https://doi.org/10.35845/kmuj.2025.23838

CASE REPORT

Recurrent focal myositis with extraocular and lower limb muscle involvement: a case report

Kamil Khan [1], Eithne Murphy [1], Claire Louise Murphy [1]

1: Department of Rheumatology, Connolly Hospital, Blanchardstown, Dublin 15, Ireland

Email: drkamilkhan@yahoo.com
Contact #: +353-830-628736

Date Submitted: October 27, 2024
Date Revised: December 02, 2025
Date Accepted: December 08, 2025

ABSTRACT

Background: Focal myositis is a rare, localized inflammatory myopathy of unknown etiology. Owing to its rarity and nonspecific presentation, it is frequently misdiagnosed, which may result in delayed or inappropriate management.

Case Presentation: We report the case of a 37-year-old woman who presented with a three-day history of pain, swelling, and tenderness involving both calf muscles, accompanied by right-sided periorbital edema and diplopia. She had no systemic symptoms or clinical features suggestive of an underlying autoimmune disorder. Laboratory investigations revealed elevated creatine kinase and inflammatory markers. Magnetic resonance imaging (MRI) of the lower limbs demonstrated extensive edema of the right gastrocnemius muscle with associated subcutaneous edema, along with mild edema of the left gastrocnemius and soleus muscles. MRI of the brain and orbits showed inflammatory changes in the right lateral rectus muscle with surrounding periorbital edema. This was her third hospital presentation over the past decade with similar clinical manifestations. An extensive diagnostic workup excluded infectious, autoimmune, metabolic, and malignant causes. A diagnosis of idiopathic recurrent focal myositis with multifocal involvement was established. While her previous two episodes resolved spontaneously without immunosuppressive therapy, the current episode required immunosuppressive treatment due to persistent symptoms and to reduce the risk of recurrence.

Conclusion: This case illustrates the recurrent and multifocal presentation of focal myositis and emphasizes the need to consider this rare entity in patients presenting with localized muscle inflammation. Early recognition is crucial to prevent unnecessary investigations and to facilitate timely and appropriate treatment, particularly in recurrent or persistent cases.

Keywords: Myositis (MeSH); Musculoskeletal Diseases (MeSH); Muscular Diseases (MeSH); Muscle, Skeletal (MeSH); Magnetic Resonance Imaging (MeSH); Biopsy (MeSH); Biomarkers (MeSH); Diagnosis (MeSH).

THIS ARTICLE MAY BE CITED AS: Khan K, Murphy E, Murphy CL. Recurrent focal myositis with extraocular and lower limb muscle involvement: a case report. Khyber Med Univ J 2025;17(4):501-5. https://doi.org/10.35845/kmuj.2025.23838

INTRODUCTION

Focal myositis (FM) is a rare, idiopathic inflammatory myopathy characterized by the rapid development of a localized mass within skeletal muscle, typically presenting with pain and tenderness but without associated muscle weakness.[1-3] Owing to its uncommon occurrence and nonspecific clinical features, FM is frequently misdiagnosed as an infectious process, deep venous thrombosis, or a neoplastic lesion.[2] Several potential etiological associations have been described in the literature, including infections, drug exposure, radiculopathy, trauma, autoimmune connective tissue diseases, and idiopathic mechanisms; however, in most cases, no definitive cause is identified.[3] The diagnosis is usually confirmed by muscle biopsy, while contrast-enhanced magnetic resonance imaging (MRI) typically demonstrates a solitary inflammatory muscle lesion. The clinical course is generally benign, with most cases resolving spontaneously with conservative and supportive management.[3]

CASE PRESENTATION

First presentation: Ten years prior to the current admission, the patient presented to a local hospital in her home country with pain and swelling of the left thigh, accompanied by periorbital swelling. She was managed conservatively with supportive treatment and made a full recovery. No official medical records from this episode were available for review.

Second presentation: In 2021, the patient was referred by her general practitioner with a several-day history of pain, swelling, and tenderness involving the right calf and thigh. The symptoms initially began in the right calf and gradually progressed proximally to involve the right thigh. Concurrently, she developed itchiness, pain, and periorbital swelling of the left eye. On presentation, she was febrile with a temperature of 38.2 °C.

She denied any systemic symptoms or features suggestive of connective tissue disease, including polymyositis or dermatomyositis. Her past medical history was unremarkable, and she was not taking any regular medications. She was a non-smoker and reported only occasional alcohol consumption.

On physical examination, there was marked swelling and tenderness of the right leg and thigh, with milder swelling noted in the left leg. Right-sided periorbital edema was present, along with diplopia on right lateral gaze. Muscle strength in the lower limbs was preserved, and the remainder of the neurological examination was normal.

Laboratory investigations revealed leukocytosis (white cell count 19.8× 10^9/L; reference range 3.5-10.5× 10^9/L), elevated C-reactive protein (143 mg/L; 5mg/L), alanine aminotransferase (126 U/L; 0–41 U/L), bilirubin (24 μmol/L; 0–17 μmol/L), and markedly raised creatine kinase (8,593 U/L). Blood cultures were negative.

Table I: Comparison table of investigations for 2nd and 3rd presentations

Investigations	2nd presentation	3rd Presentation
Haemoglobin (g/dl)	14.5 (g/dl)	12.6 (g/dl)
White Blood Cells (10^9/L)	19.8 (10^9/L)	8.9 (10^9/L)
Platelets (10^9/L)	300 (10^9/L)	331 (10^9/L)
Urea (mmol/L)	5.0 (mmol/L)	5.2 (mmol/L)
Creatinine (umol/L)	56 (umol/L)	66 (umol/L)
Serum Sodium (mmol/L)	136 (mmol/L)	139 (mmol/L)
Serum Potassium (mmol/L)	4.0 (mmol/L)	4.8 (mmol/L)
Alanine Aminotransferase (IU/L)	126 (IU/L)	76 (IU/L)
Bilirubin (umol/L)	24 (umol/L)	12 (umol/L)
Gamma-Glutamyl Transferase (IU/L)	21 (IU/L)	16 (IU/L)
Creatine Kinase (ug/L)	8593 (ug/L)	4523 (ug/L)
C-reactive protein (mq/L)	143 (mq/L)	52 (mq/L)
Erythrocyte Sedimentation Rate (mm/hour)	77 (mm/hour)	62 (mm/hour
Connective Tissue Disease and Myositis Panel	Normal	Normal
Blood culture	No growth after 5 days	No growth after 5 days
Muscle biopsy	Inconclusive for myositis and no feature of mitochondrial and metabolic diseases	Presence of necrotic fibres, HLA-1 immunoreactivity, and fast fibre atrophy without predominant lymphocytic infiltrate.
Mitochondrial genome sequencing on skeletal muscle	No pathogenic mtDNA variant detected	Not repeated in this presentation.
Hepatitis viruses and HIV screen	Negative	Negative
Chest X-ray	Normal	Normal
MRI of lower limbs	Diffuse oedema in the right adductor muscle compartment and parts of the hamstring compartment. Multiple small foci of high signal in the left thigh.	Oedema in the right medial gastrocnemius and left gastrocnemius.
MRI of brain and orbits	CT brain is done which is normal	Inflammation of the right lateral rectus muscle with periorbital oedema.

Screening for connective tissue disease, vasculitis, and myositis was unremarkable. An extensive viral screen and tumor markers were also negative.

Computed tomography of the thorax and abdomen showed no abnormalities, and transthoracic echocardiography was normal. MRI of the lower limbs demonstrated extensive subcutaneous, perifascial, and interfacial edema in the right thigh, with diffuse edema of the adductor compartment and parts of the hamstring compartment, predominantly involving the right adductor magnus muscle. Multiple small foci of high signal intensity were also noted within the muscles of the left thigh.

A radiology-guided muscle biopsy from the right adductor muscle was performed but yielded inconclusive results. Subsequent molecular genetic analysis of the same biopsy sample excluded sporadic or maternally inherited mitochondrial DNA disease, as no pathogenic or likely pathogenic mitochondrial DNA variants were detected. Given the severity of the presentation, she was empirically treated for suspected necrotizing fasciitis with intravenous flucloxacillin, benz-ylpenicillin, clindamycin, and cipro-floxacin. She did not receive corticosteroids or other immunosuppressive therapy. Her hospital stay lasted two weeks, after which she was discharged home following complete clinical recovery.

Third presentation: In 2022, one year after her second admission, the patient presented again with a similar clinical picture. She reported pain and swelling of the right calf, along with pain over the medial aspect of the right thigh. This was accompanied by right-sided

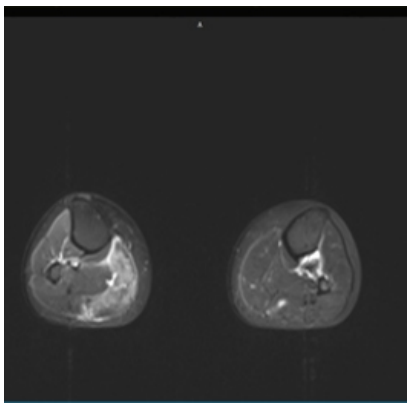

Figure 1: Axial section of both calves.

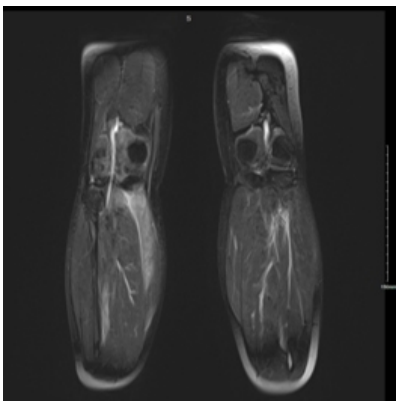

Figure 2: Coronal section of both calves, involving mainly the right gastrocnemius.

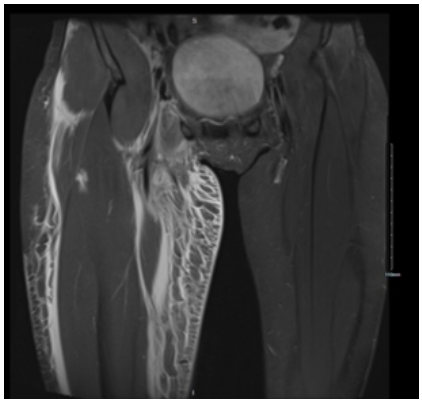

Figure 3: MRI coronal section of bilateral thigh with adductor compartment involvement on the right side.

periorbital swelling and diplopia; two days later, the left eye also became involved. Systemic examination was unremarkable apart from inflammatory changes in the lower limbs and periorbital edema with diplopia, predominantly affecting the right eye. She was reviewed by the ophthalmology team, who recommended MRI of the orbits and measurement of serum IgG4 levels. Routine laboratory investigations, including connective tissue disease screening, vasculitis workup, myositis panel, and serum IgG4 level, were all within normal limits. Doppler ultrasonography demonstrated a below-knee deep vein thrombosis (DVT) in the right leg, which was considered secondary to the underlying inflammatory muscle process. She was treated with Apixaban for three months for DVT management and Paracetamol for symptomatic pain relief. Detailed laboratory results are summarized in Table 1.

MRI of the lower limbs revealed extensive edema involving the right medial gastrocnemius muscle and the right adductor compartment, with patchy edema noted in the posterior compartment of the left calf (Figures 1-3). MRI of the brain and orbits showed inflammation of the right lateral rectus muscle with associated periorbital edema.

An open biopsy of the right gastrocnemius muscle was performed. Histopathological examination demonstrated hypertrophic and atrophic muscle fibers not confined to perifascicular regions, with a small group of atrophic fibers showing nuclear bag and pin-prick changes. There was no evidence of regenerating fibers or rimmed vacuoles. Blood vessels showed no features of vasculitis. Periodic acid–Schiff (PAS) and PAS-diastase staining revealed no excess glycogen. Gomori trichrome staining showed no ragged-red fibers, and no cytochrome c oxidase (COX)-negative fibers were identified. Immunohistochemical analysis showed absence of CD3, CD4, and CD8 T lymphocytes, as well as CD20 B lymphocytes and CD68 macrophages. Overall, the findings were consistent with a myopathic process.

On follow-up, due to incomplete clinical recovery, persistent mild periorbital swelling, and mildly elevated creatine kinase levels, she was commenced on Methotrexate 15 mg weekly along with Folic acid 5 mg weekly. She has remained clinically well for over one year, with no further disease recurrence.

DISCUSSION

Focal myositis is a distinct and uncommon form of inflammatory myopathy with a generally favorable prognosis. It can occur at any age and affects males and females equally. Although any skeletal muscle may be involved, there is a particular predilection for the muscles of the lower limbs.1 Clinically, it typically presents as a localized, rapidly enlarging intramuscular mass associated with pain and swelling, often mimicking neoplastic or infectious processes. Systemic symptoms are usually absent.[2]

The exact etiology of focal myositis remains unclear; however, it has been associated with various triggering factors, including trauma, autoimmune disorders, infections, neurological conditions, malignancy, and idiopathic causes. In some reported cases, focal myositis has preceded or evolved into polymyositis, suggesting a possible pathogenic relationship between these entities.[2,3]

Focal myositis has been reported in association with a wide range of autoimmune conditions, including Behçet's disease, psoriasis, systemic lupus erythematosus, primary Sjögren's syndrome, scleroderma, granulomatosis with polyangiitis, juvenile idiopathic arthritis, and Crohn's disease.[3-5] Infectious etiologies have also been implicated, with bacterial infections, most commonly Staphylococcus and Streptococcus species, being the predominant causes. However, focal myositis has been described in association with viral, fungal, and parasitic infections. Fungal myositis, in particular, is rare and typically occurs in immunocompromised individuals. [6]

In addition, focal myositis has been reported in patients with underlying neurological conditions, including radiculopathy and peripheral neuropathy.[7,8] Drug-induced focal myositis has also been described, with cases reported following exposure to medications such as statins and anti–tumor necrosis factor (anti-TNF) agents.[9,10] Initial evaluation with MRI is particularly valuable, as it typically demonstrates a well-defined

intramuscular lesion without involvement of adjacent structures such as bone or surrounding soft tissues, thereby helping to exclude alternative diagnoses. Characteristic MRI findings include reduced signal intensity on T1-weighted images and increased signal intensity on T2-weighted images, with enhancement following gadolinium contrast administration (Figures 1-3).[2,3]

Histopathological examination of muscle biopsy specimens commonly reveals muscle fiber necrosis, fibrosis, a mixture of atrophic and hypertrophic fibers, regenerating fibers, and inflammatory infiltrates with macrophages and a predominance of $CD4^+$ T lymphocytes, along with upregulation of major histocompatibility complex (MHC) class I expression.[2] Electromyography typically demonstrates a myopathic pattern.

Published case series have reported normal inflammatory markers and creatine kinase levels in many patients with focal myositis. However, patients presenting with elevated C-reactive protein or erythrocyte sedimentation rate in combination with markedly raised creatine kinase levels are more likely to experience disease recurrence and, rarely, progression to a systemic inflammatory myopathy.1 Although focal myositis usually affects a single muscle and resolves spontaneously, multifocal and recurrent forms have been described.[11]

Various treatment strategies have been attempted for idiopathic focal myositis, with inconsistent and often limited success. When focal myositis occurs secondary to an identifiable underlying condition, management should be directed toward the primary disease. In cases associated with autoimmune disorders, such as vasculitis or connective tissue disease, corticosteroids are commonly used, often in combination with immunosuppressive agents such as Methotrexate.[13]

Long-term follow-up is essential for patients with focal myositis to monitor for disease recurrence and to detect potential progression to a systemic inflammatory myopathy, including polymyositis.[14]

CONCLUSION

Focal myositis may present as a focal, multifocal, or recurrent condition. Although the overall prognosis is favorable, rare progression to systemic inflammatory myopathies, such as polymyositis, has been reported. Therefore, long-term follow-up is recommended to monitor for recurrence and to ensure early detection of systemic disease.

REFERENCES

1. Auerbach A, Fanburg-Smith JC, Wang G, Rushing EJ. Focal Myositis: A Clinicopathologic Study of 115 Cases of an Intramuscular Mass-like Reactive Process. Am J Surg Pathol 2009;33(7):1016-24. https://doi.org/10.1097/PAS.0b013e31819e63fe
2. Gordon Smith A, Urbanits S, Blaivas M, Grisold W, Russell JW. Clinical and pathologic features of focal myositis. Muscle Nerve 2000;23(10):1569-75. https://doi.org/10.1002/1097-4598(200010)23:10<1569::aid-mus14>3.0.co;2-m
3. Devic P, Gallay L, Streichenberger N, Petiot P. Focal myositis: A review. Neuromusc Disord 2016;26(11):725-33. https://doi.org/10.1016/j.nmd.2016.08.014
4. Gallay L, Hot A, Allenbach Y, Maucort-Boulch D, Comarmond C, Marques C, et al. Occurrence of focal myositis during Behçet's disease: The identification of a specific vasculitis-associated focal myopathy. Neuropathol Appl Neurobiol 2023;49(2):1-8. https://doi.org/10.1111/nan.12900
5. Liewluck T, Ernste FC, Tracy JA. Frequency and spectrum of myopathies in patients with psoriasis. Muscle Nerve 2013;48(5):716-21. https://doi.org/10.1002/mus.23812
6. Crum-Cianflone NF. Nonbacterial myositis. Curr Infect Dis Rep 2010;12(5):374-82. https://doi.org/10.1007/s11908-010-0118-z
7. González Toledo GR, Pérez Pérez H, Brage Martín L, Castro López-Tarruella V. Bilateral triceps surae muscle focal myositis after a cauda equina syndrome. Neurología 2021;36(8):647-9. https://doi.org/10.1016/j.nrleng.2020.11.007
8. Gross R, Degive C, Dernis E, Plat M, Dubourg O, Puéchal X. Focal Myositis of the Calf following S1 Radiculopathy. Semin Arthritis Rheum 2008;38(1):20-7. https://doi.org/10.1016/j.semarthrit.2007.09.004
9. Masnammany AM, Lau W, Wong P-KK, Manolios N. Anti tumor necrosis factor induced focal myositis. Proc Singap Healthc 2022;31:1-4. https://doi.org/10.1177/2010105822114775
10. Wagner M, Mühldorfer-Fodor M, Prommersberger KJ, Schmitt R. Statin-induced focal myositis of the upper extremity. A report of two cases. Eur J Radiol 2011;77(2):258-60.https://doi.org/10.1016/j.ejrad.2010.12.031
11. Heffner Jr RR, Armbrustmacher VW, Earle KM. Focal myositis. Cancer 1977;40(1):301-6. https://doi.org/10.1002/1097-0142(197707)40:1<301::aid-cncr2820400142>3.0.co;2-n
12. Barron SA, Heffner RR. Polymyositis Beginning as a Focal Process. Vol. 38, Archives of Neurology. American Medical Association (AMA); 1981. pp. 439-42. https://doi.org/10.1001/archneur.1981.00510070073013
13. Nagafuchi H, Nakano H, Ooka S, Takakuwa Y, Yamada H, Tadokoro M, et al. Recurrent bilateral focal myositis. Intern Med 2016;55(22):3369-74. https://doi.org/10.2169/internalmedicine.55.7172
14. Sato N, Okamoto S, Mori T, Watanabe R, Hamano Y, Kawamura J, et al. Recurrent acute myositis after allogeneic bone marrow transplantation for myelodysplasia. Hematology 2002;7(2):10912. https://doi.org/10.1080/10245330290022115

AUTHORS' CONTRIBUTION

The Following authors have made substantial contributions to the manuscript as under:

KK: Management of the disease, drafting the manuscript, approval of the final version to be published

EM & CLM: Identification, diagnosis and management of the disease, drafting the manuscript, critical review, approval of the final version to be published

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

CONFLICT OF INTEREST

Authors declared no conflict of interest, whether financial or otherwise, that could influence the integrity, objectivity, or validity of their research work.

GRANT SUPPORT AND FINANCIAL DISCLOSURE

Authors declared no specific grant for this research from any funding agency in the public, commercial or non-profit sectors

DATA SHARING STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request

KMUJ web address: www.kmuj.kmu.edu.pk
Email address: kmuj@kmu.edu.pk