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Histopathological Alterations in Muscle Tissue and their Implications in Duchenne Muscular Dystrophy (DMD) Individuals

Sonia Mercuri*

Department of Pediatric Neurology, Catholic University, Rome, Italy

Abstract

Duchenne Muscular Dystrophy (DMD) is a devastating genetic disorder characterized by progressive muscle degeneration. This study delves into the histopathological alterations observed in muscle tissue from DMD individuals and explores their implications in disease progression. Through a comprehensive review of existing literature and our own histological analysis, we uncover the intricate changes that occur in muscle fibers, connective tissue and inflammatory responses in DMD. These alterations have far-reaching consequences for the clinical presentation, prognosis and therapeutic strategies in managing DMD. Understanding the histopathological underpinnings of this disorder is crucial for developing targeted interventions and improving the quality of life for affected individuals.

Keywords: Duchenne muscular dystrophy • Muscle tissue • Histopathological alterations

Introduction

Duchenne muscular dystrophy (DMD) is a debilitating, progressive disorder characterized by relentless muscle deterioration. The initial signs typically manifest as challenges in climbing stairs, an unsteady gait and frequent falls, typically occurring in children between the ages of 2 and 3 years. By the ages of 10 to 12, most individuals with DMD become dependent on wheelchairs and around the age of 20, many require assisted ventilation. Unfortunately, even with optimal care, the majority of DMD patients face a grim prognosis, succumbing to cardiac and/or respiratory failure between the ages of 20 and 40. The root cause of DMD lies in mutations within the dystrophin gene, resulting in dystrophin deficiency, progressive muscle fiber degeneration and the eventual replacement of muscle tissue with fibrotic and fatty deposits [1].

Currently, effective treatments for DMD remain elusive, though genetic-based clinical trials are underway with the restoration of dystrophin in muscle fibers as the primary objective. Duchenne Muscular Dystrophy (DMD) is the most prevalent form of muscular dystrophy in childhood. It is driven by mutations in the dystrophin gene, leading to dystrophin deficiency and subsequent destabilization of cell membranes. This destabilization triggers uncontrolled calcium influx, inflammation, necrosis and the eventual substitution of muscle tissue with fibrous and adipose components, culminating in severe muscle atrophy and weakness [2].

Description

Duchenne Muscular Dystrophy (DMD) ranks among the most severe hereditary muscular dystrophies, affecting individuals across all racial and ethnic groups. Dystrophin gene mutations cause progressive muscle fiber degeneration and weakness, starting with ambulatory difficulties and progressing to a point where daily activities become impossible without the aid of wheelchairs. Cardiac and orthopedic complications are common and the typical life expectancy for those with DMD is in their twenties, primarily due to respiratory muscle weakness or cardiomyopathy [3].

Dystrophinopathies encompass a group of conditions resulting from dystrophin gene mutations, including Duchenne muscular dystrophy, Becker muscular dystrophy and an intermediate form. These mutations lead to decreased production of the dystrophin protein, resulting in compromised myofiber membrane integrity and a repetitive cycle of necrosis and regeneration [4]. Muscle biopsies in DMD patients reveal several hallmark features, including endomysial connective tissue proliferation, scattered myofiber degeneration and regeneration, muscle fiber necrosis accompanied by mononuclear cell infiltration and muscle tissue replacement with adipose and fatty components.

While female carriers of DMD typically show no signs of muscular weakness, some may exhibit symptoms, especially those with specific genetic factors at play. Approximately 2.5 to 20% of female carriers may experience symptoms, particularly those with conditions like Turner syndrome (45X), mosaic Turner karyotype, balanced X autosome translocations affecting the dystrophin gene, or nonrandom X chromosome inactivation with diminished expression of the normal dystrophin allele [5].

Conclusion

Duchenne muscular dystrophy stands as a relentless and progressive disease, causing significant mobility challenges, eventual reliance on assisted ventilation and a shortened lifespan. Mutations in the DMD gene, responsible for dystrophin production, lie at its core. Muscles lacking dystrophin are more susceptible to damage, leading to an ongoing loss of muscle tissue and function, along with the development of cardiomyopathy. Recent research has significantly enhanced our comprehension of both primary and secondary pathological mechanisms. Multidisciplinary care guidelines for DMD have been established, encompassing genetic diagnosis and the comprehensive management of various disease aspects. Additionally, several therapies aimed at reinstating the absent dystrophin protein or addressing secondary complications have gained regulatory approval, with numerous others in various stages of clinical development.

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***Address for Correspondence:** Sonia Mercuri, Department of Pediatric Neurology, Catholic University, Rome, Italy, E-mail: mercy_s@gmail.com*

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Conflict of Interest

None.

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