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A 38 Week and 2 Day G1P0A0 Primigravida in the First Stage of Labor with Thalassemia: A Case Report

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ABSTRACT

Thalassemia is one of the most prevalent inherited blood disorders worldwide. We reported a case of 20-year-old G1P0A0 pregnant woman diagnosed with thalassemia intermedia since childhood based on chronic anemia, facies Cooley, and family history. She had 6 times red blood cell transfusion throughout current pregnancy. On 38 week 2 day gestation, she was admitted with chief complaint of painful regular uterine contractions and bloody show. Physical and laboratory findings were low hemoglobin, hematocrit, MCV, and MCH. Thalassemia can be alpha or beta type, depending on the affected globin chain. Clinical severity varies. The main therapies are repeated blood transfusions and iron chelation. During pregnancy, optimal evaluation and management are crucial to prevent maternal and fetal complications. This patient will undergo cesarean section and needs thorough high-risk antenatal care.

KEYWORDS: primigravida, thalassemia, labor, anemia, transfusion

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INTRODUCTION

Thalassemia is one of the most common inherited single gene disorder globally. A WHO report estimated that at least 5.2% of population are thalassemia carriers, and 1.1% of couples are at risk of having a child with thalassemia [1]. In Indonesia, thalassemia trait frequency was reported 3–10% for β-thalassemia, 1.2-11% for α-thalassemia, and 1.5-36% for HbE [2]. Thalassemia has multisystemic complications. During pregnancy, both maternal and fetal adverse outcome may happen [3]. In pregnancy, complications such as preeclampsia, heart failure, hemolytic anemia are reported high [4]. Regarding the fetus, intrauterine growth restriction, preterm delivery, and small for gestational age are also common [2]. Therefore, a thorough understanding about this disease is important.

CASE PRESENTATION

A 20-year-old G1P0A0 woman came to the emergency unit of R. Goeteng Teroenadibrata General Hospital, Purbalingga on July 6th, 2022 with chief complaint of regular painful uterine contractions since a day before admission. The contractions were felt for about 1 minute every 30 minutes. She also complained of bloody show that was noticed at 3 pm. She was diagnosed to have thalassemia when she was 18 months old.

She is the only child of the family. No history of thalassemia was acknowledged by her father. However, one of her mother's cousins had thalassemia. Despite diagnosed early, she did not routinely undergo blood transfusions except during current pregnancy that she received transfusion for 6 times. She never underwent iron chelation either during or prior pregnancy. She was routinely visiting antenatal care. Last menstrual period was on October 11th 2021. Expected date of confinement was on July 18th 2022, means she was on 38 weeks 2 days of gestation.

On physical examination, she looked pale. Facies Cooley was observed. Her conjunctiva was anemic. Vital signs showed blood pressure 124/70 mmHg, pulse rate 78/minute, respiratory rate 20/minute, temperature 36.6°C and O2 saturation 98% in room air. Cardiac and pulmonary examinations were normal. Abdominal examination revealed symmetrical enlargement of breast, no abdominal tenderness, and presence of fetal heart sound. Vaginal touché demonstrated cervix dilatation of 1 cm, effacement 50%, station -2, with bloody show (+) although no membrane rupture yet.

Complete blood count revealed low hemoglobin (9.6 g/dL), hematocrit (29%), MCV (65 fL), MCH (22 pg), and lymphocyte (22%); but high monocyte count (10%)

although still within normal limit. Serologic test for HBsAg and SARS-CoV-2 antigen were negative.

With these clinical and laboratory findings, she was diagnosed with G1P0A0 in the first stage of labor with thalassemia. The decision was agreed by her and her family that this pregnancy will be terminated. Because spontaneous vaginal delivery was predicted difficult regarding her cephalopelvic disproportion history, cesarean section is planned, preceded by blood product transfusion.

DISCUSSION

Our patient was diagnosed by history taking, physical examination and ancillary test. She had chronic microcytic hypochromic anemia since childhood, although the diagnosis of thalassemia was made when she first underwent hemoglobin electrophoresis test at 10 years old. The diagnosis was strengthened by presence of facial bone deformity (Facies Cooley), family history of cousin affected with thalassemia, and peripheral blood smear with characteristic finding (Figure 1). The low hemoglobin level in complete blood count also supported the diagnosis. Among thalassemia patients, this patient's clinical feature is categorized into thalassemia intermedia patient. She required occasional red blood cell transfusion, started just during current pregnancy.

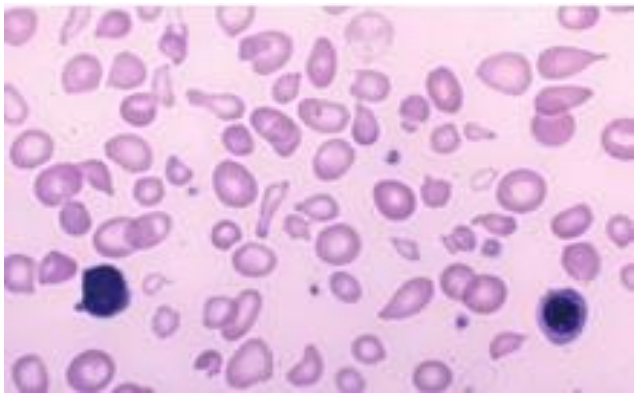

Figure 1. Peripheral blood smear of thalassemia patient. Note microcytic hypochromic picture with presence of target cell [5].

In terms of genetic, thalassemia is classified into alpha or beta thalassemia. The alpha type affects alpha globin chain synthesis, whereas beta type disturbs beta globin production [6]. Alpha mutation inheritance is more complex since it involves four genes arranged in two clusters, compared to beta gene that consists of one gene on each homolog chromosome 11. In beta type, more than 150 various point mutations have been recognized. Normal alpha/non-alpha chain ratio of 2:2 becomes imbalanced, generating excess of unmatched alpha or non-alpha chain, which lead to ineffective erythropoiesis. Clinical spectrum varies widely, from almost asymptomatic like in thalassemia minor or carrier state, up to life-threatening condition as in thalassemia major. The main principle of treatment is transfusional therapy in major type to overcome ineffective erythropoiesis, but iron chelation is also compulsory to prevent iron overload complication [7]. Regarding pregnancy, optimal preconception evaluation and counselling are absolute requirements that involve clinical, cardiological, endocrinological, and liver function aspects [8].

CONCLUSIONS

A case of G1P0A0 woman in labor with thalassemia intermedia has been discussed thoroughly regarding the etiology, classification, pathophysiology, diagnosis, and management strategy during pregnancy and delivery. Thalassemia imposes adverse risk towards maternal and fetal wellbeing. Comprehension about this disease is substantial to prevent morbidity for both mother and child.

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