CASE STUDY

90. Beyond Genetics: Clinico-Pathological Recognition of Ullrich Muscular Dystrophy in a Resource-Limited Setting

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Background: Ullrich congenital muscular dystrophy (UCMD) is a rare collagen VI–related myopathy caused by mutations in COL6A1, COL6A2, or COL6A3 genes. Although traditionally considered an autosomal recessive disorder, recent reports have identified autosomal dominant variants that cause the UCMD phenotype. Clinically, it is characterized by early-onset muscle weakness, proximal joint contractures, and distal joint hyperlaxity. This condition frequently progresses to restrictive respiratory involvement, while typically sparing cognitive, sensory, and autonomic functions. Given this predictable trajectory, early integration of palliative care is increasingly emphasised to optimize quality of life and reduce crisis-driven interventions. UCMD is extremely rare, with an estimated prevalence of fewer than 1 in 1,000,000 births worldwide, though the exact frequency remains unknown. A definitive diagnosis requires genetic confirmation and immunohistochemistry, but such resources are often limited in low- and middle-income countries.

Case summary: A 15-year-old male, born of a third-degree consanguineous marriage, presented to the internal medicine department with recurrent lower respiratory tract infections and respiratory distress.

He was first evaluated at age 4 for progressive motor regression, difficulty ambulating, and multiple joint contractures. Over the next decade, he had progressive motor decline and restrictive pulmonary disease, while cognitive, sensory, autonomic, and cardiac functions remained preserved.

Physical examination revealed proximal muscle weakness, contractures of elbows and ankles, lumbar lordosis, café-au-lait spots, and a waddling gait. Muscle biopsy demonstrated features consistent with a dystrophic process and mild non-specific myopathic changes. A clinical diagnosis of UCMD was established based on characteristic clinical findings and histopathological correlation due to the unavailability of genetic testing and collagen VI immunohistochemistry. The internal medicine team integrated palliative care principles in his management through holistic assessment, symptom management, caregiver support, advanced care planning, and multidisciplinary coordination.
This case illustrates UCMD's diagnostic challenges, natural history marked by progressive musculoskeletal and respiratory decline, and the imperative for early integration of palliative care principles in management.

Conclusion: UCMD is a rare genetic disorder that is often underdiagnosed due to limited awareness and diagnostic challenges, particularly in resource-limited settings. Early diagnosis is essential to initiate supportive care, which can significantly improve quality of life. This case highlights the diagnostic value of comprehensive clinical evaluation and muscle biopsy in identifying UCMD. Increased awareness and expanded diagnostic testing programs are key to uncovering additional cases. Wider access to genetic testing is critical for diagnostic confirmation. Continued research into the molecular and genetic basis of UCMD will help to improve diagnosis and develop novel therapies. Owing to its rare occurrence, comprehensive epidemiological studies are needed to get more knowledge about its global incidence and natural history. Early palliative care integration is recommended to optimize patient and family well-being.

Figure 1. Clinical Features of Ullrich Congenital Muscular Dystrophy

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