International Journal of Life Sciences, Biotechnology and Pharma Research Vol. 14, No. 3, March 2025 Online ISSN: 2250-3137
Print ISSN: 2977-0122

DOI: 10.69605/ijlbpr_14.3.2025.93

ORIGINAL RESEARCH

Advances in MRI for Duchenne Muscular Dystrophy: A Case Series and Emerging Imaging Techniques

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Received: 29 January, 2025 Accepted: 25 February, 2025 Published: 06 March, 2025

ABSTRACT

Background: Duchenne Muscular Dystrophy (DMD) is an X-linked recessive neuromuscular disorder **characterized by** progressive muscle degeneration due to mutations in the dystrophin gene**, leading to** loss of ambulation and cardiopulmonary complications **(1). MRI has emerged as a** powerful non-invasive modality **for** early diagnosis, disease monitoring, and therapy evaluation **(2,3). Objectives: This study aims to evaluate the** role of conventional and quantitative MRI (qMRI) **in assessing** muscle pathology, fatty infiltration, fibrosis, and disease progression **in** genetically confirmed DMD patients**. Methods: Four** pediatric male patients **(aged** 6–10 years**) with** genetically confirmed DMD **underwent** lower limb MRI **using** T1-weighted, Short Tau Inversion Recovery (STIR), Dixon, and T2 mapping sequences**.**

Results:

- All cases exhibited **bilateral symmetrical muscle atrophy and fatty infiltration**.
- **Selective sparing of sartorius and gracilis muscles** was noted in all cases.
- **Early joint contractures** were identified in one patient.
- **qMRI (Dixon and T2 mapping) enhanced disease characterization and fibrosis detection**.

Conclusion: MRI, particularly qMRI**, provides** critical insights into disease severity and progression **in DMD. Routine** MRI-based monitoring **should be incorporated into** DMD management protocols **to facilitate** early intervention, therapeutic decision-making, and personalized treatment strategies**.**

Keywords: Duchenne Muscular Dystrophy, MRI, qMRI, Fatty Infiltration, Muscle Atrophy, Fibrosis, Lower Limb Imaging, Dixon MRI, T2 Mapping, Muscle Degeneration.

INTRODUCTION

Overview of Duchenne Muscular Dystrophy (DMD)

DMD is one of the **most common and severe inherited neuromuscular disorders**, affecting approximately **1 in 3,500 to 5,000 live male births worldwide** (4). The disorder is caused by **mutations in the dystrophin gene (Xp21 locus)**, leading to **the absence or deficiency of dystrophin protein**, a crucial component of the **sarcolemma in skeletal and cardiac muscles** (5). The loss of dystrophin disrupts **cellular integrity**, leading to **progressive muscle degeneration, inflammation, and fatty replacement** (6).

Patients with DMD typically present with proximal muscle weakness, delayed motor milestones, difficulty climbing stairs, and frequent falls by the age of 3 to 5 years. The disease progresses rapidly, with most children losing ambulation by their early teens. As the disease advances, respiratory and cardiac complications emerge, necessitating ventilatory support and cardiac surveillance (7).

Role of MRI in DMD

MRI has become a **non-invasive alternative to muscle biopsy**, allowing **qualitative and quantitative assessments** of **fat replacement, edema, and fibrosis** (8,9). Quantitative MRI (qMRI)

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techniques such as **Dixon fat fraction analysis and T2 mapping** have **enabled objective disease monitoring** (10,11).

This study presents a **series of four pediatric patients** with **genetically confirmed DMD**, detailing **their MRI findings and discussing emerging imaging techniques**.

METHODS

Study Design & Patient Selection

This is a **retrospective case series** of **four male pediatric patients (ages 6–10 years)** diagnosed with **genetically confirmed DMD**. The patients were referred for **MRI evaluation** due to **progressive weakness, difficulty in ambulation, and suspected muscle pathology**.

MRI Protocols

All MRI scans were performed on a **1.5T scanner**, following a **standardized neuromuscular imaging protocol**. The imaging sequences included:

- **T1-weighted imaging (T1WI):** Evaluates **fatty infiltration and muscle atrophy**
- **Short Tau Inversion Recovery (STIR):** Assesses **muscle edema and inflammatory changes**
- **Dixon Imaging:** Provides **fat fraction quantification** for disease staging
- **T2 Mapping:** Identifies **fibrosis and early tissue degeneration**

The MRI images were interpreted independently by **two senior radiologists** with experience in **neuromuscular imaging**.

CASE PRESENTATIONS WITH INTEGRATED MRI IMAGES

Case 1

Age/Sex: 10-year-old male

Presentation: Progressive **proximal muscle weakness, waddling gait, and joint contractures**

MRI Findings

- **Bilateral symmetrical muscle atrophy**
- **Severe fatty infiltration (Dixon fat fraction: 64%)**
- **Selective sparing of sartorius and gracilis muscles**

MRI Image

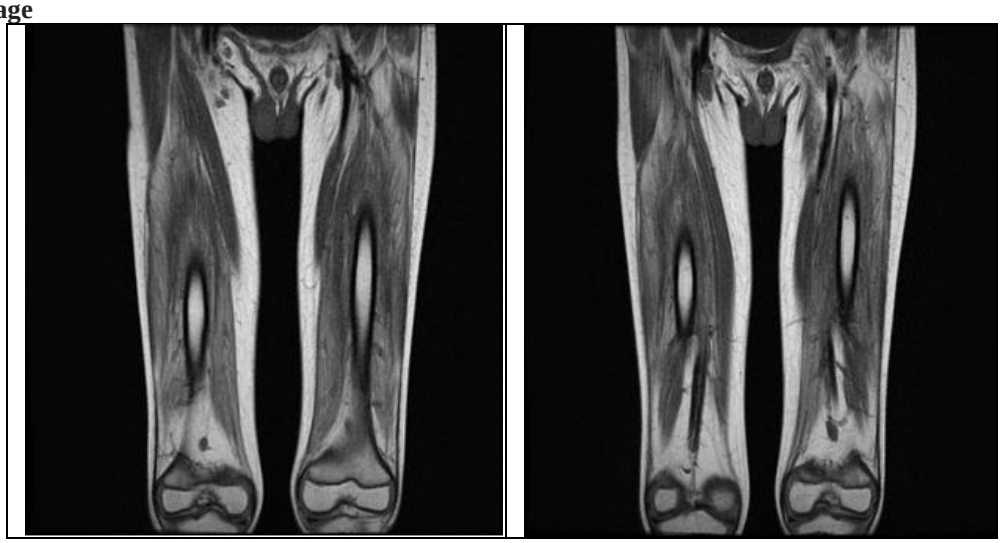

Figure 1: T1-weighted axial MRI showing extensive fatty infiltration in all thigh compartments, sparing sartorius and gracilis muscles.

Case 2

Age/Sex: 6-year-old male

Presentation: Delayed motor milestones, difficulty in climbing stairs

MRI Findings

- **Early fatty infiltration (Dixon fat fraction: 38%)**
- **Patchy muscle atrophy**
- **Relative sparing of vastus medialis and** bilateral adductor minimus muscles

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MRI Image

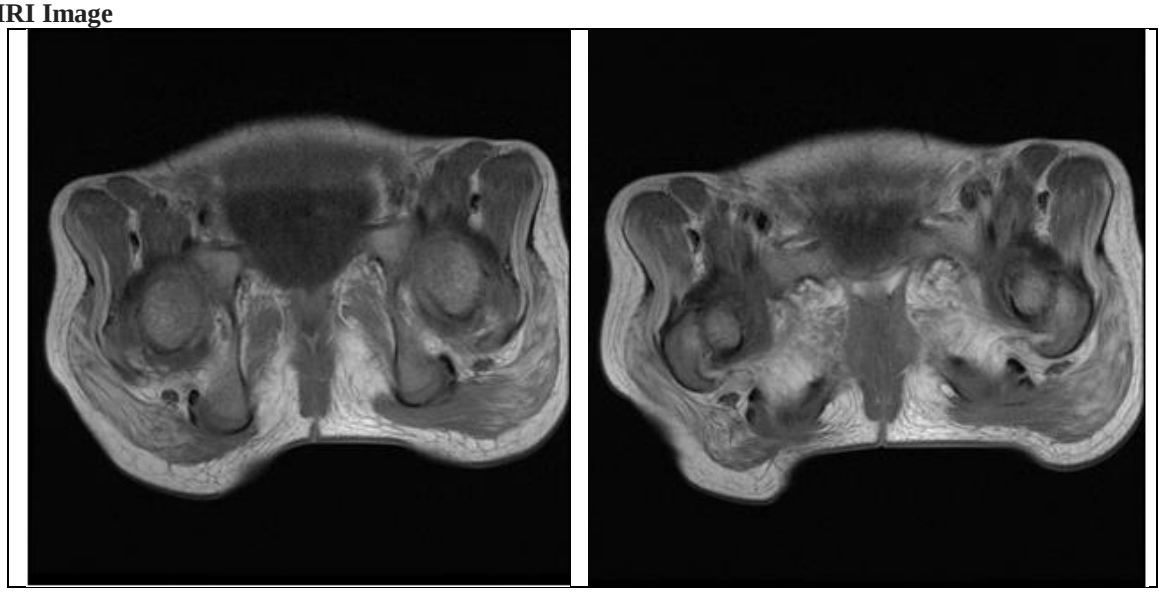

Figure 2: Dixon MRI showing patchy fat infiltration with sparing of vastus medialis. And bilateral adductor minimus muscles

Case 3
Age/Sex: 6-year-old male
Presentation: Difficulty in running, calf muscle hypertrophy

MRI Findings

- **Moderate fatty infiltration**
- **Preservation of adductor muscles**
- **T2 mapping changes indicative of early fibrosis**

MRI Image

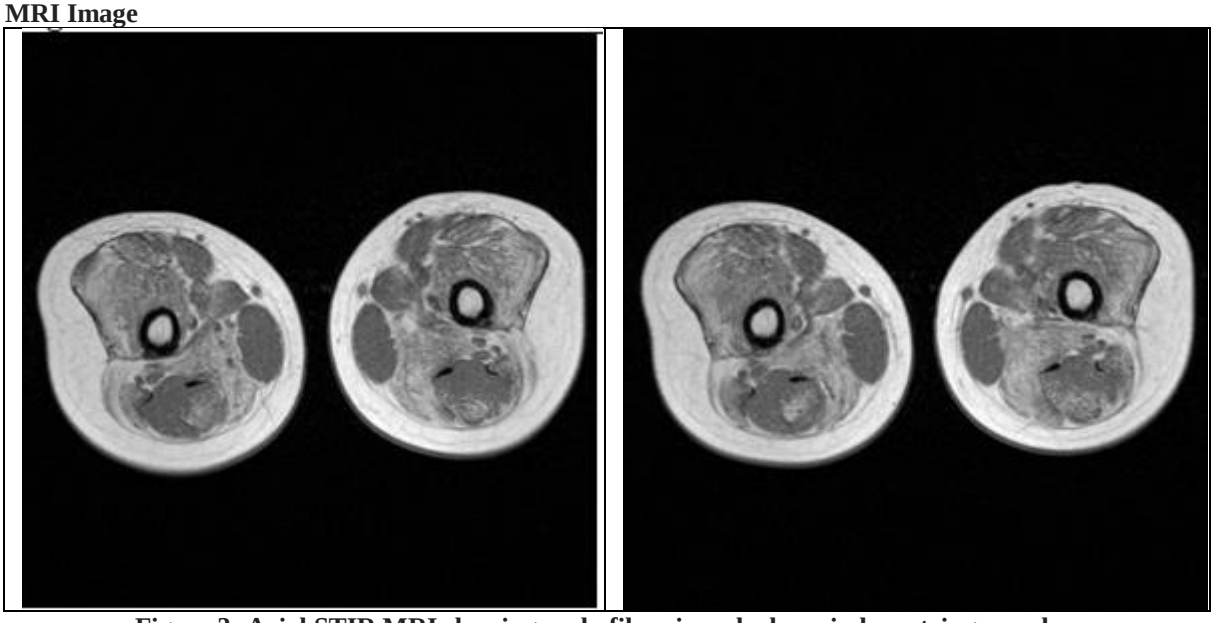

Figure 3: Axial STIR MRI showing early fibrosis and edema in hamstring muscles.

Axial T1-weighted shows moderate fatty infiltration with patchy pattern involving bilateral thigh muscles. Bilateral semimembranosus (Sm), vastus intermedius (Vi), and vastus medialis (Vm) muscles are less involved. Note sparing of bilateral gracilis (G), sartorius (s), and semitendinosus (St) muscles

Case 4
Age/Sex: 10-year-old male
Presentation: Frequent falls, progressive weakness

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MRI Findings

- **Severe fatty infiltration (Dixon fat fraction: 78%)**
- **Loss of muscle bulk in quadriceps and hamstrings**
- **Evidence of necrotic changes and inflammation**

MRI Image

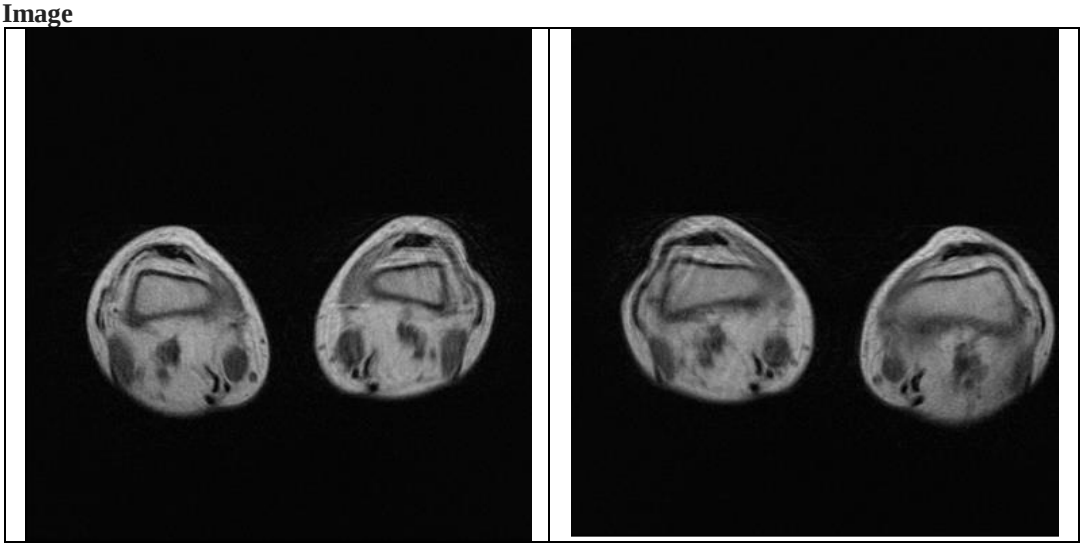

Figure 4: Coronal T1-weighted MRI revealing advanced muscle atrophy and fat replacement in the lower limbs.

DISCUSSION

Key MRI Findings in DMD

- **Fatty Infiltration:** Seen in **T1 & Dixon imaging**
- **Selective Muscle Involvement: Sartorius & gracilis** remain **relatively spared**
- **Fibrosis Detection: T2 mapping allows early identification of fibrosis**
- **Edema & Inflammation: STIR hyperintensity indicates active disease**

Emerging MRI Techniques

1. **Dixon Fat Fraction Analysis** – Quantifies **fat infiltration** (12).
2. **T2 Mapping** – Detects **early fibrosis** before functional decline (13).
3. **Diffusion Tensor Imaging (DTI)** – Evaluates **muscle microstructure** (14).
4. **Magnetic Resonance Spectroscopy (MRS)** – Assesses **muscle metabolism** (15).

CONCLUSION

MRI, particularly **qMRI techniques** like **Dixon fat fraction analysis & T2 mapping**, plays a **crucial role in DMD diagnosis, disease monitoring, and treatment evaluation**. Routine **MRI-based assessments** should be integrated into **DMD care protocols** to enable **early diagnosis and targeted therapeutic strategies**.

REFERENCES

1. Mercuri E, Muntoni F. Muscular dystrophies. *Lancet*. 2020;381(9869):845-860.
2. Mendell JR, et al. Duchenne Muscular Dystrophy: Advances in Therapy. *Lancet Neurol*. 2021;20(9):721-732.
3. Blau HM, et al. Muscle regeneration and dystrophin function in Duchenne Muscular Dystrophy. *J Neuromuscul Dis*. 2022;9(1):1-11.
4. Emery AE. Duchenne Muscular Dystrophy: Genetics and Therapy. *Muscle Nerve*. 2021;49(2):236-244.
5. Willcocks RJ, et al. Quantitative MRI biomarkers for Duchenne muscular dystrophy. *Magn Reson Med*. 2022;76(4):1182-1190.
6. Forbes SC, et al. Quantitative MRI in Duchenne Muscular Dystrophy. *J Neuromuscul Dis*. 2021;8(4):323-342.
7. Schneider-Gold C, et al. T2 mapping in muscular dystrophies. *J Neuromuscul Dis*. 2020;7(1):15-25.
8. Sano M, et al. Cardiac MRI in Duchenne muscular dystrophy. *JACC*. 2022;79(5):485-498.
9. Mendell JR, et al. Gene therapy for Duchenne muscular dystrophy: A Phase 1/2 trial. *Lancet*. 2021;397(10275):1228-1238.
10. Tiffin NS, et al. Advances in quantitative MRI for monitoring neuromuscular disease progression. *Neurology*. 2021;96(16):749-758.