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Investigating Duchenne's Muscular Dystrophy

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Signs and Symptoms

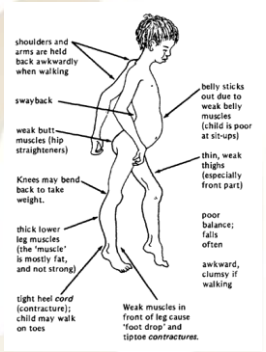
Young boys usually present with disordered or delayed motor development, muscle weakness, and possible speech delays in early childhood (Ruiten et al, 2014). The first symptoms almost always show up prior to the age of six years. Parents may notice their young child with DMD having a hard time standing up, walking, or climbing stairs, and many will eventually need a wheelchair to get around (Brennan, 2014). Initial blood tests obtained are remarkable for an elevated creatine kinase (CK) and then diagnosis is typically confirmed with a genetic test showing a mutation in the dystrophin gene (Ruiten et al, 2014).

Eventually with progressive muscle dysfunction, cardiac complications occur. Cardiomyopathy occurs in at least 90% of patients with DMD, but shows up at varying ages (Ashwath, Jacobs, Crowe, Ashwath, Super, & Bahler, 2014) Most of these patients initially develop heart disease characterized by fibrosis in the basal inferolateral wall of the left ventricle and then the lateral free wall (Ashwath et al, 2014).

Some other symptoms that occur as DMD progresses include a waddling gait, walking on toes, scoliosis, shortness of breath, fatigue, difficulty concentrating, and problems with learning and memory.

Although intelligence is not affected by DMD, some children develop learning disabilities or behavioral issues (Brennan, 2014).

Of note, DMD is not usually a painful disease process, although it can cause muscle cramping at times. Most of those affected maintain function of their bladder and bowel (Brennan, 2014).

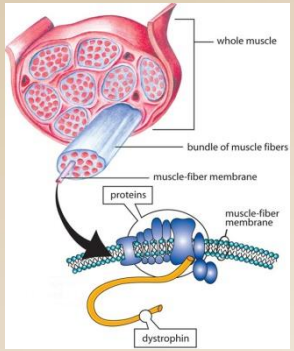


Underlying Pathophysiology

When dystrophin is affected by a mutation, it's function is compromised and several changes occur. Refer to the following list and chart for these changes (Kaspar et al, 2009):

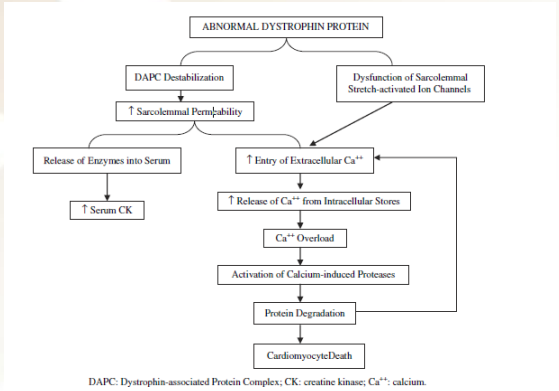
- the sarcolemma becomes fragile and susceptible to tears from muscle contraction (Kaspar et al, 2009)
- extracellular calcium enters the muscle fiber through tears(Kaspar et al, 2009)
- calcium activated proteases are activated (Kaspar et al, 2009)
- protein degradation occurs (Kaspar et al, 2009)
- release of intracellular muscle proteins leads to increased creatine kinase (CK) levels in the blood (Kaspar et al, 2009)
- gradual muscle cell unit death and macrophage invasion (Do, 2014
- dead muscle is replaced by a fatty infiltrate and clinically appears as pseudohypertrophy (Do, 2014)

The changes discussed, occur in all types of muscle all over the body, including cardiac muscle. Because cardiomyocytes contract many more times a day than skeletal muscle fibers do, the process is accelerated in cardiac muscle, leading to cardiomyocyte death and fibrosis and thus dilation of the heart (Kaspar et al, 2009).



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). Premature death often occurs in the second or third decade of life due to cardiac dysfunction and respiratory muscle weakness (Ashwath et al., 2014).

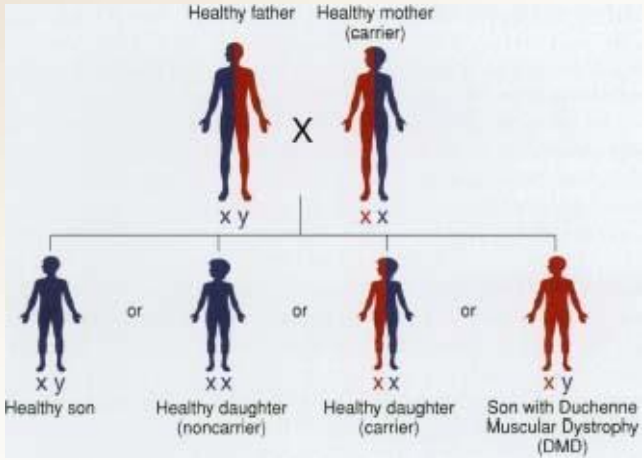


DAPC: Dystrophin-associated Protein Complex; CK: creatine kinase; Ca^{++}: calcium.

Introduction

Duchenne Muscular Dystrophy (DMD) is an X-linked neuromuscular disorder characterized by progressive, generalized weakness, and wasting of muscle (Kaspar, Allen, & Monetnaro, 2009). DMD is the most common form of childhood muscular dystrophy, and it affects 1 in every 4,000 male newborns (Ruitin, Straub, Bushby, & Guglieri, 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 (Kaspar et al, 2009). Because the appearance of symptoms usually emerge in the early childhood years, nurses typically have the chance to work closely with patients and families throughout disease course of DMD

Many times patients with DMD are admitted with heart failure and respiratory failure once the muscles of these organ systems become too weak. Although there is no cure, supportive therapy is available to help these patients live through complications of this disease process. New technology is also allowing these patients to have an increasingly normal life outside of the hospital. These patients hold a very special place in the hearts of many nurses. They overcome many hardships and still seem to be the most uplifting children and young adults.



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 allow some expression of the dystrophin gene lead to less severe forms of muscular dystrophy, while 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). As a professional nurse, it is important to advocate for a multidisciplinary team and continuity of care for a patient with DMD. The American Academy of Pediatrics recommends that DMD patients regular yearly evaluations, as well as evaluations when cardiac symptoms occur (Muscular Dystrophy Association, 2015). Physical therapy is an important consult these patients should have. The primary goals of physical therapy are to prevent 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 families living with DMD can be good resources for families with a new diagnosis; local support groups and online discussion boards can be excellent tools for a nursing professional to provide (Brennan, D., 2015)

Education can also play a big factor in supporting families of patients with DMD. It is important that patients with DMD stand and walk on a regular basis and stay active while possible; this keeps bones strong, the spine straight, and joints limber (Brennan, 2015). Nursing 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 expectancies for children with DMD are increasing, growing recognition, within healthcare, of the need to support these individuals as they transition into adult life is being realized (Hamdani, Mistry, & Gibron, 2014). Today, many young adults living with DMD are able to attend college, have a career, and even start families (Brenan, 2015).

In Conclusion

Although no cure 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. Genetics cannot be altered at this point, but nurses can do a great deal else to help these patients. Nurses should have an understanding of the pathophysiology behind the 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 these individuals may looks deteriorated on the outside, but they are of 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 vey rewarding for nurses who care for these special patients.

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