Summer 2015

Investigating Duchenne’s Muscular Dystrophy

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Duchenne Muscular Dystrophy (DMD) is an X-linked neuromuscular disorder characterized by progressive, generalized muscle weakness, and wasting of muscle (Kaspar, Allen, & Montenegro, 2009). DMD is the most common of the muscular dystrophies, and it affects 1 in every 4,000 male newborns (Battie, Bradin, Bughby, & Guglielmi, 2014). Diagnosis is usually made between the ages of three and six and these with DMD do not typically live past the age of thirty (Kaspar, 2009). Because the appearance of symptoms usually begins in the early childhood years, nurses typically have the chance to work closely with parents and families throughout disease course of DMD.

**Significance of Pathophysiology**

As muscle units die and the total number of functional muscle units decreases, weakness occurs, and eventually leads to contractures (Do, 2014). This process is responsible for the overt symptoms and gradual progression of the disease. This progression occurs in an ascending fashion, starting with minimal weakness, leads to a wheelchair bound patient, and ends up causing respiratory and cardiac failure in the end stages of the disease (Do, 2014). The changes discussed, occur in all types of muscle all over the body, including the heart. Because cardiomyocytes contract many more times a day than skeletal muscle fibers do, the process is accelerated in the heart, leading to cardiomyopathy death and heart failure and then failure of the heart (Kaspar, 2009).

**Overview of Pathophysiology**

All types of muscular dystrophy involve a mutation affecting the dystrophin gene. The dystrophin gene is located on the short arm of chromosome X, close to p21 locus (Do, 2014). Different mutations result in different forms and severities of muscular dystrophy. Mutations that are more severe and result in extremely low levels of dysfunctional dystrophin cause DMD (Kaspar et al., 2009). Two examples of mutations that result in DMD are those that interfere with the translation reading frame or promoter sequences; both of these mutations lead to unstable, ineffective proteins (Do, 2014).

**Implications for Nursing Care**

Experts have identified, that a multidisciplinary approach to caring for those with DMD is key (Muscular Dystrophy Association, 2015). A dedicated professional nurse, it is important to advocate for the diagnosis and continuity of care for a patient with DMD. The American Academy of Pediatrics recommends the child and family yearly evaluations, as well as evaluation what occurs when symptoms occur (Muscular Dystrophy Association, 2015). Physical therapists report that the patients should have the primary goals of physical therapy to avoid contractures and scoliosis, while allowing greater motion of joints (Muscular Dystrophy Association, 2015). As with most diseases, one of the greatest services a nursing professional can offer is support for the patient and family. Being a source of support or consulting others for support is an important part of caring for a patient with DMD, especially in the early stages. Other nursing interventions that can be of great importance can be done in the care of patients with DMD (Kaspar, Allen, & Montenegro, 2009). Education can also play a big factor in supporting families of patients with DMD. It is important for patients with DMD and their family to understand the disease process, as well as allow an understanding of how to make adaptations for this debilitating disease, patients continue to strive even though most odds are against them. Severely cannot be altered at this point, but nurses can do a deal deal to help these patients. Nurses should have an understanding of muscular dystrophy and have the ability to also be able to educate families on health maintenance and healthy practices that will prolong some complications of DMD. It is also important to remember individuals may look deteriorated on the outside, but they are normal intelligence and need human interaction just as anyone else. Sometimes just spending a minute of time conversing with these individuals can mean all the world to them. It can be very rewarding for nurses who care for these special patients.

**References**


