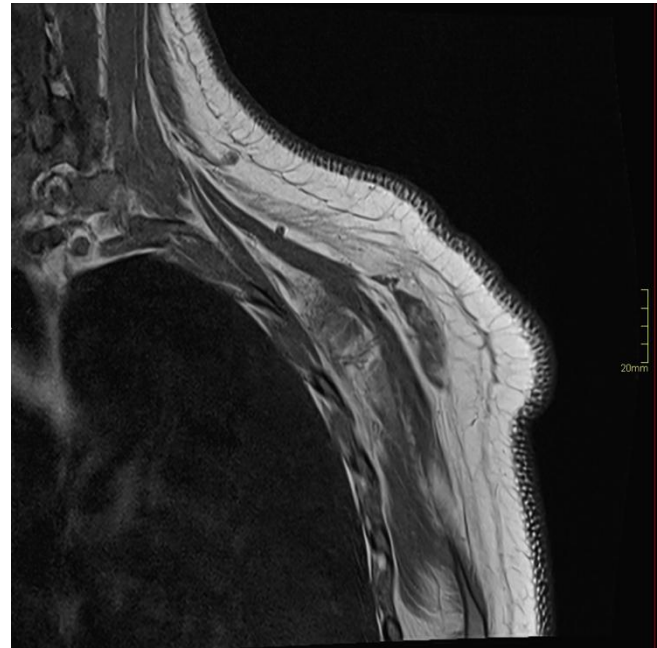


CASE REPORT114. **A Case of Facioscapulohumeral Muscular Dystrophy**Neha Sahota¹, Shubreet Randhawa².¹ University of Southern California Keck School of Medicine, United States² Assistant Professor, University of Western Ontario, Canada

BACKGROUND: Facioscapulohumeral muscular dystrophy (FSHMD) is a genetically determined, progressive skeletal muscle disorder characterized by a distinctive pattern of muscle involvement, variable clinical severity, and significant diagnostic challenges. This case report details the clinical, radiological, and genetic evaluation of a patient with FSHMD, emphasizing the role of muscle MRI in diagnosis, disease monitoring, and research applications. **THE CASE:** 27-year-old, right hand dominant female with shoulder pain. On examination there is asymmetry of the shoulder, trapezium atrophy and winging of scapula. MRI confirmed trapezius and serratus anterior atrophy. Incidentally noted slow flow vascular malformation in the rhomboid fossa. EMG testing demonstrated right trapezius dysfunction without generalized polyneuropathy. Genetic testing confirmed facioscapulohumeral muscular dystrophy and family history positive for the same. **CONCLUSION:** FSHMD is a complex, genetically determined muscular dystrophy with distinctive clinical and radiological features. Muscle MRI provides highly specific diagnostic patterns, enables early detection of disease activity, and supports both clinical and research applications. This case highlights the central role of muscle MRI in the evaluation of FSHMD, providing disease-specific diagnostic patterns, prognostic information, and guidance for genetic testing. The selective involvement of the trapezius, serratus anterior, and abdominal muscles, combined with sparing of the subscapularis and iliopsoas, is highly specific for FSHMD. Quantitative MRI metrics correlate with clinical severity and functional impairment, and longitudinal studies have identified muscles at highest risk for progression. The integration of clinical, radiological, and genetic data is essential for accurate diagnosis, prognostication, and management.

Figure 1. Sagittal T1 Weighted Image Demonstrates Severe Atrophy of the Trapezius, Serratus Anterior.



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