Non-Immune Hemolytic Anemia (Hemoglobinopathy)

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Abstract
Hemoglobinopathy is an autosomal recessive disorder that is classified into 2 major groups, namely thalassemia syndromes and structural variants of the hemoglobin chain. Thalassemia is a group of genetic disorders caused by decreased speed of chain synthesis or. The second group, structural abnormalities in the hemoglobin chain, often causes a condition called sickle cell (sickle cell anemia), this structural abnormality occurs due to the substitution of adenine by thymine which causes the conversion of the amino acid glutamate to valine. 7% of the world's population are carriers of hemoglobinopathies. This type of research is literature research or literature review. Based on data from the Indonesian Thalassemia Foundation (YTI) in 2008-2017, 5.8% of thalassemia carriers were found from 12,038 people examined, and 28.62% of 4,137 people according to the results of family screening, thalassemia in 2009-2017. The prognosis of thalassemia disease depends on medication adherence and the type of thalassemia suffered. Death in patients with sickle cell anemia is usually due to co-morbidities such as renal and other organ failure. Early and adequate symptom management can improve the prognosis. This disorder is autosomal recessive which is classified into 2 major groups, namely thalassemia syndrome and structural variants of hemoglobin chains that often produce sickle cell shape

Keywords: Hemoglobinopathy; Thalassemia; Sickle Cell Anemia.
**Introduction**

Anemia is a condition where the concentration of hemoglobin or the number and size of red blood cells is below the normal value so that it can interfere with oxygen transportation throughout the body (Organization 2014). Anemia caused by increased erythrocyte destruction is called hemolytic anemia. Hemolytic anemia occurs due to erythrocyte destruction that is faster than the ability of bone marrow erythropoiesis, this type of anemia is divided into two, namely immune hemolytic anemia and non-immune hemolytic anemia (Tanto et al. 2014). One of the disorders of non-immune hemolytic anemia lies in gene abnormalities, resulting in abnormal hemoglobin production, this disorder is called hemoglobinopathy (Manukiley and Perdani 2017). Hemoglobinopathy is an autosomal recessive disorder that is classified into 2 major groups, namely thalassemia syndromes and structural variants of the hemoglobin chain (Payandeh et al. 2014). Thalassemia is a group of genetic disorders caused by the decreased speed of or chain synthesis. The second group, structural abnormalities in the hemoglobin chain, often causes a condition called sickle cell anemia, these structural abnormalities occur due to the substitution of adenine by thymine which causes the conversion of the amino acid glutamate to valine (Kaushansky 2010).

About 7% of the world's population are carriers of the hemoglobinopathy trait. Hemoglobinopathy is most common in the Mediterranean area and most parts of Asia and Africa. Germany is one of the countries experiencing an increasing prevalence of hemoglobinopathies, about 9 million immigrants from this country are at risk of hemoglobinopathies and about 4.5% of the entire population has a carrier gene for hemoglobinopathies (Kohne 2011). In addition, according to a study in Western Iran, -thalassemia minor hemoglobinopathies are the second most common cause of microcytic hypochromic anemia (Payandeh et al. 2014). Based on data from the Indonesian Thalassemia Foundation (YTI) in 2008-2017, there were 5.8% of thalassemia carriers from 12,038 people examined, and 28.62% of 4,137 people according to the results of thalassemia family screening in 2009-2017. In addition, based on data from the RSCM in 2016, there were 9,131 thalassemia patients registered in Indonesia (Salsabila, Perdani, and Irawati 2019). Initially, hemoglobinopathy, a type of sickle cell structural disorder, was an evolution of Sub-Saharan African and black populations so that falciparum malaria could not infect them (Kaushansky 2010). But of course, the subsequent development of this sickle cell structure can cause the sufferer to experience hemolytic anemia.

Hemoglobinopathy has a variety of clinical manifestations ranging from asymptomatic to lethal. In addition, complications caused by the presence of hemoglobinopathies can affect several organs. This shows the importance of understanding hemoglobinopathies in order to be able to diagnose as early as possible and provide appropriate treatment so as to minimize the poor prognosis in patients with hemoglobinopathies, especially thalassemia and sickle cell.
Method

This type of research is literature research or literature review. Literature review is a search and literature research by reading books, journals, and other publications related to research topics, to produce a particular topic or issue, such as to produce a scientific paper, such as thesis, thesis, and dissertation.

Result and Discussion

Definition

Hemoglobinopathy is an inherited disorder of globin, the protein component of hemoglobin (Hb). This disorder is autosomal recessive which is classified into 2 major groups, namely thalassemia syndrome and structural variants of hemoglobin chains that often produce sickle cell shape.

Thalassemia

Thalassemia is a type of hemoglobinopathy caused by a decrease in the rate of synthesis of or chains. This genetic disorder is caused by mutation of one or more globin chains, resulting in an imbalance in globin chain production, ineffective erythropoiesis, hemolysis, and ultimately anemia (Hoffbrand et al. 2019; Kaushansky 2010).

Sickle cell

Sickle cell anemia is a group of hemoglobin disorders caused by sickle β-globin gene, resulting in the structure of sickle hemoglobin (Hb S). This disease occurs due to homozygous mutations or a combination of heterozygous mutations of Hb S and thalassemia variants such as Hb C, Hb D, or Hb E (Hoffbrand et al. 2019; Kaushansky 2010).

Figur 1. Genetika Talasemia α

The second type, thalassemia, occurs due to the absence of (β ) chains or few (β+) chains. The absence of globin chains causes an imbalance of globin chains. Excess chains precipitate in erythroblasts and mature erythrocytes, causing ineffective erythropoiesis and severe hemolysis. The greater the excess of the chain, the greater the anemia. The production of chains helps to “clean up” excess chains and relieves anemia, a condition called Hb A2 (α 2γ2) (Hoffbrand et al. 2019).
Sickle Cell Anemia

Sickle cell disease occurs due to a hemoglobin disorder caused by inheritance of the sickle globin gene. Homozygous sickle cell anemia (Hb SS) is the most severe syndrome. Molecular pathology of sickle cell anemia occurs due to a single base change in DNA that codes for an amino acid at position six in the -globin chain where adenine is replaced by thymine so that the amino acid glutamate that should be formed turns into valine (Hoffbrand et al. 2019). Heterozygous sickle cell or trait sickle cell (Hb AS) is a benign condition without anemia with Hb S varying from 25% to 45% of total hemoglobin. An estimated 300 million people carry the trait around the world (Hoffbrand et al. 2019; Kaushansky 2010). In addition, sickle cell anemia may result from a combination of hemoglobin S with other genetic defects of hemoglobin. The most common combinations are /Hb S and Hb C/Hb S thalassemia. Hemoglobin C (Hb C) disease is caused by the substitution of lysine for glutamic acid in the globin chain at the same point as the substitution on Hb S.

![Figur 2. Sickle cell anemia pathology](image)

Sickle cell anemia pathology

In addition to the above abnormalities, there are also other abnormalities in hemoglobin such as hemoglobin D disease, which is a group of variants that have the same electrophoretic mobility, patients in the heterozygous state do not show symptoms while the homozygous disorder suffers from mild hemolytic. In addition, there is hemoglobin E disease. In the homozygous state, there is mild hypochromic microcytic anemia but if there is a combination of /Hb E thalassemia, clinically it resembles homozygous B° thalassemia.

Risk Factor

Hemoglobinopathies are generally autosomal recessive disorders so that heredity factors greatly determine the offspring who are at risk of getting defective genes in hemoglobin such as thalassemia or other hemoglobin defects (Rodiani and Anggoro 2017). If a pregnant woman is found to have a hemoglobin disorder then her partner should be checked to see if she also carries the defect. If both carry the defect, their child will be at risk for serious defects, especially -thalassemia major. Therefore, it is important out antenatal diagnosis using the amplification refractory mutation system to carry(ARMS) and PCR as well as PCD Ddel analysis for sickle cell anemia (Hoffbrand et al. 2019). According to Mendel’s Law, the transmission of thalassemia to the next generation can be seen in the figure below (12).
Pathophysiology

Thalassemia

Hemoglobin consists of protein and non-protein. The non-protein structure is heme which consists of iron and protoporphyrin, while the protein structure is a globin chain which consists of 2 alpha (α) chains and 2 betas (β) chains. Decreased synthesis of globin chains can reduce the concentration of hemoglobin and red blood cells, causing symptoms of anemia. Defects in decreased synthesis of globin chains, either or chains, are called thalassemias. This defect in globin chain synthesis will result in an imbalance of globin chains in the hemoglobin molecule so that it will form an insoluble homotetramer, resulting in precipitation in red blood cells. This triggers hemolysis of red blood cells in the spleen and infective erythropoiesis. The formation of ineffective erythropoiesis occurs because red blood cells are formed imperfectly due to defects in their globin chains (Kaushansky 2010).

Sickle Cell
A defect in the globin chain on chromosome 11 in which adenine is substituted for thymine so that the amino acid glutamate changes to valine. The amino acid glutamate is hydrophilic while the amino acid valine is hydrophobic, in the presence of this defect hemoglobin becomes deoxygenated (Deoxy Hb S). HbS undergoes polymerization into long molecules. This resulted in a change in cell membranes where an influx of Ca$^{2+}$ and release of potassium, followed by H$_2$O so that the cells become dehydrated and cell concentration increases. In the end, the cell will be sickle-shaped (sickle cell). This sickle cell form will be destroyed by macrophages in the spleen which is called extravascular hemolytic / spleen hemolytic anemia. This happens because the cells have an abnormal shape. In addition, this sickle cell shape can cause a vaso-occlusive crisis. Cells will be blocked in the capillaries and cause ischemia in these tissues. The blockage stimulates the adhesion and aggregation of inflammatory factors such as white blood cell adhesion, activation of inflammatory cytokines that will exacerbate occlusion and ischemia. Hemolysis can also occur in blood vessels, thereby disrupting NO regulation and causing endothelial dysfunction (Gardner 2018; Hoffbrand et al. 2019; Kaushansky 2010).

Clinical Manifestations And Complications

Thalassemia
1. Severe anemia at the age of 6 months after birth when the chain changes to. Especially for the thalassemia major.
2. Enlargement of the liver and spleen due to excessive destruction of erythrocytes, and extramedullary hemopoiesis.
3. Widening of the bones due to bone marrow hyperplasia which causes thalassemia facies and thinning of the cortex in the bones with a tendency to fracture and protrusion of the skull bones with the appearance of "standing hair".
4. There can be the accumulation of iron, especially in thalassemia major. This occurs due to repeated transfusions.
5. Infectious complications. Infections are very susceptible to occur in children with anemia such as pneumococci and meningococci.
6. Complications of osteoporosis. It occurs frequently in diabetic patients with endocrine disorders and with bone marrow expansion caused by infective erythropoiesis (Hoffbrand et al. 2019).
Non-Immune Hemolytic Anemia (Hemoglobinopathy)

Figur 6. a. The skull is prominent in a child with thalassemia major.
b. X-ray of the skull of a child with thalassemia major. There is an image of “hair standing up” (Hoffbrand et al. 2019)

Sickle Cell

Symptoms of anemia depend on Hb S which is relatively easier to release oxygen to tissues compared to Hb A. Clinical expression of Hb SS varies widely, such as crises that can clog blood vessels (vaso-occlusive), visceral sequestration crises, aplastic, and hemolytic crises.

1. Vaso-occlusive
   Crisis This crisis is triggered by factors such as infection, acidosis, dehydration, and deoxygenation. Infarction can cause pain in the bones that can lead to “hands and feet” syndrome. This crisis is most severe in the brain because it can cause a stroke.

2. Visceral sequestration
   Crisis This crisis is caused by sickle formation in the organs and collection of blood. Dating patients with dyspnea, Po$_2$, decreased arterial chest pain, and pulmonary infiltrates on X-ray beam. Splenic sequestration usually manifests as an enlarged spleen, decreased hemoglobin, and abdominal pain.

3. Aplastic
   Crisis This crisis occurs when there is a complication of parvovirus infection or folate deficiency and is characterized by a sudden decrease in hemoglobin, usually requiring transfusion.

4. Hemolytic crisis
   There is an increase in hemolysis and a decrease in hemoglobin and an increase in reticulocytes. Other clinical symptoms can include complications that have occurred
such as lower leg ulcers as a result of vascular stasis and ischemia, enlarged spleen in infants, pulmonary hypertension, and increased tricuspid regurgitation rate. In addition, complications are often found in the form of proliferative retinopathy and osteomyelitis can also occur if there are complications due to Salmonella spp infection. (Hoffbrand et al. 2019)

**Figur 8.** 15 year old child with necrosis and ulcer formation complicated by sickle cell anemia. (Hoffbrand et al. 2019)

**Enforcement Of Diagnosis**

1. Preparations to remove peripheral blood

   Patients with thalassemia found severe hypochromic microcytic anemia and an increased percentage of reticulocytes with nucleated erythrocytes, target cells, and basophilic dots.

**Figur 9.** Preparation Of Peripheral Blood Smear In Thalassemia Major
Figure 10. a. it appears that there are erythrocyte cells in the form of sickles. In figure b is an example of morphological images of homozygous Hb C disease that shows many target cells, and dark-islanded spherical cells. (Hoffbrand et al. 2019; Kaushansky 2010)

2. **High-performance liquid chromatography (HPLC)**

HPLC or hemoglobin electrophoresis method. This method is the first-line method for diagnosing hemoglobin disorders. This method can also help in the diagnosis of hemoglobin disorders.

3. **DNA analysis**

DNA analysis was used to identify defects in each allele. Samples are obtained by biopsy of the chorionic villi and occasionally amniotic fluid cells. The DNA was then analyzed using the polymerase chain reaction (PCR) method. PCR is a method that can be carried out using primer pairs that only amplify a single allele or using consensus primers that amplify all alleles followed by restriction digestion to detect specific alleles.
Example by Hb S; On HbS, the Del enzyme detects changes in adenine to thymine. The image below represents the antenatal diagnosis by Del PCD analysis. In this case, the DNA taken from the chorionic villi samples showed the presence of Hb As.

![Image of DNA samples](image)

**Figure 11.**

A pre-implantation genetic diagnosis can be made to avoid hemoglobin abnormalities in the next generation including the implementation of *in vitro fertilization* conventional, followed by removal of one or two cells from the blastomere on the third day. PCR is used to detect thalassemia mutations so that blastomeres with DNA defects can be avoided and cells with normal genes can be selected for implantation (Hoffbrand et al. 2019; Kaushansky 2010).

**Classification**

Hemoglobinopathy is an autosomal recessive disorder that is classified into 2 major groups, namely thalassemia syndromes and structural variants of the hemoglobin chain (Payandeh et al. 2014). Thalassemia is a group of genetic disorders caused by the decreased speed of or chain synthesis. The second group, namely structural abnormalities in the hemoglobin chain often causes a condition called *sickle cell anemia*, this structural abnormality occurs due to the substitution of adenine by thymine which causes the conversion of the amino acid glutamate to valine (Kaushansky 2010). The detailed classification of various types of hemoglobinopathies can be seen in the table below (Kohne 2011).
### TABLE 2

<table>
<thead>
<tr>
<th>Thalassemia Subtype</th>
<th>Red Blood Cell Count</th>
<th>Hemoglobin Pattern</th>
<th>Cardiac Symptoms</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>α-thalassemias</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Normal findings</td>
<td>--</td>
<td>--</td>
<td>--</td>
</tr>
<tr>
<td>Heterozygous α-thalassemia</td>
<td>αα/αα</td>
<td>Hb normal, MCHC normal</td>
<td>--</td>
</tr>
<tr>
<td>Homozygous α-thalassemia</td>
<td>αα/αα</td>
<td>Hb normal, MCHC &lt; 27 g</td>
<td>--</td>
</tr>
<tr>
<td>Homozygous α-thalassemia</td>
<td>αα/αα</td>
<td>Hb normal or low, MCHC &lt; 29 g</td>
<td>--</td>
</tr>
<tr>
<td>Homozygous α-thalassemia</td>
<td>αα/αα</td>
<td>Hb normal or low, MCHC &lt; 24 g</td>
<td>--</td>
</tr>
<tr>
<td>Mixed heterozygosity, α+β-thalassemia</td>
<td>α/-β+</td>
<td>Hb 10 to 12 g/dL, MCHC &lt; 22 pg</td>
<td>HbH = 10 to 20%</td>
</tr>
<tr>
<td>Homozygous α-thalassemia</td>
<td>αα/αα</td>
<td>Hb 11 to 13 g/dL, MCHC 50 to 72 g, MCH 14 to 20 pg</td>
<td>HbHभ up to 90%, Hb Bart's up to 90%, Hb F up to 20%</td>
</tr>
</tbody>
</table>

### TABLE 3

<table>
<thead>
<tr>
<th>Hemoglobinopathies</th>
<th>Cell Type</th>
<th>RBC Count</th>
<th>Novel Hemoglobin</th>
<th>Cardiac Symptoms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sickle-cell disease</td>
<td>HbEβthal</td>
<td>Hb 6 to 9 g/dL, normochromic sickle cells</td>
<td>HbEβthal &gt; 45%</td>
<td>Acute organ syndromes, Sickle-cell crisis, Cardiac hemosiderosis</td>
</tr>
<tr>
<td>HbS heterozygosity</td>
<td>HbAS</td>
<td>Normal</td>
<td>HbS up to 40%</td>
<td>Variable sickle-cell disease</td>
</tr>
<tr>
<td>Sickle-cell β-thalassemia</td>
<td>HbSβthal</td>
<td>Hb 5 to 12 g/dL, normochromic sickle cells</td>
<td>HbSβthal &gt; 50%</td>
<td>Severe sickle-cell disease, Acute organ syndromes, Cardiac hemosiderosis</td>
</tr>
<tr>
<td>Sickle-cell β-thalassemia</td>
<td>HbSβthal</td>
<td>Hb 6 to 10 g/dL, normochromic sickle cells</td>
<td>HbSβthal &gt; 50%</td>
<td>Variable sickle-cell disease, Acute organ syndromes, Cardiac hemosiderosis</td>
</tr>
<tr>
<td>HbC disease</td>
<td>HbCC</td>
<td>Hb 10 to 12 g/dL, Target cells, MCHC &gt; 35 g</td>
<td>HbC &gt; 50%</td>
<td>Mild hydrocytic anemia</td>
</tr>
<tr>
<td>HbC disease</td>
<td>HbCC</td>
<td>Hb 10 to 12 g/dL, Target cells, MCHC &gt; 35 g</td>
<td>HbC &gt; 50%</td>
<td>Mild hydrocytic anemia</td>
</tr>
<tr>
<td>HbC heterozygosity</td>
<td>HbAC</td>
<td>Normal</td>
<td>HbC &gt; 50%</td>
<td>Variable sickle-cell disease, Acute organ syndromes, Cardiac hemosiderosis</td>
</tr>
<tr>
<td>HbE heterozygosity</td>
<td>HbEβthal</td>
<td>Hb 10 to 14 g/dL, high Hb/E ratio, MCHC &gt; 35 g</td>
<td>HbEβthal &gt; 50%</td>
<td>Mild hydrocytic anemia</td>
</tr>
<tr>
<td>HbE disease</td>
<td>HbE</td>
<td>Hb 10 to 14 g/dL, Target cells, MCHC &gt; 35 g</td>
<td>HbE &gt; 50%</td>
<td>Acute organ syndromes, Cardiac hemosiderosis</td>
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<td>HbE β-thalassemia</td>
<td>HbEβthal</td>
<td>Hb 5 to 7 g/dL, normochromic sickle cells, Target cells</td>
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Treatment

Thalassemia

a. Pharmacology
   1. Regular folic acid (eg 5 mg/day) is given if the diet is not good.
   2. Iron chelate therapy. Iron chelating therapy is used to treat iron deposits. For example, deferoxamine is inactive orally. This drug is usually given by subcutaneous infusion of 40 mg/kg for 8-12 hours, 5-7 days per week. In infants with thalassemia major can be given this therapy 10-15 minutes after the blood unit has been transfused. Side effects of this drug are high-tone deafness, retinal damage, and growth retardation. In addition, there is the drug deferipron which is an orally active iron chelating drug. This drug may cause an excretory effect of iron in the urine. The drug is given 75 mg/kg in three doses daily. Deferiprone is more effective than deferoxamine in removing cardiac iron. Side effects include arteopathy, agranulocytosis, neutropenia, and gastrointestinal disturbances. The newest oral chelating drug is deferasirox. This drug is given at 20-40 mg/kg, the excretion of iron through feces only and does not have major side effects so that this drug is widely used.
   3. Endocrine therapy for patients with organ failure or to stimulate the pituitary if puberty is delayed (Hoffbrand et al. 2019).

b. Non-Pharmacology
   1. Blood transfusions to maintain hemoglobin above 10 g/dL. Usually requires 2-3 units every 4-6 weeks
   2. Splenectomy is performed to reduce the need for blood. This treatment should be delayed until the patient is older than 6 years because of the high risk of infection.
   3. Allogeneic stem cell transplantation. The success rate (long life in patients with thalassemia major) is over 80-90% in young patients with good chelation without hepatomegaly. The donor is usually a relative of the patient because of the match to the human leukocyte antigen (HLA) (Hoffbrand et al. 2019)

Sickle cells

a. Pharmacology
   1. Hydroxycarbamide (hydroxyurea) as much as 15-20 mg/kg. Works to increase Hb F levels. This drug should not be given during pregnancy
   2. Taking folic acid (eg 5 mg once a week)
   3. Pneumococcal, vaccination Haemophilus, and meningococcal
   4. Pain controllers, such as opioids, NSAIDs, acetaminophen, or a combination. For adults and children weighing more than 50 kg, morphine can be given starting at a dose of 0.1 – 0.15 mg/kg. (Hoffbrand et al. 2019; Kaushansky 2010)
b. Non-Pharmacology

1. Prophylactic, which is to avoid crisis triggering factors such as dehydration, infection, circulation stasis, and cooling of the skin surface.

2. Transfusion, sometimes given repeatedly as prophylaxis in patients who frequently experience crises or major organ damage (eg brain). The goal is to suppress Hb S production over a period of several months or even years. The accumulation of iron that occurs may require iron chelation therapy and alloimmunization of the donated blood.

3. Stem cell transplantation has a mortality rate of less than 10%. Transplantation is only used in the most severe cases where the quality or life expectancy is very low. (Hoffbrand et al. 2019; Kaushansky 2010)

Prognosis

Thalassemia

The prognosis in patients with β-thalassemia major is increased when treated adequately with transfusions and iron chelates. In studies reporting long-term use of desferrioxamine to be effective against the development of heart disease and maintaining sustained iron reduction, it was estimated that serum ferritin levels of less than 2500 mcg/L during 12 years of follow-up had a heart disease-free survival rate of 91%. If ferritin levels exceed these levels, the heart disease-free life expectancy is estimated to be less than 20%. In the second study, the survival and iron-free relationship were measured based on the value of iron storage in the liver. Patients who maintain an iron concentration of at least 15 mg of iron per gram of liver have a 32% chance of surviving to 25 years of age. No heart disease developed in patients who maintained liver iron levels below this threshold. This provides clear evidence that adequate transfusion and iron chelation are associated with longevity and good quality of life. On the other hand, poor adherence or unavailability of chelating agents is still associated with poor survival prospects. (Kaushansky 2010)

Sickle Cell

Since 1968, sickle cell disease (SCD) mortality has decreased. This coincided with the introduction of the pneumococcal conjugate pneumococcal vaccine (PVC7). There was a 61% reduction in infant mortality, 67% in children aged 1 to 4 years, and a 35% decrease in children aged 5 to 19 years. The average life expectancy of patients with Hb SS disease in the United States is 42 years for women and 48 years for men. Meanwhile, in Jamaica, the survival rate is 53 years for men and 58 years for women. As sickle cells age, the cause of death changes from infectious etiology. for those associated with end-organ damage, such as renal failure. (Kaushansky 2010)
Communication, Information, and Education (IEC)

IEC is very important for the prevention of hemoglobin disorders. Prevention can be achieved by prospective genetic counseling, i.e. screening the total population while children are in school and bringing warnings about the potential risks of marriage to other careers. Such an approach may be difficult in such a large population that prenatal diagnostic programs have been developed for the prevention of serious hemoglobin defects in later offspring (Kaushansky 2010). Prenatal diagnosis genetic counseling is important for couples who are at risk of having children with major hemoglobin defects. If a pregnant woman is found to have a hemoglobin defect, her partner should be examined to find out if she also carries the defect. If both are found to carry hemoglobin defects, the offspring are at risk for serious defects, such as thalassemia major. (Hoffbrand et al. 2019)

KIE in patients with hemoglobinopathy or a defect in their hemoglobin is in the form of advice to take medication according to a doctor’s prescription and routinely carry out examinations to monitor the development of complications that occur. For example, thalassemia patients who receive blood transfusions in order to suppress the symptoms of anemia have a risk of iron accumulation, so they must take iron-chelating drugs as recommended by the doctor. If not done, serious complications can occur in organs such as the heart, liver, etc. due to the accumulation of iron.

Conclusion

Hemoglobinopathy is an inherited disorder of globin, the protein component of hemoglobin (Hb). This disorder is autosomal recessive which is classified into 2 major groups, namely thalassemia syndrome and structural variants of hemoglobin chains that often produce sickle cell shape. This defect causes clinical manifestations in the form of hemolytic anemia with microcytic hypochromic morphology. Abnormalities in this globin chain have many classifications depending on which gene or chromosome is mutated/defected and has a different prognosis. Management of hemoglobinopathies should be carried out as carefully as possible to get a better prognosis. And the importance of IEC which aims as a preventive measure to reduce the risk of giving birth to offspring with this defect and increase the life expectancy of patients with hemoglobinopathies.
REFERENCE

Aulia. Faktor Risiko Penurunan dan Klasifikasi Thalassemia - Direktorat P2PTM [Internet]. 2017


Kaushansky, Kenneth. 2010. Williams Hematology. McGraw-Hill Medical,


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