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Marfan Syndrome in Athletes

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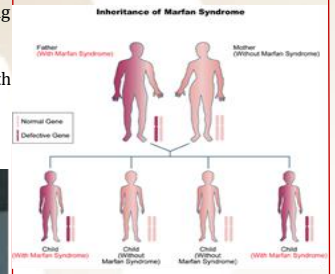
Introduction

Genetic disorders are widely misunderstood in our society and can lead to early mortality. Marfan syndrome (MFS) is a genetic disorder that affects connective tissue (Harris, Croce, & Tian, 2014). Antoine Marfan, a French pediatrician, first described this disease in 1896 (Elshershari & Harris, 2014). MFS can manifest in several different organ systems. The cardiovascular complications of aortic dilation and dissection often account for the morbidity associated with this disease (Harris et al., 2014). Understanding the inheritance, pathophysiology, and treatment of MFS is important for the advanced practice nurse (APN). Prevalence of the disease is approximately two per 10,000 individuals, but it is thought that MFS is under diagnosed and authorities suspect MFS may affect one in 3,000 (Pitcher, Emberson, Lacro, Sleeper, & Stylianou, 2015). MFS is most notable for affecting Abraham Lincoln and causing the death of Flo Hyman, a member of the 1986 US Olympian team (Davis, Dyar, Vargas, & Grossfeld, 2015). By obtaining a detailed health history of all athletes and recognizing the rare phenotype expressed by children withMFS, the APN can prevent the mortality associated with MFS, especially in the athletic population.

Presentation of the Case

Lamar has played basketball his entire life. His height, long arms, and long fingers have propelled him to the top of every team and league he has ever participated in. Lamar is 6'8", has bilateral lens discoloration, and slight scoliosis of the spine. As a child he also had a detached retina, which required repair. Lamar has to have an echocardiogram done as part of his yearly physical to play division one basketball.

After the screening, he is referred to a cardiologist because the test reveals a dilated aortic root. Genetic testing reveals a mutation in the gene FBN1, found on chromosome 15. These factors as scored by the Ghent Criteria, are enough to diagnose Lamar with Marfan syndrome. The phenotype of Marfan syndrome can be very variable (Braverman, 2015).



Pathophysiology

The underlying pathophysiology of Lamar's diagnosis revolves around the alteration of the FBN1 gene. The disease is usually transmitted in an autosomal dominant fashion. FBN1 is responsible for encoding the glycoprotein fibrillin. Fibrillin is the structural component of microfibrils. Microfibrils serve as building blocks in the human body and ligaments found in eye lenses, the aorta, and other connective tissues (Chen, 2015). Defects in fibrillin cause weakness in these tissues and hence the abnormalities observed in patients with this disease. Transforming growth factor-beta receptor (TGFBR) also plays an important role in Marfans. Fibrillin-1 binds to a form of TGFBR and keeps it sequestered throughout the body. Insufficient amounts of fibrillin cause an excess of TGFBR in the lungs, heart valves, and aorta. It is unclear how high levels of TGFBR cause the pathology associated with the disease.

Significance of Pathophysiology

The significance of this pathophysiology explains the symptoms commonly seen. The defect in fibrillin coupled with the overabundance of TGFBR causes problems in connective tissue throughout the body (Greenemeir, 2014). In Lamar's case, the dilated aortic root can lead to an emergent situation known as aortic dissection. This can be a life-threatening emergency. Lamar suffered a retinal detachment as a young child because of Marfans, and the inability of the connective tissue of the eye to hold his retina in place (Marfans Research Foundation, 2009). As people age and weaken with Marfan syndrome, the more likely they are to suffer consequences as connective tissue weakens as well.

Conclusion

When diagnosed, Marfan patients can have a normal life expectancy with crucial lifestyle modifications in place. Undiagnosed, Marfan syndrome can unfortunately lead to an early, unexpected death. The APN has a responsibility to inform themselves on the symptoms expressed by this rare disease. Becoming a knowledgeable APN can benefit the young athletes in their community.

Nursing Implications

The most crucial step for the advanced practice nurse (APN) in dealing with Marfan syndrome is the diagnosis. Becoming familiar with the Ghent criteria can help the APN recognize the signs of Marfan in the general public and athletes in particular. The Ghent nosology involve a scoring system where systemic features of Marfans are graded (Wright & Connolly, 2015). In athletes with Marfan-like qualities of superior height and extended extremity length, an echocardiogram should be performed. This test can give the APN a view of the heart using sound waves (Greenemeier, 2014). Gene testing can be performed, but is usually only done so in families that have a high occurrence of Marfan. Once the disease is diagnosed the APN can focus on the medical management of the disease with the patient. Physical activity should be modified in order to reduce the stress placed on the aorta (The Marfan Foundation, 2015). Medications such as beta-blockers or angiotensin receptor blockers are prescribed to lower the workload of the heart or aorta. The APN needs to stress to athletes and their families that this does not reduce the risk of strenuous exercise on the aorta. The APN must keep in mind the emotional toll that this may place on an athlete. Removing their ability to participate in strenuous sports such as basketball may result in depression.

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