PRENATAL DIAGNOSIS OF EXCENCEPHALY : A CASE REPORT

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ABSTRACT

Prenatal ultrasonographic diagnosis of 18 weeks gestational age fetus with Exencephaly is reported. Transabdominal ultrasound showed absence of fetal skull with presence of a large volume of brain tissue. The condition is rare and has a lethal outcome. Some authors have called Exencephaly as the variant or precursor of Anencephaly.

INTRODUCTION

Exencephaly is a rare,serious form of neural tube defects, which expresses by acrania (partial or total) and present of a large volume of brain tissue (with developmental anomaly). The incidence is very low and may confuse with the term "Anencephaly", which is absent of both cranial vault and cerebral tissue. Both conditions are serious and incompatible with life. Early diagnosis of these lethal abnormalities are required to minimize parenteral anguish and to reduce maternal morbidity from termination of pregnancy. Ultrasonographic diagnosis can be made as early as 10-11 weeks gestational age, especially by endo-vaginal ultrasound.

CASE REPORT

A 26-year-old woman (gravida 5, para 3, abortus 1, living children 3) last for 1 year; was referred from a health centre because of absence of fetal movement and negative fetal heart sound on examination. She had 5 months pregnancy. Threre was no significant family history of genetic or structural abnornalities.

Transabdominal ultrasonographic examination with 3.5 MHz,convex-sector realtime transducer showed a single, viable fetus with head presentation. The gestational age by femur length measurement was corresponding with 18 weeks. There was absence of normal fetal cranium with a large volume of brain tissue was noted, floating in amniotic fluid. (Fig.1) There was also deformity of cervical and thoracic spines (Fig.2), and mild polyhydramnios was revealed.The fetal face, heart, kidney, bladder, stomach and extremities appeared to be normal.

As the condition is serious and not compatible with life, pregnancy termination was done with maternal consent. Visual inspection of the abortus revealed a 300 grams non-viable fetus which confirmed the skeletal findings.

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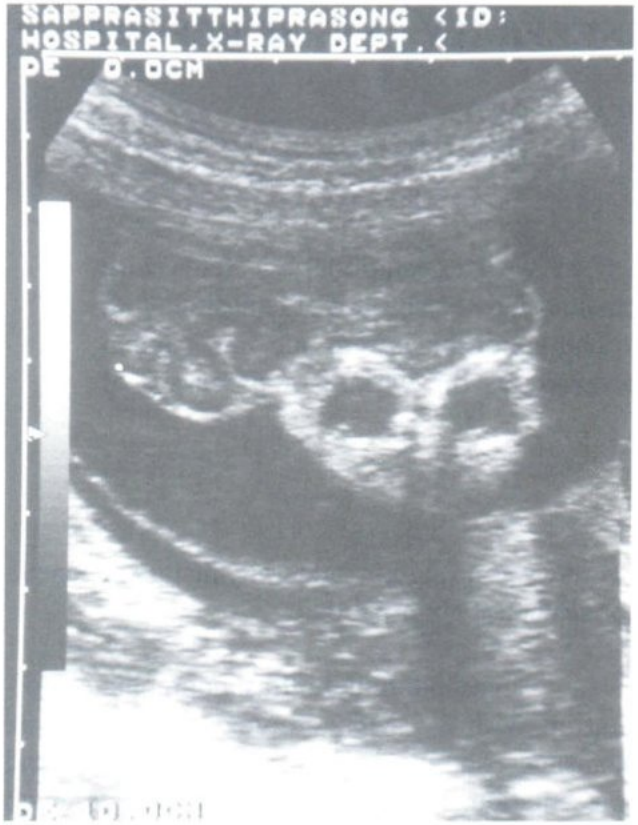

Fig.1 Coronal view at head of exencephalic fetus at 18 weeks gestational age by trans-abdominal ultrasonography. There is absence of normal fetal skull above the orbits, but a large amount of fetal brain tissue is noted and floating in amniotic fluid.

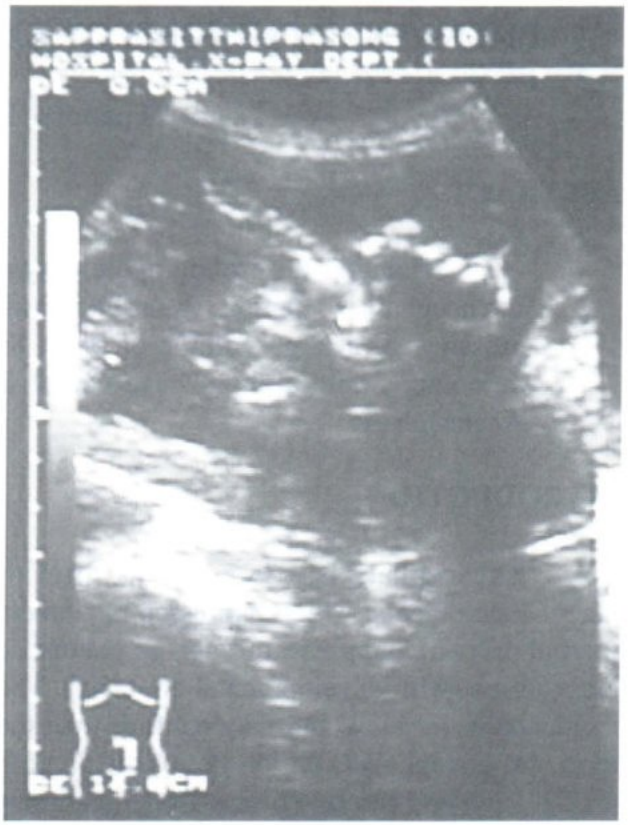

Fig.2 Sagittal view at spines of exencephalic fetus shows anomalous, deformed cervical and thoracic spines

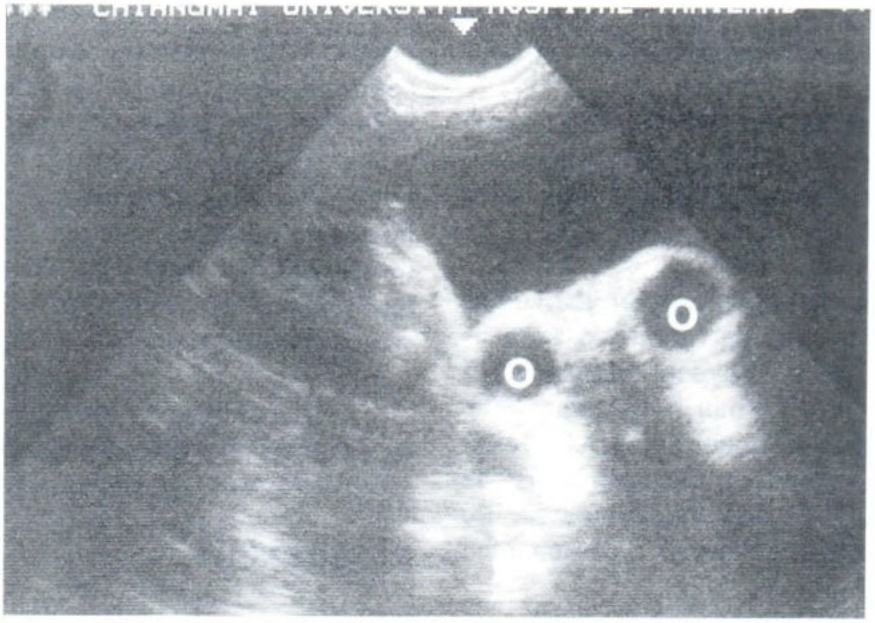

Fig.3 Coronal view at head of anencephalic fetus reveals absence of fetal skull and brain tissue. The orbits appear to be prominent, resembling like wearing sunglasses "Spectacle sign". (Theera Tongsong. Textbook&Atlas of obstetric ultrasound : P.B. Foreign Book Centre L.P.,1995:237)

DISCUSSION

Neural tube defects (NTD) are conditions that caused by failure of closure of the rostral portion of the neural tube during the sixth week of gestational age. Lack of closure of part or all of the neural tube leads to serious anomalies, some of which are incompatible with life, and resulted in abnormal forebrain development and a defective bony calvaria. The incidence of NTD is relatively high;1.2-1.7:1000 lifebirths. NTD is a multifactorial hereditary disorder, and may be suspected by increased meternal serum ∝ -feto-protein level, which are caused by leakage of this protein across the membrane covering the defect. The presence of NTD significantly increases the mother's risk of having another infant with NTD, and therefore warrants close monitoring of subsequent pregnancies. The NTD have many anomalous presentations, including anencephaly, exencephaly, iniencephaly, cephalocele and spina bifida.

Exencephaly is a rare cranial malformation that related to anencephaly,and the incidence is very low.In both disorders,the cranial bones are absent and the primary distinction between the two is the presence of a large amount of disorganized brain tissue in the former. It has been shown in animal studies that anencephaly can be resulted from exencephaly presumably from progressive reduction in the volume of the abnormal brain matter because of mechanical or chemical trauma. This same process have been documented in human fetuses. Some authors have mentioned that exencephaly is the variant of anencephaly which varying amount of brain tissue appear to be presented. If the entire brain is present but not confined within the skull,the term Acrania is used.

The prenatal ultrasonographic appearance of exencephaly has been reported infrequently, and those were previously described are

1. Absent calvarium with presence of large

mass of disorganized cerebral tissue without cranial bony covering.

2. Brain tissue volume is nearly normal but disorganized and may present with pseudosulci.

3. Preservation of bony base of the skull and normal facial structures.

The differential diagnoses are the conditions that have very thining calvarium, such as osteogenesis imperfecta or hypophosphatasia,but these conditions have normal brain tissue. The associated anomalies are abnormal vertebra, cleft lip,cleft palate and club foot. Exencephaly is one of serious anomaly that is incompatible with life and termination of pregnancy is recommended when diagnosed.

Mostly, reliable diagnosis was made at the beginning of the second trimester. Early diagnosis (first trimester) of exencephaly and other anomalies is clearly possible, by ultrasonography especially with the help of vaginal and other high resolution probes. There was one reported case of prenatal diagnosis at 10-11 weeks gestational age. However, caution is warranted because the fetal cranium is not completely calcified before 10-11 weeks gestational age. Therefore finding other than absent calvarium should also be sought.

Anencephaly is the most severe and most common form of NTD; occuring in 1 per 1000 births in the United states and 5 in 1000 births in Irerland and Wales. Female fetuses are affected more often than male (4:1). Anencephaly is a condition occuring with the absence of both normal formed brain and skull. (Fig.3) The base of skull and a portion of the occipital bone, which are formed in cartilage may be presented. The membranous neurocranium, which forms cranial vault, is absent .The rudimentary brainstem and a portion of the basal ganglia are usually presented. The facial bones are nearly normal. Anencephaly may be associated with rachischisis (congenital splaying of posterior spinal elements) depending on the extent of the neural tube defect.

The other associated anomalies are spina bifida, hydronephrosis, diaphragmatic hernia, congenital heart diseases and omphalocele. Anencephaly is also associated with polyhydramnios, presumably due to defective fetal swallowing. Ultrasonographic diagnosis of anencephaly may be made as early as 12 weeks on transabdominal ultrasound and it may be detected earlier with endovaginal ultrasound (may be as early as 8 weeks gestational age).

In conclusion,I have been reported a case of Exencephaly with prenatal ultrasonic diagnosis in a 18 weeks gestational age fetus.

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