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CASE REPORT

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Diaphragmatic myoclonus post cerebellar hemorrhage: a case report

Xiuxiu Tan[1] and Junjie Yang[2*]

Abstract

Background Diaphragmatic myoclonus is a rare motor disorder that affects muscle tone. It is characterized by involuntary movements of the abdominal wall and rhythmic, repetitive contractions of the accessory or respiratory muscles, all of which are innervated by the cervical nerve roots.

Case description We reviewed the case of a 57-year-old male patient who underwent surgery for a left cerebellar hemorrhage. He exhibited persistent myoclonus in the palate, jaw, and thoracoabdominal region. Following treatment, there was a significant reduction in flutter amplitude in these areas.

Conclusion The clinical rarity and variability of presentations often make diagnosis challenging and delayed. It is believed that this condition stems from abnormal excitation within the central nervous system or neural pathways that involve the phrenic nerve. Another potential mechanism is the direct irritation of the diaphragm. Ultrasound, chest fluoroscopy, and electromyography (EMG) can support the diagnosis. Various pharmacological and surgical treatments have been tried, yet specific treatment guidelines are still lacking.

Keywords Diaphragmatic myoclonus, Posterior post cerebellar hemorrhage, Palatal tremor, Hypertrophic olivary degeneration

Background

Diaphragmatic myoclonus, a rare motor disorder, was first identified by Leeuwenhoek in his patient. This disorder is characterized by rhythmic, repetitive, involuntary contractions of the diaphragm and other respiratory muscles innervated by the cervical nerve roots [1]. Additionally, it manifests various symptoms such as upper abdominal pulsations, sleep disturbances, dyspnea, hyperventilation, hiccups, abdominal pain, and gastroesophageal reflux[2, 3]. Recorded in individuals of all ages and genders, this disorder exhibits highly variable clinical presentations. The disorder's rare clinical manifestations contribute to diagnostic challenges, frequently resulting in delayed or ineffective treatment [4].

Case presentation

A 57-year-old male was admitted to hospital, following over two months of unexplained loss of consciousness. A head CT scan showed a left cerebellar hemorrhage, leading to emergency surgery involving left cerebellar hemorrhage removal, craniectomy decompression, and dural relaxation suture. After surgery, The patient exhibited confusion with a Glasgow Coma Scale score of 9 (E4VTM5). Occasionally, he opened his eyes spontaneously and tracked movements, but did not follow commands. Noticeable continuous flutter affected the palate,

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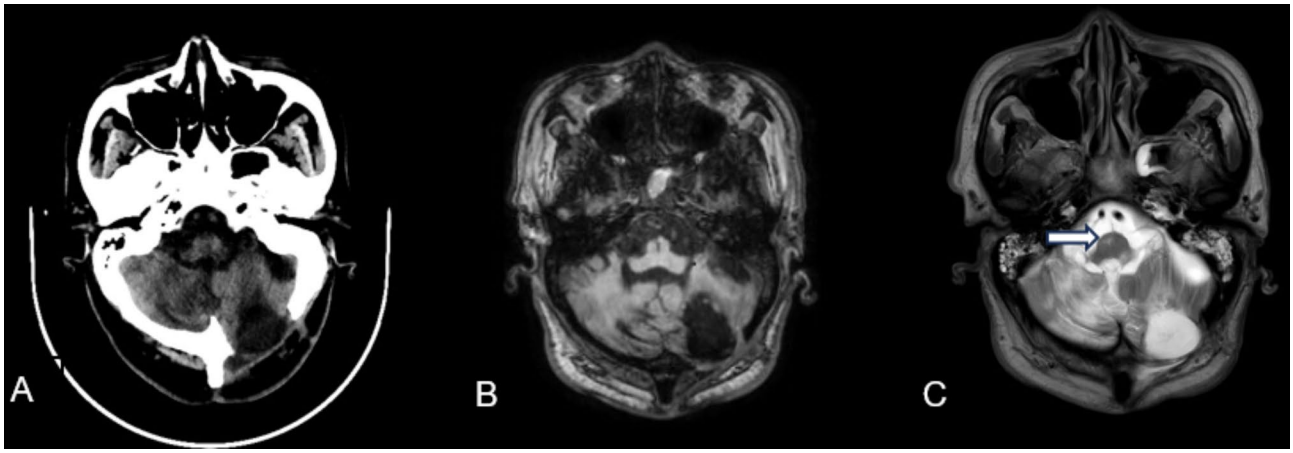

Fig. 1 Imaging manifestations; (**A**) Head CT indicated postoperative left cerebellar hemorrhage; (**B**) T1 and (C) T2 show bilateral cerebellar softening foci, White arrows in (**C**) indicate the abnormal inferior olive hypersignal

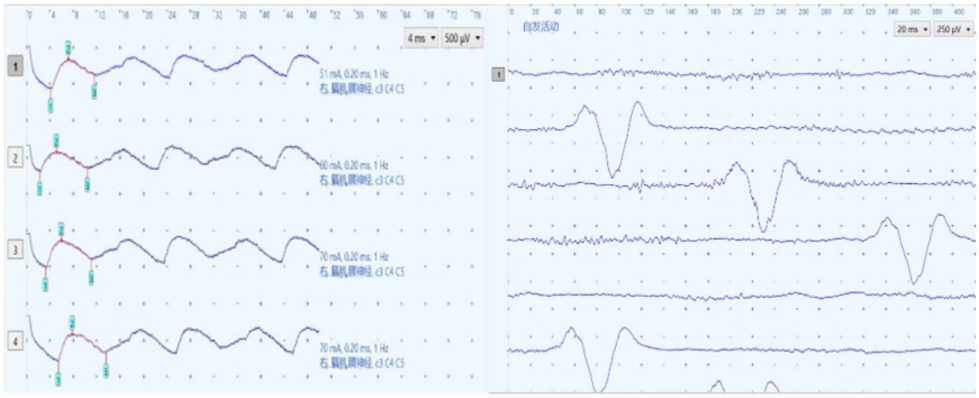

Fig. 2 EMG and NCV shows rhythmic discharge in the right diaphragm

jaw, and chest-abdominal region. His medical history included unmonitored and untreated hypertension.

In terms of auxiliary inspection, The head CT (Fig. 1) revealed a postoperative left cerebellar hemorrhage, bilateral cerebellar softening, left basal ganglia softening, multiple old and new lacunar infarctions in the thalami and basal ganglia, and multiple ischemic foci in the deep frontal and parietal lobes, The head MRI (Fig. 1) showed postoperative left cerebellar hemorrhage, bilateral cerebellar softening, multiple old lacunar infarctions in the thalami, basal ganglia, and brainstem with softening foci, and multiple ischemic foci in the frontal and parietal lobes. Among them, T2 phase suggests the presence of inferior olive body hypertrophy.Diaphragm electromyography and NCV(nerve conduction velocity)(Fig. 2) revealed diaphragmatic myoclonus. Additionally, chest and abdominal X-ray (Additional file 1) showed bilateral diaphragmatic myoclonus, more pronounced on the left side. Diaphragm ultrasound (Additional file 2) showed bilateral diaphragmatic myoclonus. The video(Additional file 3) shows jaw tremor. Rehabilitation included speech, cognitive, motor, and symptomatic support therapies. Medications included felodipine for hypertension and ambroxol for expectoration.

Bilateral temporal nerve electrical stimulation and benztropine were administered to treat diaphragmatic myoclonus. The tracheostomy tube has been removed, enabling the patient to engage in simple verbal communication. However, the patient continues to exhibit reduced limb muscle strength, memory loss, and other impairments in higher brain functions, which impair his ability to return to work. Continuous pulsations persist in the jaw and chest-abdominal areas, though their amplitude has significantly decreased.

Discussion

Studies by Rigatto et al. suggest that the pathophysiology of diaphragmatic myoclonus involves abnormal excitation of the phrenic nerve via central nervous system or neural conduction pathways [5, 6]. The diaphragm receives innervation from two neural circuits: the medullary respiratory center and the corticospinal tract originating from the cerebral motor cortex. The medullary respiratory center manages involuntary breathing, and

the corticospinal tract regulates voluntary breathing. Cortical inhibition of the brainstem respiratory center's influence on the diaphragm accounts for the suppressed diaphragmatic activity during activities such as eating, speaking, or voluntary breath-holding [7]. Thus, diaphragmatic contractions caused by abnormal brainstem activity can potentially be suppressed through voluntary effort.

Diaphragmatic or respiratory myoclonus is thought to be linked to palatal tremor and considered a variant thereof [3, 7, 8]. Palatal tremor is categorized into three subtypes: progressive ataxia and palatal tremor (PAPT), symptomatic palatal tremor (SPT) associated with brainstem or cerebellar lesions, and essential palatal tremor (EPT), which lacks identifiable brain disease. The origin of EPT remains unidentified, whereas SPT is attributed to lesions within the Guillain-Mollaret triangle (GMT) in the brainstem and cerebellum [9]. The Guillain-Mollaret triangle (GMT) is composed of the ipsilateral red nucleus (RN) in the midbrain, the ipsilateral inferior olivary nucleus (ION) in the medulla, and the contralateral dentate nucleus (DN) in the cerebellum. The central tegmental tract (CTT) connects the RN and ION. The superior cerebellar peduncle (SCP) links the ipsilateral RN with the contralateral DN, while the inferior cerebellar peduncle connects the ION to the contralateral DN. Symptomatic palatal tremor (SPT) typically manifests as hypertrophic olivary degeneration (HOD), a condition in which damage within the GMT triangle causes anterograde vacuolar degeneration and enlargement of remote ION neurons. However, not every lesion within this triangle results in HOD. HOD may develop from lesions affecting the cerebellum, SCP, RN, or CTT—the neural pathways leading to the ION. To date, no reports exist of HOD resulting from damage to the medulla or the inferior cerebellar peduncle—the efferent pathways from the ION [10].This finding confirms that HOD arises from neuronal degeneration caused by the loss of upstream neural inputs to the inferior olivary nucleus in the medulla. Following a cerebellar hemorrhage, our patient developed palatal tremor, diaphragmatic myoclonus, jaw tremor, and limb paralysis, with notable lesions in the GMT pathway. The MRI showed inferior olive hypersignal andprolonged T1 and T2 signals in the left medulla, strongly indicative of HOD. In HOD, myoclonus may affect the palate, pharynx, larynx, tongue, jaw, facial, and ocular muscles, though it seldom involves the diaphragm. The patient exhibited both palatal tremor and diaphragmatic myoclonus, maintaining stable intensity and similar flutter amplitude, unaffected by sleep.

Previous treatments for diaphragmatic myoclonus, as reported in small-scale studies, include phenytoin, carbamazepine, and valproate, which have been shown to stabilize neuronal membranes and reduce neuronal discharge. These medications have had past success. These medications may be effective due to their ability to lower the action potential of the phrenic nerve [11, 12]. However, no studies have definitively confirmed the efficacy of a specific drug, since the data primarily come from case reports, some of which are decades old. Pharmacological treatments are selected based on individual circumstances and do not consistently work for all patients. Initially, this patient did not respond to valproate; however, after switching to benztropine, there was a decrease in myoclonus amplitude. In addition to pharmacological approaches, other treatments for diaphragmatic myoclonus include phrenic nerve block or transection. However, phrenic nerve block is associated with symptom recurrence, whereas transection can lead to paralysis of the affected diaphragm. Electromyography-guided botulinum toxin injection represents a relatively new treatment approach [13]. However, few cases of botulinum toxin injection treatment have been reported, and neither its efficacy nor the appropriate dosage has been established.

Conclusion

Diaphragmatic myoclonus, a rare clinical condition, often presents with non-specific symptoms. The rarity of this condition means that no treatment guidelines currently exist. Case reports indicate that patients with palatal tremor rarely exhibit concurrent diaphragmatic myoclonus. However, our patient presented with significant palatal tremor, jaw tremor, and diaphragmatic myoclonus. The patient occasionally experienced paroxysmal tremors in both upper limbs. However, impaired higher brain functions precluded cooperation for a thorough muscle strength examination. Previous studies have demonstrated that diaphragmatic myoclonus can occur independently or as a secondary response to central nervous system diseases, abdominal trauma, or drug use. Both medical and surgical treatments have been employed; however, the lack of data on efficacy, side effects, and long-term outcomes makes choosing the appropriate treatment challenging.

Abbreviations

DM	Diaphragmatic myoclonus
ICU	Intensive care unit
GCS	Glasgow coma scale
EMG	Electromyography
NCV	Nerve conduction velocity
PAPT	Progressive ataxia and palatal tremor
SPT	Symptomatic palatal tremor
EPT	Essential palatal tremor
GMT	Guillain-Mollaret triangle
RN	Red nucleus
ION	Inferior olivary nucleus
DN	Dentate nucleus
CTT	Central tegmental tract
TBST	SCP superior cerebellar peduncle
HOD	Hypertrophic olivary degeneration

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12883-024-03809-7.

Supplementary Material 1

Supplementary Material 2

Supplementary Material 3

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Author contributions
Xiuxiu Tan discovered this case, collected clinical data, and wrote initial manuscript draft.Junjie Yang expanded the content and polished it.

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Data availability
No datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate
Not applicable.

Consent for publication
I confirm that the patient has given written consent that their personal or clinical details and any identifying images will be published in this study. A copy of the written consent is available for review by the editor of this journal.

Competing interests
The authors declare no competing interests.

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