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A CASE REPORT – ANENCEPHALY WITH CLEFT LIP,PALATE : RELEVANCE OF ANTENATAL EXAMINATION

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Abstract

Anencephaly is a lethal embryological malformation of the central nervous system in which the brain and the cranial vault are grossly malformed due to defect in the neurulation process[1]. It can be detected during the pregnancy with ultrasonography and maternal serum alpha-fetoprotein (MSAFP). We present a rare case of a 19 year old primipara , with no antenatal care, who delivered postdate by cesarean section , an anencephalous female child with cleft lip and palate .This case insists on the relevance of antenatal examination and care for early diagnosis and appropriate management during pregnancy.

Introduction

Anencephaly, a part of the neural tube defects, is a serious developmental defect of the central nervous system where the cerebrum and cerebellum are reduced or absent but hindbrain is present[2] . This defect results when the neural tube fails to close during the third to fourth weeks of embryological development .Failure at the cephalic end produces anencephaly and failure lower down produces spina bifida. It can be diagnosed in utero on ultrasound examination and elevated maternal serum levels of alpha-fetoprotein. Folic acid has been shown to be an efficacious preventive agent that reduces the potential risk of anencephaly and other neural tube defects by approximately two – thirds.

Case details

A 19 year old primipara presented to the emergency duty labour room with 9 completed months of gestation with complaints of pain in abdomen since 5 hours with no other complaints. She did not undergo any prior antenatal checkups and her date of last menstrual period was not known. The patient was from the lower economic strata and was an agricultural laborer. She was not exposed to any teratogen or mutagenic agents or radiation during pregnancy .There was no history of chronic illness or any drug exposure. No history of fever or rashes during pregnancy. No history of iron or folic acid intake. The only significant history obtained wasthat of first degree consanguineous marriage. The clinical examination revealed a full term singleton pregnancy with mild uterine contractions, vertexpresentation, diminished liquor and fetal heart sounds were not audible .On per vaginumexamination, the diagnosis of face presentation was made with absent membranes and thinly meconium stained liquor. An urgent ultrasound examination was ordered for estimation of gestational age, confirmation of intrauterine fetal demise, estimation of fetal weight and confirmation of anencephaly. It revealed a postdate 40+3 weeks gestation anencephalous fetus with intrauterine fetal demise. Labor was induced with PGE1 gel and augmented with oxytocin and progress of labor was monitored. Patient was posted for emergency lower segment cesarean section for obstructed labor and a dead female child delivered weighing 3.1kg with evidence of anencephaly with cleft lip and cleft palate (Figure 1 &2) .The postpartum period was uneventful and patient was discharged.

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Figure 1

Figure 2

Discussion

Neural tube defects are birth defects of the brain , spine or spinal cord . The two most common NTDs are anencephaly and spina bifida , anencephaly being the most common with incidence of 1:1000 to 1: 20000[3] . In the normal human embryo the neural plate arises approximately 18 days after fertilization . During the 4th week of development , the neural plate invaginates along the embryonic midline to form the neural groove[4] .The neural tube is formed as closure of the neural groove progresses from the midline toward the ends in both the directions with completion between day 24 for cranial end and day 26 for the caudal end. Anencephaly results from the failure of the neural tube closure at the cranial end of the developing embryo. Absence of the brain and calvaria may be partial or complete.

Epidemiological studies show variation in prevalence rates. The incidence is 6 times more common in the white race compared to the black race and the anencephalous fetuses born were predominantly female[5,6] . A variety of environmental factors appear to be influential in the closure of the neural tube . Folate antimetabolites , maternal diabetes[7] , maternal obesity , mycotoxins in contaminated corn meal , arsenic and maternal hyperthermia in early development have been identified as stressors that increase the risk of NTDs , including anencephaly . There are reports in favour of the relation between the exposure of both parents to agriculture work and increased risk of anencephaly[8]. Most cases of anencephaly follow a multifactorial pattern of inheritance with interaction of multiple genes as well as environmental factor . Genes involved in folate metabolism are believed to be important – one such gene of methyltetrahydrofolate reductase has been shown to have significant association with the risk of NTD[9].

Fetuses with anencephaly can be correctly identified at 11-13 weeks of gestation on ultrasound examination. A first trimester scan definitely allows reliable diagnosis and active management of anencephaly .Ultrasound examination shows incomplete development of frontal and occipital bone , well developed maxillary , zygomatic and mandibular bone . MSAFP screening during the second trimester of pregnancy is an effective screening tool for identification of a vast majority of cases of anencephaly[10] . Amniotic fluid alphafetoprotein testing during the late first trimester and second trimester of pregnancy is adiagnostic biochemical test for anencephaly.

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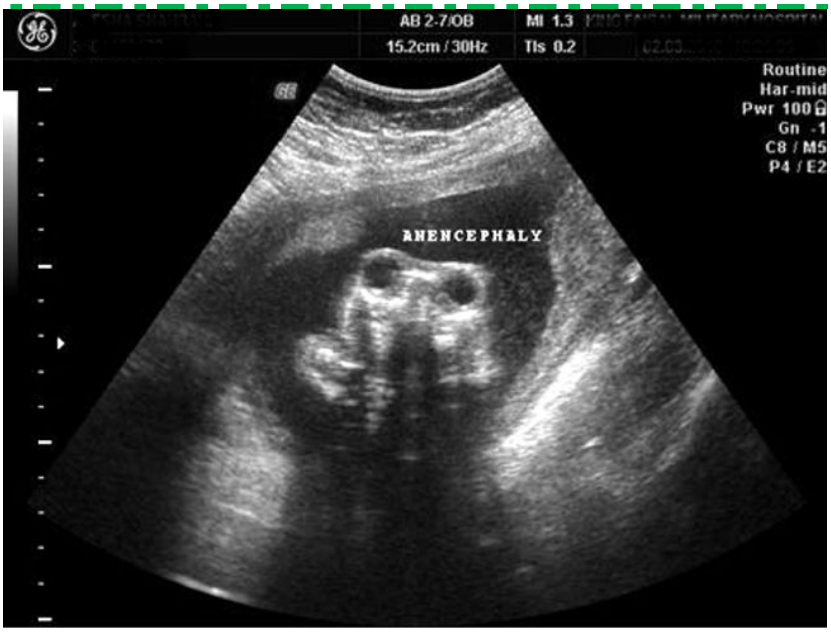

The complications of anencephaly include polyhydramnios , the patient may experience significant discomfort from abdominal distension . The risk of Preterm labour is increased .Because the pituitary gland may be absent in fetuses of anencephaly, spontaneous precipitation of labour may be delayed , therefore , the risk of pregnancy progressing to postterm is significant .The rate of abnormal fetal presentations during delivery is increased .

Anencephaly is lethal in all cases and a significant proportion of all anencephalic fetuses are stillborn or are aborted spontaneously. The neonates prognosis when born alive is extremely poor, the newborn may be blind, deaf, unconscious and death of a live child is unavoidable and most often occurs during early neonatal period[11].

The recurrence risk for NTDs in general is 2-4 % in subsequent pregnancy .The preventive measures include supplementation with folic acid before pregnancy and in the first month[12,13] – for women with history of NTD in a child -4mg per day ; for all others -0.4mg (400mcg) per day . Another measure to be used is the fortification of both wheat and maize flour with folic acid[13] .A secondary line of prevention is to detect the abnormality as soon as possible during pregnancy , obtained by the implementation of prenatal diagnosis[13]. The late presentation of gross congenital anomaly , a situation associated with many complications both physical , of the pregnancy and labor , and psychological , for the parents is extreme .To avoid this situation health education should include issues regarding congenital malformation delivered by trained experts[14] .

Conclusion

Anencephaly may be diagnosed as early as 11 weeks by transvaginal sonography and also an abnormally elevated maternal serum AFP . Anencephaly with associated malformations may be associated with aneuploidy and therefore karyotyping may be indicated . The recurrence risk for future pregnancies is 2-4% . Preconceptual supplementation of folic acid may reduce the risk by upto 70%. Anencephaly is lethal , with no cure . Hence , the aim should be to create public awareness for early antenatal checkups promoting early diagnosis and management

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