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 (Kapoor, Allen, & Monnet, 2009). DMD is the most common form of childhood muscular dystrophy, and it affects 1 in every 4,000 male newborns (Bhatti, Reade, Bashir, & Guglielmi, 2014) Diagnosis is usually made between the ages of three and six and those with DMD do not typically live past the age of thirty (Kapoor et al, 2009). Because the appearance of symptoms usually begins in the early childhood years, nurses typically have the chance to work closely with patients 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 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 disease usually affects 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 cardiac muscle, leading to cardiomyopathy death and the eventual death of the heart (Kapoor et al, 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 (Kapoor 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).

References


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In Conclusion

Although no sure has been offered for this debilitating disease, patients who are affected by DMD can overcome many obstacles and still live a decent quality of life with current healthcare support. These patients continue to strive even though most odds are against them. Severe cannot be altered at this point, but nurses can do a great deal to help these patients. Nurses should have an understanding of the disease, treatments, and complications of DMD as well as 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 feel deteriorated on the outside, but they are an external intelligence and need human interaction just as everyone else. Sometimes just spending a time of conversation with these individuals can mean all the difference in the world. This can be very rewarding for nurses who care for these special patients.

Implications for Nursing Care

Experts have identified that a multidisciplinary approach to care for those with DMD is key (Muscular Dystrophy Foundation of America, 2013). A nursing professional’s role is not only to be involved in the overall plan of care and continuity of care for a patient with DMD. The American Academy of Pediatric Nursing recommends that "Ongoing follow-up care is usually necessary yearly evaluations, as well as evaluations when new symptoms occur (Muscular Dystrophy Association, 2015). Physical therapy and occupational therapy, although costly, are usually required for many of these patients. The primary goals of the therapy is to prevent contractures and scoliosis, while allowing greater motion of joints (Muscular Dystrophy Association, 2015). As well as medical, one of the greatest skills 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 families living with DMD can be good resources for families with a new diagnosis; local support groups and online patient support groups can also be excellent tools for a nursing professional to provide (Brennan, D., 2015).

Education: Educators can also play a big factor in supporting families of patients with DMD. it is common that parents with DMD stand and walk on a regular basis and their children who have DMD, are very strong, the spine is strong, and joints are healthy (Kapoor et al, 2014). Physical therapists and professionals should also teach the importance of a balanced healthy diet; this prevents weight problems from having additional effects on the underlying disease process of DMD (Brennan, 2015).

All of these things along with medical management improve the outlook of life for these individuals. Because life expectancy for children with DMD is increasing, growing recognition, within healthcare systems, to support these individuals as they transition into adult life is being realized (Brennan, D., 2014). Today, many young adults living with DMD are able to leave their childhood homes, go to college, have a career, and even start families (Brennan, D., 2014).