



When Elevated Troponin Tells a Different Story: A Diagnostic Quest in a Duchenne Muscular Dystrophy Carrier

Matthew A. Goldfine, OMS-I¹; Carl Bryce, MD, Clinical Associate Professor¹; Jennifer Huang-Tsang, DO, FACC²
¹Midwestern University Arizona College of Osteopathic Medicine, Glendale, Arizona; ²Marshall Cardiology, Tucson, Arizona



Objectives

- Review diagnostic challenges of elevated troponin in the absence of ischemia
- Identify postpartum phenotypic expression in female dystrophinopathy carriers
- Emphasize early genetic confirmation in high-risk individuals
- Integrate osteopathic principles into longitudinal neuromuscular and cardiac care

Introduction

Duchenne muscular dystrophy (DMD) is an X-linked disorder caused by pathogenic variants in the *DMD* gene, resulting in dystrophin deficiency and progressive skeletal and cardiac muscle degeneration.¹

Although classically affecting males, 2.5-19% of female carriers demonstrate skeletal muscle involvement due to skewed X-chromosome inactivation or mosaic dystrophin expression¹ (figure 1).

Cardiac abnormalities occur in up to 17.2% of manifesting carriers.¹

Pregnancy is increasingly recognized as a physiologic stressor capable of unmasking disease expression.²

Despite consensus surveillance recommendations, female carriers remain a clinically underrecognized and inconsistently monitored population.³

Case Description

Initial Postpartum Presentation

28-year-old female with strong family history of DMD presented with:

- Progressive myalgia and exertional dyspnea
- Elevated creatine kinase: 3660 U/L (reference range 25-200 U/L)
- Elevated high-sensitivity troponin T: 0.08 ng/mL (reference range 0-0.01 ng/mL)
- Normal ECG
- Normal transthoracic echocardiogram
- Normal lower extremity MRI

Diagnostic Delay Timeline

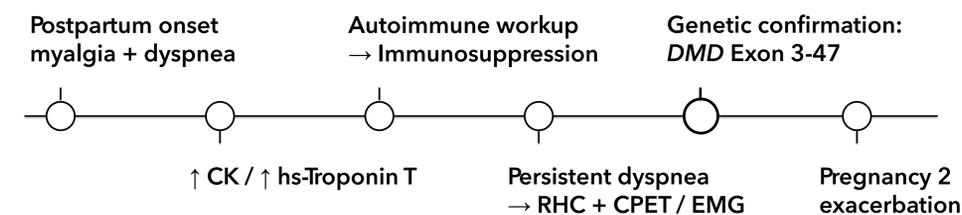


Figure 2. Diagnostic progression from postpartum biomarker elevation to genetic confirmation of dystrophinopathy.

Case Description

Rheumatologic Evaluation

- Transient anti-Scl-70 positivity
- Raynaud's phenomenon
- Gastroesophageal reflux
- Edematous hands
- Presumed inflammatory myositis

Treatment course

- Methotrexate → no improvement
- Mycophenolate (8 months) → minimal response
- Symptoms improved after discontinuing immunosuppression

Advanced Evaluation

- Right heart catheterization with cardiopulmonary exercise testing → oxygen extraction defect with no signs of pulmonary hypertension
- Electromyography → diffuse, non-irritable myopathy
- Genetic testing → DMD exon 3-47 deletion
- Cardiac MRI → mild left ventricular dilation with inferolateral hypokinesia, no myocarditis

Symptoms recurred postpartum following a second pregnancy. The diagnostic progression is summarized in figure 2.

Discussion

Differential Diagnosis

- Acute coronary syndrome
- Inflammatory myositis
- Postpartum cardiomyopathy
- Genetic myopathy
- Acute myocarditis

High-sensitivity troponin T may be elevated in dystrophinopathies due to early, subclinical myocardial involvement rather than ischemia.⁵

Normal echocardiography does not exclude early myocardial disease. Cardiac MRI demonstrated structural change consistent with evolving cardiomyopathy.

Pregnancy-associated hemodynamic and metabolic stress likely amplified dystrophin-deficient muscle vulnerability.²

Misattribution to autoimmune disease resulted in prolonged immunosuppression without benefit.

Per ENMC consensus, manifesting carriers warrant structured cardiac surveillance with periodic echocardiography or MRI.³

Conclusions

Female DMD carriers may present with isolated troponin elevation in the absence of ischemia.⁵

Pregnancy may unmask or exacerbate phenotypic expression.²

Transient autoimmune serologies may delay accurate diagnosis and expose patients to unnecessary immunosuppression.

Early genetic testing facilitates appropriate surveillance and counseling.³

Heightened awareness is essential to prevent delayed recognition of dystrophinopathy in women.³

Clinical Pearls

- **Troponin elevation does not equal myocardial infarction**
- **Family history may outweigh transient serologies**
- **Pregnancy can unmask dystrophinopathy**
- **Manifesting carriers require structured cardiac monitoring**
- **Osteopathic principles support functional optimization**

Acknowledgements

The authors thank the patient for consenting to share this case and the clinical staff for their assistance in data collection.

References

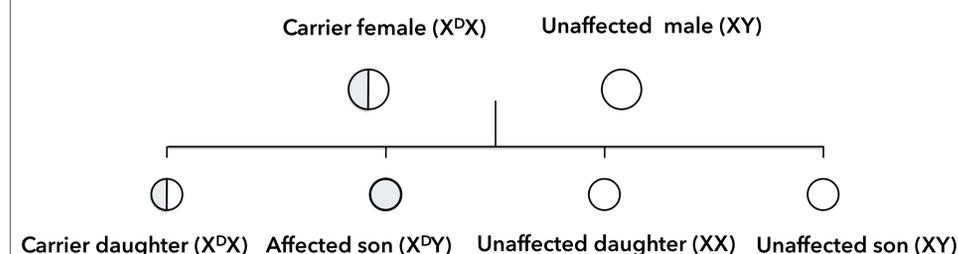


Figure 1. X-linked inheritance demonstrating carrier females and affected males in dystrophinopathy. Skewed X-inactivation may result in symptomatic expression in carriers.¹

Osteopathic Integration

Structural considerations:

- Thoracic inlet restriction
- Rib dysfunction affecting ventilation
- Diaphragmatic restriction

Although OMT does not alter the genetic mutation, lymphatic pump techniques have been shown to increase lymphatic flow and immune mediators and may be considered as supportive care to help manage respiratory infections and peripheral edema within multidisciplinary management.⁴