Diaphragmatic Plication for Brown-Vialetto-Van Laere Syndrome: A Rare Presentation with Diaphragmatic Paralysis

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ABSTRACT

Brown-vialetto-van laere syndrome (BVVLS) is a rare autosomal recessive neurodegenerative disorder characterised by ponto-bulbar palsy and sensorineural hearing loss. Diaphragmatic paralysis is an uncommon manifestation of BVVLS, and there is limited literature on this association. Furthermore, there is no literature regarding diaphragmatic plication on BVVLS with diaphragmatic paralysis. The authors report a 17-year male diagnosed with BVVLS at the age of 12 years. In September 2023, the patient presented with complaints of widespread gas-related issues and dyspnoea. Computerised tomography showed an elevated left diaphragm with no additional findings. Fluoroscopy suggested a possible diaphragmatic paralysis. The patient underwent a uniportal videothoracoscopic left diaphragmatic plication. The chest tube was removed on the second postoperative day, and the patient was discharged on the third postoperative day. Diaphragmatic paralysis without trauma is a rare manifestation of BVVLS. This case emphasises the need for increased awareness of potential diaphragmatic involvement in BVVLS patients, as early diagnosis and intervention can provide symptomatic relief.

Key Words: *Brown-vialetto-van laere syndrome, Diaphragmatic plication, Diaphragmatic paralysis, Neurological disorders, Genetic mutation*.

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INTRODUCTION

Brown-vialetto-van laere syndrome (BVVLS) is a rare autosomal recessive neurodegenerative disorder characterised by ponto-bulbar palsy and sensorineural hearing loss.[1] It typically presents with palsies of the VII, IX, X, XI, and XII cranial nerves, developing for a short period in a healthy individual. Involvement of the III, V, and VI cranial nerves is less common.[2,3] Diaphragmatic paralysis is an uncommon manifestation of BVVLS, with limited literature documenting this association, particularly in the absence of trauma. To the authors' knowledge, there is no literature regarding diaphragmatic plication in BVVLS with diaphragmatic paralysis. Though uncommon, diaphragmatic paralysis is one of the most morbid features of BVVLS; it leads to recurrent chest infections and thus respiratory failure, which often leads to mortality.[3] The absence of a trauma history in such cases suggests possible involvement of the phrenic nerve, a vital nerve that controls the diaphragm.

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CASE REPORT

A 17-year male was diagnosed with BVVLS at the age of 12 years, who initially presented with syncope and hearing loss. Initial evaluations were conducted externally, revealing a homozygous mutation in the *SLC52A3* gene. The patient was followed up at an external centre with supportive treatment and was not experiencing seizures. These treatments were primarily based on riboflavin and steroids, aiming for stabilisation. In September 2023, he was referred to our institution with complaints of bloating, abdominal pain, abdominal distension, and dyspnoea, indicating widespread abdominal gas-related issues.

General surgical evaluations, including physical examination, thoracoabdominal tomography, and ultrasonography, revealed an elevation of the left diaphragm, leading to the referral to our thoracic surgery department. Fluoroscopy indicated an elevated left haemidiaphragm, suggesting possible diaphragmatic paralysis (Figure 1). Thoracic computerised tomography showed an elevated left diaphragm with no additional findings.

The patient underwent uniportal video-assisted thoracoscopic left diaphragmatic plication in the same month, with no additional abnormalities detected during the procedure. Postoperatively, he was transferred directly to the clinical room, and a control chest x-ray exhibited normalised diaphragm position (Figure 2). The chest tube was removed on the second postoperative day, and he was discharged on the third postoperative

day. At two weeks post-discharge, follow-up examination and a one-year follow-up chest x-ray showed no significant recurrence of diaphragmatic elevation (Figure 2), and the patient did not complain of shortness of breath.

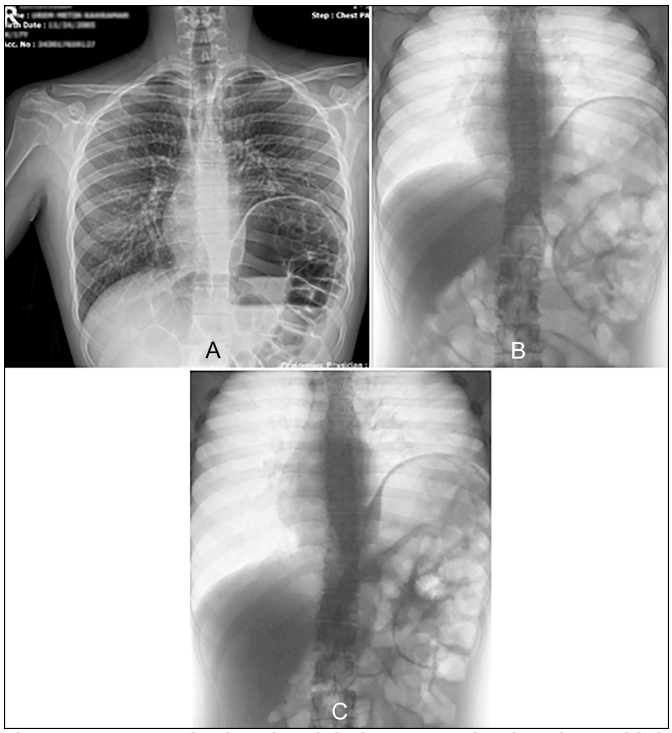

Figure 1: Preoperative imaging. (A) Chest x-ray showing elevated left diaphragm. (B) Fluoroscopy in the expiration phase. (C) Fluoroscopy in the inhalation phase. Please note that while the right diaphragm moves during breathing, the left diaphragm remains stationary.

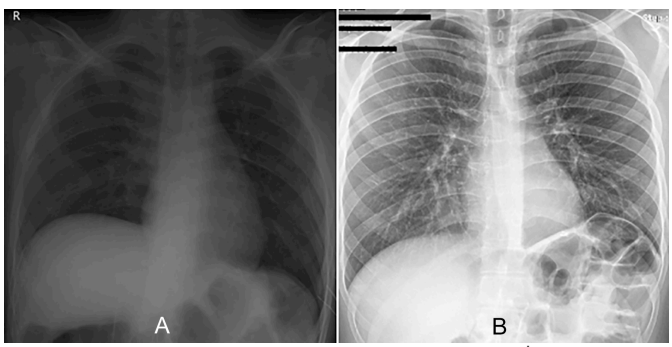

Figure 2: Postoperative imaging. (A) Postoperative 1st day, portable chest x-ray showing reduced left diaphragm. (B) Postoperative 1st year, standard chest x-ray, showing reduced left diaphragm.

DISCUSSION

BVVLS, first defined by Brown in 1894, Vialetto in 1936, and Van Laere in 1966, is a rare and progressive neuropathy seen in less than 1 in 1000000 cases.[3]

Its clinical presentation can vary in severity, with the average age of onset generally between 4.1 and 8.2 years.[4]

In the present case, the patient was a 17-year boy who had been diagnosed at an external centre at the age of 12 years. He had been completely healthy until the age of 12 years, when he developed sudden hearing loss and episodes of fainting.

Diaphragmatic involvement in BVVLS, which has a very limited coverage in the literature, is much rarer. In a study conducted by Woodcock *et al*.,[5] left phrenic nerve paralysis was detected in an 8-month girl who was previously healthy and later presented with complaints of apnoea and cyanosis and diagnosed with BVVLS. As mentioned in the same study, respiratory problems due to weakness of the respiratory muscles and diaphragmatic paralysis have been previously described in infants.

In this case, the patient complained of shortness of breath, which started in the last few weeks *i.e*. 5 years after the diagnosis of BVVLS, when the patient was 17 years. There was no history of trauma (fall, crash, surgical intervention, or traffic accident, *etc*.) before the onset of the presenting complaint. He had no respiratory complaints before. In addition, the radiological images accessed from the interhospital data bank revealed no evidence of diaphragmatic elevation on a chest radiograph taken 2 years ago.

In a study by Anand *et al*.,[6] a 22-month previously healthy female infant presenting with an acute onset of stridor and muscle weakness was diagnosed with BVVLS. Although radiological imaging was not mentioned in the study, non-invasive ventilation was used due to diaphragmatic weakness indicated by paradoxical breathing and stridor. Diaphragmatic plication was not performed on the patient; however, high-dose riboflavin administration and non-invasive ventilation were reported to give positive results.

In this case, apart from hearing loss and cognitive problems, which were the initial symptomatology, there were no symptoms such as general muscle weakness, fainting, or seizures, *etc*. The patient had no additional respiratory symptoms other than shortness of breath, which started late due to diaphragmatic paralysis. As a result of diaphragmatic plication, the patient's breathlessness resolved completely. However, considering the patient's condition (neurodegenerative nature of the disease, deafness, cognitive problems, *etc*.), this assessment remained subjective. Although spirometry was attempted for a more objective evaluation, patient compliance was not achievable.

Diaphragmatic paralysis is a rare manifestation of BVVLS, particularly without trauma. This case emphasises the need for increased awareness of potential diaphragmatic involvement in BVVLS patients, as early diagnosis and timely intervention can provide symptomatic relief. For choice of surgical technique, the uniportal video-assisted thoracoscopic approach was preferred as the least invasive method in thoracic surgery. This approach offers advantages in minimising operative morbidity, enhancing postoperative pain control, facilitating early discharge, and improving patient compliance.[7] On the other hand, the advantages of video-assisted thoracoscopy over thoracotomy—such as acceptable postoperative pain levels, faster recovery, and earlier discharge—make it the preferred approach for relatively straightforward procedures such as diaphragmatic plication.

In conclusion, given the potential progression of BVVLS, especially involving speech and cognition, BVVLS patients presenting with respiratory symptoms should undergo regular chest imaging. Even in the absence of such symptoms, annual chest x-rays may be recommended for surveillance.

The rarity of late-onset diaphragmatic involvement in BVVLS underscores the need for further investigation into the neurological implications of the syndrome.

PATIENT'S CONSENT:
Written informed consent for publication of the data related to this case was obtained from the patient's father, as the patient was 17 years of age and had cognitive impairment.

COMPETING INTEREST:
The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:
OY: Study design, data collection, data analysis, interpretation, and manuscript drafting.
AK: Data analysis, literature review, and critical revision of key sections.
MI: Data acquisition, data analysis, and critical revision.
AY: Overall supervision, interpretation of findings, and critical revision.
All authors approved the final version of the manuscript to be published.

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