Thalasemmia Essay, Research Paper

Over 2,000,000 American’s are carriers of the genetic trait for Thalassemia, a fatal blood

disease. It is also the most common single gene disorder in the world. This disease which

prevents normal hemoglobin production in the blood, is curently uncurable, and until

recently only let its victim live no longer then the first decade of their life. Thalassemia is a

serious genetic disease that afflicts children and adults all around the world.

In order to understand Thalassemia, one must understand the physiolgy of blood.

Blood, the carrier of nutrition and waste in our bodies, contains a protein called

hemoglobin. Hemoglobin is solely responsible for the transport of oxygen from the lungs

to cells through out the body. It is imperative that hemoglobin is readily avaible in the

blood to ensure cells can function properly. There are three types of hemoglobin. One is

Hemoglobin A, which is the most prevalent in adults. Hemoglobin A is composed of two

alpha globins and two beta globins. There are two mior hemoglobins names A2 and F.

Hemoglobin A2 is composed of 2 alpha and two delta globins. Finally, hemoglobin F,

predominatntly found in infants, is composed of 2 alpha globins and 2 gamma globins. In a

normal human being the globins that make up hemoglobin would be produced at certain

times in a human’s life. Initially, while it is a fetus, the human would contain high amounts

of hemoglobin F, and thus be producing alpha and gamma globins in majority. However

once a baby is born, gamma globin production drops and is countered by beta globin

production, thus allowing hemoglobin A to be created. Delta globins also increase once a

human is growing but they are rather insignificantly low in numbers.

In a thalassemia patient, the genes that code for the production of certain globlins

are either mutated or destroyed. This misinformation in the genetic code, leads to an

abnormal ratio of globins leaving too many unpaired globins and the expression of

thalassemia. In thalamessia, usually one type of globins is produced at normal levels while

is pair is not. The globin produced in normal levels and those that do not becom paired

for red cell aggregates which prove to be harmful to red blood cells. These aggregrates,

destroy the cell membrance of corpuscles which leads to hemolysis, the destruction of red

cells, or eeythropoices, the abnormal growth of red blod cells. The amount at which these

red cell aggregrates and the properties of them define which type and the serverity of

thalassemia a patient has.