Thalassemia disorders essay sample

ASSIGN BUSTER

At age eleven, I remember my doctor announcing that I had B thalassemia trait minor and I was a carrier. It did not strike me as hard as it did when I found out that a friend’s aunt has recently passed away from being defeated by B Thalassemia Major. In the years to come, I understood that the only way I could make sure I had happy healthy children, was to dive into genetic counseling with whoever was to become my husband. When the time came I was over joyed to find out that my spouse was not a carrier and went on to having two amazing boys. One who is also a carrier of B Thalassemia Trait. The disorder that causes me mild anemia was passed from my paternal grandmother, to my father, to me and my youngest son. I am fortunate and very grateful for the diagnosis at age eleven. I now can ensure that youngest son will go on to raising his very own happy healthy children.

Thalassemia is a group of related blood disorders that cause abnormal hemoglobin production. It is a genetic disorder that is recessive; therefore both parents must be carriers to pass on the disorder in its major form. Thalassemia is categorized into two types, major and minor. Individuals suffering from the major form, tend to need frequent blood transfusions in order to survive. (Garrison & Peterson) Other names for Thalassemia disorders are: Mediterranean Anemia, Sickle Cell Anemia, and Cooley’s anemia (named after the first physician who diagnosed it). Thalassemia disorders are commonly found in people from Mediterranean decent, South East Asia, South Asia, the Middle East, China and the Caribbean. It appears that Thalassemia arose in the areas of the world where Malaria was an epidemic. In South Africa many people with Malaria developed Sickle cell Anemia. (Garrison & Peterson)

With increased migration of people from these countries into the United States; inter-marriage increasing between people from different ethnic backgrounds, more Hispanics and Latinos are affected by Thalassemia disorders.

“ Thalassemia is the most common inherited single gene disorder in the world.” (www. thalassemia. us) The Thalassemia’s can be broken down into two main types, Thalassemia A and Thalassemia B, each yielding sub disorders.

To understand more about the distinction of these disorders, we must first look at hemoglobin. Hemoglobin contains proteins called globins. Hemoglobin production involves two sets of genes. The genes are produced on different chromosomes which in turn produce two different pairs of proteins, alpha globin and beta globin. Each will contain two alpha and two beta proteins. (http://medical-dictionary. thefreedictionary. com/hemoglobin) When hemoglobin is functioning properly it binds and releases oxygen. This occurs when the two alpha proteins are connected to the two beta proteins. The gene for alpha globin is located at chromosome 16 and the gene for beta globin is located at chromosome 11. (Garrison & Peterson) (www. webmd. com/atozguide/thalassemia-topic-overview, 2011)

Parents determine the genes that their children inherit, therefore, “ Thalassemia will occur if one or more of the genes fail to produce protein.” (www. nhlbl. nih. gov/health-topic/topics/Thalassemia) A defective beta globin will result in Beta Thalassemia and a defective alpha protein will result in Alpha Thalassemia.

There are several types of Thalassemia A. The A types include the following: Silent Carrier State – this form is difficult to detect and generally causes no health issues. (www. thalassemia. us) Hemoglobin Constant Spring – this form gets its name from where it was found in Jamaica. Like Silent Carrier, individuals do not experience health issues. (www. thalassemia. us) Thalassemia Trait- In Thalassemia Trait there is an increased lack of alpha proteins. It is often confused with iron deficiency anemia. Individuals have mild anemia because of smaller red blood cells. (www. thalassemia. us) Hemoglobin H Disease- Because of the lack of alpha proteins, severe anemia is reported as well as health conditions including ; enlarged spleen, bone deformities and fatigue. In this disorder hemoglobin H destroys remaining red blood cells that are created by beta globin. (www. thalassemia. us) Hemoglobin H Constant Spring- In order for a person to have this disorder one parent passes the gene and the other parent passes the trait. These patients experience severe anemia and are more prone to enlarged spleen and frequent viral infections. (www. thalassemia. us)

The most devastating of all the Thalassemia A disorders is Hydrops Fetalis or Alpha Thalassemia Major. Babies born with this disorder die shortly after birth. Individuals with Thalassemia Major have no alpha genes, causing gamma globins to create abnormal hemoglobin called Hemoglobin Bart. If Thalassemia A is detected in utero, a technique which allows in utero blood transfusions to be performed may save the life of the unborn baby. This is a rare occurrence, but has been done. (www. thalassemia. us)

A variation of Hemoglobin Constant Spring is called Homozygous Constant Spring. Two parents are passing the gene to this child. This condition is

similar to Hemoglobin H disease, but less severe than Hemoglobin H Constant Spring. (www. thalassemia. us)

In Beta Thalassemia, if one beta globin is defected the disorder will result in Beta Thalassemia Minor. If both genes are defective, yielding no production of beta globin, the individual will have Beta Thalassemia Major. (Garrison & Peterson)

(www. ncbi. nin. gov/pubmedhealth/PMH0001613)

There are times where the clinical symptoms of thalassemia are not so bad resulting in a condition known as Thalassemia Intermedia. E-Beta Thalassemia is caused by one beta globin mutation and hemoglobin E causing a structural alteration in the globin chain. This combination will cause an intermediate form of hemolytic anemia. It is more common in Asians, Cambodians, , Laos and the people of Thailand. (Garrison & Peterson)

In some forms of thalassemia individuals may experience “ simultaneous anemia” and “ iron overload”. This is caused by red blood cells that have been destroyed prematurely. When excess iron is released from destroyed red blood cells, they collect in the tissue of the organs such as, the liver, joints, pancreas, heart and the pituitary gland. Frequent blood transfusions can also cause iron overload. Prior to 1970, iron overload was the main reason most children with major thalassemia died in their late teens to early twenties. (Garrison & Peterson)

An article from the Hematology Journal stated that “ the Italian Society of Hematology is taking new measures to manage iron overload in thalassemia major patients. “ Super conducting quantum inference devices, magnetic resonance imaging and oral iron chelators (deferipone and deferasisiox) are all being used. Guidelines are being set for these practices. (www. hemotolgy. com, 2008)

Diagnosing Thalassemia disorders can be done through various methods. Red blood cell indices are helpful in determining whether a patient has beta thalassemia. A hemoglobin electrophoresis with a finding of elevated Hgb A2 and F is noted. Both will e increased in Beta Thalassemia trait without iron deficiency and will be normal or decreased in Alpha thalassemia. It will read isolated in iron deficiency anemia. Iron deficiency can be ruled out by using free erythrocyte protoporphyrin transferrin saturation. Ferritian Saturation is a screening test in children who have hypo chronic microcytic anemia. (McPhee & Papadalis)

The Mentzer Index also helps distinguish between thalassemia and iron deficiency. This index divides the red blood cell count and puts them into a mean. The mean is called the Mean Corpuscular Volume or MCV. If the results are less than thirteen, Thalassemia is more likely. If the result is higher than thirteen, iron deficiency is more likely. When an MCV indicates numbers greater than eighty, the individual does not carry trait. Numbers less than eighty will indicate that the patient is not iron deficient, but may be a trait carrier. Thalassemia Intermedia diagnosis is made after a period of clinical observation. (McPhee & Papadalis)

In order for Thalassemia Major patients to survive, they need blood transfusions. Those who are chronically transfused need aggressive monitoring to maintain their well-being. Because blood transfusions can lead to iron overload, patients will need to be tested. Ferritin testing is effective for monitoring iron overload. There are several factors that can affect test results. Some of these factors are: inflammation, infection of the liver, breakdown of red blood cells, mistakes in sample handling, vitamin c deficiency and too much alcohol intake the night before testing. (McPhee & Papadalis)

A liver biopsy is the second measure in testing. However, it is not the choice of testing for thalassemia patients. The test is invasive and there is a longer recovery time. Direct sampling from the body tissue helps measure the iron overload. The third and most accurate for of testing is MRI based technology with R2 and T2 Techniques. Measuring iron overload in the liver is done with the R2 Technique and assessing cardiac damage from iron overload is done with the T2 Technique. (McPhee & Papadalis)

Treatment for Hemoglobin H disease is folate supplementation and avoidance of medicinal iron and oxidative drugs like sulfonamides. (www. thalassemia. com)

In the United States there are two approved iron chelators used for removing iron from iron overload. Desferal(deferoxamine) which is administered by subcutaneous infusion. The process takes eight to twelve hours and is done five to seven nights a week. In 2005, oral chelator, Engrade (deferansirox) was used. It is taken one time per day. (McPhee & Papadalis)

Symptoms of Thalassemia disorder vary upon the type of disorder. In the minors, individuals appear clinically normal. They have a normal life expectancy and have normal performance status. Some have mild microcytic anemia. With the majors B type for example, at birth children appear to be normal, but after six months they develop severe anemia due to the hemoglobin switches from F to A. These children will have growth failure, bone deformities, hepatosplendomegaly (enlargement of the liver and spleen) and are jaundice. They will survive into childhood but with bony deformities and hepatosplendomegaly. (McPhee & Papadalis)

In Italy, experimental bone marrow transplants are being researched and have proved the reversibility of cirrhosis of the liver in six patients cured of Thalassemia. Exact bone marrow donors are needed for this procedure. As with all surgeries there is a risk of infection or even death. Researchers are looking toward gene therapy as a possible curative approach. Gene therapy and fetal hemoglobin is a technique done, where the scientist insert a normal fetal gene into the stem cell of the unborn child having thalassemia. It is believed that one day it will be possible to cure an unborn child. (www. medicalnewstoday, 2010)

The best way to prevent this disorder is by educating others about it. Parental diagnosis prior to pregnancy, and genetic counseling have been saving lives. Knowing your own body by getting regular checkups and labs in addition to asking family members about their medical history is the best prevention for passing on a possible genetic disorder.

Bibliography:

(n. d.). Retrieved from www. thalassemia. com.

(2008). Hematology Journal, doi: 10. 3324/haematol. 12413.

(2008, April). Retrieved from www. hemotolgy. com.

(2010, 7 15). Retrieved from www. medicalnewstoday.

www. webmd. com/atozguide/thalassemia-topic-overview. (2011, July).

Garrison, C. D., & Peterson , C. M. (n. d.). The Iron Disorders Guide to Anemia. http://medical-dictionary. thefreedictionary. com/hemoglobin. (n. d.). McPhee, S. A., & Papadalis, M. A. (n. d.). 2011 Current Medical Diagnosis and Treatment. www. ncbi. nin. gov/pubmedhealth/PMH0001613. (n. d.).

www. nhlbl. nih. gov/health-topic/topics/Thalassemia. (n. d.).

www. thalassemia. us. (n. d.).