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Case Report

Diagnostic problems of pregnancy in a mother with fetal anencephaly

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ABSTRACT

Anencephaly is a rare but fatal central nervous system anomaly involving failure ofthe neural tube to close during fetal development in the mother's womb. The numberof anencephaly cases varies, globally estimated to be between 0.5-2 per 1,000 births. Anencephaly cannot be treated but can be detected early. A 38-year-old woman presented with complaints of abdominal tightness that had been felt since 10 hours before admission, with a diagnosis of G5P4004 30-31 weeks, live singleton, latent phase 1, anencephaly. Cito sectio caesarea and pomeroy tubectomy were performed. The baby girl weighed 1800 grams, 40 cm in length, APGAR score 3/5, with anencephaly. The baby died 2 hours after birth. The prognosis for babies born with anecephaly is poor. Termination of pregnancy is theonly option to prevent the birth of a child with anencephaly, but it is still controversial in Indonesia.

Keywords: Pregnancy, Anencephaly, Neural tube defects, Folic acid

INTRODUCTION

Pregnancy is an important stage in the human life cycle for reproduction. Many factors influence pregnancy, starting from preconception, pregnancy and ending at birth. There are some conditions that make pregnancy not develop properly, resulting in congenital defects. One of the rare but deadly congenital abnormalities is anencephaly.[1-3]

Anencephaly is a rare but fatal congenital central nervous tract anomaly. Itis caused by failure of the neural tube to close at the skull bone during the third or fourth week (26-28 days) post fertilization.[4-8] The mechanism of anencephaly is that brain tissue that is not protected by the meninges, skull bones, and skin will beexposed to amniotic fluid (high concentration of urea in amniotic fluid) resulting inmechanical damage that causes brain tissue to degrade gradually (risk of friction with the uterine wall, placenta, and fetal parts). This is a lethal condition with nothing that can be done, which can lead to fetal death while in the womb, during birth, or after the baby is born. If some babies are born alive, most will die within hours and a few may survive a few days after birth.[1,4,5,8]

The number of anencephaly cases varies, globally estimated to be between 0.5-2 per 1,000 births.[2,9] Southeast Asia has a prevalence of 2-16 per 10,000 births, but Indonesia has no data.[10] Diagnosis is made by ultrasound examination (USG) and amniocentesis. Routine ultrasound that can be done in the first trimester is usually during week 11 to week 14.[4,12] At 15 to 22 weeks of pregnancy, an amniocentesis examination is performed which is found to be elevated by maternal serum alpha fetoprotein (AFP) levels and ultrasound. If elevated AFP levels are found at amniocentesis, there is a possibility that the fetus has a neural tube defect.[11] Further, examination is confirmed by ultrasound. Although the cause of anencephaly is still unclear, there are various factors such as environment, genetics, mother's condition, and baby's condition.[13] There is a strong correlation between high rates of neural tube defects and diets low in folate from several studies, so daily folic acid supplementation of 4 mg during the periconceptional period reduces the likelihood of recurrent neural tube defects.[3,14-16]

CASE REPORT

A 38-year-old woman came to the emergency room with complaints of abdominal tightness that had been felt since 10 hours before entering the hospital, accompanied by blood mucus, no water discharge from the birth canal,active fetal movements, a history of intercourse with her husband 1 day earlier, thepatient was known to have never had a routine pregnancy check-up with a health worker. From the physical examination, it was found that the patient was well conscious, appeared mildly ill, blood pressure 100/70 mmHg with a pulse rate of 90 beats per minute, respiratory rate 18 beats per minute, oxygen saturation 98% room water, axilla body temperature 36.8 ºC, pre-pregnancy weight 61 kg, currentweight 73 kg, height 158 cm, with a body mass index of 24.4 kg/m^2. Obstetric examination obtained uterine fundus height 24 cm, uterine contractions 2 times in 10 minutes for 15 seconds, fetal heart rate 155 times per minute.

Figure 1: Frog eyes on ultrasound.

Figure 2: Baby born with anencephaly.

On vaginal toucher examination obtained 2 cm opening, 30% effacement, protruding intact amniotic fluid, unclear denominator, no palpable umbilical cord. Laboratory examination showed hemoglobin 10.7 g/dl, leukocytes 10,780 g/dl, platelets 217,000/ul,hematocrit 32%, blood glucose 91 mg/dl. Transabdominal ultrasonography showed frog eyes. The final diagnosis was G5P4004 30-31 weeks T/H, latent phase1, anencephaly. The patient was planned for cito section caesarea and pomeroy tubectomy. The therapy given at the emergency room was lactated ringer fluid infusion, ceftriaxone injection. After SC surgery, the baby girl weighed 1800 grams, 46 cm in length, APGAR score 3/5, with anencephaly. The baby died 2 hours after birth.

DISCUSSION

Anencephaly is a failure in the closure of the neural tube that occurs during fetal development in the mother's womb.[3,4] It is one of the most common types of neural tube defects, after spina bifida. The mechanism of anencephaly is that braintissue that is not protected by the meninges, skull bones, and skin is exposed to amniotic fluid (high concentration of urea in amniotic fluid) resulting in mechanicaldamage that causes brain tissue to gradually degrade (risk of friction with the uterine wall, placenta, and fetal parts).[12]

Anencephaly is multifactorial and the cause remains unclear. It is known that there are various factors viz: environment, genetics, maternal condition, and infant condition. Environmental factors include folic acid deficiency, exposure to chemicals, use of anticonvulsant drugs, low socioeconomic status, and fever. Genetic factors from both father and mother. Maternal factors with obesity, pre- gestational/gestational diabetes, and maternal race. And for infant factors from casestudies it is said that female babies have a greater risk factor than male babies in a ratio of 3:1, but so far the exact cause is unknown. One of the most important things in the pathogenesis of anencephaly is from nutritional factors in the form of folic acid deficiency. It has been shown that folic acid can reduce the risk of neural tube closure failure. [3,13–15,17] In this patient, the risk factor was environmental factors such as lack of folic acid intake during pre-conception and pregnancy. The Centers for Disease Control and Prevention (CDC) recommends that women planning to become pregnant consume 400 µg of folic acid daily for one month before pregnancy.[18] For women who have a history of having children with neural tube defects, the recommended dose is 4 mg (4000 µg) per day.[3,14-16,18]

Anencephaly can be detected early, diagnosis is made by ultrasound and amniocentesis. In the first trimester ultrasound, the "mickey mouse sign" will be found which is caused by two semicircular structures hovering above the surface ofthe fetus, resembling the round ears of "mickey mouse". A large amount of brain tissue disappears during the second trimester, which will be depicted as "frog face" or "frog eyes" as no brain tissue is visible above the eyes. In the second trimester, amniocentesis examination revealed elevated maternal serum alpha-fetoprotein levels which may indicate anencephaly.[1-3,5,12] In this case the patient did not have a routine pregnancy check and never did an ultrasound so it was not detected early. In accordance with the

literature, frog eyes were found on the mother's ultrasound results when examined on arrival.

The prognosis for babies born with anencephaly is poor as it is untreatable and therefore termination of pregnancy is the only option to prevent the birth of a child with anencephaly.[19-21]

From the literature it is found that the life of a baby with anencephaly does not last long, death can occur while the fetus is still in the womb, during birth, or after the baby is born. If some babies are born alive, most will die within a few hours and a few can survive a few days after birth.[11] In this case, the baby died 2 hours after birth.

Termination of pregnancy remains a controversial issue in many countries. Some countries allow termination of pregnancy in women carrying babies with disorders where the death of a child cannot be prevented during or shortly after birth. In consideration of saving the woman from the physical and mental health hazards of prolonged pregnancy or delivery of a stillborn or dying baby. Some countries reject termination of pregnancy due to human and religious rights.[19-21] Likewise, in Indonesia, termination of pregnancy is still a controversial case.

CONCLUSION

Anencephaly is a lethal congenital defect characterized by failure of the neural tube to close at the skull bone. Early detection is based on obstetric ultrasound examination in the first and second trimester, if in the second trimester there is an increase in the mother's serum alpha fetoprotein level, it is confirmed byultrasound. Many factors can cause anencephaly, from research it is found that thisincident is associated with folic acid deficiency during preconception and early pregnancy. The prognosis for anencephaly cases is poor. To prevent anencephaly, folic acid consumption is recommended at a dose of 400 µg for women planning to become pregnant per day for one month before pregnancy and 4 mg (4000 µg) for women who have a history of giving birth to children with previous neural tube defects per day for one month before pregnancy. Termination of pregnancy in cases of anencephaly is controversial in Indonesia. There are still many people who do not understand and do not want to check their pregnancy to the nearest health facilities. Therefore, it is necessary to educate the community on the importance of pre-conception preparation, during pregnancy, and also after birth. So that they cangive birth to healthy babies and safe mothers.

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