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17-Years-Old Teenager with Anencephaly

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ABSTRACT

Background: Anencephaly is the most common type of Neural Tube Defect (NTD). Babies born with anencephaly usually die within hours or a few days after birth. The prevalence of anencephaly cases reaches 5.1 per 10,000 births in the world. There is no therapy for patients with anencephaly, but this condition can be detected early by ultrasonography. ***Case:*** 17-year-old girl with abdominal complaints that are getting bigger. She has pregnant and the fetus had suspected anencephaly. The patient's vital signs were good and laboratory examination showed a slight increase in leukocytes, proteinuria, and leukocyturia. The ultrasound examination results gestational age was 29/30 weeks, and the radiological examination by a radiologist expert had similar results. The baby girl was born with a weight of 1,568 grams and an Apgar score of 2-3. On examination, prominent brain tissue was visible. The baby was treated for three days at Trenggalek General Hospital and survived for four days. ***Conclusion:*** The aetiology and pathophysiology of anencephaly are still unclear, but risk factors such as environmental, genetic, maternal and fetal conditions encourage this condition. Detection of NTDs can be done early in pregnancy using ultrasound. Early prevention must be carried out, such as regular consumption of folic acid to prevent the occurrence of NTDs. In this case, the occurrence of NTDs is thought to be due to the condition of the mother who is too young, socioeconomic factors, low consumption of folic acid and lack of knowledge and information about the importance of prevention in NTD cases.

Keywords: anencephaly; neural tube defect; pregnancy

INTRODUCTION

Anencephaly is a disorder of the central nervous system and the most common abnormality in Neural Tube Defect (NTD) cases. Anencephaly is described as the total or partial absence of the calvaria and brain tissue but still has the structures of the brainstem, cerebellum, and diencephalon [1]. Neural defects are generally distinguished by the failure of the neural tube to close during the early stages of embryonic development [2]. Babies born with anencephaly usually die within hours or a few days after birth [3].

Several studies have reported various risks associated with anencephaly, including socioeconomic status, education level of the prospective mother, age, occupation, smoking, alcohol, history of previous births, infections during early pregnancy, diabetes, obesity, consumption of caffeine and drugs during early pregnancy [4]. If an abnormality is found during a routine examination, pregnant women are advised to go to an advanced health facility to detect abnormalities and further treatment [5].

Until now, there is no therapy for patients with anencephaly, but this condition can be detected by early ultrasonography (USG) [2].

CASE REPORT

A 17-year-old teenager was brought by her mother to a general practitioner with complaints that her child's stomach had been getting bigger for two weeks. Furthermore, the doctor conducted a limited essential ultrasound examination and found suspicion of anencephaly in the fetus. The doctor took the patient to the nearest obstetrics and gynaecology practice and confirmed it as anencephaly. The patient was referred to the Primary Health Care to get a referral to the Hospital. At the obstetric polyclinic, the patient had no complaints and did not know the first day of the last menstrual period (LMP). The patient is a grade 2 senior high school student and during pregnancy tends to cover up her pregnancy because she is not married. During pregnancy, the patient never checked her pregnancy and did not take additional vitamins such as iron, calcium and folic acid. The patient denied taking certain drugs during pregnancy and found no history of chronic disease or surgery.

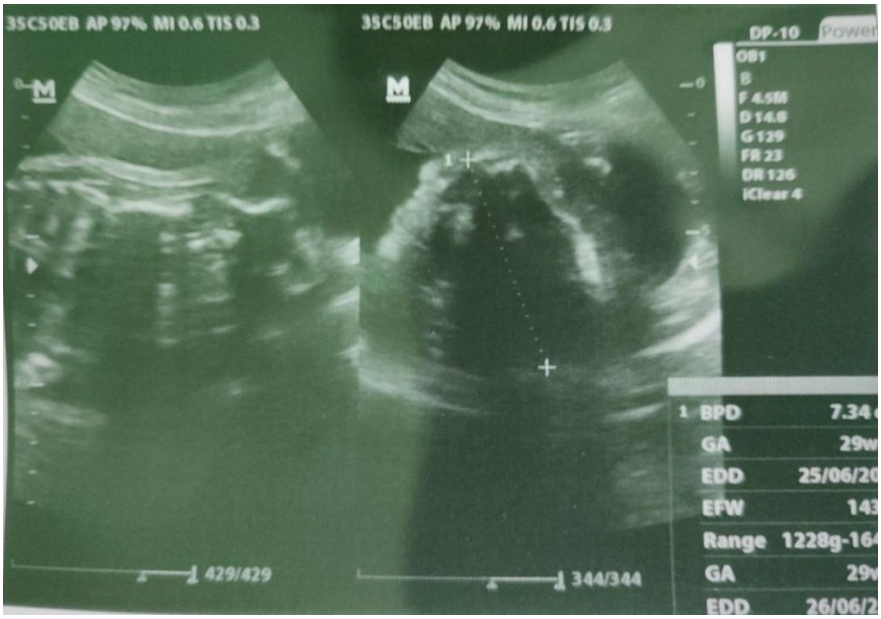

FIGURE 1: Ultrasound results showed anencephaly.

The general condition and vital signs were good; physical examination revealed an enlarged abdomen with a Uterine Fundal Height of about 19 cm. Laboratory examination revealed slightly increased leukocytes, proteinuria and leukocyturia. Furthermore, a gynaecological obstetrician specialist carried out an ultrasound examination, obtained a gestational age of around 29/30 weeks with suspected anencephaly and an ultrasound confirmation by a radiologist obtained similar results.

The patient was admitted to the delivery room for vaginal labor. In termination of pregnancy, an action is required in the form of a buccal induction of 100 mg of misoprostol which is carried out for two days. The baby was delivered vaginally after the eighth misoprostol induction; the baby girl weighs 1,568 grams with an Apgar score of 2-3. On examination, there was an abnormality of the skull, and protruding brain tissue was seen. The baby was treated for three days at Trenggalek General Hospital and then allowed to go home. The baby survived for four days and then died. Meanwhile, the baby's mother was treated for two days in the hospital after giving birth and was allowed to go home because her condition was stable.

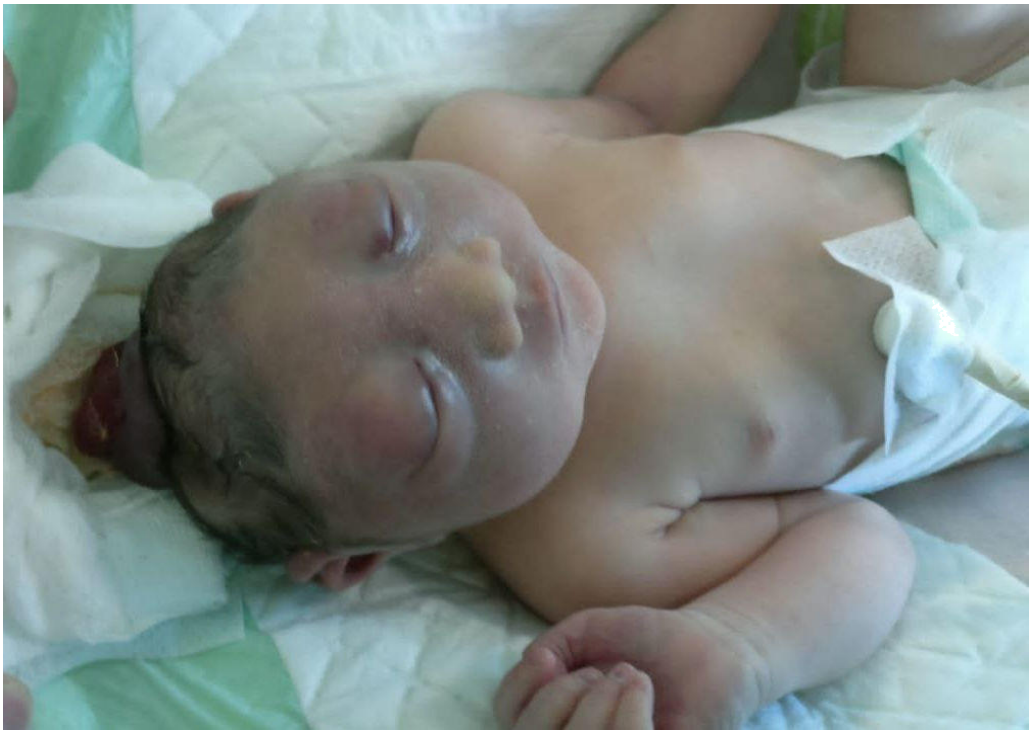

FIGURE 2: The shell does not cover the brain tissue.

DISCUSSION

NTD is one of the most common inherited disorders in humans. This disorder is formed during embryonic development and affects the quality of life from birth. The presentation of NTD varies wildly, depending on the localization of the lesion. NTDs are divided into Open NTDs, such as craniorachischisis, anencephaly and myelomeningocele, while Close NTDs, such as malformations of fat, bone or membranes in the spine. Anencephaly is a defect that only affects the skull part of the Neural Tube so that the child's brain tissue is visible but is not covered by the calvarium bone [6].

The prevalence of anencephaly cases reaches 5.1 per 10,000 births in the world. The Australian continent has the highest anencephaly rate, 8.6 per 10,000 live births. The Asian continent is in second place with the African continent, reaching 6.5 per 10,000 live births, while the lowest population is the Americas reaching 4.3 per 10,000 live births [3]. Each country has a different prevalence regarding the incidence of anencephaly [1]. Research conducted by Kondo et al. in 2019 in Japan evaluated the incidence of Open NTD. It stated that the incidence of anencephaly from 2000 to 2015 experienced a decrease in prevalence each year, with an average incidence rate of only 0.4 per 10,000 live births [7]. Another study belonging to Gong et al. in 2017 stated that the prevalence rate of Anencephaly in China from 2006 - 2015 reached 4.92 per 10,000 live births [8]. The prevalence of anencephaly is high in India; research belonging to Praktja Bhide et al. in 2018 stated that the incidence of anencephaly could reach 21.10 per 10,000 live births from 25 hospitals in India [9]. In Indonesia, no data shows the overall prevalence of anencephaly.

Although the aetiology and pathophysiology of anencephaly are still unclear, several risk factors, such as environmental, genetic, maternal and fetal conditions, can cause anencephaly [1]. One of the most critical factors in the pathogenesis of anencephaly is environmental factors such as folic acid deficiency. Anencephaly fetuses described in vivo studies with mice lacking monofunctional mitochondrial 10-formyl-tetrahydrofolate synthetase (Mthfd1l) exhibit delayed neural tube closure as well as dysfunctional folate receptor 1 (Folr1/folate binding protein 1/folate receptor α) which causes the appearance of clinically severe NTD in a trial population. This deficiency condition can be prevented by administering folic acid because folic acid acts as an essential regulator of neural tube morphogenesis. In addition to its role as a vitamin for DNA synthesis, folic acid has a metabolism that allows the biosynthesis of nucleic acids and the proliferation and growth of body cells [10].

Several sources stated that genetic factors play a role in anencephaly's occurrence, such as the Methylenetetrahydrofolate reductase (MTHFR) gene involved in folic acid metabolism. The enzymatic activity of this gene can be reduced if there are two polymorphisms, namely 677C-T and 1298A-C, which cause enzyme thermolability and decrease in enzymatic activity so that the risk of neural tube defects is 2-4 times higher [1]. Other sources say that the paired box 3 (Pax3) gene mutation also contributes to NTD. The Pax3 gene is known to be responsible for neurulation, which is the process of forming the neural tube that becomes the future of the nervous system [11]. In addition, other factors play a role in this, namely maternal and fetal factors. Maternal factors can contribute to events such as diabetes, hyperinsulinemia and a body mass index (BMI) of 30 kg/m2 or higher. On the other hand, a weight loss diet in the early stages of pregnancy and before pregnancy can increase the risk of NTD due to low intake of nutrients, including folic acid. The fetal factor that influences it is the female sex, which until now is still unknown how the mechanism works [11].

A low prevalence of NTD is reported in most developed countries and increases in developing countries. This condition, among others, is caused by low fortification of folic acid; poor health and nutritional adequacy of a person, especially during pregnancy; lack of information, mentality and costs related to planned pregnancies; and the lack of preconception care services for expectant mothers [12]. In addition to this, welfare factors also play a role which can be seen in developing countries where the majority of the population is middle to lower income and at risk of potential predisposing factors such as infection, low socioeconomic status and education, minimal environmental protection and low access to health services [13]. This was proven in a study by Gashaw et al. in Ethiopia in 2021 which concluded that annual family income of less than 24000 ETB (Birr Ethiopia) was associated with an increased prevalence of NTD [13]. Another study belonging to Abel et al. in 2018 also stated that a family's annual cash income of less than USD 1,300, USD1,300–USD 1,800, and USD 1,801–2,700 increases risk factors compared to income higher than USD 2,700 [14].

Maternal age affects the incidence of NTD, one of which is anencephaly. Saleem et al.'s research published in 2010 stated that maternal age < 30 years had an NTD rate of 71.4%, while maternal age > 30 years only reached 28.4% [15]. A report conducted by Eurocat (European Surveillance of Congenital Anomalies) in 2010, which collected NTD cases in 2000-2008 in Europe, stated that maternal age <25 years increased the risk of NTD events compared to ages 25-29. Maternal ages 30-34 years can reduce the incidence of NTD compared to ages 25-29 [16]. Another study by Emanuele et al. in Brazil in 2015 described that maternal age <20 years had an incidence of anencephaly reaching 37.9%, while maternal age > 35 years reached 10.3% [17]. Unlike the results of these three studies, Bourouba et al. explained that maternal age between 25-35 years has the highest NTD rate, reaching 52%, so it can be concluded that maternal age has no relationship with the incidence of NTD [18].

Anencephaly is caused by congenital inhibition of mesenchymal migration in the fourth week of gestation which results in underdevelopment of the calvarium and abnormalities of the cortical structures. There is a need for an ultrasound examination as the main and simple diagnostic tool that helps assess structural abnormalities in the fetus, such as anencephaly so that early recognition of this diagnosis allows early decision-making, such as counseling regarding anencephaly conditions and risk options for termination of pregnancy if desired and available [19].

CONCLUSION

Anencephaly is a disorder of the central nervous system and the most common disorder in NTD cases. The aetiology and pathophysiology of anencephaly are still unclear, but risk factors such as environmental, genetic, maternal and fetal conditions encourage this condition. Detection of NTDs can be done early in pregnancy using ultrasound pregnancy screening performed by doctors and can be easily accessed by all patients. Early prevention must also be carried out, such as regular consumption of folic acid because it plays an essential role in the development and growth of the fetus and has been proven to prevent the occurrence of NTDs. In this case, the occurrence of NTDs is thought to be due to the condition of the mother who is too young, socioeconomic factors, low consumption of folic acid and lack of knowledge and information about the importance of prevention in NTD cases.

REFERENCES

[1] Munteanu O, Cîrstoiu MM, Filipoiu FM, Neamţu MN, Stavarache I, Georgescu TA, et al. The etiopathogenic and morphological spectrum of anencephaly: A comprehensive review of literature. Romanian Journal of Morphology and Embryology 2020; 61:335–43. https://doi.org/10.47162/RJME.61.2.03.

[2] Shilpa K, Ranganath P, S S. Frequency of Anencephaly and Its Associated Anomalies. International Journal of Anatomy and Research 2018; 6:5294–300. https://doi.org/10.16965/ijar.2018.190.

[3] Salari N, Fatahi B, Fatahian R, Mohammadi P, Rahmani A, Darvishi N, et al. Global prevalence of congenital anencephaly: a comprehensive systematic review and meta-analysis. Reprod Health 2022;19. https://doi.org/10.1186/s12978-022-01509-4.

[4] Abebe M, Afework M, Emamu B, Teshome D. Risk Factors of Anencephaly: A Case–Control Study in Dessie Town, North East Ethiopia. Pediatric Health Med Ther 2021; Volume 12:499–506. https://doi.org/10.2147/phmt.s332561.

[5] Edwards L, Hui L. First and second trimester screening for fetal structural anomalies. Semin Fetal Neonatal Med 2018; 23:102–11. https://doi.org/10.1016/j.siny.2017.11.005.

[6] Isaković J, Šimunić I, Jagečić D, Hribljan V, Mitrečić D. Overview of Neural Tube Defects: Gene–Environment Interactions, Preventative Approaches and Future Perspectives. Biomedicines 2022;10. https://doi.org/10.3390/biomedicines10050965.

[7] Kondo A, Akada S, Akiyama K, Arakawa M, Ichi S, Inamoto Y, et al. Real prevalence of neural tube defects in Japan: How many of such pregnancies have been terminated? Congenit Anom (Kyoto) 2019; 59:118–24. https://doi.org/10.1111/cga.12333.

[8] Gong T-T, Wu Q-J, Chen Y-L, Jiang C-Z, Li D, Li J, et al. Changing trends in the prevalence of anencephaly in Liaoning province of Northeast China from 2006-2015: data from a population-based birth defects registry. n.d.

[9] Bhide P, Kar A. A national estimate of the birth prevalence of congenital anomalies in India: Systematic review and meta-analysis. BMC Pediatr 2018;18. https://doi.org/10.1186/s12887-018-1149-0.

[10] Balashova OA, Visina O, Borodinsky LN. Folate action in nervous system development and disease. Dev Neurobiol 2018; 78:391–402. https://doi.org/10.1002/dneu.22579.

[11] Munteanu O, Cîrstoiu MM, Filipoiu FM, Neamţu MN, Stavarache I, Georgescu TA, et al. The etiopathogenic and morphological spectrum of anencephaly: A comprehensive review of literature. Romanian Journal of Morphology and Embryology 2020; 61:335–43. https://doi.org/10.47162/RJME.61.2.03.

[12] Berhane A, Belachew T. Trend and burden of neural tube defects among cohort of pregnant women in Ethiopia: Where are we in the prevention and what is the way forward? PLoS One 2022;17. https://doi.org/10.1371/journal.pone.0264005.

[13] Gashaw A, Shine S, Yimer O, Wodaje M. Risk factors associated to neural tube defects among mothers who gave birth in North Shoa Zone Hospitals, Amhara Region, Ethiopia 2020: Case control study. PLoS One 2021;16. https://doi.org/10.1371/journal.pone.0250719.

[14] Gedefaw A, Teklu S, Tadesse BT. Magnitude of Neural Tube Defects and Associated Risk Factors at Three Teaching Hospitals in Addis Ababa, Ethiopia. Biomed Res Int 2018;2018. https://doi.org/10.1155/2018/4829023.

[15] Saleem M, Ali M, Hussnain Q, Razzak Malik A, Mahamood R. Prevalence and Associated Risk Factors for Neural Tube Defects in Patients at Sheikh Zayed Hospital Rahim Yar Khan. 2010.

[16] M Haeusler, V Nelen, I Barisic, E Garne. Prevalence of Neural Tube Defects in younger mothers in Europe. 2010.

[17] Pelizzari E, Valdez CM, dos Santos Picetti J, da Cunha AC, Dietrich C, Fell PRK, et al. Características de fetos avaliados por suspeita de anencefalia: Um estudo de coorte de base populacional do Sul do Brasil. Sao Paulo Medical Journal 2015; 133:101–8. https://doi.org/10.1590/1516-3180.2013.8012608.

[18] Bourouba R, Houcher B, Akar N. Risk factors of neural tube defects: A reality of Batna region in Algeria. Egyptian Journal of Medical Human Genetics 2018; 19:225–9. https://doi.org/10.1016/j.ejmhg.2017.10.003.

[19] Hall JW, Denne N, Minardi JJ, Williams D, Balcik BJ. Check the head: Emergency ultrasound diagnosis of fetal anencephaly. Western Journal of Emergency Medicine 2016; 17:460–3. https://doi.org/10.5811/westjem.2016.5.30326.