Marfan syndrome

Marfan Syndrome Wesley Thompson Jim Hutchins Biomedical Core 1110 November 19, 2011 Marfan syndrome is a variable, autosomal-dominant disorder in the connective tissue with distinct physical characteristics. The principal features affect the cardiovascular system, eyes and skeleton. This condition affects the connective protein that forms the structural support for tissues outside the cell. This disease is a result of a faulty makeup of chromosomes. Since MFS (Marfan Syndrome) is an autosomal-dominant disorder, only one copy of a faulty gene is necessary to cause manifestation of the trait (Polzin, 2005).

Recent advances and further development in the study of genetics have traced this disease to certain gene mutations. The most common being the fibrillin and TGFBR1 (Transforming Growth Factor, Beta Receptor 1) genes. Without early detection and attentive surveillance of MFS, mortality and morbidity increase significantly. In the article written by Elizabeth Gonzales, she discusses diagnostic criteria, genetic factors, and emerging theories of MFS. The article discusses the prevalence of the disease and reports that it affects about 1 in 10, 000 people.

It is equally common in all ethnicities, social classes, and countries (Marfan Syndrome, n. d.). This disease is typically diagnosed through genetic testing of the blood and the identification of certain physical characteristics that are commonly prevalent in MFS subjects. Although Marfan Syndrome is characterized by a triad of symptoms, most patients only have a few manifestations. Loose joints, long thin extremities, and aortic aneurysms are the primary symptoms of MFS. These clinical manifestations may be present at birth or develop later in life (Marfan Syndrome, n. d.).

Her explanation of the management of this disease was very specific. She explains that MFS management requires holistic care including paying particular attention to cardiovascular, orthopedic, and ocular issues. Aside from inspecting and taking care of the physical needs of a patient, psychological and genetic issues need to be addressed. If close attention is not given, those with MFS are at a high risk of acquiring a bacterial infection during pre and postsurgical operations due to an abnormal aortic valve. A treatment such as instituting antibiotic prophylaxis will decrease the probability that an infection like this will ensue.

The article then goes on to explain the prognosis and that there is no cure for Marfan Syndrome. However, with intervention, early detection, and attentive follow up, those with MFS can have a normal life span(Judge & Dietz, 2008). Without intervention and treatment, the life span is approximately 37 years. The article “ Marfan Syndrome: clinical diagnosis and management” discusses the genetic and physical aspects of the diagnosing and treatment/management of MFS. Clinical diagnosis is made using Ghent nosology, which defines the major criteria for the diagnosis of Marfan Syndrome.

Very few patients match up to these strict measures which is clearly the reason why this method diagnoses or excludes MFS in 86% of cases. Ghent nosology can be very complex to interpret due to the requirement of evaluating over 30 clinical features. One example of a positive sign in the Ghent nosology process is when the thumb and pinky fingers overlap when encircling the wrist. The main aspects in the clinical management of Marfan

Syndrome include inspection and care of the cardiovascular, ocular, musculoskeletal, respiratory, and central nervous systems.

Each of these systems must be carefully examined in order to insure that the skin stays intact. The article “ Cardiovascular Problems in Pregnant Women with the Marfan Syndrome” focuses on the potentially catastrophic problems that MFS poses in pregnant women. One problem is the potential for an acute aortic dissection which is often lethal (Pyeritz, 1980). Another is the risk of birthing a child who inherits the syndrome from the mother. The fetus has a surprising 50% chance of inheriting the mutant gene.

The dissection of the aorta in pregnant women is certainly a mystery and it is unknown exactly why it happens. Some hypothesize that estrogen inhibits the production of collagen and elastin fibers in the aorta. In some cases where the mother has substantial dilation of the aorta, therapeutic abortion or surgical intervention should be considered (Pyeritz, 17). Surgical intervention is in itself a risk because cardiac surgery often results in increased fetal loss. In cases where mothers show normal results on cardiovascular examinations, vaginal delivery is an option.

In conclusion, Marfan Syndrome (MFS) is a connective tissue disorder that primarily affects the connective protein that structurally supports cell tissue. It is prevalent in the cardiovascular, musculoskeletory, ocular, and respiratory systems. It is caused by the mutation of chromosomes in DNA. Without proper care and attentiveness, MFS is potentially fatal. Marfan Syndrome is particularly dangerous to women who are pregnant because of

the risk of substantial dilation of the aorta, and increased risk of the fetus inheriting the disease.

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