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CASE REPORT: ANENCEPHALY IN AN UNIDENTIFIED BABY

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Abstract

Anencephaly is a congenital disorder affecting the central nervous system, characterized by the absence of a major portion of the brain, skull, and scalp, with an estimated prevalence of 0.5–2 per 1,000 births globally. This condition is a subset of Neural Tube Defects (NTDs), which affect approximately 300,000 newborns annually. This study aims to investigate the pathological and forensic aspects of a case of anencephaly in a female infant found deceased in Lingsar, West Lombok, on June 23, 2023. The methods employed case study included external and internal examinations to identify the structural abnormalities and potential cause of death. The external examination revealed open wounds on several parts of the body, and discoloration of the chest and abdominal cavities. Internal examination confirmed the absence of brain tissue, consistent with anencephaly. The findings indicate that the cause of death was directly attributed to the severe congenital anomaly, with no evidence of external factors contributing to mortality. The study concludes that anencephaly, while rare, remains a significant congenital disorder with profound implications for prenatal care and early detection. This case emphasizes the necessity of improving maternal nutrition, particularly folic acid supplementation, as a preventive strategy to reduce the incidence of NTDs. The findings also underscore the importance of interdisciplinary collaboration in handling forensic and medical investigations of congenital disorders, aiding in better understanding and management of similar cases in the future.

Keywords: Anencephaly, Baby, Brain defect, neural tube defect, Pregnancy

Introduction

Anencephaly is a congenital disorder that occurs in the central nervous system (Salari et al., 2022; Verity & Firth, 2003). This disorder is caused by the absence of hemispheres in the cerebrum and cerebellum, spinal cord, and pyramidal pathways (Encha-Razavi, 2015; ten Donkelaar et al., 2023). Babies with anencephaly have little scalp, cranium and brain but usually have a brain stem. Anencephaly is a severe disorder or defect that can result in spontaneous abortion in pregnancies of varying ages, being stillborn or being born alive but only surviving a few hours.(Simanjuntak, Saranga, & Munir, 2021)

Anencephaly means "having no brain" and is included in the Neural Tube Defect (NTD) condition due to the neural tube in the rostral part not closing entirely during pregnancy (Fellman & Eriksson, 2000). Every year, it is estimated that 300,000 babies are born with NTD conditions, where cases of anencephaly occur in 0.5-2 per 1,000 births. NTD conditions can be prevented by adequate blood folate concentrations in the mother before conception and during pregnancy.(Erdiana, 2021)

In Indonesia, one of the causes of death in babies is congenital abnormalities, with a percentage of 1.4% in babies aged 0-6 days and increasing to 18.1% in babies aged 7-28 days. In 2006, the prevalence of congenital abnormalities in babies in Indonesia was

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59.3 per 1,000 live births. This means that Indonesia is one of the countries with a relatively high prevalence of babies with congenital abnormalities among countries in Southeast Asia.(Matthew, Wilar, & Umboh, 2021) This study was an observational descriptive study with a case study approach and describe the death of anencephalic baby based on external and internal examination in Mataram Bhayangkara Hospital. This study aims to investigate the pathological and forensic aspects of a case of anencephaly in a female infant found deceased in Lingsar, West Lombok, on June 23, 2023.

Case Report

A corpse of female new born baby was found in Lingsar, West Lombok. When arriving in Bhayangkara Hospital, Mataram on Monday, June 26th, 2023, the body was covered with a cardboard speaker and wrapped in a yellow police body bag (see Figure 1). The observation was carried out through external and internal examination.

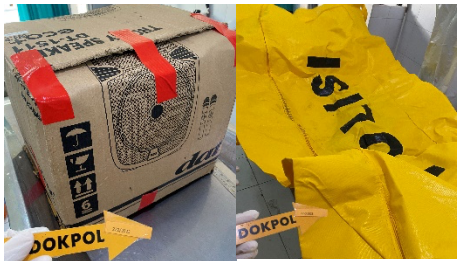

Figure 1. Corpse's wrap

External examination results showed that the body of a female baby aged nine months gestation with a body length of 61 cm and a body weight of 1.8 kg with the right arm severed, see Figure 2.

Figure 2. Right hand severed

There were open wounds on several parts of the body, such as the neck, left and right chest, back, right buttocks, left upper arm, groin and right calf, see Figure 3.

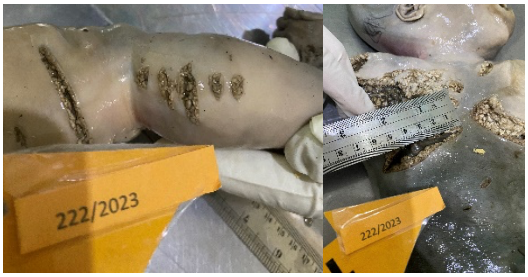

Figure 3. Open wounds on several parts of the body

On internal examination, it was seen that the chest and abdominal cavity were blackish (Figure 4).

Figure 4. Blackish color in chest and abdominal cavity

No brain, brain tissue, and brain stem was found on the head cavity but only a cranium, see Figure 5.

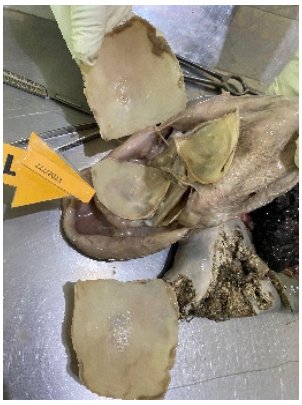

Figure 5. No brain was found

While supporting examination results, the lungs are mosaic. The lung buoyancy test shows that the left lung floats while the right lung sinks, see Figure 6.

Figure 6. Lung buoyancy test

Discussion

Several types of brain defects with the exact causes include anencephaly. The primary thing that occurs in some of these cases is due to failure of fetal development. The most severe congenital disabilities are spina bifida and anencephaly. Anencephaly is an abnormality that is congenital or a birth defect in the head, especially the cerebrum (Munteanu et al., 2020; Panduranga et al., 2012), cranium and scalp, which happen when the neural tube fails to close properly in the nasal (rostral) direction, which usually occurs during fetal development in the 24th day.(Erdiana, 2021; Mathews, Honein, & Erickson, 2002) No foramen magnum can be said as meroacrania. If the defect extends beyond the

foramen magnum, it is holoacrania, and if it is associated with spina bifida, it is called holoacrania with rachischisis.

Anencephaly is a fatal congenital disability, so usually, people with anencephaly are born under term, die soon after birth, or only live a few hours after birth(Book & Rayner, 1950). Anencephalic fetuses born alive are in direct proportion to fetuses that die before birth, with evidence from an extensive series of stillborn babies, where the average induction-to-delivery interval was 8.5 hours.(Obeidi, Russell, Higgins, & O'Donoghue, 2010) In cases of anencephaly, usually the brain stem, cerebellum, diencephalon, so in the case in Monteagudo study(Monteagudo & Society for Maternal Fetal Medicine, 2020), 35% of intrapartum fetal deaths. In the case of live birth babies with anencephaly, most will experience neonatal death in the first birth, but some survive up to 1 year 17 weeks with active care and family support.

Anencephaly can usually be checked at 10 to 14 weeks of gestation.(Erdiana, 2021; Mathews et al., 2002) Monteagudo(Monteagudo & Society for Maternal Fetal Medicine, 2020), detected all cases through the 16th and 34th weeks with an average fetal age of 21 weeks via ultrasound. Trimester ultrasound explains that the natural look of anencephaly is the "frog eyes" indication due to the nonappearance of observable brain tissue above the eye socket. During pregnancy, if there is hydramnios or an expansion in the volume of amniotic fluid where hydramnios is related to anencephaly in 30 to 50% of cases.

Many elements that cause anencephaly are thought to be genetic factors, such as chromosomal changes and the methylene tetrahydrofolate reductase gen, which has been proven to be associated with anencephaly(Erdiana, 2021). Monteagudo(Monteagudo & Society for Maternal Fetal Medicine, 2020) explained that there was 1 case in a group with a different of the TRIM36 gen on chromosome 5q22, which was perceived to cause anencephaly, anencephaly was also caused by a recessive anencephalic gen.(Coffey & Jessop, 1957) Other factors are environmental influences such as malformations in the family of two identical twin sisters(Coffey & Jessop, 1957), social status, drugs such as antiepileptic drugs, valproate, isotretinoin can cause an increased risk of giving birth to anencephalic fetuses(Monteagudo & Society for Maternal Fetal Medicine, 2020), anencephaly caused by abortion(Book & Rayner, 1950), maternal nutrition such as a lack of folic acid during fetal development, where many women do not get enough folic acid from folic acid fortification(Mathews et al., 2002), disease during pregnancy. Chu andToh. (2003) and Berry, et al. (2022) explained that cases of anencephaly were more at risk in mothers in the older age group, but Matthew, et al. (2021), Book and Rayner (1950), and Abebe et al. (2021) explained that the mother's age had no effect or no effect at all.

Apart from that, the cause of anencephaly is exposure to nitrates in cleaning products, spray paint or paint thinner, and pesticides at home, at work or in the community. Maternal diseases such as diabetes, hyperinsulinemia, and a BMI of 30 kg/m2 or higher, weight loss (dieting) and taking weight loss products have a more significant possibility of anencephaly. (Monteagudo & Society for Maternal Fetal Medicine, 2020) and (Berry et al., 2022) explained that women who have anencephalic children for the first time have a higher risk of having a second child experiencing anencephaly compared to the rest of the population and that anencephaly usually occurs in mothers who have diabetes but are dependent on insulin in the first trimester of pregnancy.

Conclusions

Based on external examination, it was found that there were open wounds on several parts of the body. Based on internal examination, it was discovered that the chest and abdominal cavity had experienced advanced decay. In the head cavity, no brain was found, neither brain tissue nor brain stem. The lung float test showed positive results, so it can be concluded that the baby was born alive but soon died due to the severe abnormalities or defects he experienced namely anencephaly. Case reports of anencephaly in Lombok should be properly recorded to determine the birth rate of babies with the disorder so that it can be used as a database.

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