Journal Review

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Brief Overview

Marfan syndrome is a rare condition that occurs in about 1 in 5,000 people. Most often an effect of genetics, Marfan syndrome affects the connective tissue in one's body, and the disease can result in a myriad of effects on various organs of the body such as the heart, eyes, skin, lungs, blood vessels, and major bones.

Causes for Marfan Syndrome

Marfan syndrome is usually an effect of a gene mutation, specifically a defect in the gene FBN1, and its mutation results in the hindrance of the production of proteins, Fibrillin-1 proteins, used to construct the body's connective tissue. Fibrillin-1 proteins are responsible for binding to each other and other molecules to produce thin, threadlike filaments otherwise known as microfibrils. Connective tissue is comprised of fibers containing microfibrils that allows for the strength and flexible structure of connective tissue. Due to the mutation in the FBN1 gene, the production of microfibril is reduced and results in the unstable nature of tissue in the body. Most often, people diagnosed with Marfan syndrome receive the disorder from a parent, and those with the syndrome possess a 50 percent chance of passing it down to their children. For 25 percent of those diagnosed with Marfans, these people did not inherit the gene from either parent and their condition is due to unspecified causes.

Symptoms/Effects of Marfan Syndrome

Marfan syndrome most often results in both physiological and anatomical abnormalities, such as extremely tall builds and long, thin arms, legs, toes, and fingers. Marfan syndrome does not necessarily target just one specific area of the body as it acts on the connective tissue in the body, which is located all throughout the human body; due to its great diversity even amongst those in the same family, Marfan syndrome possesses a multitude of effects. Common symptoms include: a tall and thin body, loose joints, crowded teeth, scoliosis or kyphosis, stretch marks on the skin, and/or a sunken or protruding chest/breastbone. While symptoms can range from mild to more dangerous, Marfan syndrome can be fatal for some.

Marfan Syndrome Effect on Cardiovascular Areas

For those with fatal conditions of the disease, most are due to the problems with connective tissue affecting the aorta, which is crucial to supplying blood to the rest of the body. Marfan syndrome can hinder the aortic root, where the pressure of blood leaving the heart can result in a bulge in the walls of the aorta; this occurrence is known as an aortic aneurysm. Another common obstruction for the heart is an aortic dissection, in which a tear forms on the inner layer of the aorta, and as blood passes through the tear, the tear grows and causes for the inner and middle layers of the aorta to pull apart of diverge further and can ultimately lead to a fatal rupture. Lastly, those with the Marfan condition have futile heart valve tissue, which causes risk for heart failure as the heart must work harder and overtime to account for faulty heart tissue and functioning.

Marfan Syndrome Effect on Eyes

Occurring very often for those with Marfan syndrome, ectopia lentis or lens dislocation proves to be a large obstruction in the lives of those with this condition. It causes for the eye lens to move out of its place due to the weak support structures surrounding it. In addition, marfan syndrome can also lead to retinal tears or detachments. The optic nerve of the eye can be damaged as risk for glaucoma and cataracts increases for those with Marfan syndrome, even in the younger stages.

Diagnosing and Treating Marfan Syndrome

Due to Marfan syndrome having a wide range of symptoms and targeted areas of the body, doctors typically diagnose Marfan syndrome through echocardiograms to test for heart valve conditions and aortic size. Other diagnosing tests include genetic testing and specific exams for eyes, such as slit-lamp exams and eye pressure testing.

While there is no way to eradicate marfan syndrome as it is primarily an effect of genetics, there are steps that can be taken to help alleviate some of the outcomes that are a result of this genetic condition. For those who suffer from heart problems, medication can help alleviate symptoms. There are specific surgeries in place to help targeted areas affected by Marfan syndrome such as aortic repairing and replacement of part of the aorta, braces or surgical procedures for those with scoliosis, correction of the breastbone, and eye lens replacement.

Marfan Syndrome and Athletes

Some athletes can be suspected to have Marfan syndrome due to many athlete's tall and slender builds, especially in track and basketball. However, Marfan syndrome can prove to be a great hindrance for the careers of many athletes. For those suspected to have Marfan syndrome,

athletes can continue with their sport as long as they adhere to their doctor's rules, maintain routine check-ups, and do not experience any massive health scares. On the contrary, when Marfan syndrome is diagnosed in an athlete, it prevents athletes from further pursuing their athletic goals as those with Marfan syndrome are prohibited from competing in sports unless they are low intensity. These low intensity sports include golf, bowling, and other ones that meet the low body exertion qualification. Since Marfan syndrome can result in weakened hearts, athletes participating in high intensity sports are exposed to high levels of dynamic and isometric activity that can cause an increase in damage of the aortic wall and potential aortic dissection. It's important that when diagnosed to Marfan syndrome, athletes step away from sports that lead to stress and exertion of the body, but it is still recommended that they maintain simple aerobic exercises to maintain bone and heart health.

Closing/Conclusion

Marfan syndrome is a condition commonly attributed to a mutation in the genes and is commonly a hereditary disease. It results in issues with connective tissue and results in problems with the eyes and heart and in abnormal physical traits. Due to it usually being caused by genetics, there is no way to fully treat Marfan syndrome, but there are some treatment steps in place for its precise ailments.

Works Cited

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