Pathology Defined: Anatomy and Physiology

Thalassemia is a blood disorder that is inherited through genes. It causes the body to have/produce less healthy erythrocytes and hemoglobin. Hemoglobins are iron-rich proteins in the erythrocytes. They also carry CO2 (carbon dioxide) from the body to the lungs where it's expirated (Locicero, Zieve, Conaway, 2018). There are two types of hemoglobin, which means that there are two types of thalassemia. The two types of hemoglobin are alpha globin and beta globin (Locicero et al., 2018). That means that the two types of thalassemia are alpha thalassemia and beta thalassemia. The subcategories of alpha and beta thalassemia are thalassemia major and thalassemia minor (Locicero et al., 2018).

Patient Population

The inherited blood disorder affects mainly Asian, Chinese, Mediterranean, and African American ethnicities. The alpha thalassemia mostly occurs within people from Southeast Asia, Middle East, China and the African descent (Locicero et al., 2018). The beta thalassemia often occurs in people of Mediterranean origin (Locicero et al., 2018). Carriers of the thalassemia genes may not know that they carry the genes. The symptoms do not show up within the carriers. If both of the parents contain the thalassemia genes, their child will most likely have thalassemia (Miller, 2015).

Signs and Symptoms

In beta thalassemia major, infants become pale, have feeding problems, diarrhea, irritability, recurrent fevers, and large abdomen. Patients that have beta thalassemia intermedia showed signs of paleness, jaundice, and leg ulcers (Cao & Galanello, 2010). If an unborn baby has the most severe form of alpha thalassemia major, this causes a stillbirth. Some more

symptoms with children include, facial bone deformities, fatigue, failure of growth, shortness of breath, and jaundice (LoCicero et al., 2018).

Differential Diagnosis

There are 2 types of Thalassemia as stated in the first paragraph. There is Alpha Thalassemia and Beta Thalassemia. But there are also more sub-categories to those two types. In alpha thalassemia, there are, silent alpha thalassemia, alpha thalassemia minor, hemoglobin H disease, and alpha thalassemia major. In beta thalassemia, there are, beta thalassemia minor, beta thalassemia major, and beta thalassemia intermedia. (Miller, 2015). When there is one missing gene in a child, that means that the child is a silent alpha thalassemia carrier. There are no signs or symptoms, but that means their child will have the thalassemia disease (Miller, 2015). An alpha thalassemia minor is also known as having an alpha thalassemia trait means that there are two missing or mutated genes (Miller, 2015). When there are three mutated or missing genes, that is called hemoglobin H disease. The signs and symptoms could range from average to serious (Miller, 2015). If your child has four absent or mutated genes, it is what you call alpha thalassemia major, or also known as hydrops fetalis. This type of thalassemia can cause your fetus to die before its delivery date or die a little while after birth (Miller, 2015). If a person has one of their beta globin genes mutated, their condition is called beta thalassemia minor, or also called beta thalassemia trait (Miller, 2015). In beta thalassemia major, or also known as Cooley's anemia, both beta globin genes are mutated. These mutations are very severe. If a baby has beta thalassemia major, the baby would seem normally healthy for their first 2 years of life (Miller, 2015). The child would soon have a life-threatening anemia that would need regular blood transfusions. Finally, in beta thalassemia intermedia, both their beta globins are mutated, but

their mutations are not as severe as it is in beta thalassemia major. This type of beta thalassemia does not regularly require blood transfusions (Miller, 2015).

Diagnostic Exams related to pathology

Carrier screening was initiated to countries that had a high population with beta thalassemia. Another way to diagnose someone with thalassemia is geno/phenotype correlations. But using this type of diagnosis is a very limited practical use (Ioannides, 2013). This is due to the complications of identifying other genome studies within the human body (Ioannides, 2013).

Medical Terms related to pathology

An erythrocyte is a another word for a red blood cell (Chabner, 2017). Erythrocyte is related to this pathology because thalassemia occurs within an erythrocyte. Same as the word Thrombocyte, it is a another term for platelet (Chabner, 2017). The word is related to thalassemia because patients tend to have a low platelet count (Chabner, 2017). Another medical term related to our pathology is anemia. Anemia is the underlying diagnosis in thalassemia. Hemoglobin in this pathology means that patients are not able to produce enough hemoglobin, or protein to transport oxygen, within their blood. Leukocyte means white blood cell, in which patients who have thalassemia often have an elevated white blood cell count (Chabner, 2017).

What is the prognosis?

Within patients with thalassemia minor, their prognosis has a very well life expectancy. Patients that have thalassemia intermedia, their life expectancy is also very well (Galanello & Origa, 2010). Patients who do have severe hemosiderosis in thalassemia intermedia are susceptible to cardiac problems. A great deal of transfusion-dependent patients have increased their life expectancy with the help of the DFO, or also called deferoxamine (Galanello & Origa,

2010). Patients who have beta thalassemia major usually have a life expectancy of 17 years. They usually die by the age of 30 years (Muncie & Campbell, 2009).

Synthesis of recent peer review academic article related to the pathology

A hereditary hemoglobin disorder, also known as haemoglobinopathies, include beta thalassemia. There have been a high cardiac mortality rate with patients who have haemoglobinopathies (Ioannides, 2013). There are two main denominations of the cardiovascular phenotype in haemoglobinopathies. It is the underlying molecular defects and the therapy. Within the beta thalassemia, the most basic deficiency is the quantitative and the expenditure of the hemoglobin chain. This leads to cardiovascular complications with the left and right ventricular abnormal and pulmonary hypertension (Ioannides, 2013).

References

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