Case Report

Anencephaly: A Case Report

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Abstract

Anencephaly occurs in 1.4-4.7/10,000 deliveries and is thought to result from failed closure of the anterior neuropore at 24-26 days post fertilization. It is characterized by congenital absence of the major portion of brain, skull and scalp. Thus the cranial neural tissue is exposed. In some conditions, some development of cerebral hemispheres can occur but this exposed tissue may be destructed because of hemorrhage. Thus, it leads to nonfunction of the cerebrum. The etiology of this is still unknown. The diagnosis can be made by various prenatal methods as simple as an ultrasound. This report is being made because it may afford material for a review of embryological and gross anatomy findings in a case of anencephaly, which could help in a thorough evaluation and early diagnosis.

Keywords: Acrania, Alpha-fetoprotein test, Anencephaly, Calvaria, Neurulation, Polyhydromnios

INTRODUCTION

Anencephaly is congenital absence of a major portion of the brain, skull and scalp. It results due to the defective neurulation process, which is defined as the process of neural tissue formation from the ectoderm. In anencephaly the abnormality occurs in neurulation of the cranial part.[1] Due to this the neural tissue is exposed and is not covered with the skull. The development of the cerebral hemispheres is also absent.[2] If at all any amount of neural tissue is formed, it may show destructive changes like hemorrhage. It can be diagnosed *in-utero* on ultrasound examination and by elevated maternal serum levels of alpha fetoprotein (AFP). It is, usually, associated with polyhydramnios. About 65% of the cases of anencephaly die in utero, and some may be delivered prematurely. Infants are born with anencephaly show permanent unconscious, due to lack of functioning cerebral cortex and varying degrees of brain stem functions causing brain death.

MATERIALS AND METHODS

A pregnant woman of G2P1L1 aged 28 years presented 29 weeks of gestation without any prior antenatal checkups. Previous history shows one vaginal delivery at home with a normal child. On examination, abdomen was over-distended with fundal height of 36 weeks/37 cm, abdominal girth was 83 cm. On sonography live fetus with anencephaly was detected. There was no history of iron and folic acid intake. No history of any chronic illness, drug or radiation exposure identified. Labor was induced with prostaglandin E1 and she delivered vaginally a stillborn anencephalic female baby with cephalic presentation, weighed 1000 g. The baby died 48 h later. The postpartum period was uneventful and the patient was discharged.

OBSERVATIONS

Fetuses with anencephaly are correctly identified at 12-13 weeks of gestation. Ultrasound findings can be normal until the onset of ossification has definitely failed. A first-trimester scan definitely allows a reliable diagnosis and active management of anencephaly.

On the observation, the fetus showed absence of calvaria, short neck, low-set ears and protruded eye ball (Figure 1). Ultrasound scan showed incomplete development of frontal and occipital bone, well-developed maxillary, zygomatic, mandibular bone (Figures 2a,b and 3). Thoracic cage was normal, there was spina bifida in the region of C1, L4-5 and S1-5. We confirmed the sonographic findings following the dissection of the head, spinal cord, thorax and abdomen. Spina bifida occulta was confirmed, which can be correlated with embryological basis of teratological

insult during 3-4 weeks of intrauterine life involving the development of neural tube before the closure of anterior and posterior neural pores. There was the absence of brain tissue with normal spinal cord. Abdominal organs were normal, and there was no other associated congenital anomaly.

DISCUSSION

Neural tube defects are birth defects of the brain, spine, or spinal cord. They happen in the 1st month of pregnancy, often before a woman even knows that she is pregnant. The two most common neural tube defects are spina bifida and anencephaly. Among common neural defects the anencephaly is one of the most common, the incidence of anencephaly is 1:1000-1:20000.[3] Epidemiology studies demonstrate variation in prevalence rates. The highest incidence is in Great Brittan and Irland, and the lowest is in Asia, Africa and South America. Anencephaly occurs 6 times more frequent in white than in blacks, females are more often affected than males.[4,5]

Figure 1: Fetus with anencephaly, low set ears and protruded eye ball

In a normal human embryo, the neural plate is formed approximately 18th days after fertilization. During the 4th week of development, the neural plate invaginates to form the neural groove.[6] The neural tube is formed due to closure of the neural groove by fusion of neural folds. The process is initiated at a single site and extends towards the rostral and caudal neuropores. Closure completed by day 24 for the cranial end and day 26 for the caudal end.

Anencephaly results from the failure of neural tube closure at the cranial end of the developing embryo leading to incomplete development of calvaria and brain. Babies with anencephaly are either stillborn or die shortly after birth. The incidence of anencephaly shows a multifactorial pattern of inheritance, with interaction of multiple genetic and environmental factors. The specific genes which cause the neural tube defects are not been identified still. One such gene methylene tetrahydrofolate reductase has been shown to be associated with the rise of neural tube defects.[7] Anencephaly can be diagnosed prenatally with a high degree of certainty. The initial screening for anencephaly and other neural tube defects are performed by testing for high levels of maternal serum alpha-fetoprotein in the second trimester of pregnancy and by ultrasonography in the third trimester of pregnancy.[8]

Fetus with neural tube defects lacks functioning cerebrum that rules out the possibility of ever gaining

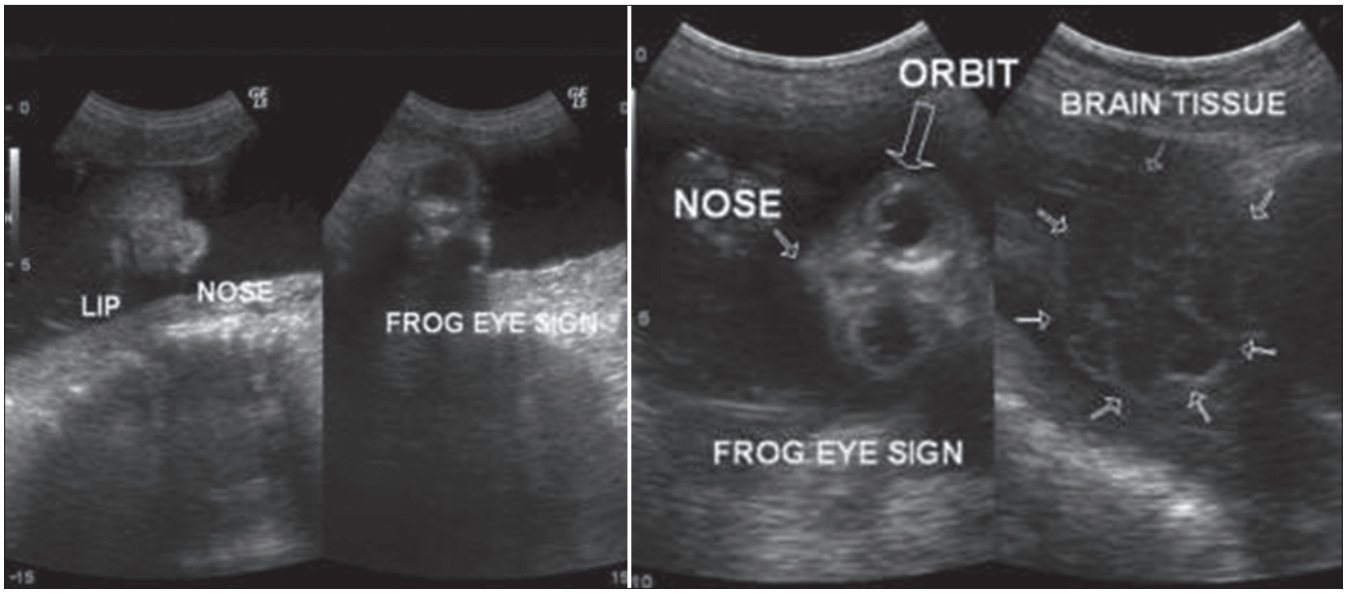

Figure 2: (a and b) Ultrasound images showing the absent calvaria and exposed brain tissue

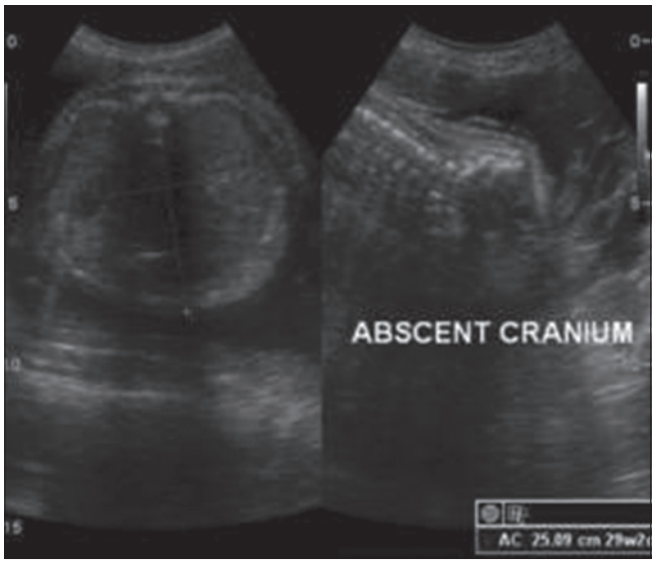

Figure 3: Ultrasound showing the eye and lips

consciousness. They will be blind, deaf and unable to feel pain. Some individuals with anencephaly may be born with a rudimentary brainstem, which controls autonomic and regulatory function. Hence, reflex actions such as respiration and responses to sound or touch may be present.

The preventive measures include diet supplementation with folic acid before pregnancy and in the 1st month.[9,10] This can decrease both the frequency and severity of the condition.[9] Another measure to be used is the fortification of both wheat and maize flour with folic acid.[10] A secondary line of prevention is to detect the abnormality as soon as possible during the pregnancy, obtained by the implementation of the program of the prenatal diagnosis. The knowledge very help for diagnose and treating of neural tube defects.

CONCLUSION

Anencephaly may be diagnosed by transvaginal sonography as early as 11 weeks. All anencephalic foetuses will have an abnormally elevated maternal serum AFP. Isolated anencephaly is rarely associated with aneuploidy, and therefore, amniocentesis for karyotype is not indicated. The recurrence risk for future pregnancies is 2-5%. Preconceptual supplementation with folic acid may reduce the recurrence risk by up to 70%. Hence, the aim should be focused to create awareness among the people about the preventable causes lide nutritional deficiency, exposure to teratogen so that the recurrence of this condition can be reduced by early diagnosis and termination of pregnancy.

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