Hereditary Spherocytosis

Fear comes from the unknown. When someone you love becomes ill and doctors cannot figure out what is wrong, fear sets in. This happened to my family when my grandfather became ill a few days after I was born. Today, I will inform you about hereditary spherocytosis, a semi-rare blood condition that affects my grandfather, my aunt, and myself.

Hereditary spherocytosis occurs when a protein in the red blood cell’s surface membrane becomes defected. This causes the defected blood cell to take on a spherical shape instead of a biconcave disk shape. The defected cells are referred to as spherocytes. The decrease in surface area makes it difficult for the spherocytes to receive an adequate amount of oxygen. This leads to a decrease in oxygen throughout the entire body. The cell’s fragile cytoskeleton causes the cells to break down faster. A regular cell’s lifespan is anywhere from 90-120 days, where as a spherocyte’s lifespan is only 30 days. The spleen has to work harder to recycle and break down the excessive amount of old blood cells.

Now that you all know the cause and definition of hereditary spherocytosis, I will now discuss the symptoms and side effects, and give an insight to my family’s struggle with the disorder. Spherocytosis usually becomes present in infancy or early childhood, but sometimes is not diagnosed until a person reaches middle-age. My grandfather was diagnosed with hereditary spherocytosis in the summer or 1990 at the age of 57. He was hospitalized due to a rapid onset of acute gallbladder pain. During surgery, the physician discovered that gangrene had taken over his gallbladder. He quickly ordered a team of doctors to perform a variety of tests. Six months later, the doctors discovered he had hereditary spherocytosis. This was an answer to many unanswered questions throughout his lifetime. Unexplained jaundice, frequent headaches, pallor, abdominal pain, shortness of breath, and fatigue often occurred about once every 1-2 months throughout my his earlier years. A normal viral infection would take weeks, instead of days, to get over. I began experiencing all of the same symptoms when I was 15. At the age of 16, I became sick with mononucleosis, and it took me 6 months to recover from. As I became progressively worse, my grandmother decided to take me to my grandfather’s hematologist. At first, I was not tested for hereditary spherocytosis, because doctors have never known the condition to skip a generation. Doctors thought that since my mom did not test positive for the condition when the doctors tested her and her sister in 1990, that I had no chance of having it. The hematologist finally tested me, and called me that evening to tell me he indeed saw spherocytes on the smear.

High levels of bilirubin in the bloodstream cause the jaundice appearance. The excess bilirubin is the result of the rapid breakdown of red blood cells, known as hyperbilirubinemia. Too much bilirubin can cause gallbladder pain, and eventually gallstones. Anemia is another primary symptom. The anemia is a result of too few red blood cells, which is highly likely because of their rapid breakdown. The anemia can also result from a sudden stop of production of red blood cells in the bone marrow.The bone marrow is already overworking to try and produce a normal amount of red blood cells, so if a person contracts a normal viral infection, it can slow the red blood cell production down to a dangerously low level.This is known as an aplastic crisis. An enlarged spleen is also very common in people with hereditary spherocytosis.When the spleen becomes clogged with broken red blood cells, it causes the spleen to swell. The enlargement of the spleen is known as splenomegaly, a dangerous condition because there is an increased chance of rupturing the spleen, causing internal bleeding. Now that I’ve informed you of the symptoms, I will talk to you about some common and uncommon ways to manage the disorder, and what I have learned works best for me.

Anemia causes fatigue, irritability, pallor, and bruising. A diet high in iron, daily iron supplements, and annual iron infusions along with vitamins B-12 and folic acid can all reverse symptoms of anemia. I eat a diet high in folic acid, I take B-12 supplements, and try to get enough iron from dietary sources. But because I am a vegetarian, my body does not have a high reserve of iron, so every 4-8 months I receive an intravenous iron infusion. If I have an episode of anemia, I usually am jaundice also, since both are the result of blood cells breaking down faster than the body can replenish them. An intravenous sugar water infusion helps calm the blood cells and stops them from bursting in such high numbers. It also reduces the size of the spleen and replenishes the body’s oxygen levels. There is little medical evidence that shows the sugar water infusion to be an effective treatment, but my hematologist swears by this method. There is no preventative treatment to protect the gallbladder from getting gallstones. Many people with hereditary spherocytosis will eventually have to have their gallbladder removed. My grandfather and my aunt have both had their gallbladder removed, and I’m sure someday I will have to have mine removed.

I am not sure the challenges that lay ahead of me, but for now I am just thankful that I am a happy, healthy, normal 22 year old. There are millions of people suffering with conditions way worse than the highly manageable hereditary spherocytosis. I hope that you have gained some knowledge of what causes hereditary spherocytosis, the symptoms, and some ways to manage the condition. I also feel that through a personal insight of how the condition has affected my family, you will have a better understanding of what it is like to have hereditary spherocytosis.

Works Cited

Gonzalez, MD, Gus. "Spherocytosis, Hereditary." *EMedicine - Medical Reference*. Medscape, 27 Aug. 2009. Web. 17 July 2010. <http://emedicine.medscape.com/>.

"Medical Dictionary Definitions of Popular Medical Terms Easily Defined on MedTerms." *Hereditary Spherocytosis Definition*. MedicineNet, Inc. Web. 17 July 2010. <http://www.medterms.com/>.

Perrell, William R. "My Grandfather's Struggle With Hereditary Spherocytosis." Interview. Print.

Pineda, MD, Luis F. "Hereditary Spherocytosis." Interview. Print.