celiac disease, and no additional pathology was detected on colonoscopy.

In the patient's biopsy examination, almost total villous atrophy was observed in the mucosa. There was an increase in intraepithelial lymphocytes, with a cellular count revealing 33 intraepithelial lymphocytes per 100 enterocytes. Additionally, accompanying crypt hyperplasia was noted. These findings were determined to be compatible with type 3b celiac disease according to the Marsh classification (Figure 3-5).



Figure 1: Preoperative cervical T2 weighted sagittal magnetic resonance imaging (MRI) scan.



Figure 2: Postoperative cervical T2 weighted sagittal magnetic resonance imaging (MRI) scan.

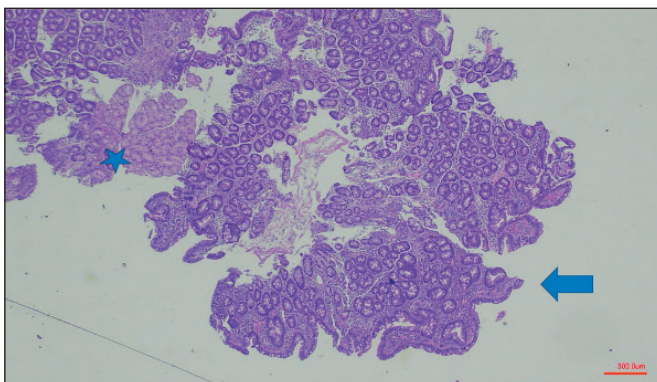


Figure 3: Villi with mild atrophy, star-shaped Brunner's glands (Hematoxylin and Eosin, x20).

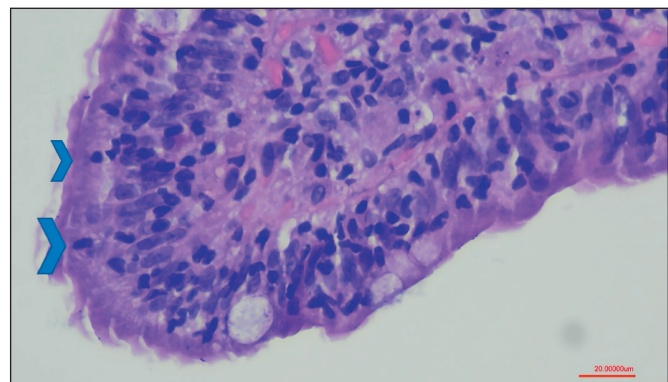


Figure 4: Increased lymphocytes in the villus (arrowhead indicates lymphocyte) (Hematoxylin and Eosin, x400).

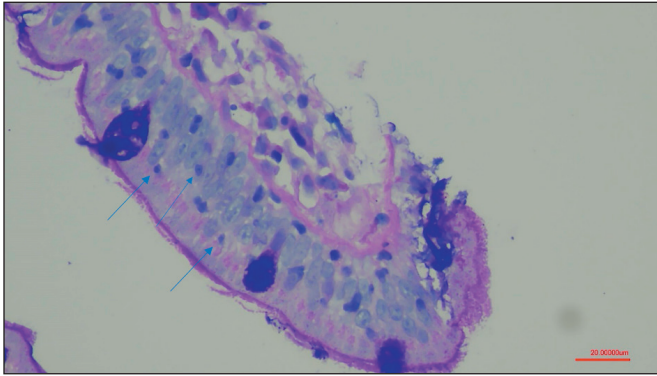


Figure 5: Increased lymphocytes in the villus (arrowheads indicate lymphocytes) (PAS, x400).

A gluten-free diet was initiated for the patient, and replacement therapy was provided with iron, folate, vitamin B12 and vitamin D. Following treatment, the patient experienced a marked improvement in abdominal symptoms, although neurological complaints partially persisted. The patient was enrolled in a regular follow-up program.

The patient's history and clinical course offer important clues for understanding the pathophysiological interactions between these two diseases. In particular, the patient's history of vitamin deficiency and its possible effects on the symptoms of CM1 are noteworthy in terms of the aetiology and management of neurological complications.

■ DISCUSSION

The coexistence of CM1 and celiac disease may share potential pathophysiological links, although this association may also occur coincidentally. Both conditions have been associated with autoimmune and connective tissue-related mechanisms. In celiac disease, chronic immune activation and systemic inflammation can lead to microvascular and neuronal damage, whereas in CM1, altered CSF dynamics and possible connective tissue weakness contribute to cerebellar tonsillar descent (4). Moreover, chronic malabsorption in celiac disease causes deficiencies in vitamins B12, E, D, folate and minerals, such as copper, which may independently cause neurological manifestations including peripheral neuropathy, ataxia and balance disorders, mimicking or exacerbating CM1-related findings. Therefore, it should be considered that some neurological symptoms in such patients may result from systemic metabolic and nutritional deficiencies rather than directly from CM (7).

CM1 is characterized by developmental/anatomical alterations of the craniocervical junction and disturbances in CSF flow dynamics. The current framework regarding classification and phenotypic variations has become significantly clearer in recent years (12,13). Particularly in symptomatic cases accompanied by syringomyelia, posterior fossa decompression

is a commonly utilized surgical treatment, and clinical and radiological outcomes may differ among techniques (10,13). Whether duraplasty is performed has been reported to affect short- and medium-term outcomes, but the choice may vary depending on the patient and anatomy (6). The variability in postoperative syringomyelia regression and rates of symptomatic improvement also highlight the importance of clinical follow-up (3).

In this study, the simultaneous diagnosis of celiac disease at the Marsh 3b level via duodenal biopsy and malabsorption in a patient with persistent neurological symptoms after CM1 surgery suggests possible systemic factors contributing to the symptom burden. Vitamin and mineral deficiencies caused by malabsorption due to celiac disease may increase the symptomatic severity of CM1; specifically, folate and B12 deficiencies could exacerbate the findings of peripheral neuropathy and myelopathy, while vitamin D deficiency may be associated with musculoskeletal pain and prolonged fatigue complaints (11). The potential of these deficiencies to intensify CM1-related symptomatology could provide a plausible biological basis for the resistant neurological findings observed in our case. In particular, investigating coexisting malabsorption/autoimmune diseases in patients for whom the expected clinical improvement after CM surgery is not achieved is critically important, not only to prevent unnecessary surgical interventions but also to inform the planning of symptomatic treatment (2,5,6).

In the literature, although the descent of the cerebellar tonsils more than 5 mm below the foramen magnum is typically considered CM1, it is emphasized that there are different pathologies that can lead to similar radiological findings. Park et al. reported that conditions such as posttraumatic craniocervical arachnoiditis, dural bands, spontaneous intracranial hypotension and idiopathic intracranial hypertension can mimic CM1 malformation. In our case, the presence of malabsorption syndromes, such as celiac disease, highlights that symptomatic assessment may be altered and underscores the need for such factors to be taken into account, especially when making surgical decisions (10,13). In cases diagnosed with CM and accompanied by malabsorption syndromes, such as celiac disease, we recommend that replacement and dietary therapies be initiated first and that the surgical decision be reconsidered based on the follow-up of neurological symptoms.

In light of these findings, it is recommended to screen for celiac disease in selected patients diagnosed with CM1 who present with clue symptoms such as unexplained abdominal pain or diarrhoea accompanied by anaemia and ferritin, folate/B12 vitamin or vitamin D deficiencies. The screening panel should include anti-tissue transglutaminase immunoglobulin A (IgA) antibodies, anti-endomysial IgA antibodies, anti-deamidated gliadin peptide IgA/IgG antibodies and total IgA level, and, if needed, diagnosis should be confirmed via duodenal biopsy (9). On the other hand, in celiac patients with unexplained or atypical neurological complaints, MRI to rule out craniocervical junction pathologies may also be of clinical value.

CONCLUSION

This case report highlights the rare but clinically significant coexistence of CM1 and celiac disease and underscores the necessity of an integrated and multidisciplinary approach in managing such comorbidities. In particular, systemic causes, such as celiac disease, should be excluded in patients who continue to experience symptoms after CM surgery. The presence of CM should be kept in mind in the differential diagnosis of atypical neurological symptoms in individuals with celiac disease. Furthermore, this case report demonstrates the importance of screening for celiac disease in CM patients presenting with gastrointestinal symptoms. For such complex cases, holistic treatment strategies that improve patients' quality of life should be developed through the collaboration of different disciplines such as neurosurgery, gastroenterology and nutrition expertise.

Declarations

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Availability of data and materials: The data presented in this study are available from the corresponding author on reasonable request.

Disclosure: The authors declare no conflict of interest.

Ethics approval and consent to participate: According to the authors' institutional policies, single-patient case reports that do not reveal identifying information do not require formal IRB approval.

All procedures were performed in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments.

Informed consent was obtained from the patient for publication of this case report and any accompanying images; a copy is available for review by the Editor.

AUTHORSHIP CONTRIBUTION

Study conception and design: TT, SO

Data collection: ACD, GA

Analysis and interpretation of results: EC, TT

Draft manuscript preparation: GA, TT

Critical revision of the article: TT, SO

Other (study supervision, fundings, materials, etc.): n/a

All authors (TT, GA, SO, EC, ACD) reviewed the results and approved the final version of the manuscript.

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