Thalassemia

Thalassemia is a blood disorder that is hereditary in nature. It is the most common disorder in the world that is hereditary and characterized by a single gene defect. Thalassemia was first discovered in individuals who inhabit South Asia and the Mediterranean region. The word thalassemia means sea, a translation from Greek. The syndrome of Cooley anemia, a severe thalassemia, was first described in 1925, to be caused by the complete absence of the production of beta-globin (Giardina & Hilgartner, 1992). Mild forms of thalassemia have also been discovered such as beta thalassemia minor, thalassemia intermedia, and alpha thalassemia. The characteristics of this disorder include few red blood cells and less hemoglobin in the red blood cells, which leads to transportation of low concentration of oxygen, thus causing fatigues in most of the patients. Hemoglobin is a protein that is rich in iron and has a role in transporting oxygen to various parts of the body, and also transports carbon dioxide to the lungs for exhalation. Most people suffering from thalassemia experience mild or severe cases of anemia, which is a disorder caused by low numbers of red blood cells, and low amounts of hemoglobin in the red blood cells (Giardina et al, 2008). Types and causes of ThalassemiaThere are different types of thalassemia such as alpha and beta thalassemia, Mediterranean, and Cooley’s anaemia.

Alpha thalassemiaThis disorder is commonly reported in people from the Middle East, Mediterranean region, India, North Africa, and Central Asia. This is an inherited disorder that reduces the production of hemoglobin, an iron-rich protein that is found in the red blood cells and are responsible for the transportation of oxygen to cells throughout the body. The reduction in the

levels of hemoglobin leads to little amount of oxygen reaching the cells in the body system. There are two categories of alpha thalassemia i. e. HbH disease and Hb Bart syndrome (hemoglobin Bart hydrops fetalis syndrome) or alpha thalassemia major. Alpha thalassemia usually results from deletions of two genes namely HBA1 and HBA2. It also results when a gene related to the alpha globin protein mutates. Silent carriers of the disorder have only one missing gene and do not exhibit any symptoms. Alpha thalassemia minor is caused when one misses two genes, and may have mild anemia. People found missing three genes develop hemoglobin H disease, which causes moderate to severe anemia (NHLBI, 2012). The most severe symptom or effect of alpha thalassemia is stillbirth. Other symptoms of alpha thalassemia include anemia as a result of shortage of red blood cells, pale skin, fatigue, weak joints and muscles, and other serious complications, though a good number of people diagnosed with alpha thalassemia show no signs or symptoms (DeBaun et al, 2007).

A human being requires two genes, one from each parent, to make sufficient beta globin protein chains for the normal functioning of the body system. Alteration of one or both of these genes results to beta thalassemia. Alteration of one gene makes one to be a carrier, a condition called beta thalassemia minor, characterized by mild anemia. Alteration of the two genes results into thalassemia major (Cooley’s anemia), characterized by severe anemia, or beta thalassemia intermedia, which cuses moderate anemia (Piomelli & Loew, 1990).

Inheritance Pattern for Beta Thalassemia

Fig. 1 Illustration of the inheritance of alpha thalassemia (NHLBI, 2012).

Who is at risk? The groups of people who are at risk of developing thalassemia are people of Chinese, African American, Asian, Mediterranean, or Filipino origin. The people at risk also include those with a family history of the disorder. This is because thalassemia is hereditary, i. e. it is passed from the parents to the offsprings. It is commonly transmitted from parents who have mutated/defective genes responsible for production of hemoglobin (Wing-Yan et al, 2010). Signs and symptoms of ThalassemiaThe major cause of thalassemia's signs and symptoms are the lack of or limited oxygen in the bloodstream. This occurs because the body system can not produce of synthesis sufficient hemoglobin and red blood cells. The type or severity of the disorder determines the level of severity of the symptoms. Silent carriers of the alpha thalassemia trait show no sign or symptoms of the disease because the body can surpass the lack of alpha globin protein as minor, and continue with the normal body processes. Mild anemia is exhibited by people having alpha or beta thalassemia trait, though some may not show symptoms apart from feeling fatigued (Giardina et al, 2008). Beta thalassemia sufferers show mild to moderate anemia plus a host of other health problems including slowed growth and development or maturity delay, complications of the bone such as expansion of the bone marrow making the bones become brittle or break easily. Another health complication associated with beta thalassemia is enlargement of the spleen because the spleen has to function extra hard in order to fight infection and remove waste substances. This results in severity of anemia, and can be dealt with by removing the spleen once it becomes too large. Beta

thalassemia major (Cooley’s disease) or hemoglobin H disease causes severe signs and symptoms of severe anemia together with other health complications. These include loss of appetite, slow development or growth, puberty delays, fatigue, shortness of breath, pale skin, dark urine due to degeneration of rd blood cells, jaundice, bone complications in the face, and enlargement of the liver, spleen or heart (NHLBI, 2012). Tests for ThalassemiaPeople with thalassemia minor do no necessarily need to do blood tests. Most instances of thalassemia can be diagnosed through blood tests, specific hemoglobin tests, complete blood count, or mutational analysis when hemoglobin electrophoresis fails. The types and levels of hemoglobin in the blood can be diagnosed by the hemoglobin test. CBS tests, on the other hand, measures the levels of hemoglobin and red blood cells. Positive CBC tests show less hemoglobin and few red blood cells. Diagnosis in early childhood can reveal the symptoms of moderate and severe thalassemias, including anemia (NHLBI, 2012). Genetic studies can also be carried out to diagnose thalassemia because of the hereditary nature of the disease. This entails doing blood tests on family members as well as evaluating the family history. Prenatal testing is also encouraged for expectant mothers whose partners or relatives are thalassemia sufferers or carriers. Treatment of ThalassemiassThe type and severity of thalassemia determines the nature of treatment for the disorder. Little or no treatment is necessary for carriers or people with alpha or beta traits. Standard treatment procedures are normally applied for moderate to severe forms of thalassemias, which include iron chelation, blood transfusion, and supplements of folic acid. Blood transfusion is the main treatment for moderate to severe thalassemias, as a source of red blood cells with normal

and healthy hemoglobin. Repeated transfusions are required to maintain the standard level of hemoglobin and red blood cells. This procedure enables one to feel normal, continue with their daily activities, and live through to maturity or adulthood. The limitations of blood transfusion include risk of getting viruses such as hepatitis and other infections, and it is also expensive. Regular blood transfusions also cause a build up of iron in the blood, causing damage to the heart, liver or spleen. This condition is called iron overload and can be prevented by iron chelation therapy (Giardina et al, 2008). Iron chelation therapy involves the use of two medicines, deferoxamine which is in liquid form and administered under the skin. However, it is painful and can cause side effects such as hearing or visual complications. The other medicine used in this therapy is deferasirox which is an everyday pill but causes side effects such as diarrhea, nausea, vomiting, headaches, fatigue, or joint pains (Olivieri & Brittenham, 1997). Folic acid supplements are also effective in treating thalassemias because they assist in building up the reserves of healthy red blood cells. It is advisable to take these supplements alongside other treatment option such as blood transfusion or iron chelation therapy. Coping with the disorderPeople living with thalassemias need to cope up with the disorder by adopting strategies such as strictly following the treatment procedures, get ongoing medical care, take initiatives and steps of healthy living, and seek help and support from the relevant people or institutions. This disorder can lead to heart failure resulting deaths at an early age, therefore, people suffering from thalassemia need to seek treatments as early as possible. Untreated thalassemia can also lead to liver failure and development of infections (Giardina et al, 2008). Genetic and prenatal counseling should be

adopted by people with a family history of thalassemia, and are planning to get children. People are encouraged to take frequent tests on the complete blood counts, iron levels, vision and hearing tests, and check for build up for iron, tests for assessing functions of the liver, heart, spleen and for viral infections (Rund & Rachmilewitz, 1995). Future research on treatmentOther treatments are still under screening and trials and include blood and marrow stem cell transplants, insertion of genes of normal hemoglobin or stimulation of fetal hemoglobin production. Research is still underway to invent ways of increase the production of healthy red blood cells with normal hemoglobin. There are also attempts to find new medicines for iron removal, trials to evaluate the possibility of transplanting normal stem cells and bone marrow, as well as gene therapy to add normal genes as a substitute for thalassemia genes.