Hypoparathyroidism Meets the Unexpected: A Diagnostic Dilemma of Coeliac Disease and Fahr's Syndrome

Abdul Salam, Muhammad Ishaq Shah, Khalid Shahab and Hammad Ahmed Saif

Medical C Unit, Hayatabad Medical Complex, Peshawar, Pakistan

ABSTRACT

A 34-year man with long-standing recurrent seizures presented with complaints of lower limb swelling, weight loss, and stiffness in his shoulders and back. The patient had a history of recurrent idiopathic pericardial effusion, severe hypocalcaemia, and hypoalbuminaemia. Eventually, he was diagnosed with primary hypoparathyroidism and Fahr's syndrome, characterised by bilateral basal ganglia calcifications, which were responsible for his recurrent seizures. An extensive work-up ruled out other potential causes. However, despite ongoing treatment with calcium carbonate, calcitriol, and a protein-rich diet, the patient exhibited significant systemic complications caused by persistently low calcium and albumin levels. Against all odds, the authors uncovered coeliac disease responsible for his persistent hypoalbuminaemia and hypocalcaemia. This case underscores the complexity of hypoparathyroidism presentation and management because of vague and rare associations that make the clinical picture more complicated.

Key Words: *Hypoparathyroidism, Hypoalbuminaemia, Fahr's syndrome, Coeliac disease*.

How to cite this article: Salam A, Shah MI, Shahab K, Saif HA. Hypoparathyroidism Meets the Unexpected: A Diagnostic Dilemma of Coeliac Disease and Fahr's Syndrome. *JCPSP Case Rep* 2025; **3**:242-244.

INTRODUCTION

Hypocalcaemia manifests in many ways, from asymptomatic laboratory findings to severe life-threatening complications.[1] The most common cause of hypocalcaemia is hypoparathyroidism, either primary or secondary. Although widely known for its acute symptoms, *i.e.*, spasms and tetany, hypocalcaemia can also result in a variety of neurological, psychiatric, cardiac, and musculoskeletal manifestations in the longer run.[2-4] These vague manifestations can sometimes obscure the clinical picture, causing a diagnostic challenge.

A case of a 34-year man with severe, long-standing hypocalcaemia and hypoalbuminaemia, resistant to standard treatment and causing many unexpected complications is presented. This case highlights the complexity and necessity of a thorough clinical evaluation and multidisciplinary approach in such patients to reveal rare associations. Additionally, the authors discuss the occurrence of idiopathic musculoskeletal manifestations in this patient.

CASE REPORT

A 34-year-old man presented to the outpatient department complaining of lower limb swelling, weight loss, fatigue, apathy, and stiffness in his back and right shoulder.

Correspondence to: Dr. Khalid Shahab, Medical C Unit, Hayatabad Medical Complex, Peshawar, Pakistan
E-mail: dr.khalidshahab@gmail.com

Received: January 23, 2025; Revised: May 05, 2025; Accepted: May 25, 2025
DOI: https://doi.org/10.29271/jcpspcr.2025.242

His medical records showed a history of long-standing seizures and recurrent moderate-to-severe pericardial effusion (Figure 1), with no definitive cause found, for which he had been prescribed anti-epileptics and a course of steroids, respectively.

The patient's family history was unremarkable, and his past surgical history included bilateral cataract surgery.

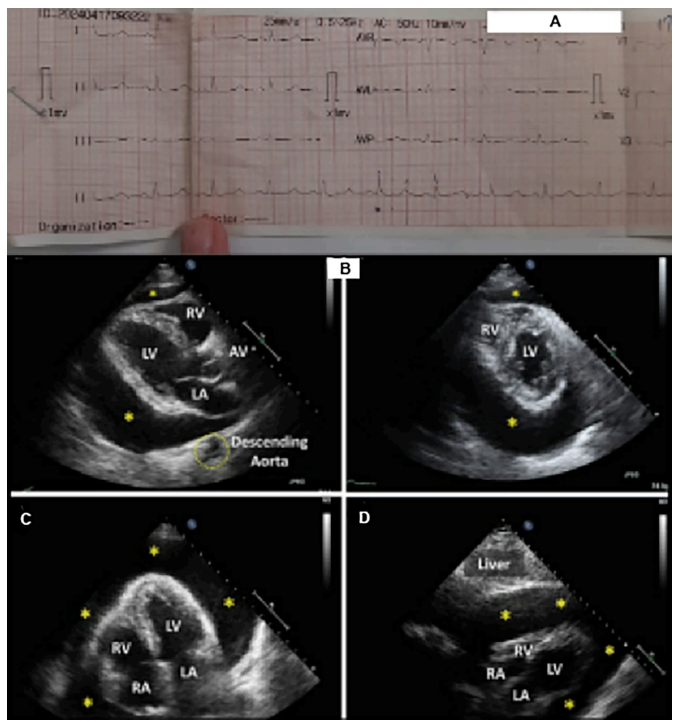

Figure 1: (A) ECG of the patient showing long QT-interval; (B-D) Echocardiographic images of the patient in different angles showing significant pericardial effusion (marked with asterisks).

On examination, he appeared lean, with darker skin complexion, normal conjunctivae and sclera, with a body mass index (BMI) of 18 kg/m^2, a blood pressure (BP) of 110/70 mmHg, a respiratory rate of 22 breaths/min, and a heart rate of 90 beats/min. Also, he exhibited stiffness in the dorsolumbar spine and right shoulder. Moreover, pitting-type oedema in both legs up to the knees was found, with clear lungs on auscultation and no visceromegaly.

During routine BP checking, an incidental finding of the Trousseau's sign led to further evaluation, which showed a positive Chvostek sign with severely low calcium levels (4.7 mg/dl), raised phosphate (6.33 mg/dl), low parathyroid hormone (4.14 pg/ml), and normal vitamin D levels (36.7 ng/ml), *i.e*. findings consistent with primary hypoparathyroidism. A CT of the brain revealed calcification in the bilateral basal ganglia (Figure 2A), suggesting Fahr's syndrome, with hypoparathyroidism to be a likely cause.

Despite prominent stiffness on physical examination, the MR imaging studies yielded no significant abnormalities (Figure 2B).

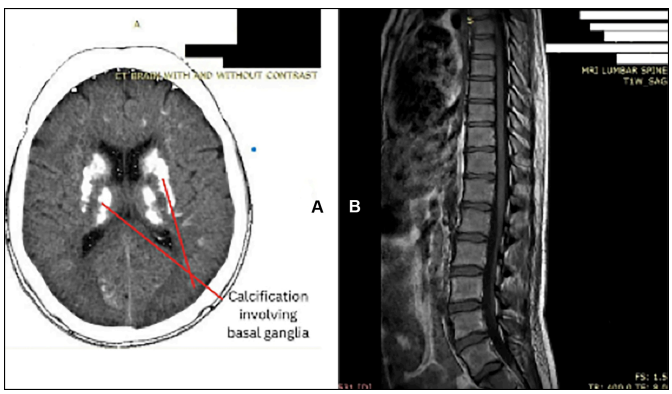

Figure 2: (A) CT brain showing calcifications in the bilateral basal ganglia; (B) MR spine showing normal radiological findings.

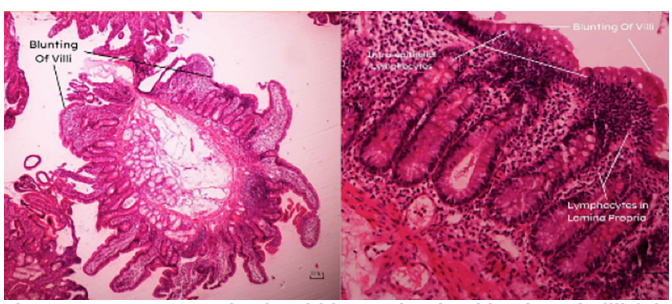

Figure 3: D2 segment duodenal biopsy showing blunting of Villi (A); Lymphocytic infiltration in the lamina propria, intraepithelial lymphocytes, and blunted villi (B).

In addition, his albumin levels (2.07 g/dl) were very low, so an extensive workup was done, including renal, liver, and thyroid function tests, with unremarkable results; however, some notable findings are mentioned in Table I. The mild proteinuria found was considered non-significant and unlikely to be solely responsible for low albumin levels. Suspecting malnutrition to be a contributing factor, the patient was advised to follow a protein-rich diet with supplemental calcium, calcitriol (400 IU/day), and B-proteins (40-50 g/day). However, no improvement was seen during the follow-up (July 2024). Thus, malabsorption was considered to be a possibility, and subsequently, investigations were performed (Table I), which were suggestive of coeliac disease, *i.e*., mucosal erosions, villous blunting, and intra-epithelial lymphocytes on D2 segment biopsy (Figure 3) (August 2024).

The patient was commenced on a gluten-free diet, and during follow-up (October 2024), there was a significant improvement in his albumin (6.2 g/dl) and calcium levels (8.1 mg/dl) along with improvement in his generalised stiffness and fatigue and BMI of 19.7 kg/m^2. Furthermore, no event of pericardial effusion or seizure activity has been reported yet.

Table I: Investigations.

Tests	Results	Reference ranges
Urinalysis	1+ proteinuria	
Urine albumin-to-creatinine ratio	60 mg/g	<30mg/g
24-hour urinary protein	504 mg/1200 ml/24 hour	<150 mg/1500-2000 ml/24 hour
Tests	**Results**	**Interpretations**
Stool fat staining	Positive	
Anti-TTG IgA	1.7 AU/ml	Negative <10 AU/mL
Anti-TTG IgG	<0.5 AU/ml	Negative <10 AU/mL
Oesophagogastroduodenoscopy	Antral gastropathy	

DISCUSSION

Hypocalcaemia, widely known for its acute symptoms, can also cause vague chronic manifestations as exhibited by this patient, posing a diagnostic challenge. The long-standing seizure history is eventually linked to Fahr's syndrome, a rare association of hypoparathyroidism.[2] Its pathophysiology remains elusive but is postulated to involve disruption of calcium- phosphorus metabolism, leading to calcium deposition in soft tissues, especially the basal ganglia.[2] Fahr's syndrome can present with a variety of movement disorders and behavioural symptoms, emphasising the need to consider hypoparathyroidism as a possible cause in such patients.

This patient had undergone bilateral cataract surgery 6 years before, suggesting undiagnosed hypocalcaemia at that time, as few cases reported in the literature show an association of posterior capsular cataracts with hypoparathyroidism.[5] The literature also links pericardial effusion to hypoparathyroidism.[3] Despite these manifestations, the patient's condition was overlooked until more prominent neurological symptoms developed, which led to his diagnosis of hypoparathyroidism. This is an unusual case, and the purpose of reporting this is to highlight the rare above-mentioned associations and complications of hypoparathyroidism to sensitise the medical fraternity.

Furthermore, the patient's spine stiffness mimicked ankylosing spondylitis, and a few cases have reported the association of hypoparathyroidism with syndesmophyte formation and ossification of the vertebral column and sacroiliac joints.[4] However, this patient exhibited no radiographic evidence of these changes, which is an odd feature. Moreover, his stiffness subsided with improving calcium levels, emphasising the potential role of calcium in musculoskeletal functioning.

The most perplexing feature of this case was the long-standing hypoalbuminaemia and hypocalcaemia, resistant to standard therapy and causing various complications throughout the patient's life. A duodenal biopsy confirmed the diagnosis of coeliac sprue.[6] However, a notable aspect was the negative serology for coeliac sprue, along with normal iron studies and vitamin B-12 levels and haemoglobin levels at the lower limit of the normal range, which caused a delay in diagnosis and management. This highlights the importance of tissue biopsy even in serology-negative patients to decide on a diagnosis of coeliac disease. Also, the need to keep a low threshold for such rare associations in these patients that can complicate the situation is highlighted.

The coexistence of hypoparathyroidism with coeliac sprue complicates the diagnosis and management, emphasising the need for a multidisciplinary approach for optimal patient care.

PATIENT'S CONSENT:
Written informed consent was obtained from the patient to publish this case report and accompanying images.

COMPETING INTEREST:
The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:
AS: Managed the patient and contributed substantially to the concept and design of the work.
MIS: Drafted the initial manuscript.
KS: Supervised clinical management and revised the manuscript.
HAS: Conducted literature review and helped in final editing.
All authors approved the final version of the manuscript to be published.

REFERENCES

1. Cherchir F, Oueslati I, Yazidi M, Chaker F, Mizouni H, Feki M, *et al*. Long-term complications of permanent hypoparathyroidism in adults: Prevalence and associated factors. *Endocrine* 2024; **84(3)**:1164-71. doi: 10.1007/s12020-024-03765-9.
2. Moushumi L, Rajarshi M. Primary hypoparathyroidism misdiagnosed as epilepsy - A case report: Seizures, hypocalcaemia and cerebral calcification. *EJIFCC* 2014; **25(2)**: 195-8.
3. Riesenberg K, Pick N, Levy I, Borer A, Schlaeffer F. Pericardial effusion accompanying isolated hereditary hypoparathyroidism. *Isr Med Assoc J* 1999; **1(3)**:194.
4. Okazaki T, Takuwa Y, Yamamoto M, Matsumoto T, Igarashi T, Kurokawa T, *et al*. Ossification of the paravertebral ligaments: A frequent complication of hypoparathyroidism. *Metabolism* 1984; **33(8)**:710-3. doi: 10.1016/0026-0495(84)90209-9.
5. Liao X, Huang X. Characteristic cataract associated with idiopathic hypoparathyroidism. *J Clin Exp Ophthalmol* 2016; **07(04)**. doi: 10.4172/2155-9570.1000588.
6. Kumar V, Valeski JE, Wortsman J. Celiac disease and hypoparathyroidism: Cross-reaction of endomysial antibodies with parathyroid tissue. *Clin Diagn Lab Immunol* 1996; **3(2)**:143-6. doi: 10.1128/cdli.3.2.143-146.1996.