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Cyclic Vomiting in an Adolescent With Muscular Dystrophy: A Case Report Highlighting Menstrual-Timing Diagnostic Challenges

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Case Report

Keywords: premenstrual disorder, probable premenstrual syndrome, muscular dystrophy, cyclic vomiting, adolescent, menstrual timing, case report

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Cyclic Vomiting in an Adolescent With Muscular Dystrophy: A Case Report Highlighting Menstrual-Timing Diagnostic Challenges

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Abstract

Background

Premenstrual disorders can be overlooked in adolescents with chronic neuromuscular disease because nausea, vomiting, and abdominal pain may be attributed to pre-existing gastroesophageal reflux or other gastrointestinal comorbidity. We describe an adolescent with muscular dystrophy and recurrent gastrointestinal symptoms showing a reproducible premenstrual pattern.

Case presentation

A 12-year-9-month-old girl with childhood-onset muscular dystrophy, progressive neuromuscular scoliosis, restrictive ventilatory impairment, pulmonary hypertension, myocardial fibrosis, chronic non-atrophic gastritis, and gastroesophageal reflux was evaluated during admission for scoliosis management. Variants in COL6A1 and SYNE1 had been reported as part of her underlying neuromuscular history. Menarche occurred at 11 years 10 months. Subsequently, she experienced abdominal pain, nausea, and vomiting 3–4 days before the expected onset of menstruation; episodes lasted 2–3 days and improved after menstrual onset. The episodes were initially managed as reflux or gastritis exacerbations, with limited symptomatic relief. Detailed menstrual history and targeted gynecologic, endocrine, and pelvic evaluation did not identify a new endocrine or pelvic structural explanation for the cyclic symptoms. Because prospective daily

symptom ratings across at least two cycles were unavailable, the working diagnosis was probable premenstrual syndrome (PMS) rather than definitively established PMS. Naproxen was prescribed as individualized symptomatic treatment for abdominal pain; omeprazole, ondansetron, fluid and electrolyte correction, and brief adjunctive traditional Chinese medicine were also provided. Symptoms improved after menstrual onset during multimodal management, but the independent contribution of each intervention could not be determined.

Conclusions

In adolescents with neuromuscular disease, recurrent vomiting and abdominal pain should not automatically be attributed to chronic reflux or gastrointestinal dysmotility. A menstrual history and prospective symptom diary may help identify a premenstrual pattern while alternative causes are evaluated.

Keywords: premenstrual disorder; probable premenstrual syndrome; muscular dystrophy; cyclic vomiting; adolescent; menstrual timing; case report

Introduction

Inherited muscular dystrophies are clinically heterogeneous disorders that may involve skeletal muscle, joints, the spine, respiratory system, heart, and, in some patients, the gastrointestinal tract. Collagen VI–related myopathies associated with COL6A1, COL6A2, or COL6A3 span a spectrum from severe Ullrich congenital muscular dystrophy to milder Bethlem myopathy and may include progressive weakness, contractures, and scoliosis [1,2]. SYNE1 variants have been reported in association with Emery–Dreifuss muscular dystrophy-like phenotypes and cardiac involvement [3,4].

Premenstrual disorders are clinical conditions characterized by recurrent physical, emotional, cognitive, or behavioral symptoms during the late luteal phase that improve after menstrual onset. Diagnosis relies primarily on the timing, recurrence, and functional impact of symptoms. Prospective daily symptom recording across at least two cycles is recommended to confirm the temporal pattern and to distinguish PMS from other cyclic conditions; sex-hormone concentrations are not used alone to establish the diagnosis [5–8]. Management is individualized and can include education, lifestyle measures, selective

serotonin reuptake inhibitors, hormonal contraceptives, and selected symptom-directed therapies [6,8,9].

In adolescents with neuromuscular disease, chronic reflux, gastritis, constipation, reduced mobility, and respiratory impairment may complicate interpretation of nausea or vomiting. A reproducible relationship between symptoms and menstruation may therefore be overlooked. We report an adolescent with muscular dystrophy and chronic reflux who developed recurrent premenstrual gastrointestinal symptoms. The clinical focus is the diagnostic challenge and the importance of menstrual timing. The reported genetic findings are considered part of the underlying neuromuscular phenotype and are not interpreted as proof of a causal relationship with the premenstrual symptoms.

Case Presentation

Patient information and underlying disease

A 12-year-9-month-old girl from Sichuan Province, China, was admitted for progressive thoracolumbar scoliosis. She was born at term by spontaneous vaginal delivery to non-consanguineous parents. The available record did not contain formal Tanner staging. Pubertal development was described as otherwise unremarkable, although formal Tanner staging was not available. No similar myopathy or spinal deformity was reported in the family.

At 3–4 years of age, the patient developed gait abnormality and frequent falls followed by progressive lower-limb weakness. A muscle biopsy at approximately 4 years of age showed fibers of unequal size, degenerative, necrotic, and regenerative fibers, and increased interstitial connective tissue, findings considered compatible with muscular dystrophy. A hereditary myopathy panel performed at 12 years 7 months reported variants in COL6A1 and SYNE1. These findings are reported as part of the patient's underlying neuromuscular history; no genotype–phenotype inference is made in this case report.

The patient also had multiple joint contractures, chronic non-atrophic gastritis, gastroesophageal reflux, severe restrictive ventilatory impairment, pulmonary hypertension, myocardial fibrosis, hyperlipidemia, and progressive neuromuscular scoliosis. She had previously received nocturnal oxygen after an episode of respiratory failure with hypercapnia and was undergoing halo-pelvic traction for scoliosis. These comorbidities were clinically

important because they provided plausible non-gynecologic explanations for nausea, vomiting, and abdominal pain.

Menstrual history and symptom pattern

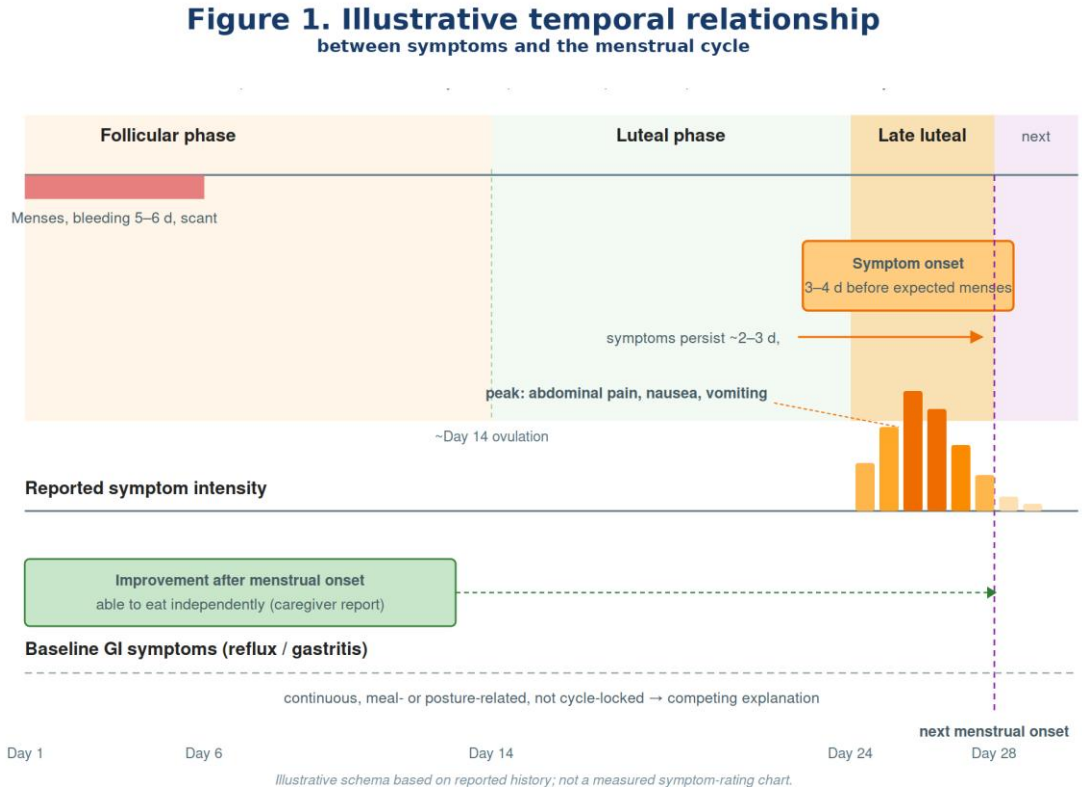


Figure 1. Illustrative temporal relationship between symptoms and the menstrual cycle. The diagram is based on retrospective caregiver-reported history and is not a measured symptom-rating chart. The patient's cycles were irregular, and the timing of ovulation was not directly measured.

Menarche occurred at 11 years 10 months. Menses were irregular, lasted 5–6 days, and were described as scant. From menarche onward, the patient repeatedly developed abdominal pain, nausea, and vomiting 3–4 days before the expected onset of menstruation. Episodes lasted approximately 2–3 days and improved after menstrual onset.

The episodes were initially interpreted as exacerbations of chronic gastritis or gastroesophageal reflux. Omeprazole and ondansetron provided limited symptomatic relief. The available record did not include prospective daily symptom ratings, formal symptom-scale scores, or a complete assessment of emotional, cognitive, or breast symptoms. Typical cramping

menstrual pain was not documented; therefore, the available information does not establish coexisting dysmenorrhea. The reported temporal pattern is shown in Figure 1.

Symptom characterization

The evidence supporting a premenstrual pattern is summarized below. Positive features were documented from history; several symptom domains and confirmatory assessments were not available in the record, which is why the diagnosis remained provisional.

Domain	Documented in this patient	Not available / not assessed
Physical symptoms	Abdominal pain, nausea, vomiting	Breast symptoms; bloating not documented
Symptom timing	Onset 3–4 days before expected menses; resolution after menstrual onset; duration 2–3 days; reported as recurrent	Number of cycles prospectively documented
Emotional, cognitive, behavioral	Not documented	Mood, anxiety, irritability, concentration, sleep
Functional impact	Reduced oral intake reported during episodes	Structured impact on school, rehabilitation and daily activity

Table 1. Symptom characterization: documented features versus missing evidence.

Physical examination and targeted assessment

At evaluation, the patient was conscious, with oxygen saturation of 90%–94% without supplemental oxygen. She had marked thoracolumbar scoliosis, limb contractures, reduced muscle strength and tone, and mild abdominal tenderness without guarding. The pin-track sites at the bilateral iliac crests were mildly erythematous with white exudate.

The following investigations were undertaken to assess potential endocrine, pelvic, gastrointestinal, and neuromuscular explanations for the symptoms. They were not considered confirmatory tests for PMS.

Assessment	Result	Clinical implication	Timing	Limitation
Serum electrolytes	Sodium 134.80 mmol/L; chloride 94.00 mmol/L	Mild abnormalities compatible with recurrent vomiting and reduced intake	During the current admission	Non-specific; may reflect vomiting of any cause
Thyroid testing	Total triiodothyronine 0.69 nmol/L; TSH 0.490 mIU/L	No clear thyroid explanation for the cyclic gastrointestinal symptoms	During the current admission; cycle day not recorded	Interpretation limited by unavailable cycle-day-specific ranges
Reproductive hormone panel	FSH 8.29 IU/L; LH 6.61 IU/L; estradiol 29.60 pmol/L; testosterone <0.10 μg/L; prolactin and progesterone reported as within the local reference range	Obtained because of irregular and scant menstruation; not diagnostic of PMS	During the current admission; cycle day not recorded	Not used alone to confirm or exclude PMS; formal Tanner staging unavailable
Pelvic ultrasonography	No adnexal mass or pelvic effusion; right ovary reported as normal; left ovary not visualized	No new pelvic structural lesion was identified in the available examination	Non-acute; relative to peak symptoms not recorded	Left ovary not visualized; intermittent torsion cannot be excluded
Spine radiography	Cobb angle 75°	Severe progressive neuromuscular scoliosis	During the current admission	Establishes comorbidity severity only
Echocardiography and cardiac MRI	Right-ventricular enlargement, moderate pulmonary hypertension, and subepicardial late gadolinium enhancement	Significant cardiopulmonary comorbidity	During the current admission	Establishes comorbidity severity only
Pulmonary function testing	Severe restrictive ventilatory defect	Advanced neuromuscular respiratory involvement	During the current admission	Establishes comorbidity severity only

Table 2. Targeted diagnostic assessment and its limitations.

Multidisciplinary assessment and working diagnosis

The recurrent premenstrual timing prompted review of the menstrual history and targeted assessment of competing causes. Chronic gastritis and gastroesophageal reflux remained relevant competing explanations; no new pelvic structural explanation was identified in the available evaluation. The hormone panel was interpreted cautiously because formal Tanner

staging, cycle-day-specific reference ranges, and a complete endocrine assessment were not available in the record.

Specialty / input	Key question addressed	Impact on the diagnostic decision
Gynecology / adolescent medicine	Could the symptoms reflect a premenstrual or other gynecologic disorder?	Reviewed menarche, irregular bleeding and the temporal relationship of symptoms to menses; recommended prospective symptom recording
Gastroenterology	Could chronic gastritis or reflux account for the recurrent vomiting?	Retained as a competing explanation; no new structural gastrointestinal cause documented
Endocrinology-oriented testing	Is there a thyroid or reproductive-endocrine cause of irregular menstruation?	No new endocrine cause established; results did not confirm or exclude PMS
Pelvic ultrasonography	Is there a pelvic structural lesion or adnexal pathology?	No adnexal mass or pelvic effusion; left ovary not visualized, so torsion could not be fully excluded
Traditional Chinese medicine consultation	Can adjunctive care relieve nausea and vomiting?	Provided supportive care only; independent efficacy could not be determined

Table 3. Multidisciplinary input and its impact on the diagnostic decision.

Because the symptoms were reported to recur before menstruation and improve after menstrual onset, the clinical team considered a probable premenstrual disorder, described here as probable PMS. Prospective daily symptom ratings over at least two subsequent cycles were recommended to confirm the temporal pattern, but such records were not available. Therefore, the diagnosis remained provisional.

Differential diagnosis

The recurrent symptoms required distinction from a premenstrual disorder, reflux or chronic gastritis, dysmenorrhea, cyclic vomiting syndrome, and acute pelvic pathology. A premenstrual disorder is suggested by a reproducible late-luteal pattern with improvement after menstrual onset. Reflux is more often associated with meals, posture, regurgitation, or heartburn. Dysmenorrhea is characterized primarily by cramping pain during or around menstruation and may be accompanied by nausea or vomiting; typical menstrual cramping was not documented in this case. Cyclic vomiting syndrome can cause stereotyped episodes with symptom-free intervals and may be

triggered by menstruation [13]. Because the left ovary was not visualized and the timing of ultrasonography relative to peak symptoms was not recorded, intermittent ovarian torsion could not be completely excluded by the available non-acute examination.

Therapeutic Intervention and Follow-up

The patient's chronic neuromuscular, cardiopulmonary, and orthopedic treatments continued under the relevant specialist teams, including halo-pelvic traction, nocturnal oxygen, pulmonary rehabilitation, and medications for cardiopulmonary management. These measures were not introduced as treatment for the cyclic premenstrual symptoms.

Naproxen was prescribed as individualized symptomatic treatment for abdominal pain. It should not be interpreted as a diagnostic test or as evidence of PMS-specific efficacy. Nonsteroidal anti-inflammatory drugs have stronger evidence for dysmenorrhea than for the overall symptom complex of PMS; however, typical dysmenorrhea was not documented in this patient.

Omeprazole and ondansetron were continued for coexisting reflux or gastritis and for nausea or vomiting, respectively. Mild hyponatremia and hypochloremia were managed with fluid and electrolyte correction.

Adjunctive traditional Chinese medicine, including modified Pingwei San and auricular therapy, was provided as supportive care for nausea and vomiting. Because the evidence for such interventions in premenstrual disorders is limited and heterogeneous [10,11], no specific therapeutic effect can be inferred from this case.

After menstrual onset, the parent reported that nausea and vomiting improved and that the patient was able to eat independently. Because spontaneous postmenstrual improvement and multiple simultaneous interventions occurred together, the independent effects of any individual intervention could not be determined. Longer follow-up with prospective symptom recording is needed.

List of abbreviations

FSH: follicle-stimulating hormone; GERD: gastroesophageal reflux disease; LH: luteinizing hormone; MRI: magnetic resonance imaging; NSAID: nonsteroidal anti-inflammatory drug; PMS: premenstrual syndrome; TSH: thyroid-stimulating hormone.

Discussion

This case illustrates a potential attribution bias in an adolescent with complex neuromuscular disease. Chronic gastritis, reflux, reduced mobility, respiratory impairment, and ongoing scoliosis treatment all provided plausible explanations for nausea and vomiting. However, the repeated onset of symptoms before menstruation and improvement after menstrual onset suggested a possible premenstrual disorder.

The diagnostic interpretation must remain cautious. Premenstrual disorders are clinical diagnoses, and symptom timing, recurrence, and functional impact are more informative than a single hormone panel [5–8]. In this patient, the absence of prospective daily ratings across at least two cycles, formal Tanner staging, and complete assessment of emotional or cognitive symptoms precluded definitive confirmation. “Probable PMS” or “probable premenstrual disorder” is therefore more appropriate than an unequivocal diagnosis.

The reported molecular findings should be interpreted cautiously. The COL6A1 and SYNE1 variants are presented as part of the patient’s underlying neuromuscular history, without inferring a specific genotype–phenotype relationship or a causal relationship with the menstrual gastrointestinal symptoms. The clinical contribution of this case is not a proposed genetic link between muscular dystrophy and PMS; rather, it is the need to assess menstrual timing when recurrent gastrointestinal symptoms occur in an adolescent with complex chronic disease.

The therapeutic interpretation similarly requires restraint. Naproxen has a clearer role in menstrual pain than in the broader syndrome of PMS, and its use in a patient with gastritis and reflux warrants individualized risk assessment. Improvement after naproxen cannot distinguish a premenstrual disorder from nonspecific abdominal pain or unrecorded dysmenorrhea. Likewise, improvement observed during traditional Chinese medicine and conventional supportive care cannot establish independent efficacy of any herbal or acupuncture-related intervention.

The practical clinical lesson is that menstrual timing should be assessed when an adolescent with chronic neuromuscular or gastrointestinal disease develops recurrent vomiting or abdominal pain. A practical approach is outlined below:

1. Characterize the cycle: document age at menarche, cycle interval, bleeding duration, flow, and the timing of symptoms relative to menses.

2. Record symptoms prospectively: use a daily symptom diary for at least two cycles, including physical, emotional, cognitive, and behavioral symptoms.

3. Assess impairment and warning signs: document effects on eating, sleep, school, rehabilitation, and daily activities; assess acute abdominal and pelvic warning signs.

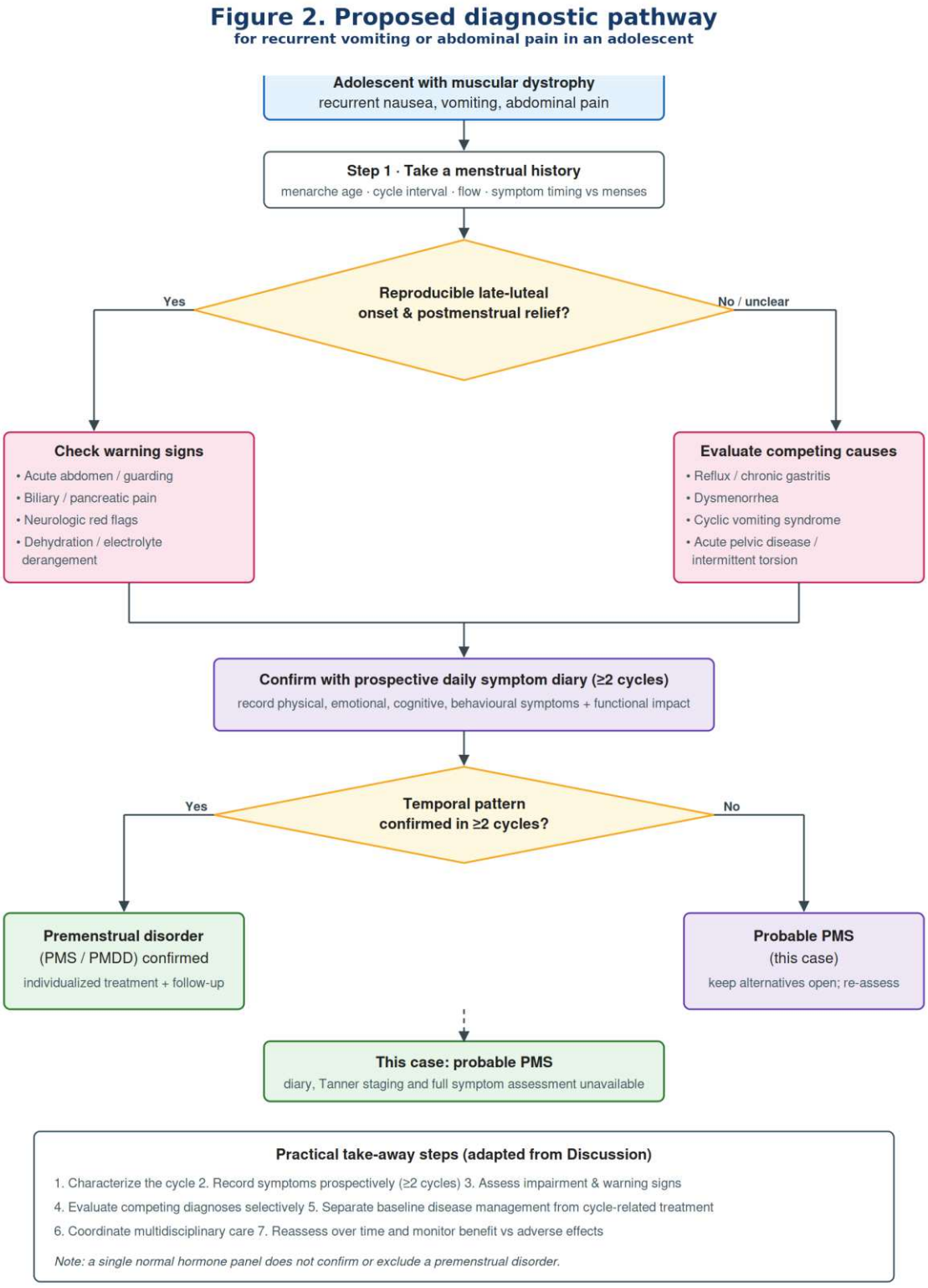
4. Evaluate competing diagnoses selectively: use targeted gastrointestinal, endocrine, pelvic, and neurologic assessments. Do not treat normal hormone results as diagnostic of PMS.

5. Separate baseline disease management from cycle-related treatment: distinguish chronic medications from interventions introduced for cyclic symptoms.

6. Coordinate multidisciplinary care: gynecology or adolescent medicine can coordinate with gastroenterology and neuromuscular specialists; endocrinology should be involved when menstrual or endocrine abnormalities warrant it.

7. Reassess over time: confirm recurrence across subsequent cycles and monitor treatment benefits and adverse effects. A summary of this approach is presented in Figure 2.

Figure 2. Proposed diagnostic pathway for recurrent vomiting or abdominal pain in an adolescent with chronic neuromuscular disease. The pathway emphasizes early menstrual-history assessment, warning-sign evaluation, assessment of competing causes, and prospective daily symptom recording over at least two cycles.



Limitations

First, prospective daily symptom ratings across at least two cycles were unavailable; therefore, the diagnosis remained provisional. Second, the available record did not contain formal Tanner staging or a complete assessment of emotional, cognitive, or behavioral

symptoms. Third, the left ovary was not visualized, and the timing of ultrasonography relative to peak symptoms was not documented; a non-acute examination cannot exclude intermittent torsion. Fourth, several interventions were administered concurrently with spontaneous postmenstrual improvement, preventing attribution of the outcome to any individual treatment. Fifth, the genetic findings are reported only at the gene level, without detailed genotype–phenotype interpretation. Finally, follow-up was short and the long-term recurrence pattern remains unknown.

Conclusion

In adolescents with muscular dystrophy and multisystem comorbidity, recurrent nausea, vomiting, and abdominal pain should not automatically be attributed to chronic reflux or other baseline disease manifestations. A reproducible premenstrual pattern may indicate a premenstrual disorder, but confirmation should rely on prospective symptom recording and careful evaluation of competing conditions. In this patient, the most defensible interpretation is probable PMS with postmenstrual symptom improvement during multimodal symptomatic management.

Caregiver Perspective

The patient's mother reported that her daughter had experienced motor difficulties since early childhood and that recurrent nausea and vomiting caused substantial anxiety because treatment for presumed reflux was not consistently helpful. Recognition of the repeated premenstrual pattern provided a possible explanation for the episodes and helped the family plan observation and supportive care around the menstrual cycle. The caregiver understood that the diagnosis remained provisional and that longer prospective follow-up was needed.

Statements

Ethics statement

This study was conducted in strict accordance with the ethical principles of the Declaration of Helsinki. The research protocol was reviewed and approved by the Medical Research Ethics Committee of West China Second University Hospital, Sichuan University (Ethics ID: Medical Research 2026 Ethics Approval No. (270), Approval Date: July 20, 2026).Data availability statement.

All de-identified data relevant to the case are included in the article. Additional information may be available from the corresponding author on reasonable request, subject to institutional and privacy requirements.

Consent for publication

Written informed consent for publication of this case report, including any accompanying images and identifying information, was obtained from the patient's parents (legal guardians).

Competing interests

The authors declare no competing interests.

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Author contributions

QX conceptualized the report, drafted the manuscript, and coordinated submission. LL curated the clinical data, critically revised the manuscript, and approved the final version. Both authors approved the submitted version.

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