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Muscle Histopathological Changes and Consequences in Duchenne Muscular Dystrophy (DMD) Patients

Sonia Mercuri*

Department of Pediatric Neurology, Catholic University, Rome, Italy

Editorial

Duchenne muscular dystrophy (DMD) is a severe, progressive disease that causes muscle wasting. The first symptoms are difficulties climbing stairs, a waddling gate, and frequent falls; patients present with these symptoms between the ages of 2-3 years1. Most patients become wheelchair dependent between the ages of 10 and 12 and require assisted ventilation around the age of 201. With optimal care, the majority of DMD patients die between the ages of 20 and 40 from cardiac and/or respiratory failure.

Mutations in the dystrophin gene cause dystrophin deficiency, muscle fibre degeneration, and progressive fibrotic muscle replacement in Duchenne Muscular Dystrophy (DMD). There are currently no effective treatments for Duchenne muscular dystrophy (DMD), though genetic-based clinical trials are being piloted. The restoration of dystrophin in muscle fibres is the endpoint of the majority of these trials.

The most common muscular dystrophy in childhood is Duchenne Muscular Dystrophy (DMD). Mutations in the dystrophin gene cause the disease, resulting in dystrophin deficiency and subsequent cell membrane instability. This instability causes uncontrollable calcium influx, inflammation, necrosis, and muscle replacement with fibrotic tissue and fat, resulting in severe muscle wasting and weakness [1,2].

One of the most severe forms of inherited muscular dystrophy is Duchenne muscular dystrophy (DMD). It is the most common hereditary neuromuscular disease with no preference for any race or ethnic group. Dystrophin gene mutations cause progressive muscle fibre degeneration and weakness. This weakness may begin with difficulty ambulating but progresses to the point where affected patients are unable to carry out daily activities and must use wheelchairs. Cardiac and orthopaedic complications are common, and death typically occurs in one's twenties as a result of respiratory muscle weakness or cardiomyopathy.

Dytrophinopathies are diseases caused by mutations in the dystrophin gene, which include Duchenne muscular dystrophy, Becker muscular dystrophy, and an intermediate form. Mutations cause a reduction in the production of the dystrophin protein, resulting in a loss of myofiber membrane integrity and repeated cycles of necrosis and regeneration. Endomysial connective tissue proliferation, scattered degeneration and regeneration of myofibers, muscle fibre necrosis with a mononuclear cell infiltrate, and muscle replacement with adipose tissue and fat will all be seen in a muscle biopsy.

Female carriers show no signs of muscular weakness; however, symptomatic female carriers have been reported. Approximately 2.5 percent to 20% of female carriers may be affected. Female carriers who have Turners syndrome (45X) or a mosaic Turner karyotype, balanced X autosome translocations with breakpoints within the dystrophin gene and preferential inactivation of the normal X, or females with a normal karyotype but non-random X chromosome inactivation with diminished expression of the normal dystrophin allele can become symptomatic.

Duchenne muscular dystrophy is a severe, progressive muscle-wasting disease that causes movement difficulties and, eventually, the need for assisted ventilation and death. Mutations in DMD (encoding dystrophin) cause the disease by preventing dystrophin production in muscle. Muscles lacking dystrophin are more vulnerable to injury, resulting in progressive loss of muscle tissue and function, as well as cardiomyopathy. Recent research has significantly increased our understanding of the primary and secondary pathogenetic mechanisms. Guidelines for multidisciplinary care for Duchenne muscular dystrophy have been established, which address obtaining a genetic diagnosis and managing the various aspects of the disease. Furthermore, several therapies aimed at restoring the missing dystrophin protein or addressing secondary pathology have received regulatory approval, and many more are in clinical development [3-5].

Conflict of Interest

None.

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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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